NATIONAL OPEN UNIVERSITY OF NIGERIA

SCHOOL OF SCIENCE AND TECHNOLOGY

COURSE CODE: BIO 307

COURSE TITLE: EVOLUTION

BIO 307: EVOLUTION

TEAM WRITERS: **Mr Kofi O. Renner**

Nigerian Institute for Oceanography and
Marine Research,
No 3 Wilmot Point Road, Bar-beach Bus-stop,
Victoria Island,
Lagos, Nigeria.

Mr O. S. Fakayode

Nigerian Institute for Oceanography and
Marine Research,
No 3 Wilmot Point Road, Bar-beach Bus-stop,
Victoria Island,
Lagos, Nigeria.

BIO 307 COURSE GUIDE

Introduction

Evolution (307) is a first semester course. It is a two credit unit compulsory course which all students offering Bachelor of Science (BSc) in Biology must take.

Evolution is an important area of study for scientists. It is described as series of changes across successive generations in the heritable characteristics of biological populations. Processes in evolution give rise to diversity at every level of biological organisation, which include species, individual organisms and at the molecular level, such as proteins and DNA. Since this course evolution involve changes across generation of life (either plant or animal), we will focus on the various heritable characteristics in evolution of both plants and animals and how their genetic make-up supports their existence.

This course deals with the historical concept of evolution, Population genetics, and gene frequency/equilibrium. Hardey Weinberg Principle, Polymorphism, Mutation; origin and types. Polyploidy, isolation mechanism, adaptation; origin of life; evolution of organic molecules, Polymer synthesis, origin of species, Evidence of evolution; fossils (carbon dating), comparative anatomy, Taxonomy, Comparative-biochemistry, physiology, immunology, cell biology. Evolution of the plants, role of oxygen, multicellular development, Phylogeny and geological periods and epochs.

What you will learn in this course

In this course, you have the course units and a course guide. The course guide will tell you briefly what the course is all about. It is a general overview of the course materials you will be using and how to use those materials. It also helps you to allocate the appropriate time to each unit so that you can successfully complete the course within the stipulated time limit.

The course guide also helps you to know how to go about your Tutor-Marked-Assignment which will form part of your overall assessment at the end of the course. Also, there will be tutorial classes that are related to this course, where you can interact with your facilitators and other students. Please I encourage you to attend these tutorial classes.

This course exposes you to Evolution, a sub-discipline and very interesting field of Biology.

Course Aims

This course aims to enable you to know/understand different theories of evolution postulated and genetic basis of evolution in animals and plant related to their development.

Course Objectives

To achieve the aim set above, there are objectives. Each unit has a set of objectives presented at the beginning of the unit. These objectives will give you what to concentrate and focus on while studying the unit and during your study to check your progress.

The Comprehensive Objectives of the Course are given below. At the end of the course/after going through this course, you should be able to:

- Differentiate one evolutionary mechanism from another
- Define biological evolution?
- Explain how theories of microevolution and macroevolution related?
- Understand the various mechanisms governing evolution
- Explain in detail the mechanisms of evolution.
- Discuss extensively on Isolation mechanisms
- Discuss pre and post zygotic isolation
- How reproductive isolation important to speciation and what forms can it take?
- Explain mutation, its types and what causes it.
- Discuss the outcomes of evolution and how these outcomes affect the changes in our environment, related to species existence.
- Define Population Genetics
- Describe the History of Population Genetics
- Define genetic frequency
- Explain the significance of genetic frequency
- Relate genetic frequency and the Hardy-Weinberg Principle
- Define the Ecology and discuss its significance to Evolution
- Differentiate the types of Ecology
- Define polymorphism
- State and discuss types of polymorphism
- Describe variation and understand the basis for variation: Evolution
- Highlight the importance of natural selection
- Explain genetic recombination

Working through the Course

To successfully complete this course. You are required to read each study unit, read the textbooks and other materials provided by the National Open University.

Reading the reference materials can also be of great assistance.

Each unit has self –assessment exercise which you are advised to do. At certain periods during the course you will be required to submit your assignments for the purpose of assessment.

There will be a final examination at the end of the course. The course should take you about17 weeks to complete.

This course guide provides you with all the components of the course, how to go about studying and how you should allocate your time to each unit so as to finish on time and successfully.

The Course Materials

The main components of the course are:

1 The Study Guide

2 Study Units

3 Reference/ Further Readings

4 Assignments

5 Presentation Schedule

Study Units

The study units in this course are given below:

BIO 307 EVOLUTION (2 UNITS)

MODULE: 1 THEORIES OF EVOLUTION

Unit 1: History of Evolutionary Thought

Unit 2: Mechanisms of Evolution

Unit 3: Isolation Mechanisms

Unit 4: Mutation

Unit 5: Outcome of Evolution

MODULE 2: POPULATION GENETICS

Unit 1: History of Population Genetics

Unit 2: Processes of Population Genetics

Unit 3: Hardey-Weinberg Principle

Unit 4: Gene Frequency/Equilibrium

MODULE 3: POLYMORPHISM

Unit 1: Ecology

Unit 2: Introduction to Polymorphism

Unit 3: Examples of Polymorphism

MODULE 4: VARIATION: TYPES AND CAUSES

Unit 1: Evolution and Variation

Unit 2: Genetic variation

Unit 3: Reshuffling of genes

Unit 4: Polyploidy

MODULE 5: EVOLUTION OF LIFE

Unit 1: Origin of Life

Unit 2: Evidence of evolution, adaptation and Speciation

Unit 3: Classification and phylogeny

Unit 4: Geological periods and epoch

Unit 5: Evolution of the plants

In module one, unit one and two deal with the history of Evolutionary Thought and mechanism of evolution, unit three to five deal with isolation mechanisms, mutation and outcome of evolution.

Module Two is concerned with the history of population genetics as well as their processes. Application of Hardey-Weinberg Principle in analyzing population genetics for gene frequency, sex linkage, equilibrium, and heterozygote frequency.

Module Three unit one to three deal with ecology as it related to polymorphism with examples. Polymorphism is related to biodiversity, genetic variation and adaptation and is common in nature.

In Module Four, the first two units deal with Evolution and Variation with further explanation on genetic variation. Unit three and four; reshuffling of genes and polyploidy is further explained to understand genetic variation in evolution.

Module Five, unit one and two focused on Origin of Life, evidence of evolution, adaptation and Speciation in organisms. Classification and phylogeny, Geological periods and epoch and Evolution of the plants are extensively discussed in unit three, four and five.

Each unit will take a week or two lectures, will include an introduction, objectives, reading materials, self assessment question(s), conclusion, summary, tutor-marked assignments (TMAs), references and other reading resources.

There are activities related to the lecture in each unit which will help your progress and comprehension of the unit. You are required to work on these exercises which together with the TMAs will enable you to achieve the objective of each unit.

Presentation Schedule

There is a time-table prepared for the early and timely completion and submissions of your TMAs as well as attending the tutorial classes. You are required to submit all your assignments by the stipulated date and time. Avoid falling behind the schedule time.

Assessment

There are three aspects to the assessment of this course.

The first one is the self-assessment exercises. The second is the tutor-marked assignments and the third is the written examination or the examination to be taken at the end of the course.

Do the exercises or activities in the unit applying the information and knowledge you acquired during the course. The tutor-marked assignments must be submitted to your facilitator for formal assessment in accordance with the deadlines stated in the presentation schedule and the assignment file.

The work submitted to your tutor for assessment will account for 30% of your total work.

At the end of this course you have to sit for a final or end of course examination of about a three hour duration which will account for 70% of your total course mark.

Tutor Marked Assignment

This is the continuous assessment component of this course and it accounts for 30% of the total score. You will be given four (4) TMAs by your facilitator to answer. Three of which must be answered before you are allowed to sit for the end of the course examination.

These answered assignments must be returned to your facilitator.

You are expected to complete the assignments by using the information and material in your reading references and study units.

Reading and researching into the references will give you a wider view point and give you a deeper understanding of the subject.

1 Make sure that each assignment reaches your facilitator on or before the deadline given in the presentation schedule and assignment file. If for any reason you are not able to complete your assignment, make sure you contact your facilitator before the assignment is due to discuss the possibility of an extension. Request for extension will not be granted after the due date unless there is an exceptional circumstance.
2 Make sure you revise the whole course content before sitting for examination. The self-assessment activities and TMAs will be useful for this purposes and if you have any comments please do before the examination. The end of course examination covers information from all parts of the course.

Course Marking Scheme

Assignment	**Marks**
Assignment 1-4	Four assignments, best three marks of the four count at 10% each - 30% of course marks.
End of course examination	70% of overall course marks
Total	100% of course materials

Facilitators/ Tutors and Tutorials

Sixteen (16) hours are provided for tutorials for this course. You will be notified of the dates, times and location for these tutorial classes.

As soon as you are allocated a tutorial group, the name and phone number of your facilitator will be given to you.

These are the duties of your facilitator:

- He or she will mark and comment on your assignment
- He will monitor your progress and provide any necessary assistance you need.
- He or she will mark your TMAs and return to you as soon as possible.

(You are expected to mail your tutored assignment to your facilitators at least two days before the schedule date).

Do not delay to contact your facilitator by telephone or e-mail for necessary assistance if

- You do not understand any part of the study in the course material.
- You have difficulty with the self assessment activities.
- You have a problem or question with an assignment or with the grading of the assignment.

It is important and necessary you attend the tutorial classes because this is the only chance to have face to face contact with your facilitator and to ask questions which will be answered instantly. It is also a period where you can point out any problem encountered in the course of your study.

Summary

Evolution (307) is a course that introduces you to the concepts and principles of how living things gradually change from one form into another over the course of time, the origin of species and lineages by descent of living forms from ancestral forms, and the generation of diversity. This emphasizes the appearance of new, physically distinct life forms that can be grouped with similar appearing life forms in a taxonomic hierarchy. It commonly is referred to as macroevolution. It can also be defined as changes in the genetic composition of a population with the passage of each generation.

On the completion of this course, you will have an understanding of basic knowledge of historical theories of evolution, population genetics, genetic variation, polymorphism and evolution of life etc you will be able to understand the concept behind evolution. In addition you will be able to answer the following questions:

- Discuss three schools of thought for the History of Evolution?
- For or against; Charles Darwin's Theory of Evolution?
- State and elaborate on three of these mechanisms?
- What are the different types of mutation?
- How important is mutation to the evolutionary process?

- Identify keys areas of biological and environmental conservation where a genetic approach benefits outcomes of management?
- Explain polymorphism and niche diversity?
- Discuss mechanism of balancing selection?
- What are the mechanisms that affect genetic variation?
- Describe polyploidy in animals and plants?
- List 5 probable stages involved in the origin of life?
- The RNA- first hypothesis for the origin of cells is supported by the discovery of---------

The list of questions you are expected to answer is not limited to the above list.

I believe you will agree with me that Evolution is a very interesting field of biology.

I wish you success in this course.

MODULE 1: THEORIES OF EVOLUTION

Unit 1: History of Evolutionary Thought

Unit 2: Mechanisms of Evolution

Unit 3: Isolation Mechanisms

Unit 4: Mutation

Unit 5: Outcomes of Evolution

UNIT 1: HISTORY OF EVOLUTIONARY THOUGHT

CONTENTS

UNIT 1: HISTORY OF EVOLUTIONARY THOUGHT

1.0 Introduction

Evolution has been described as series of changes across successive generations in the heritable characteristics of biological populations. Processes in evolution give rise to diversity at every level of biological organisation, which include species, individual organisms and at the molecular level, such as proteins and DNA. Approximately 3.7billion years ago, life on Earth originated and then evolved from a universal common ancestor. The divergence and repeated speciation of life can be traced through shared sets of biochemical and morphological traits, or by shared DNA sequences. Existing patterns of biodiversity have been shaped both by speciation and by extinction. The theory of evolution by natural selection was formulated by Charles Darwin, who was the first to recognise natural selection as an important cause of evolution.

2.0 Objectives

At the conclusion of this unit, students should be able to:

- Give an account of the history of evolution,
- Describe the different schools of thought of the evolutionary process,
- Differentiate one school of thought of the evolutionary process from the other
- Understand Darwin's theory of natural selection.

3.0 Main Focus

3.1 Evolution Pre-Darwin

Contrary to many assumptions, evolutionary theory did not begin in 1859 with Charles Darwin and *The Origin of Species*. Rather, evolution-like ideas had existed since the times of the Greeks, and had been in and out of favour in the periods between ancient Greece and Victorian England. Indeed, by Darwin's time the idea of evolution - called "descent with

modification" - was not especially controversial, and several other evolutionary theories had already been proposed. Darwin may stand at the beginning of a modern tradition, but he is also the final culmination of an ancient speculation.

3.2 Greek Evolution

Many examples of societies that postulated the history of evolution include the Greeks, who did not specifically refer to their concepts as "evolution", they did have a philosophical notion of descent with modification. Several different Greek philosophers subscribed to a concept of origination, arguing that all things originated from water or air. Another common concept was the idea that all things descended from one central, guiding principle. Aristotle suggests a transition between the living and the nonliving, and theorizes that in all things there is a constant desire to move from the lower to the higher, finally becoming the divine.

3.3 Medieval Evolution

Medieval theories argued that all living things came into existence in unchanging forms due to divine will, was notably in opposition to the concept of evolution. Medieval thinking was also, oddly enough, confused by the idea of spontaneous generation, which stated that living things can appear fully formed from inorganic matter. In this view, maggots came from rotting meat; frogs came from slime, etc. This sort of a concept prevented both genetic thinking and speculation about evolution or descent with modification. Nevertheless, a few philosophers theorized about some sort of teleological principle by which species might derive from a divine form.

3.4 Immanuel Kant

Immanuel Kant the German philosopher Immanuel Kant developed a concept of descent that is relatively close to modern thinking; he did in a way anticipate Darwinian thinking. Based on similarities between organisms, Kant speculated that they may have come from a single ancestral source. In a thoroughly modern speculation, he mused that "an orang-utan or a chimpanzee may develop the organs which serve for walking, grasping objects, and speaking-in short, that lie may evolve the structure of man, with an organ for the use of reason, which shall gradually develop itself by social culture"

3.5 Biological Conceptions of Evolution.

Preceding discussion has focused on the philosophical components of evolutionary theory, but precursors exist for its biological aspects as well. Indeed, as mentioned above, by Darwin's time the concept of descent with modification was hardly controversial - it was only the mechanism, the rate of modification, and the ultimate origin of life that were being debated. Darwin's major breakthrough consisted in providing a plausible mechanism to drive change in organisms.

3.6 Carolus Linnaeus.

Carolus Linnaeus (1707-1778), is considered the father of modern taxonomy for his work in hierarchical classification of various organisms. At first, he believed in the fixed nature of species, but he was later swayed by hybridization experiments in plants, which could produce new species. However, he maintained his belief in special creation in the Garden of Eden, consistent with the Christian doctrine to which he was quite devoted. He still saw the new species created by plant hybridization to have been part of God's plan, and never considered the idea of open-ended, undirected evolution not mediated by the divine.

3.7 Erasmus Darwin.

Charles Darwin's grandfather Erasmus Darwin (1731-1802) was also a distinguished naturalist with his own intriguing ideas about evolution. While he never thought of natural selection, he did argue that all life could a have a single common ancestor, though he struggled with the concepts of a mechanism for this descent. He also discussed the effects of competition and sexual selection on possible changes in species. Like Lamarck, Erasmus Darwin subscribed to a theory stating that the use or disuse of parts could in itself make them grow or shrink, and that unconscious striving by the organism was responsible for adaptation.

3.8 Jean-Baptiste. Lamarck

Jean-Baptiste Lamarck's (1744-1829) theory of evolution was a good try for his time, but has now been discredited by experimental evidence and the much more plausible mechanism of modification proposed by Darwin. Lamarck saw species as not being fixed and immutable, but rather in a constantly changing state. He presented a multitude of different theories that he believed combined to explain descent with modification of these changing species. Lamarck subscribed to a number of what we now know to be false beliefs about inheritance. First, like Erasmus Darwin, he argued for strong effects of the use and disuse of parts, which he thought would make the relevant parts change size or shape in accordance with their use. Second, Lamarck believed that all organisms fundamentally *wanted* to adapt themselves to their environment, and so they strove to become better adapted. The belief most commonly associated with Lamarck today is his idea of the inheritance of acquired characteristics. This theory stated that an organism could pass on to its offspring any characteristics it had acquired in its lifetime. For example, if a man exercised and thus developed strong muscles, his offspring would then have strong muscles at birth.

3.9 Thomas Malthus.

Thomas Malthus' (1766-1834) theory of population growth was in the end what inspired Darwin to develop the theory of natural selection. According to Malthus, populations produce many more offspring than can possibly survive on the limited resources generally available. According to Malthus, poverty, famine, and disease were natural outcomes that resulted from overpopulation. However, Malthus believed that divine forces were ultimately responsible for such outcomes, which, though natural, were designed by God.

3.10 Charles Darwin and Alfred Russel Wallace.

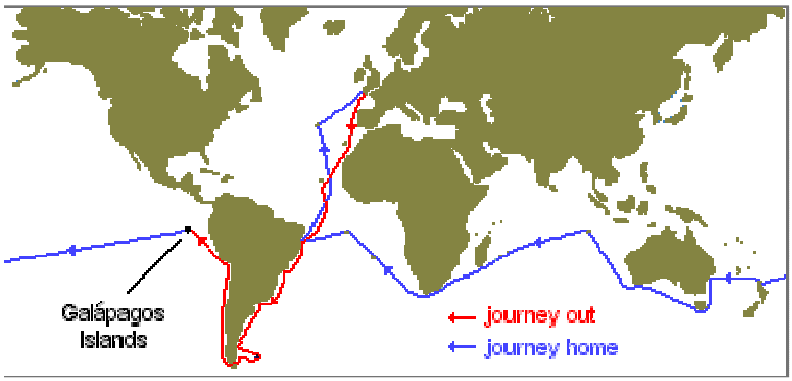
Figure 1. Charles Darwin
1809-1882

Alfred Wallace
1823-1913

Charles Darwin and Alfred Russel Wallace both independently developed the idea of the mechanism of natural selection after reading Thomas Malthus' *Essay on the Principle of*

Population (1798). However, Darwin had been turning the problem over in his mind for some twenty years before he first published *The Origin of Species*. Moreover, Darwin was much more willing to explore the implications of natural selection, particularly in relation to humans, than Wallace was. In addition, Wallace was a champion of rather radical social causes and later openly embraced spiritualism - all elements that resulted in the downplay of his role in the discovery of natural selection.

Darwin's was a part of an expedition on board the HMS Beagle in 1831. He embarked on a 5 week visit to the Galápagos Islands in the Eastern Pacific Ocean. It was there that he made the observations that eventually led him to comprehend what causes plants and animals to evolve, but he apparently did not clearly formulate his views on this until 1837. At the time he left the Galápagos Islands, he apparently still believed in a traditional Biblical creation of all life forms.



Darwin's Beagle Diary
audio recording of excerpts from Darwin's popular account of his voyage of exploration.

Figure 2.

The Galápagos Islands have species found in no other part of the world. Darwin was surprised that the birds were slightly different from one island to another. He realized that

the key to why this difference existed was connected with the fact that the various species live in different kinds of environments.

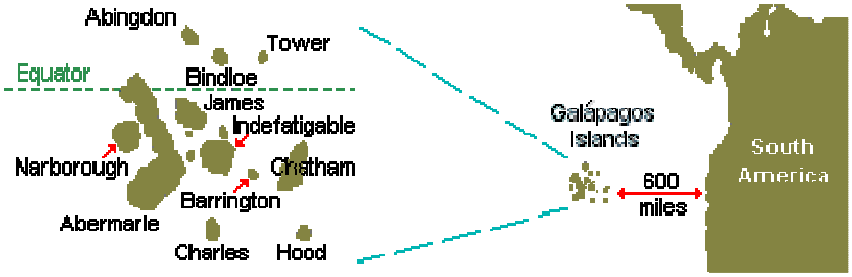
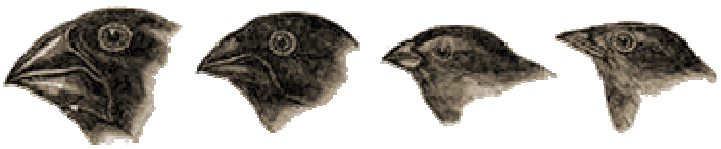


Figure 3.

On his return to England, Darwin and an ornithologist identified 13 species of finches that he had collected on the Galápagos Islands. This was surprising since he knew of only one species of this bird on the mainland of South America, nearly 600 miles to the east, where they had all presumably originated. He observed that the Galápagos species differed from each other in beak size and shape. He noted that the beak varieties were associated with diets based on different foods. He concluded that when the original South American finches reached the islands, they dispersed to different environments where they had to adapt to different conditions. Over many generations, they changed anatomically in ways that allowed them to get enough food and survive to reproduce. This observation was verified by intensive field research in the last quarter of the 20th century.



Galapágos Creatures--some of the unusual animals that Darwin saw on the Galapágos islands. Video clip from "Evolution: Constant Change and Common Threads". (Howard Hughes Medical Institute, 2005)

Figure 4

Today the term adaptive radiation is used to refer to this sort of branching evolution in which different populations of a species become reproductively isolated from each other by adapting to different ecological niches and eventually become separate species.

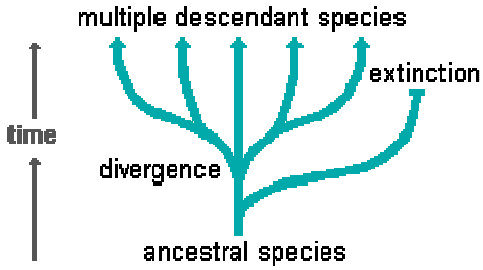


Figure 5

Darwin understood that any population is made up of individuals that are all slightly different from one another. Individuals having variation that gives them an advantage in staying alive long enough to successfully reproduce are the ones that pass on their traits more frequently to the next generation. Subsequently, their traits become more common and the population evolves. Darwin called this "descent with modification."

The Galápagos finches provide an excellent example of this process. Among the birds that ended up in arid environments, the ones with beaks better suited for eating cactus got more food. As a result, they were in better condition to mate. Similarly, those with beak shapes that were better suited to getting nectar from flowers or eating hard seeds in other environments were at an advantage there. In a very real sense, nature selected the best adapted varieties to survive and to reproduce. This process has come to be known as natural selection.

In 1859, Darwin's publication of *On the Origin of Species* explained natural selection in detail and in a way that lead to an increasingly wide acceptance of Darwinian evolution. Thomas Henry Huxley applied Darwin's ideas to humans, using palaeontology and comparative

anatomy to provide strong evidence that humans and apes shared a common ancestry. Some were disturbed by this since it implied that humans did not have a special place in the universe. Due to the fact that the exact mode for reproductive heritability and the origin of new traits remained a mystery, Darwin developed his provisional theory of pangenesis. In 1865 Gregor Mendel reported that traits were inherited in a predictable manner through the independent assortment and segregation of elements. All plants and animals receive their specific characteristics from their parents by inheriting particular combinations of genes. Molecular biologists have discovered that genes are, in fact, segments of DNA molecules in our cells.

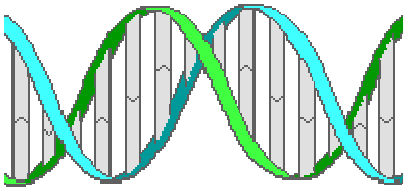


Figure 6 section of a DNA molecule

Mendel's laws of inheritance eventually supplanted most of Darwin's pangenesis theory. August Weismann made the important distinction between germ cells and somatic cells of the body, demonstrating that heredity passes through the germ line only. Hugo de Vries connected Darwin's pangenesis theory to Wiesman's germ/somatic cell distinction and proposed that Darwin's pangenes were concentrated in the cell nucleus and when expressed they could move into the cytoplasm to change the cells structure. De Vries was also one of the researchers who made Mendel's work well-known, believing that Mendelian traits corresponded to the transfer of heritable variations along the germ line. To explain how new variants originate; De Vries developed a mutation theory that led to a temporary rift between those who accepted Darwinian evolution and biometricians who allied with de Vries. At the turn of the 20th century, pioneers in the field of population genetics, such as J.B.S. Haldane,

Sewall Wright, and Ronald Fisher, set the foundations of evolution onto a robust statistical philosophy. The false contradiction between Darwin's theory, genetic mutations, and Mendelian inheritance was thus reconciled.

An evolutionary synthesis in the 1930s connected natural selection, mutation theory, and Mendelian inheritance into a unified theory that applied generally to any branch of biology. The modern synthesis was able to explain patterns observed across species in populations, through fossil transitions in palaeontology, and even complex cellular mechanisms in developmental biology. The publication of the structure of DNA by James Watson and Francis Crick in 1953 demonstrated a physical basis for inheritance. Molecular biology improved our understanding of the relationship between genotype and phenotype. Advancements were also made in phylogenetic systematics, mapping the transition of traits into a comparative and testable framework through the publication and use of evolutionary trees. Theodosius Dobzhansky an evolutionary biologist in 1973 penned that "nothing in biology makes sense except in the light of evolution", because it has brought to light the relations of what first seemed disjointed facts in natural history into a coherent explanatory body of knowledge that describes and predicts many observable facts about life on this planet. Since then, the modern synthesis has been further extended to explain biological phenomena across the full and integrative scale of the biological hierarchy, from genes to species.

4.0 Conclusion

The student shall be able to give a thorough account of the History of evolution as well as to discuss major highlights of evolution especially Pre-Darwinian era and to establish similarities and differences amongst the different schools of thought. The student should be

able to discuss for and against the different theories of evolution which are thought to have led to our current existence.

5.0 Summary

The concept of evolution has been the centre of debate for over a century. Evolution has been called the cornerstone of biology, and for good reasons. It is possible to do research in biology with little or no knowledge of evolution. Most biologists do. But, without evolution biology becomes a disparate set of fields. Evolutionary explanations pervade all fields in biology and bring them together under one theoretical umbrella. After the publication of *On the Origin of Species* in 1859, the idea that life had evolved was an active source of academic debate centred on the philosophical, social and religious implications of evolution. Nowadays, the modern evolutionary synthesis is accepted by a vast majority of scientists. However, evolution remains a contentious concept for some theists.

6.0 Tutor-Marked Assignment

- What do you understand by History of Evolutionary Thought?
- Discuss three schools of thought for the History of Evolution?
- For or against; Charles Darwin's Theory of Evolution?

7.0 Reference/Further Readings

The Descent of Man by Charles Darwin; The Origin of Species by Charles Darwin; Evolution from *The Internet Encyclopedia of Philosophy;* A History of Evolutionary Thought; Timeline of Evolutionary Thought; Lamarck and his Theory of Evolution by Thomas E. Hart from *The Victorian Web.*

UNIT2: MECHANISMS OF EVOLUTION

CONTENTS

1.0 Introduction

Based on the Neo-Darwinian perspective, evolution occurs when there are changes in the frequencies of alleles within a population of interbreeding organisms. A good example is the allele for black colour in a population of moths becoming more common. Mechanisms that can lead to changes in allele frequencies include natural selection, genetic drift, genetic hitchhiking, mutation and gene flow.

2.0 Objectives

At the end of this unit, the student should be able to:

- Differentiate one evolutionary mechanism from another
- Define biological evolution?
- How are theories of microevolution and macroevolution related?
- Understand the various mechanisms governing evolution
- Explain in detail the mechanisms of evolution.

3.0 Evolution and its many forms

The word "evolution" does not apply exclusively to biological evolution. The universe and our solar system have developed out of the explosion of matter that began our known universe. Chemical elements have evolved from simpler matter. Life has evolved from non-life, and complex organisms from simpler forms. Languages, religions, and political systems all evolve. Hence, evolution is an appropriate theme for a course on global change. The core aspects of evolution are "change" and the role of history, in that past events have an influence over what changes occur subsequently. In biological evolution this might mean that complex organisms arise out of simpler ancestors - though be aware that this is an over-simplification not acceptable to a more advanced discussion of evolution. A full discussion of evolution requires a detailed explanation of genetics, because science has given us a good understanding of the genetic basis of evolution. It also requires an investigation of the differences that characterize species, genera, indeed the entire tree of life, because these are the phenomena that the theory of evolution seeks to explain. We will begin with observed patterns of similarities and differences among species, because this is what Darwin knew about. The genetic basis for evolution only began to be integrated into evolutionary theory in

the 1930's and 1940's. We will add genetics into our understanding of evolution through a discussion activity.

Definitions of Biological Evolution

We begin with two working definitions of biological evolution, which capture these two facets of genetics and differences among life forms. Then we will ask what is a species, and how does a species arise?

- **Definition 1:**

  Changes in the genetic composition of a population with the passage of each

  generation

- **Definition 2:**

  The gradual change of living things from one form into another over the course of time, the origin of species and lineages by descent of living forms from ancestral forms, and the generation of diversity

Note that the first definition emphasizes genetic change. It commonly is referred to as microevolution. The second definition emphasizes the appearance of new, physically distinct life forms that can be grouped with similar appearing life forms in a taxonomic hierarchy. It commonly is referred to as macroevolution.

3.1 Natural Selection

Natural selection is the process by which genetic mutations that enhance reproduction become and remain, more common in successive generations of a population. It has often been called a "self-evident" mechanism because it necessarily follows from three simple facts:

- Heritable variation exists within populations of organisms.
- Organisms produce more offspring than can survive.

- These offspring vary in their ability to survive and reproduce.

These conditions produce competition between organisms for survival and reproduction. Consequently, organisms with traits that give them an advantage over their competitors pass these advantageous traits on, while traits that do not confer an advantage are not passed on to the next generation. Natural selection is based on a concept of evolutionary fitness. This fitness is measured by an organism's ability to survive and reproduce, which determines the size of its genetic contribution to the next generation. A good example is if an organism could survive well and reproduces rapidly, but its offspring were all too small and weak to survive, this organism would make little genetic contribution to future generations and would thus have low fitness.

The idea that species could change over time was not immediately acceptable to many: the lack of a mechanism hampered the acceptance of the idea as did its implications regarding the biblical views of creation.

Charles Darwin and Alfred Wallace both worked independently of each other, travelled extensively, and eventually developed similar ideas about the change in life over time as well as a mechanism for that change: natural selection.

Ancient Greek philosophers such as Anaximander postulated the development of life from non-life and the evolutionary descent of man from animal. Charles Darwin simply brought something new to the old philosophy -- a plausible mechanism called "natural selection." Natural selection acts to preserve and accumulate minor advantageous genetic mutations. Suppose a member of a species developed a functional advantage – let us assume it grew wings and learned to fly. Its offspring would inherit that advantage and pass it on to their offspring. The inferior or disadvantaged members of the same species would gradually die

out, leaving only the superior or advantaged members of the species. Natural selection is the preservation of a functional advantage that enables a species to compete better in the wild. Natural selection is the naturalistic equivalent to domestic breeding. Over the centuries, human breeders have produced dramatic changes in domestic animal populations by selecting individuals to breed. Breeders eliminate undesirable traits gradually over time. Similarly, natural selection eliminates inferior species gradually over time. Some types of organisms within a population leave more offspring than others. Over time, the frequency of the more prolific type will increase. The difference in reproductive capability is called natural selection. Natural selection is the only mechanism of adaptive evolution; it is defined as differential reproductive success of pre- existing classes of genetic variants in the gene pool.

The most common action of natural selection is to remove unfit variants as they arise via mutation. In other words, natural selection usually prevents new alleles from increasing in frequency. This led a famous evolutionist, George Williams, to say "Evolution proceeds in spite of natural selection."

Natural selection can maintain or deplete genetic variation depending on how it acts. When selection acts to weed out deleterious alleles, or causes an allele to sweep to fixation, it depletes genetic variation. When heterozygotes are fit than either of the homozygotes, however, selection causes genetic variation to be maintained. This is called balancing selection. An example of this is the maintenance of sickle-cell alleles in human populations subject to malaria. Variation at a single locus determines whether red blood cells are shaped normally or sickled. If a human has two alleles for sickle-cell, he/she develops anaemia -- the shape of sickle-cells precludes those carrying normal levels of oxygen. However, heterozygotes that have one copy of the sickle-cell allele, coupled with one normal allele enjoy some resistance to malaria -- the shape of sickled cells makes it harder for the

plasmodia to enter the cell. Thus, individuals homozygous for the normal allele suffer more malaria than heterozygotes. Individuals homozygous for the sickle- cell are anaemic. Heterozygotes have the highest fitness of these three types. Heterozygotes pass on both sickle-cell and normal alleles to the next generation. Thus, neither allele can be eliminated from the gene pool. The sickle-cell allele is at its highest frequency in regions of Africa where malaria is most pervasive.

Balancing selection is rare in natural populations. Only a handful of other cases beside the sickle-cell example have been found. At one time population geneticists thought balancing selection could be a general explanation for the levels of genetic variation found in natural populations. That is no longer the case. Balancing selection is only rarely found in natural populations. And, there are theoretical reasons why natural selection cannot maintain polymorphisms at several loci via balancing selection.

Individuals are selected. The example I gave earlier was an example of evolution via natural selection. Dark colored moths had a higher reproductive success because light colored moths suffered a higher predation rate. The decline of light colored alleles was caused by light colored individuals being removed from the gene pool Individual organisms either reproduce or fail to reproduce and are hence the unit of selection. One way alleles can change in frequency is to be housed in organisms with different reproductive rates. Genes are not the unit of selection; neither are groups of organisms a unit of selection. There are some exceptions to this "rule," but it is a good generalization.

Organisms do not perform any behaviour that is for the good of their species. An individual organism competes primarily with others of it own species for its reproductive success. Natural selection favours selfish behaviour because any truly altruistic act increases the recipient's reproductive success while lowering the donors. Altruists would disappear from a

population as the non- altruists would reap the benefits, but not pay the costs, of altruistic acts. Much behaviour appears altruistic. Biologists, however, can demonstrate that this behaviour is only apparently altruistic. Cooperating with or helping other organisms is often the most selfish strategy for an animal. This is called reciprocal altruism. A good example of this is blood sharing in vampire bats. In these bats, those lucky enough to find a meal will often share part of it with an unsuccessful bat by regurgitating some blood into the other's mouth. Biologists have found that these bats form bonds with partners and help each other out when the other is needy. If a bat is found to be a "cheater," (he accepts blood when starving, but does not donate when his partner is) his partner will abandon him. The bats are thus not helping each other altruistically; they form pacts that are mutually beneficial.

Helping closely related organisms can appear altruistic; but this is also a selfish behaviour. Reproductive success (fitness) has two components; direct fitness and indirect fitness. Direct fitness is a measure of how many alleles, on average, a genotype contributes to the subsequent generation's gene pool by reproducing. Indirect fitness is a measure of how many alleles identical to its own it helps to enter the gene pool. Direct fitness plus indirect fitness is inclusive fitness. J. B. S. Haldane once remarked he would gladly drown, if by doing so he saved two siblings or eight cousins. Each of his siblings would share one half his alleles; his cousins, one eighth. They could potentially add as many of his alleles to the gene pool as he could.

Natural selection favours traits or behaviour that increase a genotype's inclusive fitness. Closely related organisms share many of the same alleles. In diploid species, siblings share on average at least 50% of their alleles. The percentage is higher if the parents are related. So, helping close relatives to reproduce gets an organism's own alleles better represented in the gene pool. The benefit of helping relatives increases dramatically in highly inbred species. In

some cases, organisms will completely forgo reproducing and only help their relatives reproduce. Ants, and other eusocial insects, have sterile castes that only serve the queen and assist her reproductive efforts. The sterile workers are reproducing by proxy.

The words selfish and altruistic have connotations in everyday use that biologists do not intend. Selfish simply means behaving in such a way that one's own inclusive fitness is maximized; altruistic means behaving in such a way that another's fitness is increased at the expense of one's' own. Use of the words selfish and altruistic is not meant to imply that organisms consciously understand their motives.

The opportunity for natural selection to operate does not induce genetic variation to appear -- selection only distinguishes between existing variants. Variation is not possible along every imaginable axis, so all possible adaptive solutions are not open to populations. To pick a somewhat ridiculous example, a steel shelled turtle might be an improvement over regular turtles. Turtles are killed quite a bit by cars these days because when confronted with danger, they retreat into their shells -- this is not a great strategy against a two ton automobile. However, there is no variation in metal content of shells, so it would not be possible to select for a steel shelled turtle.

Here is a second example of natural selection. Geospiza fortis lives on the Galapagos Islands along with fourteen other finch species. It feeds on the seeds of the plant Tribulus cistoides, specializing on the smaller seeds. Another species, G. Magnirostris, has a larger beak and specializes on the larger seeds. The health of these bird populations depends on seed production. Seed production, in turn, depends on the arrival of wet season. In 1977, there was a drought. Rainfall was well below normal and fewer seeds were produced. As the season progressed, the G. fortis population depleted the supply of small seeds. Eventually, only larger seeds remained. Most of the finches starved; the population plummeted from about

twelve hundred birds to less than two hundred. Peter Grant, who had been studying these finches, noted that larger beaked birds fared better than smaller beaked ones. These larger birds had offspring with correspondingly large beaks. Thus, there was an increase in the proportion of large beaked birds in the population the next generation. To prove that the change in bill size in Geospiza fortis was an evolutionary change, Grant had to show that differences in bill size were at least partially genetically based. He did so by crossing finches of various beak sizes and showing that a finch's beak size was influenced by its parent's genes. Large beaked birds had large beaked offspring; beak size was not due to environmental differences.

Natural selection may not lead a population to have the optimal set of traits. In any population, there would be a certain combination of possible alleles that would produce the optimal set of traits (the global optimum); but there are other sets of alleles that would yield a population almost as adapted (local optima). Transition from a local optimum to the global optimum may be hindered or forbidden because the population would have to pass through less adaptive states to make the transition. Natural selection only works to bring populations to the nearest optimal point. This idea is Sewall Wright's adaptive landscape. This is one of the most influential models that shape how evolutionary biologists view evolution.

Natural selection does not have any foresight. It only allows organisms to adapt to their current environment. Structures or behaviours do not evolve for future utility. An organism adapts to its environment at each stage of its evolution. As the environment changes, new traits may be selected for. Large changes in populations are the result of cumulative natural selection. Changes are introduced into the population by mutation; the small minority of these changes that result in a greater reproductive output of their bearers are amplified in frequency by selection.

Complex traits must evolve through viable intermediates. For many traits, it initially seems unlikely that intermediates would be viable. What good is half a wing? Half a wing may be no good for flying, but it may be useful in other ways. Feathers are thought to have evolved as insulation and/or as a way to trap insects. Later, proto-birds may have learned to glide when leaping from tree to tree. Eventually, the feathers that originally served as insulation now became co-opted for use in flight. A trait's current utility is not always indicative of its past utility. It can evolve for one purpose, and be used later for another. A trait evolved for its current utility is an adaptation; one that evolved for another utility is an exaptation. An example of an exaptation is a penguin's wing. Penguins evolved from flying ancestors; now they are flightless and use their wings for swimming.

3.2 Genetic Drift

Genetic drift is the change in allele frequency from one generation to the next that occurs because alleles are subject to sampling error. Due to this, when selective forces are absent or relatively weak, allele frequencies tend to drift upward or downward randomly. This drift halts when an allele eventually becomes fixed, either by disappearing from the population, or replacing the other alleles entirely. Genetic drift therefore may eliminate some alleles from a population due to chance alone. Even in the absence of selective forces, genetic drift can cause two separate populations that began with the same genetic structure to drift apart into two divergent populations with different sets of alleles. Usually, it is difficult to measure the relative importance of selection and neutral processes, including drift. The comparative importance of adaptive and non-adaptive forces in driving evolutionary change is an area of current research

Allele frequencies can change due to chance alone. This is called genetic drift. Drift is a binomial sampling error of the gene pool. This implies that the alleles that form the next

generation's gene pool are a sample of the alleles from the current generation. When sampled from a population, the frequency of alleles differs slightly due to chance alone.

Alleles can increase or decrease in frequency due to drift. The average expected change in allele frequency is zero, since increasing or decreasing in frequency is equally probable. A small percentage of alleles may continually change frequency in a single direction for several generations just as flipping a fair coin may, on occasion, result in a string of heads or tails. A very few new mutant alleles can drift to fixation in this manner.

In small populations, the variance in the rate of change of allele frequencies is greater than in large populations. However, the overall rate of genetic drift is independent of population size. If the mutation rate is constant, large and small populations lose alleles to drift at the same rate. This is because large populations will have more alleles in the gene pool, but they will lose them more slowly. Smaller populations will have fewer alleles, but these will quickly cycle through. This assumes that mutation is constantly adding new alleles to the gene pool and selection is not operating on any of these alleles.

Sharp drops in population size can change allele frequencies substantially. In the event of a population crashing, the alleles in the surviving sample may not be representative of the pre-crash gene pool. This change in the gene pool is called the founder effect, due to the fact that small populations of organisms that invade a new territory are subject to this. Many biologists feel the genetic changes brought about by founder effects may contribute to isolated populations developing reproductive isolation from their parent populations. In sufficiently small populations, genetic drift can counteract selection.

3.3 Genetic Hitchhiking

The recombination of genes allows alleles on the same strand of DNA to become separated. However, the rate of recombination is low-approximately two events per chromosome per generation. As a result of this, genes close together on a chromosome may not always be shuffled away from each other and genes that are close together tend to be inherited together. This is referred to as linkage. This is measured by finding how often two alleles occur together on a single chromosome compared to expectations, which is called their linkage disequilibrium. A set of alleles that is usually inherited in a group is called a haplotype. This can be important when one allele in a particular haplotype is strongly beneficial: natural selection can drive a selective sweep that will also cause the other alleles in the haplotype to become more common in the population; this effect is called genetic hitchhiking or genetic draft. Genetic draft caused by the fact that some neutral genes are genetically linked to others that are under selection can be partially captured by an appropriate effective population size.

3.4 Gene Flow

Gene flow is the exchange of genes between populations and between species. The presence or absence of gene flow fundamentally changes the course of evolution. As a result of the complexity of organisms, any two completely isolated populations will eventually evolve genetic incompatibilities through neutral processes, even if both populations remain essentially identical in terms of their adaptation to the environment.

If genetic differentiation between populations develops, gene flow between populations can introduce traits or alleles which are disadvantageous in the local population and this may lead to organism within these populations to evolve mechanisms that prevent mating with genetically distant populations, eventually resulting in the appearance of new species. Thus, exchange of genetic information between individuals is fundamentally important for the development of the biological species concept.

During the development of the modern synthesis, Sewall Wright's developed his shifting balance theory that gene flow between partially isolated populations was an important aspect of adaptive evolution. However, recently there has been substantial criticism of the importance of the shifting balance theory.

In some closely related species, fertile hybrids can result from interspecific matings. These hybrids can vector genes from species to species. Gene flow between more distantly related species occurs infrequently. This is called horizontal transfer. One interesting case of this involves genetic elements called P elements. Margaret Kidwell found that P elements were transferred from some species in the *Drosophila willistoni* group to *Drosophila melanogaster*. These two species of fruit flies are distantly related and hybrids do not form. Their ranges do, however, overlap. The P elements were vectored into *Drosophila. melanogaster* via a parasitic mite that targets both these species. This mite punctures the exoskeleton of the flies and feeds on the "juices". Material, including DNA, from one fly can be transferred to another when the mite feeds. Since P elements actively move in the genome, one incorporated itself into the genome of a *melanogaster* fly and subsequently spread through the species. Laboratory stocks of *melanogaster* caught prior to the 1940's lack of P elements. All natural populations today harbour them.

4.0 Conclusion

It is important to note that Darwin's book "The Origin of Species by Means of Natural Selection" did two things. It summarized all of the evidence in favour of the idea that all organisms have descended with modification from a common ancestor, and thus built a strong case for evolution. In addition Darwin advocated natural selection as a mechanism of evolution. Biologists no longer question whether evolution has occurred or is occurring. That part of Darwin's book is now considered to be so overwhelmingly demonstrated that is is

often referred to as the fact of evolution. However, the MECHANISM of evolution is still debated. We have learned much since Darwin's time and it is no longer appropriate to claim that evolutionary biologists believe that Darwin's theory of Natural Selection is the best theory of the mechanism of evolution. I can understand why this point may not be appreciated by the average non-scientist because natural selection is easy to understand at a superficial level. It has been widely promoted in the popular press and the image of "survival of the fittest" is too powerful and too convenient.

5.0 Summary

During the first part of this century the incorporation of genetics and population biology into studies of evolution led to a Neo-Darwinian theory of evolution that recognized the importance of mutation and variation within a population. Natural selection then became a process that altered the frequency of genes in a population and this defined evolution. This point of view held sway for many decades but more recently the classic Neo-Darwinian view has been replaced by a new concept which includes several other mechanisms in addition to natural selection.

6.0 Tutor-Marked Assignment

- What is/are the importance of mechanisms governing evolution
- State and elaborate on three of these mechanisms.

7.0 Reference/Further Readings

Ewens W.J. (2004). *Mathematical Population Genetics (2nd Edition).* Springer-Verlag, New York..,

Hurst LD (2009). "Fundamental concepts in genetics: genetics and the understanding of selection". *Nat. Rev. Genet.* **10** (2): 83–93.

Morjan C, Rieseberg L (2004). "How species evolve collectively: implications of gene flow and selection for the spread of advantageous alleles". *Mol. Ecol.* **13** (6): 1341–56.,

Gillespie, John H. (2001). "Is the population size of a species relevant to its evolution?". *Evolution* **55.** Barton,

N H (2000). "Genetic hitchhiking". *Philosophical Transactions of the Royal Society of London. Series B, Biological Sciences* **355** (1403): 1553–1562.

Coyne; Barton, Turelli (1997). "Perspective: A Critique of Sewall Wright's Shifting Balance Theory of Evolution". *Evolution*. 3 **51**: 643–671.

Masel J (2011). "Genetic drift". *Current Biology* **21** (20): R837-R838.,

Mitchell-Olds, Thomas; Willis, JH; Goldstein, DB (2007). "Which evolutionary processes influence natural genetic variation for phenotypic traits?". *Nature Reviews Genetics* **8** (11): 845–856.,

Nei M (2005). "Selectionism and neutralism in molecular evolution". *Mol. Biol. Evol.* **22** (12): 2318–42..,

Hartl, Daniel L. & Andrew G. Clark. (1997). *Principles of Population Genetics*. Sunderland, Mass

Crow, James F. & Motoo Kimura. (1970). *Introduction to Population Genetics Theory*. Edina, Minn.: Burgess Publishing Company.,

Graur, Dan & Wen-Hsiung Li. (2000). *Fundamentals of Molecular Evolution*. Sunderland, Mass.: Sinauer Associates.

FUTUYMA, D. 1998. *Evolutionary biology* (3ª edición). Sinauer, Sunderland.

Mayr, E. 1963. *Animal species and evolution*. Harvard University Press, Cambridge.

Levine, L. 1979. Biología del gen. Ed. Omega, Barcelona. ISBN 84-282-0551-5.

Wiens, J. 2004. SPECIATION AND ECOLOGY REVISITED: PHYLOGENETIC NICHE CONSERVATISM AND THE ORIGIN OF SPECIES. Evolution 58: 193-197. DOI: 10.1111/j.0014-3820.2004.tb01586.x

UNIT 3: ISOLATION MECHANISMS

1.0 Introduction

2.0 Objectives

3.0 Morphological species concept:

3.1 Pre-zygotic

3.1.1 Temporal isolation

3.1.2 Sexual isolation by behaviour or conduct

3.1.3 Mechanical isolation

UNIT 3: ISOLATION MECHANISMS

1.0 Introduction

The biological species concept helps us ask how species are formed, because it focuses our attention on the question of how reproductive isolation comes about. There are many barriers to reproduction. Each species may have its own courtship displays, or breeding season, so that members of the two species do not have the opportunity to interbreed, or the two species may be unable to interbreed successfully because of failure of the egg to become fertilized or to develop.

2.0 Objectives

At the end of this unit, the student should be able to:

- Discuss extensively on Isolation mechanisms
- Discuss pre and post zygotic isolation
- How is reproductive isolation important to speciation and what forms can it take?

3.0 Morphological species concept:

Oak trees look like oak trees, tigers look like tigers. Morphology refers to the form and structure of an organism or any of its parts. The morphological species concept supports the widely held view that "members of a species are individuals that look similar to one another." This school of thought was the basis for Linneaus' original classification, which is still broadly accepted and applicable today.

This concept became criticized by biologists because it was arbitrary. Many examples were found in which individuals of two populations were very hard to tell apart but would not mate with one another, suggesting that they were in fact different species. Mimicry complexes supplied further evidence against the concept, as organisms of the same species can look very different, depending upon where they are reared or their life cycle stage (some insects

produce a spring brood that looks like one host plant and a summer brood that looks like another). The morphological species concept was replaced by another viewpoint that puts more emphasis on the biological differences between species.

Biological species concept: This concept states that "a species is a group of actually or potentially interbreeding individuals who are reproductively isolated from other such groups." This definition was attractive to biologists and became widely adopted by the 1940's. It suggested a critical test of species-hood: two individuals belong to the same species if their gametes can unite with each other under natural conditions to produce fertile offspring. This concept also emphasized that a species is an evolutionary unit. Members share genes with other members of their species, and not with members of other species.

Most scientists feel that the biological species concept should be kept, but with some qualifications. It can only be used with living species, and cannot always be applied to species that do not live in the same place. The real test applies to species that have the potential to interbreed.

Most importantly, the biological species concept helps us ask how species are formed, because it focuses our attention on the question of how reproductive isolation comes about. Let us first examine types of reproductive isolation, because there are quite a few.

There are many barriers to reproduction. Each species may have its own courtship displays, or breeding season, so that members of the two species do not have the opportunity to interbreed. Or, the two species may be unable to interbreed successfully because of failure of the egg to become fertilized or to develop.

This suggests a simple and useful dichotomy, between pre-mating or prezygotic (i.e., pre-zygote formation) reproductive isolating mechanisms, and post-mating or postzygotic

isolating mechanisms. Remember that a zygote is the cell formed by the union of two gametes and is the basis of a developing individual.

Despite our increasing ability to understand the finest details of organisms, there is still debate about what constitutes a species. Definitions of species tend to fall into two main camps, the morphological and the biological species concepts.

3.1 Pre-zygotic

The mechanisms of reproductive isolation or hybridization barriers are a collection of mechanisms, behaviors and physiological processes that prevent the members of two different species that cross or mate from producing offspring, or which ensure that any offspring that may be produced is not fertile. These barriers maintain the integrity of a species over time, reducing or directly impeding gene flow between individuals of different species, allowing the conservation of each species' characteristics

Pre-zygotic isolation mechanisms are the most economic in terms of the biological efficiency of a population, as resources are not wasted on the production of a descendent that is weak, non-viable or sterile.

3.1.1 Temporal isolation

Any of the factors that prevent potentially fertile individuals from meeting will reproductively isolate the members of distinct species. The types of barriers that can cause this isolation include: different habitats, physical barriers, and a difference in the time of sexual maturity or flowering. When factors change, especially physical barriers, often, species will branch off.

An example of the ecological or habitat differences that impede the meeting of potential pairs occurs in two fish species of the family *Gasterosteidae* (sticklebacks). One species lives all

year round in fresh water, mainly in small streams. The other species lives in the sea during winter, but in spring and summer individuals migrate to river estuaries to reproduce. The members of the two populations are reproductively isolated due to their adaptations to distinct salt concentrations. An example of reproductive isolation due to differences in the mating season are found in the toad species *Bufo americanus* and *Bufo fowleri* . The members of these species can be successfully crossed in the laboratory producing healthy, fertile hybrids. However, mating does not occur in the wild even though the geographical distribution of the two species overlaps. The reason for the absence of inter-species mating is that *B. americanus* mates in early summer and *B. fowleri* in late summer. Certain plant species, such as *Tradescantia canaliculata* and *T. subaspera*, are sympatric throughout their geographic distribution yet they are reproductively isolated as they flower at different times of the year. In addition, one species grows in sunny areas and the other in deeply shaded areas.

3.1.2 Sexual isolation by behaviour or conduct

The different mating rituals of animal species creates extremely powerful reproductive barriers, termed sexual or behavior isolation, that isolate apparently similar species in the majority of the groups of the animal kingdom. In dioecious species, males and females have to search for a partner, be in proximity to each other, carry out the complex mating rituals and finally copulate or release their gametes into the environment in order to breed.

Mating dances, the songs of males to attract females or the mutual grooming of pairs, are all examples of typical courtship behavior that allows both recognition and reproductive isolation. This is because each of the stages of courtship depend on the behavior of the partner. The male will only move onto the second stage of the exhibition if the female shows certain responses in her behavior. He will only pass onto the third stage when she displays a

second key behavior. The behaviors of both interlink, are synchronized in time and lead finally to copulation or the liberation of gametes into the environment. No animal that is not physiologically suitable for fertilization can complete this demanding chain of behavior. In fact, the smallest difference in the courting patterns of two species is enough to prevent mating (for example, a specific song pattern acts as an isolation mechanism in distinct species of grasshopper of the genus *Chorthippus*. Even where there are minimal morphological differences between species, differences in behavior can be enough to prevent mating. For example, *Drosophila melanogaster* and *D. simulans* which are considered twin species due to their morphological similarity, do not mate even if they are kept together in a laboratory. *Drosophila ananassae* and *D. pallidosa* are twin species from Melanesia. In the wild they rarely produce hybrids, although in the laboratory it is possible to produce fertile offspring. Studies of their sexual behavior show that the males court the females of both species but the females show a marked preference for mating with males of their own species. A different regulator region has been found on Chromosome II of both species that affects the selection behavior of the females.

Pheromones play an important role in the sexual isolation of insect species. These compounds serve to identify individuals of the same species and of the same or different sex. Evaporated molecules of volatile pheromones can serve as a wide-reaching chemical signal. In other cases, pheromones may be detected only at a short distance or by contact

In species of the *melanogaster* group of *Drosophila,* the pheromones of the females are mixtures of different compounds, there is a clear dimorphism in the type and/or quantity of compounds present for each sex. In addition, there are differences in the quantity and quality of constituent compounds between related species, it is assumed that the pheromones serve to distinguish between individuals of each species. An example of the role of pheromones in

sexual isolation is found in 'corn borers' in the genus *Ostrinia*. There are two twin species in Europe that occasionally cross. The females of both species produce pheromones that contain a volatile compound which has two isomers, E and Z; 99% of the compound produced by the females of one species is in the E isomer form, while the females of the other produce 99% isomer Z. The production of the compound is controlled by just one locus and the interspecific hybrid produces an equal mix of the two isomers. The males, for their part, almost exclusively detect the isomer emitted by the females of their species, such that the hybridization although possible is scarce. The perception of the males is controlled by one gene, distinct from the one for the production of isomers, the heterozygous males show a moderate response to the odour of either type. In this case, just 2 'loci' produce the effect of ethological isolation between species that are genetically very similar.

Sexual isolation between two species can be asymmetrical. This can happen when the mating that produces descendants only allows one of the two species to function as the female progenitor and the other as the male, while the reciprocal cross does not occur. For instance, half of the wolves tested in the Great Lakes area of America show mitochondrial DNA sequences of coyotes. While mitochondrial DNA from wolves is never found in coyote populations. This probably reflects an asymmetry in inter-species mating due to the difference in size of the two species as male wolves take advantage of their greater size in order to mate with female coyotes, while female wolves and male coyotes do not mate.

3.1.3 Mechanical isolation

Mating pairs may not be able to couple successfully if their genitals are not compatible. The relationship between the reproductive isolation of species and the form of their genital organs was signaled for the first time in 1844 by the French entomologist Léon Dufour. Insects' rigid carapaces act in a manner analogous to a lock and key, as they will only allow mating

between individuals with complementary structures, that is, males and females of the same species (termed *co-specifics*).

Evolution has led to the development of genital organs with increasingly complex and divergent characteristics, which will cause mechanical isolation between species. Certain characteristics of the genital organs will often have converted them into mechanisms of isolation. However, numerous studies show that organs that are anatomically very different can be functionally compatible, indicating that other factors also determine the form of these complicated structures.

Mechanical isolation also occurs in plants and this is related to the adaptation and coevolution of each species in the attraction of a certain type of pollinator (where pollination is zoophilic) through a collection of morphophysiological characteristics of the flowers (called floral syndrome), in such a way that the transport of pollen to other species does not occur.

3.1.4 Gametic isolation

The synchronous spawning of many species of coral in marine reefs means that inter-species hybridization can take place as the gametes of hundreds of individuals of tens of species are liberated into the same water at the same time. Approximately a third of all the possible crosses between species are compatible, in the sense that the gametes will fuse and lead to individual hybrids. This hybridization apparently plays a fundamental role in the evolution of coral species. However, the other two-thirds of possible crosses are incompatible. It has been observed that in sea urchins of the genus *Strongylocentrotus* the concentration of spermatocytes that allow 100% fertilization of the ovules of the same species is only able to fertilize 1.5% of the ovules of other species. This inability to produce hybrid offspring,

despite the fact that the gametes are found at the same time and in the same place, is due to a phenomenon known as *gamete incompatibility*, which is often found between marine invertebrates, and whose physiological causes are not fully understood.

In some *Drosophila* crosses, the swelling of the female's vagina has been noted following insemination. This has the effect of consequently, preventing the fertilization of the ovule by sperm of a different species.

In plants the pollen grains of a species can germinate in the stigma and grow in the style of other species. However, the growth of the pollen tubes may be detained at some point between the stigma and the ovules, in such a way that fertilization does not take place. This mechanism of reproductive isolation is common in the Angiosperms and is called *cross-incompatibility* or *incongruence*. A relationship exists between self-incompatibility and the phenomenon of cross-incompatibility. In general crosses between individuals of a self-compatible species (SC) with individuals of a self-incompatible (SI) species give hybrid offspring. On the other hand, a reciprocal cross (SI x SC) will not produce offspring, because the pollen tubes will not reach the ovules. This is known as *unilateral incompatibility*, which also occurs when two SC or two SI species are crossed.

3.2 Post-zygotic isolation

3.2.1 Zygote mortality and non-viability of hybrids

A type of incompatibility that is found as often in plants as in animals occurs when the ovule is fertilized but the zygote does not develop, or it develops and the resulting individual has a reduced viability. This is the case for crosses between species of the frog genus, where widely differing results are observed depending of the species involved. In some crosses there is no segmentation of the zygote. In others, normal segmentation occurs in the blastula but

gastrulation fails. Finally, in other crosses, the initial stages are normal but errors occur in the final phases of embryo development. This indicates differentiation of the embryo development genes (or gene complexes) in these species and these differences determine the non-viability of the hybrids.

Similar results are observed in mosquitos of the Culex genus, but the differences are seen between reciprocal crosses, from which it is concluded that the same effect occurs in the interaction between the genes of the cell nucleus (inherited from both parents) as occurs in the genes of the cytoplasmic organelles which are inherited solely from the female progenitor through the cytoplasm of the ovule.

In Angiosperms, the successful development of the embryo depends on the normal functioning of its endosperm.

The failure of endosperm development and its subsequent abortion has been observed in many interploidal crosses (that is, those between populations with a particular degree of intra or interspecific ploidy and in certain crosses in species with the same level of ploidy. The collapse of the endosperm, and the subsequent abortion of the hybrid embryo is one of the most common post-fertilization reproductive isolation mechanism found in angiosperms.

3.2.2 Hybrid sterility

A hybrid has normal viability but is deficient in terms of reproduction or is sterile. This is demonstrated by the mule and in many other well known hybrids. In all of these cases sterility is due to the interaction between the genes of the two species involved; to chromosomal imbalances due to the different number of chromosomes in the parent species; or to nucleus-cytoplasmic interactions such as in the case of *Culex* described above.

Hinnies and mules are hybrids resulting from a cross between a horse and an ass or between a mare and a donkey, respectively. These animals are nearly always sterile due to the difference in the number of chromosomes between the two parent species. Both horses and donkeys belong to the genus *Equus*, but *Equus caballus* has 64 chromosomes, while *Equus asinus* only has 62. A cross will produce offspring (mule or hinny) with 63 chromosomes, that will not form pairs, which means that they do not divide in a balanced manner during meiosis. It is curious that they can cross with each other but the mule and the hinny are actually animals created by humans, as in the wild the species ignore each other and do not cross. In order to obtain mules or hinnies it is necessary to train the progenitors to accept copulation between the species or create them through artificial insemination.

The sterility of many of the interspecific hybrids among the angiosperms is a widely recognised and studied phenomenon. There are a variety of causes that can determine the interspecific sterility of hybrids in plants, these may be genetic, related to the genomes or the interaction between nuclear and cytoplasmic factors, as will be discussed in the corresponding section. Nevertheless, it should be pointed out that - on the contrary to the situation in animals - hybridization in plants is a stimulus for the creation of new species. Indeed, although the hybrid may be sterile it can continue to multiply in the wild through the mechanisms of asexual reproduction, be they vegetative propagation or apomixis or the production of seeds. Indeed, interspecific hybridization can be associated with polyploidia and, in this way, the origin of new species that are called allopolyploids. *Rosa canina*, for example, is the result of multiple hybridizations. or there is a type of wheat that is an allohexaploid that contains the genomes of three different species.

3.2.3 Multiple mechanisms

In general, the barriers that separate species do not consist of just one mechanism. The twin species of *Drosophila, D. pseuoobscura* and *D. persimilis*, are isolated from each other by habitat (*persimilis* generally lives in colder regions at higher altitudes), by the timing of the mating season (*persimilis* is generally more active in the morning and *pseuoobscura* at night) and by behavior during mating (the females of both species prefer the males of their respective species). In this way, although the distribution of these species overlaps in wide areas of the west of the United States of America, these isolation mechanisms are sufficient to keep the species separated. Such that, only a few fertile females have been found amongst the other species among the thousands that have been analyzed. However, when hybrids are produced between both species, the gene flow between the two will continue to be impeded as the hybrid males are sterile. Also, and in contrast with the great vigor shown by the sterile males, the descendants of the backcrosses of the hybrid females with the parent species are weak and notoriously non-viable. This last mechanism restricts even more the genetic interchange between the two species of fly in the wild.

3.2.4 Hybrid gender

Haldane's Rule states that when one of the two sexes is absent in interspecific hybrids between two specific species and then the gender that is not produced, is rare or is sterile is the heterozygous (or heterogametic) sex. In mammals, at least, there is growing evidence to suggest that this is due to high rates of mutation of the genes determining masculinity in the Y chromosome.

It has been suggested that Haldane's Rule simply reflects the fact that the male gender is more sensitive than the female when the sex-determining genes are included in a hybrid genome. But there are also organisms in which the heterozygous sex is the female: birds and

butterflies and the law is followed in these organisms. Therefore, it is not a problem related to sexual development, nor with the sex chromosomes. Haldane proposed that the stability of hybrid individual development requires the full gene complement of each parent species, so that the hybrid of the heterozygous sex is unbalanced (i.e. missing at least one chromosome from each of the parental species). For example, the hybrid male obtained by crossing *D. melanogaster* females with *D. simulans* males, which is non-viable, lacks the X chromosome of *D. simulans*.

3.3 Genetics of isolation barriers

3.3.1 Pre-copulatory isolation mechanisms in animals

The genetics of ethological isolation barriers will be discussed first. Pre-copulatory isolation occurs when the genes necessary for the sexual reproduction of one species differ from the equivalent genes of another species, such that if a male of species A and a female of species B are placed together they are unable to copulate. Study of the genetics involved in this reproductive barrier tries to identify the genes that govern distinct sexual behaviors in the two species. The males of *Drosophila melanogaster* and those of *D. simulans* conduct an elaborate courtship with their respective females, which are different for each species, but the differences between the species are more quantitative than qualitative. In fact the *simulans* males are able to hybridize with the *melanogaster* females. Although there are lines of the latter species that can easily cross there are others that are hardly able to. Using this difference, it is possible to assess the minimum number of genes involved in pre-copulatory isolation between the *melanogaster* and *simulans* species and their chromosomal location.

In experiments, flies of the *D. melanogaster* line, which hybridizes readily with *simulans*, were crossed with another line that it does not hybridize with, or rarely. The females of the

segregated populations obtained by this cross were placed next to *simulans* males and the percentage of hybridization was recorded, which is a measure of the degree of reproductive isolation. It was concluded from this experiment that 3 of the 8 chromosomes of the haploid complement of *D. melanogaster* carry at least one gene that affects isolation, such that substituting one chromosome from a line of low isolation with another of high isolation reduces the hybridization frequency. In addition, interactions between chromosomes are detected so that certain combinations of the chromosomes have a multiplying effect. Cross incompatibility or incongruence in plants is also determined by major genes that are not associated at the self-incompatibility *S locus*.

3.3.2 Post copulation or fertilization isolation mechanisms in animals

Reproductive isolation between species appears, in certain cases, a long time after fertilization and the formation of the zygote, as happens - for example - in the twin species *Drosophila pavani* and *D. gaucha*. The hybrids between both species are not sterile, in the sense that they produce viable gametes, ovules and spermatozoa. However, they cannot produce offspring as the sperm of the hybrid male do not survive in the semen receptors of the females, be they hybrids or from the parent lines. In the same way, the sperm of the males of the two parent species do not survive in the reproductive tract of the hybrid female.[12] This type of post copulatory isolation appears as the most efficient system for maintaining reproductive isolation in many species.

In fact, the development of a zygote into an adult is a complex and delicate process of interactions between genes and the environment that must be carried out precisely, and if there is any alteration in the usual process, caused by the absence of a necessary gene or the presence of a different one, it can arrest the normal development causing the non-viability of the hybrid or its sterility. It should be borne in mind that half of the chromosomes and genes

of a hybrid are from one species and the other half come from the other. If the two species are genetically different, there is little possibility that the genes from both will act harmoniously in the hybrid. From this perspective, only a few genes would be required in order to bring about post copulatory isolation, as opposed to the situation described previously for pre-copulatory isolation.

In many species where pre-copulatory reproductive isolation does not exist, hybrids are produced but they are of only one sex. This is the case for the hybridization between females of *Drosophila simulans* and *Drosophila melanogaster* males: the hybridized females die early in their development so that only males are seen among the offspring. However, populations of *D. simulans* have been recorded with genes that permit the development of adult hybrid females, that is, the viability of the females is "rescued". It is assumed that the normal activity of these speciation genes is to "inhibit" the expression of the genes that allow the growth of the hybrid. There will also be regulator genes.

A number of these genes have been found in the *melanogaster* species group. The first to be discovered was "Lhr" (Lethal hybrid rescue) located in Chromosome II of *D. simulans*. This dominant allele allows the development of hybrid females from the cross between *simulans* females and *melanogaster* males. A different gene, also located on Chromosome II of *D. simulans* is "Shfr" that also allows the development of female hybrids, its activity being dependent on the temperature at which development occurs. Other similar genes have been located in distinct populations of species of this group. In short, only a few genes are needed for an effective post copulatory isolation barrier mediated through the non-viability of the hybrids.

As important as identifying an isolation gene is knowing its function. The *Hmr* gene, linked to the X chromosome and implicated in the viability of male hybrids between *D.*

melanogaster and *D. simulans*, is a gene from the proto-oncogene family *myb*, that codes for a transcriptional regulator. Two variants of this gene function perfectly well in each separate species, but in the hybrid they do not function correctly, possibly due to the different genetic background of each species. Examination of the allele sequence of the two species shows that change of direction substitutions are more abundant than synonymous substitutions, suggesting that this gene has been subject to intense natural selection.

The Dobzhansky-Muller model proposes that reproductive incompatibilities between species are caused by the interaction of the genes of the respective species. It has been demonstrated recently that *Lhr* has functionally diverged in *D. simulans* and will interact with *Hmr* which, in turn, has functionally diverged in *D. melanogaster* to cause the lethality of the male hybrids. *Lhr* is located in a heterochromatic region of the genome and its sequence has diverged between these two species in a manner consistent with the mechanisms of positive selection. An important unanswered question is whether the genes detected correspond to old genes that initiated the speciation favoring hybrid non-viability, or are modern genes that have appear post-speciation by mutation, that are not shared by the different populations and that suppress the effect of the primitive non-viability genes. The *OdsH* (abbreviation of *Odysseus*) gene causes partial sterility in the hybrid between *Drosophila simulans* and a related species, *D. mauritiana*, which is only encountered on Mauritius, and is of recent origin. This gene shows monophyly in both species and also has been subject to natural selection. It is thought that it is a gene that intervenes in the initial stages of speciation, while other genes that differentiate the two species show polyphyly. *Odsh* originated by duplication in the genome of *Drosophila* and has evolved at very high rates in *D. mauritania*, while its paralogue, *unc-4*, is nearly identical between the species of the group *melanogaster*. Seemingly, all these cases illustrate the manner in which speciation mechanisms originated in nature, therefore they are collectively known as “speciation genes”, or possibly, gene

sequences with a normal function within the populations of a species that diverge rapidly in response to positive selection thereby forming reproductive isolation barriers with other species. In general, all these genes have functions in the transcriptional regulation of other genes.

The *Nup96* gene is another example of the evolution of the genes implicated in post-copulatory isolation. It regulates the production of one of the approximately 30 proteins required to form a nuclear pore. In each of the *simulans* groups of *Drosophila* the protein from this gene interacts with the protein from another, as yet undiscovered, gene on the X chromosome in order to form a functioning pore. However, in a hybrid the pore that is formed is defective and causes sterility. The differences in the sequences of *Nup96* have been subject to adaptive selection, similar to the other examples of *speciation genes* described above.

Post-copulatory isolation can also arise between chromosomally differentiated populations due to chromosomal translocations and inversions. If, for example, a reciprocal translocation is fixed in a population, the hybrid produced between this population and one that does not carry the translocation will not have a complete meiosis. This will result in the production of unequal gametes containing unequal numbers of chromosomes with a reduced fertility. In certain cases, complete translocations exist that involve more than two chromosomes, so that the meiosis of the hybrids is irregular and their fertility is zero or nearly zero. Inversions can also give rise to abnormal gametes in heterozygous individuals but this effect has little importance compared to translocations. An example of chromosomal changes causing sterility in hybrids comes from the study of *Drosophila nasuta* and *D. albomicans* which are twin species from the Indo-Pacific region. There is no sexual isolation between them and the F1 hybrid is fertile. However, the F2 hybrids are relatively infertile and leave few

descendants which have a skewed ratio of the sexes. The reason is that the X chromosome of *albomicans* is translocated and linked to an autosome which causes abnormal meiosis in hybrids. Robertsonian translocations are variations in the numbers of chromosomes that arise from either: the fusion of two acrocentric chromosomes into a single chromosome with two arms, causing a reduction in the haploid number, or conversely; or the fission of one chromosome into two acrocentric chromosomes, in this case increasing the haploid number. The hybrids of two populations with differing numbers of chromosomes can experience a certain loss of fertility, and therefore a poor adaptation, because of irregular meiosis.

3.3.3. Post copulation or fertilization isolation mechanisms in plants

In plants, hybrids often suffer from an autoimmune syndrome known as hybrid necrosis. In the hybirds, specific gene products contributed by one of the parents may be inappropriately recognized as foreign and pathogenic, and thus trigger pervasive cell death throughout the plant. In at least one case, a pathogen receptor, encoded by the most variable gene family in plants, was identified as being responsible for hybrid necrosis.

3.4 Selection for reproductive isolation

In 1950 K. F. Koopman reported results from experiments designed to examine the hypothesis that selection can increase reproductive isolation between populations. He used *D. pseudoobscura* and *D. persimilis* in these experiments. When the flies of these species are kept at 16°C approximately a third of the matings are interspecific. In the experiment equal numbers of males and females of both species were placed in containers suitable for their survival and reproduction. The progeny of each generation were examined in order to determine if there were any interspecific hybrids. These hybrids were then eliminated. An equal number of males and females of the resulting progeny were then chosen to act as

progenitors of the next generation. As the hybrids were destroyed in each generation the flies that solely mated with members of their own species produced more surviving descendants than the flies that mated solely with individuals of the other species. In the table to the right it can be seen that for each generation the number of hybrids continuously decreased up to the tenth generation when hardly any interspecific hybrids were produced. It is evident that selection against the hybrids was very effective in increasing reproductive isolation between these species. From the third generation, the proportions of the hybrids were less than 5%. This confirmed that selection acts to reinforce the reproductive isolation of two genetically divergent populations if the hybrids formed by these species are less well adapted than their parents.

These discoveries allowed certain assumptions to be made regarding the origin of reproductive isolation mechanisms in nature. Namely, if selection reinforces the degree of reproductive isolation that exists between two species due to the poor adaptive value of the hybrids, it is expected that the populations of two species located in the same area will show a greater reproductive isolation than populations that are geographically separated. This mechanism for “reinforcing” hybridization barriers in sympatric populations is called the "Wallace Effect", as it was first proposed by Alfred Russell Wallace at the end of the 19th century, and it has been experimentally demonstrated in both plants and animals.

The sexual isolation between *Drosophila miranda* and *D. pseudoobscura*, for example, is more or less pronounced according to the geographic origin of the flies being studied. Flies from regions where the distribution of the species is superimposed show a greater sexual isolation than exists between populations originating in distant regions.

On the other hand, interspecific hybridization barriers can also arise as a result of the adaptive divergence that accompanies allopatric speciation. This mechanism has been experimentally

proved by an experiment carried out by Diane Dodd on *D. pseudoobscura*. A single population of flies was divided into two, with one of the populations fed with starch-based food and the other with maltose-based food. This meant that each sub population was adapted to each food type over a number of generations. After the populations had diverged over many generations, the groups were again mixed; it was observed that the flies would mate only with others from their adapted population. This indicates that the mechanisms of reproductive isolation can arise even though the interspecific hybrids are not selected against.

4.0 Conclusion

In this student the student learnt the following:

- Mechanism of isolation
- Pre and Post-zygotic isolation
- Mechanisms governing isolation mechanisms
- Genetics of isolation barriers

5.0 Summary

Isolating mechanisms are the reproductive characteristics which prevent species from fusing. Isolating mechanisms are particularly important in the biological species concept, in which species of sexual organisms are defined by reproductive isolation, i.e. a lack of gene mixture. Two broad kinds of isolating mechanisms between species are typically distinguished, together with a number of sub-types.

6.0 Tutor-Marked Assignment

- What do you understand by isolation mechanisms?

- What is the relationship between isolation mechanisms and the biological species concept?

7.0 References/Further Reading

Mayr E. (1970) Populations, Species, and Evolution. Harvard University Press, Cambridge, Mass.

Futuyma, D.J. 1986. *Evolutionary Biology*. Sunderland, Mass: Sinauer Associates, Inc.

Wessells, N.K. and J.L. Hopson. 1988. *Biology*. New York: Random House. Chapter 43.

Rosenzweig, M.L. 1995. *Species Diversity in Space and Time*. Cambridge: Cambridge University Press.

Wu, C. I.; Hollocher, H.; Begun, D. J.; Aquadro, C. F.; Xu, Y.; Wu, M. L. (1995), "Sexual isolation in *Drosophila melanogaster*: a possible case of incipient speciation", *Proceedings of the National Academy of Sciences of the United States of America* 92 (7): 2519–2523, doi:10.1073/pnas.92.7.2519, PMC 42249, PMID 7708677

West-eberhard, M.J. (1983), "Sexual Selection, Social Competition, and Speciation", *The Quarterly Review of Biology* 58 (2): 155, doi:10.1086/413215

Mendelson, T.C. (2003), "Sexual Isolation Evolves Faster Than Hybrid Inviability in a Diverse and Sexually Dimorphic enus of", *Evolution* 57 (2): 317–327, doi:10.1111/j.0014-3820.2003.tb00266.x, PMID 12683528

Perdeck, A.C. (1958), "The Isolating Value of Specific Song Patterns in Two Sibling Species of Grasshoppers (*Chorthippus brunneus* Thunb. and *C. biguttulus* L.)", *Behaviour* 12 (1-2): 1–75, doi:10.1163/156853957X00074

Casares, P. 2008. *Genética de poblaciones y evolutiva*. Capítulo 17. Genética del aislamiento reproductivo. Universidad de Oviedo, España. [1]

Coyne, J.A.; Crittenden, A.P.; Mah, K. (1994), "Genetics of a pheromonal difference contributing to reproductive isolation in *Drosophila*", *Science* 265 (5177): 1461–1464, doi:10.1126/science.8073292, PMID 8073292

LEHMAN N; Eisenhawer, A.; Hansen, K.; David Mech, L.; Peterson, R. O.; Gogan, P. J. P.; Wayne, R. K. (1991), "Introgression of coyote mitochondrial DNA into sympatric North American gray wolf populations", *Evolution* 45 (1): 104–119, doi:10.2307/2409486, JSTOR 2409486.

UNIT 4: MUTATION

1.0 Introduction

2.0 Objectives

3.0 Mutation

3.1 Causes of mutation

3.2 Classifying Mutations

3.3 Harmful mutations

3.4 Beneficial Mutations

4.0 Conclusion

5.0 Summary

6.0 Tutor-Marked Assignment

7.0 Reference/Further Reading

Unit 4 Mutation

1.0 Introduction

In molecular biology and genetics, mutations are changes in a genomic sequence: the DNA sequence of a cell's genome or the DNA or RNA sequence of a virus. They can be defined as sudden and spontaneous changes in the cell. Mutations are caused by radiation, viruses and

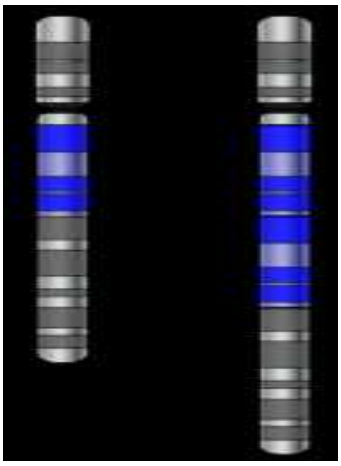
mutagenic chemicals, as well as errors that occur during meiosis or DNA replication. They can also be induced by the organism itself, by cellular processes such as hyper-mutation. A mutation is passed to the offspring stably, unless it is a dynamic mutation.

2.0 Objectives

At the end of this unit, the student should know the following:

- Definition of mutation
- What causes mutation
- Types of mutation

3.0 Mutation



Duplication of part of a chromosome

The cellular machinery that copies DNA sometimes makes mistakes. These mistakes change the sequence of a gene. This is called a mutation. There are many kinds of mutations. A point mutation is a mutation in which one "letter" of the genetic code is changed to another. Lengths of DNA can also be deleted or inserted in a gene; these are also mutations. Finally, genes or parts of genes can become inverted or duplicated. Typical rates of mutation are between 10^{-10} and 10^{-12} mutations per base pair of DNA per generation.

Most mutations are thought to be neutral with regards to fitness. Only a small portion of the genome of eukaryotes contains coding segments, although some non-coding DNA is involved in gene regulation or other cellular functions, it is probable that most base changes would have no fitness consequence.

Most mutations that have any phenotypic effect are deleterious. Mutations that result in amino acid substitutions can change the shape of a protein, potentially changing or eliminating its function. This can lead to inadequacies in biochemical pathways or interfere with the process of development. Organisms are sufficiently integrated that most random changes will not produce a fitness benefit. Only a very small percentage of mutations are

beneficial. The ratio of neutral to deleterious to beneficial mutations is unknown and probably varies with respect to details of the locus in question and environment.

Mutation limits the rate of evolution. The rate of evolution can be expressed in terms of nucleotide substitutions in a lineage per generation. Substitution is the replacement of an allele by another in a population. This is a two step process: First a mutation occurs in an individual, creating a new allele. This allele subsequently increases in frequency to fixation in the population.

The rate of evolution is $k = 2Nvu$ (in diploids)

Where

k is nucleotide substitutions,

N is the effective population size,

v is the rate of mutation and

u is the proportion of mutants that eventually fix in the population.

Mutation need not be limiting over short time spans. The rate of evolution expressed above is given as a steady state equation; it assumes the system is at equilibrium. Given the time frames for a single mutant to fix, it is unclear if populations are ever at equilibrium. A change in environment can cause previously neutral alleles to have selective values; in the short term evolution can run on "stored" variation and thus is independent of mutation rate. Other mechanisms can also contribute selectable variation. Recombination creates new combinations of alleles (or new alleles) by joining sequences with separate micro-evolutionary histories within a population. Gene flow can also supply the gene pool with variants. Of course, the ultimate source of these variants is mutation.

3.1 Causes of mutation

Two classes of mutations are spontaneous mutations and induced mutations caused by mutagens.

Spontaneous mutation

Spontaneous mutations on the molecular level can be caused by:

- Tautomerism – A base is changed by the repositioning of a hydrogen atom, altering the hydrogen bonding pattern of that base resulting in incorrect base pairing during replication.
- Depurination – Loss of a purine base (A or G) to form an apurinic site (AP site).
- Deamination – Hydrolysis changes a normal base to an atypical base containing a keto group in place of the original amine group. Examples include C → U and A → HX (hypoxanthine), which can be corrected by DNA repair mechanisms; and 5MeC (5-methylcytosine) → T, which is less likely to be detected as a mutation because thymine is a normal DNA base.
- Slipped strand mispairing - Denaturation of the new strand from the template during replication, followed by renaturation in a different spot ("slipping"). This can lead to insertions or deletions.

Induced mutation

Induced mutations on the molecular level can be caused by:

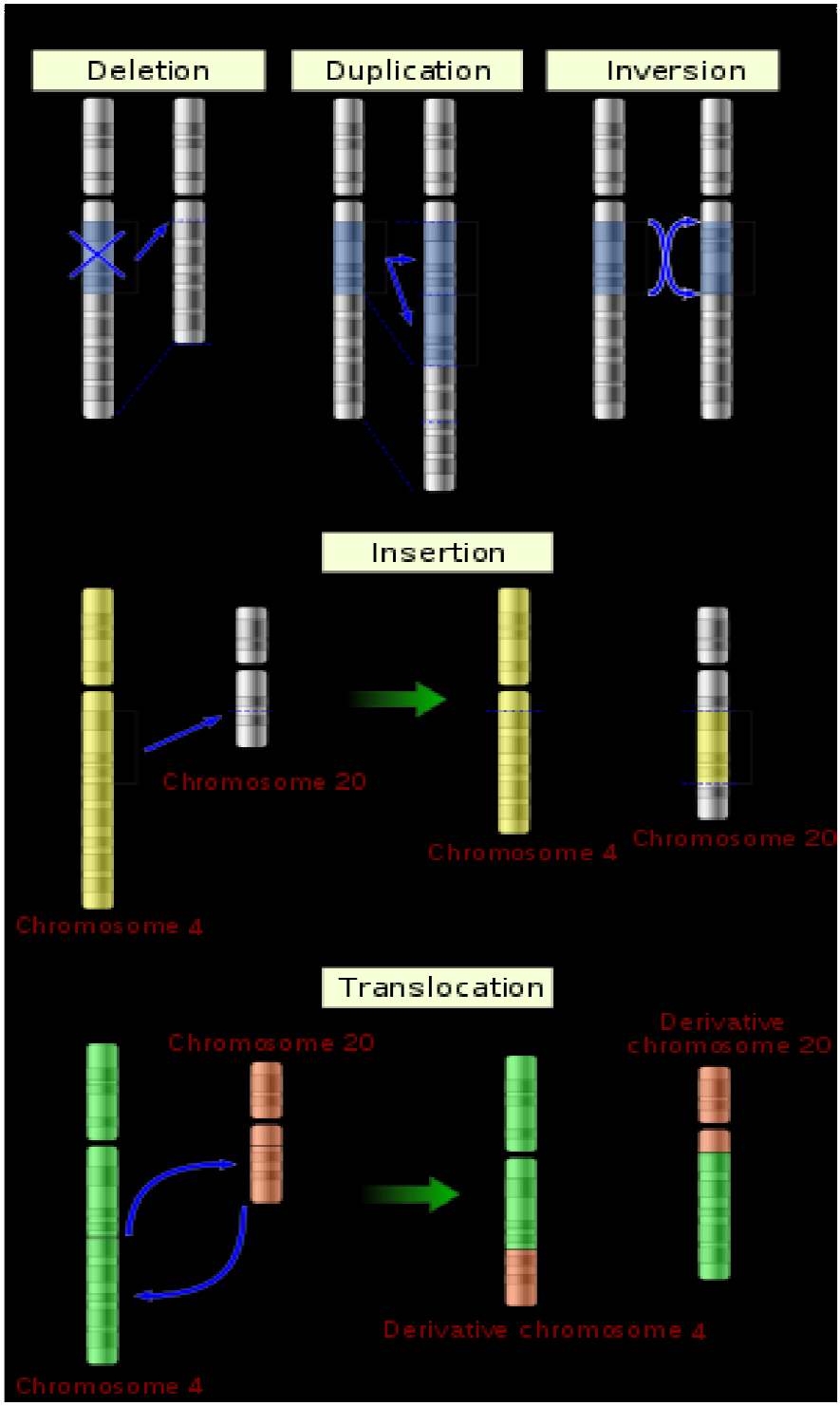
- Chemicals
    - Hydroxylamine NH_2OH
    - Base analogs (e.g. BrdU)

    - Alkylating agents (e.g. *N*-ethyl-*N*-nitrosourea) These agents can mutate both replicating and non-replicating DNA. In contrast, a base analog can only mutate the DNA when the analog is incorporated in replicating the DNA. Each of these classes of chemical mutagens has certain effects that then lead to transitions, transversions, or deletions.
    - Agents that form DNA adducts (e.g. ochratoxin A metabolites)
    - DNA intercalating agents (e.g. ethidium bromide)
    - DNA crosslinkers
    - Oxidative damage
    - Nitrous acid converts amine groups on A and C to diazo groups, altering their hydrogen bonding patterns which leads to incorrect base pairing during replication.
- Radiation
    - Ultraviolet radiation (nonionizing radiation). Two nucleotide bases in DNA – cytosine and thymine – are most vulnerable to radiation that can change their properties. UV light can induce adjacent pyrimidine bases in a DNA strand to become covalently joined as a pyrimidine dimer. UV radiation, particularly longer-wave UVA, can also cause oxidative damage to DNA.
    - Ionizing radiation
    - Radioactive decay, such as ^{14}C in DNA
- Viral infections

DNA has so-called hotspots, where mutations occur up to 100 times more frequently than the normal mutation rate. A hotspot can be at an unusual base, e.g., 5-methylcytosine.

Mutation rates also vary across species. Evolutionary biologists have theorized that higher mutation rates are beneficial in some situations, because they allow organisms to evolve and therefore adapt more quickly to their environments. For example, repeated exposure of bacteria to antibiotics, and selection of resistant mutants, can result in the selection of bacteria that have a much higher mutation rate than the original population.

3.2 Classifying Mutations



Five types of chromosomal mutations.

1. Structural Effects

The sequence of a gene can be altered in a number of ways. Gene mutations have varying effects on health depending on where they occur and whether they alter the function of essential proteins. Structurally, mutations can be classified as:

Small-scale mutations. These are mutations that affecting one or a few nucleotides. These include:

Point mutations. Point mutations involve an exchange of a single nucleotide for another one. Most common is the transition that exchanges a purine for a purine (A ↔ G) or a pyrimidine for a pyrimidine, (C ↔ T). Point mutations are often caused by chemicals or malfunction of DNA replication. A transition can be caused by nitrous acid, base mis-pairing, or mutagenic base analogs such as 5-bromo-2-deoxyuridine (BrdU). Less common is a transversion, which exchanges a purine for a pyrimidine or a pyrimidine for a purine (C/T ↔ A/G). A point mutation can be reversed by another point mutation, in which the nucleotide is changed back to its original state (true reversion) or by second-site reversion (a complementary mutation elsewhere that results in regained gene functionality). These changes are classified as transitions or transversions. An example of a transversion is adenine (A) being converted into a cytosine (C). There are also many other examples that can be found. Another Point mutation that occur within the protein coding region of a gene may be classified into three kinds, depending upon what the erroneous codon codes for:

Silent mutations. Silent mutations are those that code for the same amino acid, because of the redundancy of the genetic code. That is, there is a change in the bases but the altered triplet still inserts the same amino acid as before into the protein.

Missense mutations. Missense mutations code for a different amino acid.

Nonsense mutations. Nonsense mutations code for a "stop" and can truncate the protein.

Insertions. Insertions are mutations that add one or more extra nucleotides into the DNA. They are usually caused by transposable elements, or errors during replication of repeating elements (e.g. AT repeats). Insertions in the coding region of a gene may alter splicing of the mRNA (splice site mutation), or cause a shift in the reading frame (frameshift), both of which can significantly alter the gene product. Insertions can be reverted by excision of the transposable element.

Deletions. Deletions remove one or more nucleotides from the DNA. Like insertions, these mutations can alter the reading frame of the gene. They are irreversible.

Large-scale mutations. Large scale mutations in chromosomal structure may include:

Amplifications. Amplifications (or gene duplications) lead to multiple copies of all chromosomal regions, increasing the dosage of the genes located within them.

Deletions. Deletions of large chromosomal regions lead to loss of the genes within those regions.

Juxtaposing mutations. Mutations whose effect is to juxtapose previously separate pieces of DNA, potentially bringing together separate genes to form functionally distinct fusion genes (e.g. bcr-abl). These include:

Chromosomal translocations. Chromosomal translocations interchange genetic parts from nonhomologous chromosomes.

Interstitial deletions. Intersitional deletions involve an intra-chromosomal deletion that removes a segment of DNA from a single chromosome, thereby apposing previously distant genes. For example, cells isolated from a human astrocytoma, a type of brain tumor, were found to have a chromosomal deletion removing sequences between the "fused in

glioblastoma" (fig) gene and the receptor tyrosine kinase "ros," producing a fusion protein (FIG-ROS). The abnormal FIG-ROS fusion protein has constitutively active kinase activity that causes oncogenic transformation (a transformation from normal cells to cancer cells).

Chromosomal inversions. Chromosomal inversions involve reversing the orientation of a chromosomal segment.

Loss of heterozygosity. Loss of heterozygosity involves the loss of one allele, either by a deletion or recombination event, in an organism that previously had two different alleles.

2 Functional Effects

Loss-of-function mutations. Loss-of-function mutations are the result of gene product having less or no function. When the allele has a complete loss of function (null allele) it is often called an *amorphic mutation*. Phenotypes associated with such mutations are most often recessive. Exceptions are when the organism is haploid, or when the reduced dosage of a normal gene product is not enough for a normal phenotype (this is called haploinsufficiency).

Gain-of-function mutations. Gain-of-function mutations change the gene product such that it gains a new and abnormal function. These mutations usually have dominant phenotypes. Often called a *neo-morphic mutation*.

Dominant negative mutations. Dominant negative mutations (also called *anti-morphic mutations*) have an altered gene product that acts antagonistically to the wild-type allele. These mutations usually result in an altered molecular function (often inactive) and are characterized by a dominant or semi-dominant phenotype. In humans, Marfan syndrome is an example of a dominant negative mutation occurring in an autosomal dominant disease. In this

condition, the defective glycoprotein product of the fibrillin gene (FBN1) antagonizes the product of the normal allele.

Lethal mutations. Lethal mutations are mutations that lead to a phenotype incapable of effective reproduction.

3 By aspect of phenotype affected

Morphological mutations. Morphological mutations usually affect the outward appearance of an individual. Mutations can change the height of a plant or change it from smooth to rough seeds.

Biochemical mutations. Biochemical mutations result in lesions stopping the enzymatic pathway. Often, morphological mutants are the direct result of a mutation due to the enzymatic pathway.

4 By inheritance

The human genome contains two copies of each gene—a paternal and a maternal allele.

- **Wildtype** or **Homozygous non-mutated**. This occurs when neither of the alleles are mutated.
- A **Heterozygous mutation**. Heterozygous mutations occurs when only one allele is mutated.
- A **Homozygous mutation**. Homozygous mutation is when both the paternal and maternal alleles have an identical mutation.
- **Compound heterozygous** mutations. Compound heterozygous mutations or a **genetic compound** is when the paternal and maternal alleles have two different mutations.

5 Special classes

- **Conditional mutation**. Conditional mutation is a mutation that has wild-type (or less severe) phenotype under certain "permissive" environmental conditions and a mutant phenotype under certain "restrictive" conditions. For example, a temperature-sensitive mutation can cause cell death at high temperature (restrictive condition), but might have no deleterious consequences at a lower temperature (permissive condition).

3.3 Harmful mutations

Changes in DNA caused by mutation can cause errors in protein sequence, creating partially or completely non-functional proteins. To function correctly, each cell depends on thousands of proteins to function in the right places at the right times. When a mutation alters a protein that plays a critical role in the body, a medical condition can result. A condition caused by mutations in one or more genes is called a genetic disorder. However, only a small percentage of mutations cause genetic disorders; most have no impact on health. For example, some mutations alter a gene's DNA base sequence but do not change the function of the protein made by the gene.

If a mutation is present in a germ cell, it can give rise to offspring that carries the mutation in all of its cells. This is the case in hereditary diseases. On the other hand, a mutation can occur in a somatic cell of an organism. Such mutations will be present in all descendants of this cell, and certain mutations can cause the cell to become malignant, and thus cause cancer.

Often, gene mutations that could cause a genetic disorder are repaired by the DNA repair system of the cell. Each cell has a number of pathways through which enzymes recognize and repair mistakes in DNA. Because DNA can be damaged or mutated in many ways, the process of DNA repair is an important way in which the body protects itself from disease.

3.4 Beneficial Mutations

A very small percentage of all mutations actually have a positive effect. These mutations lead to new versions of proteins that help an organism and its future generations better adapt to changes in their environment.

For example, a specific 32 base pair deletion in human CCR5 (CCR5-Δ32) confers HIV resistance to homozygotes and delays AIDS onset in heterozygotes. The CCR5 mutation is more common in those of European descent. One theory for the etiology of the relatively high frequency of CCR5-Δ32 in the European population is that it conferred resistance to the bubonic plague in mid-fourteenth century Europe. People who had this mutation were able to survive infection thus its frequency in the population increased. It could also explain why this mutation is not found in Africa where the bubonic plague never reached. A more recent theory says the selective pressure on the CCR5 Delta 32 mutation has been caused by smallpox instead of bubonic plague.

4.0 Conclusion

At the end of this unit, the student learnt the following:

- Definition of mutation
- Causes and Classifications of mutation.

5.0 Summary

Mutation is the alteration of DNA sequence, whether it is in a small way by the alteration of a single base pair, or whether it be a gross event such as the gain or loss of an entire chromosome. It may be caused through the action of damaging chemicals, or radiation, or through the errors inherent in the DNA replication and repair reactions. One consequence

may be genetic disease. However, although in the short term mutation may seem to be a *BAD THING*, in the long term it is essential to our existence. Without mutation there could be no change and without change life cannot evolve. If it had not been for mutation the world would still be covered in primeval slime.

6.0 Tutor-Marked Assignment

- What is mutation?
- What are the different types of mutation?
- How important is mutation to the evolutionary process?

7.0 Reference/Further Reading

Types of genetic mutations. http://ghr.nlm.nih.gov/

Leroi, A. M. 2003. *Mutants: On Genetic Variety and the Human Body.* New York: Viking. ISBN 0670031100.

Maki, H. 2002. Origins of spontaneous mutations: specificity and directionality of base-substitution, frameshift, and sequence-substitution mutageneses. *Annual Review of Genetics* 36: 279-303.

Mayr, E. *What Evolution Is.* New York: Basic Books, 2001. ISBN 0465044255

Taggart, R., and C. Starr. 2006. *Biology The Unity and Diversity of Life.* Thompson Brooks/Cole

Bertram J (2000). "The molecular biology of cancer". *Mol. Aspects Med.* **21** (6): 167–223. doi:10.1016/S0098-2997(00)00007-8. PMID 11173079.

Aminetzach YT, Macpherson JM, Petrov DA (2005). "Pesticide resistance via transposition-mediated adaptive gene truncation in Drosophila". *Science* **309** (5735): 764–7. doi:10.1126/science.1112699. PMID 16051794.

Burrus V, Waldor M (2004). "Shaping bacterial genomes with integrative and conjugative elements". *Res. Microbiol.* **155** (5): 376–86. doi:10.1016/j.resmic.2004.01.012. PMID 15207870.

Sawyer SA, Parsch J, Zhang Z, Hartl DL (2007). "Prevalence of positive selection among nearly neutral amino acid replacements in Drosophila". *Proc. Natl. Acad. Sci. U.S.A.* **104** (16): 6504–10. doi:10.1073/pnas.0701572104. PMC 1871816. PMID 17409186.

UNIT 5: OUTCOMES OF EVOLUTION

CONTENTS

1.0 Introduction

Entirely, every aspect of the behaviour and form of organisms are influenced by evolution. Importantly, the most prominent are the specific behavioural and physical adaptations that are the outcome of natural selection. Certain activities are aided by these adaptations which increase fitness. Such activities include; finding food, avoiding predators or attracting mates. Organisms can also respond to selection by co-operating with each other, usually by aiding their relatives or engaging in mutually beneficial symbiosis. Evolution produces new species through splitting ancestral populations of organisms into new groups that cannot or will not interbreed in the longer term.

2.0 Objectives

Based on the theories of evolution, at the end of this study; the student should be able to discuss the outcomes of evolution and how these outcomes affect the changes in our environment, related to species existence.

3.0Main focus

The outcomes of evolution are sometimes divided into **macroevolution**. Macroevolution occurs at or above the level of species, such as extinction and speciation and **microevolution**, which refers to smaller evolutionary changes, such as adaptations, within a species or population. Generally, the outcome of long periods of microevolution is referred macroevolution. Hence, the distinction between micro- and macroevolution is not a fundamental one – the time involved is simply the difference. In macroevolution however, the traits of the entire species may be important. For example, a large amount of variation among individuals allows a species to rapidly adapt to new habitats, reducing the possibility of the species going extinct, while a wide geographic range increases the chance of speciation, by making it more likely that part of the population will become isolated. In this regard, microevolution and macroevolution might involve selection at different levels – microevolution acting on genes and organisms, against macroevolution processes which include species selection acting on entire species and affecting their rates of speciation and extinction.

It is often misconceived that evolution has goals or long-term plans; in the real sense of it however, evolution has no long-term goal and does not necessarily produce greater complexity. Inspite of the fact that complex species have evolved, they occur as a side effect of the overall number of organisms increasing and simple forms of life still remain more

common in the biosphere. A good example is that the overwhelming majority of species are microscopic prokaryotes, which form about half the world's biomass despite their small size and constitute the vast majority of Earth's biodiversity. Simple organisms therefore have been the dominant form of life on Earth throughout its history and continue to be the main form of life up to the present day, with complex life only appearing more diverse because it is more noticeable. Importantly, the evolution of microorganisms is particularly important to modern evolutionary research, since their rapid reproduction allows the study of experimental evolution and the observation of evolution and adaptation in real time.

In 1859 the English naturalist Charles Darwin published The Origin of Species. The book pointed at two major arguments: First, Charles Darwin presented a dearth of evidence of evolution. He said that all living things on earth today are the descendants of earlier species.

Second, he proposed a mechanism of natural selection to explain how evolution takes place. Evolution involves two interrelated phenomena; adaptation and speciation.

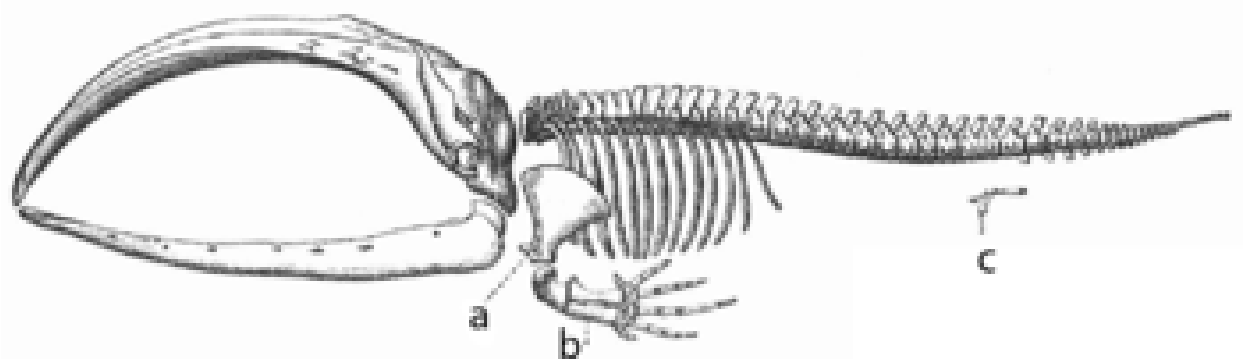
3.1 Adaptation

Over a period of time, species modify their phenotypes in certain ways that allow them to succeed in their environment. Adaptation is a process that ensures organisms are better suited to their habitat. Adaptation may also refer to a trait that is key to an organism's survival. A good example is the adaptation of horses' teeth to the grinding of grass. By using the term *adaptation* for the evolutionary process and *adaptive trait* for the product the two senses of the word may be distinguished. Adaptations are produced by natural selection. Theodosius Dobzhansky brought about the following definitions;

1. *Adaptation* is the evolutionary process whereby an organism becomes better able to live in its habitat or habitats.

2. *Adaptedness* is the state of being adapted: the degree to which an organism is able to live and reproduce in a given set of habitats.
3. An *adaptive trait* is an aspect of the developmental pattern of the organism which enables or enhances the probability of that organism surviving and reproducing.

Adaptation may cause either the loss of an ancestral feature or the gain of a new feature. An example that shows both types of change is bacterial adaptation to antibiotic selection, with genetic changes causing antibiotic resistance by either modifying the target of the drug, or increasing the activity of transporters that pump the drug out of the cell. A controversial but interesting idea is that some adaptations might increase the ability of organisms to generate genetic diversity and adapt by natural selection



A baleen whale skeleton, ***a*** and ***b*** label flipper bones, which were adapted from front leg bones: while ***c*** indicates vestigial leg bones, suggesting an adaptation from land to sea.

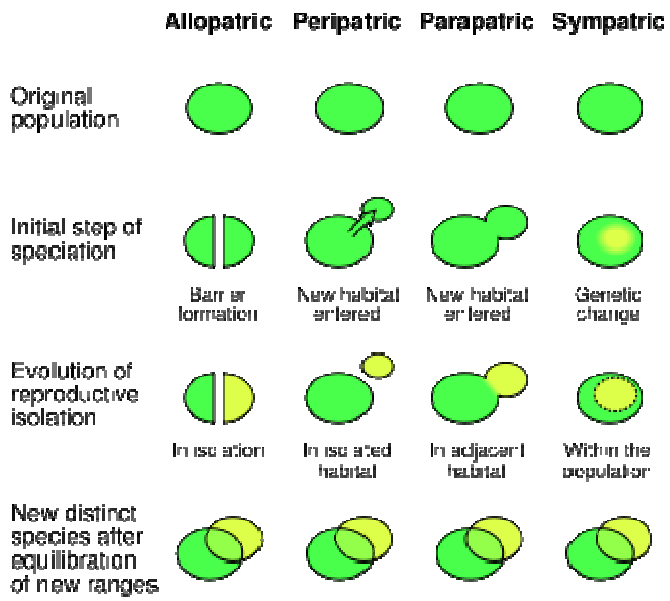
Adaptation occurs through the gradual modification of existing structures. Consequently, structures with similar internal organisation may have different functions in related organisms. This is the result of a single ancestral structure being adapted to function in different ways. The bones within bat wings, for example, are very similar to those in mice feet and primate hands, due to the descent of all these structures from a common mammalian ancestor. However, since all living organisms are related to some extent, even organs that appear to have little or no structural similarity, such as arthropod, squid and vertebrate eyes,

or the limbs and wings of arthropods and vertebrates, can depend on a common set of homologous genes that control their assembly and function; this is called deep homology.

During evolution, some structures may lose their original function and become vestigial structures. Such structures may have little or no function in a current species, yet have a clear function in ancestral species, or other closely related species. Examples include pseudogenes, the non-functional remains of eyes in blind cave-dwelling fish wings in flightless birds, and the presence of hip bones in whales and snakes. Examples of vestigial structures in humans include wisdom teeth, the coccyx, the vermiform appendix, and other behavioural vestiges such as goose bumps and primitive reflexes

An area of current investigation in evolutionary developmental biology is the developmental basis of adaptations and exaptations. This research looks at the origin and evolution of embryonic development and how modifications of development and developmental processes produce novel features. This study has shown that evolution can change a development to produce new structures, such as embryonic bone structures that develop into the jaw in other animals instead forming part of the middle ear in mammals. It is also possible for structures that have been lost in evolution to reappear due to changes in developmental genes, such as a mutation in chickens causing embryos to grow teeth similar to those of crocodiles. It is now becoming apparent that most alterations in the form of organisms are due to changes in a small set of conserved genes.

3.2 Speciation



The four Mechanisms of Speciation

When a species diverges into two or more descendant species, the process is called Speciation. In attempting to define what a species is, we find multiple ways of doing so. The choice of definition is dependent on the particularities of the species concerned. A good example is that some species concepts apply more readily toward sexually reproducing organisms while others lend themselves better toward asexual organisms. Although there are a variety of species concepts, these various concepts can be placed into one of three broad philosophical approaches: interbreeding, ecological and phylogenetic.

Barriers to reproduction between two diverging sexual populations are required for the populations to become new species. Gene flow may slow this process by spreading the new genetic variants also to the other populations. Depending on how far two species have diverged since their most recent common ancestor, it may still be possible for them to produce offspring, as with horses and donkeys mating to produce mules. Such hybrids are generally infertile. In this case, closely related species may regularly interbreed, but hybrids will be selected against and the species will remain distinct. However, viable hybrids are occasionally formed and these new species can either have properties intermediate between

their parent species, or possess a totally new phenotype. The importance of hybridisation in producing new species of animals is unclear, although cases have been seen in many types of animals, with the gray tree frog being a particularly well-studied example.

Speciation has been observed under controlled laboratory conditions and in nature. In sexually reproducing organisms, speciation results from reproductive isolation followed by genealogical divergence. There are four mechanisms for speciation. The most common in animals is allopatric speciation, which occurs in populations initially isolated geographically, such as by habitat fragmentation or migration. Selection under these conditions can produce very rapid changes in the appearance and behaviour of organisms. As selection and drift act independently on populations isolated from the rest of their species, separation may eventually produce organisms that cannot interbreed.

The second mechanism of speciation is peripatric speciation, which occurs when small populations of organisms become isolated in a new environment. This differs from allopatric speciation in that the isolated populations are numerically much smaller than the parental population. Here, the founder effect causes rapid speciation after an increase in inbreeding increases selection on homozygotes, leading to rapid genetic change.

The third mechanism of speciation is parapatric speciation. This is similar to peripatric speciation in that a small population enters a new habitat, but differs in that there is no physical separation between these two populations. Instead, speciation results from the evolution of mechanisms that reduce gene flow between the two populations. This occurs when there has been a drastic change in the environment within the parental species' habitat. One example is the grass *Anthoxanthum odoratum*, which undergoes parapatric speciation in response to localised metal pollution from mines. In this instance, plants that have resistance to high levels of metals in the soil evolve. Selection against interbreeding with the metal-

sensitive parental population produced a gradual change in the flowering time of the metal-resistant plants, which eventually produced complete reproductive isolation. Selection against hybrids between the two populations may cause *reinforcement*, which is the evolution of traits that promote mating within a species, as well as character displacement, which is when two species become more distinct in appearance.

Lastly, sympatric speciation is where species diverge without geographic isolation or changes in habitat. This is rare since even a small amount of gene flow may remove genetic differences between parts of a population. Generally, sympatric speciation in animals requires the evolution of both genetic differences and non-random mating, to allow reproductive isolation to evolve.

The cross-breeding of two related species to produce a new hybrid species is one type of sympatric speciation. This is not common in animals as animal hybrids are usually sterile. This is because during meiosis the homologous chromosomes from each parent are from different species and cannot successfully pair. However, it is more common in plants because plants often double their number of chromosomes, to form polyploids. This allows the chromosomes from each parental species to form matching pairs during meiosis, since each parent's chromosomes are represented by a pair already. An example of such a speciation event is when the plant species *Arabidopsis thaliana* and *Arabidopsis arenosa* cross-bred to give the new species *Arabidopsis suecica*. This happened about 20,000 years ago and the speciation process has been repeated in the laboratory, which allows the study of the genetic mechanisms involved in this process. Indeed, chromosome doubling within a species may be a common cause of reproductive isolation, as half the doubled chromosomes will be unmatched when breeding with undoubled organisms.

3.3 Extinction

The disappearance of an entire species is called Extinction . It is not an unusual event, however species regularly appear through speciation and disappear through extinction. Most animal and plant species that have lived on Earth millions of years ago are now extinct, and this appears to be the ultimate fate of all species. These extinctions have been continuous and have happened through the course of our history in the world, although the rate of extinction spikes in occasional mass extinction events. The Cretaceous–Tertiary extinction event, during which the non-avian dinosaurs went extinct is the most well-known, but the earlier approximately 96% of species driven to extinction during the Permian–Triassic extinction event was even more severe. The Holocene extinction event is an ongoing mass extinction associated with humanity's expansion across the globe over the past few thousand years. Present-day extinction rates are 100–1000 times greater than the background rate and up to 30% of species may be extinct by the mid 21st century. Human activities are now the primary cause of the ongoing extinction event; global warming may further accelerate it in the future.

The role of extinction in evolution is not very well understood and may depend on which type of extinction is considered. The major reasons for the continuous "low-level" extinction events, which form the majority of extinctions, may be the result of competition between species for limited resources. If one species can out-compete another, this could produce species selection, with the fitter species surviving and the other species being driven to extinction. The intermittent mass extinctions are also important, but instead of acting as a selective force, they drastically reduce diversity in a nonspecific manner and promote bursts of rapid evolution and speciation in survivors.

4.0 Conclusion

The student should be able to adequately discuss the outcomes of evolution and how it has shaped the existence of present day species in our world today. The student should also be able to elaborate on the major aspects that have determined the existence of species.

5.0 Summary

Evolutionary outcomes have shaped the way our world is today. As evolution has influenced and shaped organisms in terms of behaviour and form, natural selection has resulted in physical adaptations and specific behavioural patterns. The outcomes including adaptation, speciation and extinction have been key factors that have ensured the changes and survival of species.

6.0 Tutor-Marked Assignment

- What are the outcomes of Evolution
- What do you understand by Speciation and Adaptation
- What are the mechanisms in Speciation

7.0 Reference/Further Readings

Scott EC, Matzke NJ (2007). "Biological design in science classrooms". *Proc. Natl. Acad. Sci. U.S.A.*. 104 **1** (suppl_1): 8669–76

Hendry AP, Kinnison MT (2001). "An introduction to microevolution: rate, pattern, process". *Genetica* **112–113**: 1–8

Jablonski, D. (2000). "Micro- and macroevolution: scale and hierarchy in evolutionary biology and paleobiology". *Paleobiology* **26** (sp4): 15–52.

Mayr, Ernst 1982. *The growth of biological thought*. Harvard. p483: "Adaptation... could no longer be considered a static condition, a product of a creative past and became instead a continuing dynamic process."

Dobzhansky, T.; Hecht, MK; Steere, WC (1968). "On some fundamental concepts of evolutionary biology". *Evolutionary biology volume 2* (1st ed.). New York: Appleton-Century-Crofts. pp. 1–34.

Dobzhansky, T. (1956). "Genetics of natural populations XXV. Genetic changes in populations of *Drosophila pseudoobscura* and *Drosphila persimilis* in some locations in California". *Evolution* **10** (1): 82–92.

http://users.rcn.com/jkimball.ma.ultranet/BiologyPages/S/Speciation.html

http://anthro.palomar.edu/evolve/default.htm

MODULE 2: POPULATION GENETICS

Unit 1: History of Population Genetics

Unit 2: Processes of Population Genetics

Unit 3: Hardey-Weinberg Principle

Unit 4: Gene Frequency/Equilibrium

UNIT 1: HISTORY OF POPULATION GENETICS

Content

I.0 Introduction

2.0 Objectives

3.0 Population

3.1 History of Population Genetics

4.0 Conclusion

5.0 Summary

6.0 Tutor-Marked Assignment

7.0 Reference/Further Readings

Unit 1 History of Population Genetics

1.0 Introduction

The study of allele frequency, distribution and change under the influence of the four main evolutionary processes namely: natural selection, genetic drift, mutation and gene flow. It takes into account the factors of recombination, population subdivision and population structure. Population genetics attempts to explain adaptation and speciation.

The emergence of the modern evolutionary synthesis was due in part to Population genetics. Its primary founders were Sewall Wright, J. B. S. Haldane and R. A. Fisher, who also laid the foundations for the related discipline of quantitative genetics.

2.0 Objectives

At the end of this unit, students should be able to

- Define Population Genetics
- Describe the History of Population Genetics

3.0 Population Genetics

Population genetics is a field of biology that studies the genetic composition of biological populations and the observed changes in the genetic composition that result from the operation of various factors, including natural selection. Experts in population genetics pursue their goals by employing the following methods; development of mathematical models of gene frequency dynamics, extraction of conclusions from those models about the likely patterns of genetic variation in actual populations and testing the conclusions against empirical data. A number of the more robust generalizations to emerge from population-genetic analysis include;

Population genetics as being bound with the study of evolution and natural selection, which is often referred to as the theoretical cornerstone of modern Darwinism. This is as a result of the fact that natural selection is one of the most important factors that can affect a population's genetic composition. Natural selection occurs when some variants in a population out-reproduce other variants, as a result of being better adapted to the environment, or ‘fitter’. Presuming the fitness differences are at least partly due to genetic differences, this will cause the population's genetic makeup to be altered over time. While studying the formal models of gene frequency change, population genetics experts hope to shed light on the evolutionary process and permit the consequences of different evolutionary hypotheses to be explored in a quantitatively precise way.

In the 1920s and 1930s the field of population genetics came into light due to the work of R.A. Fisher, J.B.S. Haldane and Sewall Wright. Their achievement was to integrate the principles of Mendelian genetics, which had been rediscovered at the turn of century, with Darwinian natural selection. Many of the early Mendelians did not accept Darwin's ‘gradualist’ account of evolution, believing instead that novel adaptations must arise in a single mutational step; conversely, many of the early Darwinians did not believe in Mendelian inheritance, often because of the erroneous belief that it was incompatible with the process of evolutionary modification as described by Darwin. By working out mathematically the consequences of selection acting on a population obeying the Mendelian rules of inheritance, Fisher, Haldane and Wright showed that Darwinism and Mendelism were not just compatible but excellent bed fellows; this played a key part in the formation of the ‘neo-Darwinian synthesis’, and explains why population genetics came to occupy so pivotal a role in evolutionary theory.

Population genetics is the study of the frequency and interaction of alleles and genes in populations. A sexual population is a set of organisms in which any pair of members can breed together. This implies that all members belong to the same species and live near each other.

A good example is; all of the moths of the same species living in an isolated forest are a population. A gene in this population may have several alternate forms, which account for variations between the phenotypes of the organisms. An example might be a gene for coloration in moths that has two alleles: black and white. A gene pool is the complete set of alleles for a gene in a single population; the allele frequency for an allele is the fraction of the genes in the pool that is composed of that allele. Evolution occurs when there are changes in the frequencies of alleles within a population; for example, the allele for black colour in a population of moths becoming more common.

3.1 History of Population Genetics

Population genetics began as attempting to reconcile the Mendelian and biometrician models. A major step was the work of the British biologist and statistician R.A. Fisher. In a series of papers starting in 1918 and culminating in his 1930 book **The Genetical Theory of Natural Selection**. Fisher showed that the continuous variation measured by the biometricians could be produced by the combined action of many discrete genes, and that natural selection could change allele frequencies in a population, resulting in evolution. In 1924, a series of papers by J.B.S. Haldane worked out the mathematics of allele frequency change at a single gene locus under a broad range of conditions. Haldane also applied statistical analysis to real-world examples of natural selection, such as the evolution of industrial melanism in peppered moths, and showed that selection coefficients could be larger than Fisher assumed, leading to more rapid adaptive evolution.

Sewall Wright an American biologist, who had a background in animal breeding experiments, centred his research on combinations of interacting genes and the effects of inbreeding on small, relatively isolated populations that exhibited genetic drift. In 1932, Wright introduced the concept of an adaptive landscape and argued that genetic drift and inbreeding could drive a small, isolated sub-population away from an adaptive peak, allowing natural selection to drive it towards different adaptive peaks.

Population genetics as a discipline was based on the work of Fisher, Haldane and Wright. This integrated natural selection with Mendelian genetics, which was the critical first step in developing a unified theory of how evolution worked.

John Maynard Smith was Haldane's pupil, whilst W.D. Hamilton was heavily influenced by the writings of Fisher. The American George R. Price worked with both Hamilton and Maynard Smith. American Richard Lewontin and Japanese Motoo Kimura were heavily influenced by Wright.

Ordinary genetics in comparism with population genetics, looks at how one selects breeding stock to produce the best possible offspring. Population genetics looks at the statistical distribution of genes in a particular breeding population, such as a breed of dog, and how different kinds of selection can affect that gene distribution. Ordinary genetics is seen as predicting the phenotypic makeup of the next generation, while population genetics predicts the genetic makeup of the breed as a whole, often several generations away.

Population genetics is concerned with gene and genotype frequencies, the factors that tend to keep them constant, and the factors that tend to change them in populations. It is largely concerned with the study of polymorphisms. It directly impacts counselling, forensic medicine, and genetic screening.

The mathematics of population genetics was originally developed as the beginning of the modern evolutionary synthesis. According to Beatty (1986), population genetics defines the core of the modern synthesis. In the first few decades of the 20th century, most field naturalists continued to believe that Lamarckian and orthogenic mechanisms of evolution provided the best explanation for the complexity they observed in the living world. However, as the field of genetics continued to develop, those views became less tenable. During the modern evolutionary synthesis, these ideas were purged, and only evolutionary causes that could be expressed in the mathematical framework of population genetics were retained. Consensus was reached as to which evolutionary factors might influence evolution, but not as to the relative importance of the various factors.

Theodosius Dobzhansky, a postdoctoral worker in T. H. Morgan's lab, had been influenced by the work on genetic diversity by Russian geneticists such as Sergei Chetverikov. He helped to bridge the divide between the foundations of microevolution developed by the population geneticists and the patterns of macroevolution observed by field biologists, with his 1937 book Genetics and the Origin of Species. Dobzhansky examined the genetic diversity of wild populations and showed that, contrary to the assumptions of the population geneticists, these populations had large amounts of genetic diversity, with marked differences between sub-populations. The book also took the highly mathematical work of the population geneticists and put it into a more accessible form. Many more biologists were influenced by population genetics via Dobzhansky than were able to read the highly mathematical works in the original.

4.0 Conclusion

Students should be able to explain population genetics as we know it today. They should understand how the present theory arose from the need to reconcile Mendel with Darwin, a

need which became increasingly urgent as the empirical evidence for Mendelian inheritance began to pile up.

5.0 Summary

Population genetics is concerned with gene and genotype frequencies, the factors that tend to keep them constant, and the factors that tend to change them in populations.

6.0 Tutor-Marked Assignment

- What are the principles of population genetics
- Identify keys areas of biological and environmental conservation where a genetic approach benefits outcomes of management
- Critically review original research in population genetics

7.0 Reference Reading

Postlethwalt, John (2009). *Modern Biology*. Holt, Rinehart and Winston. pp. 317

Hartl, Daniel (2007). *Principles of Population Genetics*. Sinauer Associates. p. 95

Michael M. Desai, Daniel S. Fisher (2007). "Beneficial Mutation Selection Balance and the Effect of Linkage on Positive Selection". *Genetics* **176** (3): 1759–1798

Provine, W.B., 1971, *The Origins of Theoretical Population Genetics*, Chicago: University of Chicago Press.

http://bowlingsite.mcf.com/genetics/Genetics.html

http://www.uic.edu/classes/bms/bms655/index.html

UNIT 2: PROCESSES OF POPULATION GENETICS

Contents

1.0 Introduction

Population genetics is governed by four fundamental processes. Population genetics is intimately bound up with the study of evolution and natural selection, and is often regarded as the theoretical cornerstone of modern Darwinism. The four processes governing population genetics include; Natural selection, genetic drift, mutation and gene flow and transfer.

2.0 Objectives

- To explain classical transmission genetics and discuss this in the context of genes
- To review the overwhelming scientific evidence for biological evolution
- To explore how selection influences the genetic composition of a population

3.0 Natural Selection

Selection refers to changes in allele frequencies due to the effects of the gene on its host. Examples would be effects lowering or increasing the death rate of individuals carrying the gene, or lowering or increasing the number of its surviving offspring. Natural selection is the fact that some traits make it more likely for an organism to survive and reproduce. Population genetics describes natural selection by defining fitness as a propensity or probability of survival and reproduction in a particular environment. The fitness is normally given by the symbol **w**=1+**s** where **s** is the selection coefficient. Natural selection acts on phenotypes, or the observable characteristics of organisms, but the genetically heritable basis of any phenotype which gives a reproductive advantage will become more common in a population. In this way, natural selection converts differences in fitness into changes in allele frequency in a population over successive generations.

Prior to the advent of population genetics, many biologists doubted that small difference in fitness was sufficient to make a large difference to evolution. Population geneticists addressed this concern in part by comparing selection to genetic drift. Selection can overcome genetic drift when **s** is greater than 1 divided by the effective population size.

When this criterion is met, the probability that a new advantageous mutant becomes fixed is approximately equal to **s**. The time until fixation of such an allele depends little on genetic drift, and is approximately proportional to log(sN)/s.

3.1 Genetic Drift

Genetic drift is the result of probabilistic effects due to Mendelism or to the chance effects of mating and survival in a small population. A carrier of a particular allele may leave no surviving offspring for reasons which have nothing to do with that allele, for example accidental death. In general, the number of the surviving offspring of an individual can be thought of as a random variable, with a mean given by selection, but with still a positive probability of being zero. An allele that has a selective advantage over others may still be lost from the population due to random effects.

Genetic drift is referred to as a change in allele frequencies due to random sampling. That is, the alleles in the offspring are a random sample of those in the parents. Genetic drift may cause gene variants to disappear completely, and thereby reduce genetic variability. In contrast to natural selection, which makes gene variants more common or less common depending on their reproductive success, the changes due to genetic drift are not driven by environmental or adaptive pressures, and may be beneficial, neutral, or detrimental to reproductive success.

The effect of genetic drift is larger for alleles present in a smaller number of copies and smaller when an allele is present in many copies. Vigorous debates wage among scientists over the relative importance of genetic drift compared with natural selection.

Ronald Fisher held the view that genetic drift plays at the most a minor role in evolution, and this remained the dominant view for several decades.

In 1968 Motoo Kimura rekindled the debate with his neutral theory of molecular evolution which claims that most of the changes in the genetic material are caused by neutral mutations and genetic drift. The role of genetic drift by means of sampling error in evolution has been criticized by John H Gillespie and Will Provine, who argue that selection on linked sites is a more important stochastic force.

The population genetics of genetic drift are described using either branching processes or a diffusion equation describing changes in allele frequency. These approaches are usually applied to the Wright-Fisher and Moran models of population genetics. Assuming genetic drift is the only evolutionary force acting on an allele, after t generations in many replicated populations, starting with allele frequencies of p and q, the variance in allele frequency across those populations is

$$V_t \approx pq\left(1 - \exp\left\{-\frac{t}{2N_e}\right\}\right)$$

The *Wright-Fisher* model is an attempt to model these and similar effects. The Wright-Fisher model for dioecious populations assumes that the population is rigidly held at N_1 males land N_2 females over many generations. At the beginning of each generation, the population undergoes *random-mating* to produce a large number offspring. Of these, N_1 males and N_2 females are chosen at random to adulthood and replace the parents.

3.2 Mutation

Mutation is the ultimate source of genetic variation in the form of new alleles. Mutation can result in several different types of change in DNA sequences; these can either have no effect, alter the product of a gene, or prevent the gene from functioning. Studies in the fly *Drosophila melanogaster* suggest that if a mutation changes a protein produced by a gene,

this will probably be harmful, with about 70 percent of these mutations having damaging effects, and the remainder being either neutral or weakly beneficial.

Mutations can involve large sections of DNA becoming duplicated, usually through genetic recombination. These duplications are a major source of raw material for evolving new genes, with tens to hundreds of genes duplicated in animal genomes every million years. Most genes belong to larger families of genes of shared ancestry. Novel genes are produced by several methods, commonly through the duplication and mutation of an ancestral gene, or by recombining parts of different genes to form new combinations with new functions. Here, domains act as modules, each with a particular and independent function, that can be mixed together to produce genes encoding new proteins with novel properties. For example, the human eye uses four genes to make structures that sense light: three for color vision and one for night vision; all four arose from a single ancestral gene. Another advantage of duplicating a gene is that this increases redundancy; this allows one gene in the pair to acquire a new function while the other copy performs the original function. Other types of mutation occasionally create new genes from previously noncoding DNA.

In addition to being a major source of variation, mutation may also function as a mechanism of evolution when there are different probabilities at the molecular level for different mutations to occur, a process known as mutation bias. If two genotypes, for example one with the nucleotide G and another with the nucleotide A in the same position, have the same fitness, but mutation from G to A happens more often than mutation from A to G, then genotypes with A will tend to evolve. Different insertion vs. deletion mutation biases in different taxa can lead to the evolution of different genome sizes. Developmental or mutational biases have also been observed in morphological evolution. For example,

according to the phenotype-first theory of evolution, mutations can eventually cause the genetic assimilation of traits that were previously induced by the environment.

Mutation bias effects are superimposed on other processes. If selection would favor either one out of two mutations, but there is no extra advantage to having both, then the mutation that occurs the most frequently is the one that is most likely to become fixed in a population. Mutations leading to the loss of function of a gene are much more common than mutations that produce a new, fully functional gene. Most loss of function mutations are selected against. But when selection is weak, mutation bias towards loss of function can affect evolution. For example, pigments are no longer useful when animals live in the darkness of caves, and tend to be lost. This kind of loss of function can occur because of mutation bias, and/or because the function had a cost, and once the benefit of the function disappeared, natural selection leads to the loss. Loss of sporulation ability in a bacterium during laboratory evolution appears to have been caused by mutation bias, rather than natural selection against the cost of maintaining sporulation ability. When there is no selection for loss of function, the speed at which loss evolves depends more on the mutation rate than it does on the effective population size, indicating that it is driven more by mutation bias than by genetic drift.

3.3 Gene Flow and Transfer

Gene flow is the exchange of genes between populations, which are usually of the same species. Examples of gene flow within a species include the migration and then breeding of organisms, or the exchange of pollen. Gene transfer between species includes the formation of hybrid organisms and horizontal gene transfer.

Migration into or out of a population can change allele frequencies, as well as introducing genetic variation into a population. Immigration may add new genetic material to the established gene pool of a population. Conversely, emigration may remove genetic material.

3.4 Reproductive isolation

As barriers to reproduction between two diverging populations are required for the populations to become new species, gene flow may slow this process by spreading genetic differences between the populations. Gene flow is hindered by mountain ranges, oceans and deserts or even man-made structures such as the Great Wall of China, which has hindered the flow of plant genes.

Depending on how far two species have diverged since their most recent common ancestor, it may still be possible for them to produce offspring, as with horses and donkeys mating to produce mules. Such hybrids are generally infertile, due to the two different sets of chromosomes being unable to pair up during meiosis. In this case, closely related species may regularly interbreed, but hybrids will be selected against and the species will remain distinct. However, viable hybrids are occasionally formed and these new species can either have properties intermediate between their parent species, or possess a totally new phenotype. The importance of hybridization in creating new species of animals is unclear, although cases have been seen in many types of animals, with the gray tree frog being a particularly well-studied example.

Hybridization is, however, an important means of speciation in plants, since polyploidy is tolerated in plants more readily than in animals. Polyploidy is important in hybrids as it allows reproduction, with the two different sets of chromosomes each being able to pair with

an identical partner during meiosis. Polyploids also have more genetic diversity, which allows them to avoid inbreeding depression in small populations.

3.5 Genetic structure

Because of physical barriers to migration, along with limited tendency for individuals to move or spread, and tendency to remain or come back to natal place, natural populations rarely all interbreed as convenient in theoretical random models (Buston *et al.*, 2007). There is usually a geographic range within which individuals are more closely related to one another than those randomly selected from the general population. This is described as the extent to which a population is genetically structured (Repaci *et al.*, 2007). Genetic structuring can be caused by migration due to historical climate change, species range expansion or current availability of habitat.

3.6 Horizontal Gene Transfer

Horizontal gene transfer is the transfer of genetic material from one organism to another organism that is not its offspring; this is most common among bacteria. In medicine, this contributes to the spread of antibiotic resistance, as when one bacteria acquires resistance genes it can rapidly transfer them to other species. Horizontal transfer of genes from bacteria to eukaryotes such as the yeast *Saccharomyces cerevisiae* and the adzuki bean beetle *Callosobruchus chinensis* may also have occurred. An example of larger-scale transfers are the eukaryotic bdelloid rotifers, which appear to have received a range of genes from bacteria, fungi, and plants. Viruses can also carry DNA between organisms, allowing transfer of genes even across biological domains. Large-scale gene transfer has also occurred between the ancestors of eukaryotic cells and prokaryotes, during the acquisition of chloroplasts and mitochondria.

3.7 Complications in Population Genetics

Basic models of population genetics consider only one gene locus at a time. In practice, epistatic and linkage relationships between loci may also be important.

Epistasis

Because of epistasis, the phenotypic effect of an allele at one locus may depend on which alleles are present at many other loci. Selection does not act on a single locus, but on a phenotype that arises through development from a complete genotype.

According to Lewontin (1974), the theoretical task for population genetics is a process in two spaces: a "genotypic space" and a "phenotypic space". The challenge of a *complete* theory of population genetics is to provide a set of laws that predictably map a population of genotypes (G_1) to a phenotype space (P_1), where selection takes place, and another set of laws that map the resulting population (P_2) back to genotype space (G_2) where Mendelian genetics can predict the next generation of genotypes, thus completing the cycle. Even leaving aside for the moment the non-Mendelian aspects of molecular genetics, this is clearly a gargantuan task. Visualizing this transformation schematically:

$$G_1 \xrightarrow{T_1} P_1 \xrightarrow{T_2} P_2 \xrightarrow{T_3} G_2 \xrightarrow{T_4} G_1' \rightarrow \cdots$$

(Adapted from Lewontin 1974, p. 12). XD

T_1 represents the genetic and epigenetic laws, the aspects of functional biology, or development, that transform a genotype into phenotype. We will refer to this as the "genotype-phenotype map". T_2 is the transformation due to natural selection, T_3 are epigenetic relations that predict genotypes based on the selected phenotypes and finally T_4 the rules of Mendelian genetics.

In practice, there are two bodies of evolutionary theory that exist in parallel, traditional population genetics operating in the genotype space and the biometric theory used in plant and animal breeding, operating in phenotype space. The missing part is the mapping between the genotype and phenotype space. This leads to a "sleight of hand" (as Lewontin terms it) whereby variables in the equations of one domain, are considered parameters or *constants*, where, in a full-treatment they would be transformed themselves by the evolutionary process and are in reality *functions* of the state variables in the other domain. The "sleight of hand" is assuming that we know this mapping. Proceeding as if we do understand it is enough to analyze many cases of interest. For example, if the phenotype is almost one-to-one with genotype (sickle-cell disease) or the time-scale is sufficiently short, the "constants" can be treated as such; however, there are many situations where it is inaccurate.

Linkage

If all genes are in linkage equilibrium, the effect of an allele at one locus can be averaged across the gene pool at other loci. In reality, one allele is frequently found in linkage disequilibrium with genes at other loci, especially with genes located nearby on the same chromosome. Recombination breaks up this linkage disequilibrium too slowly to avoid genetic hitchhiking, where an allele at one locus rises to high frequency because it is linked to an allele under selection at a nearby locus. This is a problem for population genetic models that treat one gene locus at a time. It can, however, be exploited as a method for detecting the action of natural selection via selective sweeps.

In the extreme case of primarily asexual populations, linkage is complete and different population genetic equations can be derived and solved, which behave quite differently to the sexual case. Most microbes, such as bacteria, are asexual. The population genetics of microorganisms lays the foundations for tracking the origin and evolution of antibiotic

resistance and deadly infectious pathogens. Population genetics of microorganisms is also an essential factor for devising strategies for the conservation and better utilization of beneficial microbes (Xu, 2010).

4.0 Conclusion

At the conclusion of this course, students should be able to; Define population genetics and applications to other disciplines, Review basic genetic background including genes and alleles, discuss the four processes in population genetics.

5.0 Summary

Population genetics is a field of biology that studies the genetic composition of biological populations, and the changes in genetic composition that result from the operation of various factors, including natural selection. Population geneticists usually define 'evolution' as any change in a population's genetic composition over time. The four factors that can bring about such a change are: natural selection, mutation, random genetic drift, and migration into or out of the population.

6.0 Tutor-Marked Assignment

- How do genes act in a population
- What is population genetics
- What are the processes governing population genetics

7.0 Reference/Further Reading

Postlethwalt, John (2009). *Modern Biology*. Holt, Rinehart and Winston. pp. 317.

Hartl, Daniel (2007). *Principles of Population Genetics*. Sinauer Associates. p. 95

Ewens W.J. (2004). *Mathematical Population Genetics (2nd Edition).* Springer-Verlag, New York.

Futuyma, Douglas (1998). *Evolutionary Biology*. Sinauer Associates. p. Glossary.

BS Haldane (1927). "A Mathematical Theory of Natural and Artificial Selection, Part V: Selection and Mutation". *Mathematical Proceedings of the Cambridge Philosophical Society* **23**: 838-844.

Hermisson J, Pennings PS (2005). "Soft sweeps: molecular population genetics of adaptation from standing genetic variation". *Genetics* **169**: 2335-2352

http://bowlingsite.mcf.com/genetics/Genetics.html

Haldane, J.B.S., 1930-1932, 'A Mathematical Theory of Natural and Artificial Selection', *Proceedings of the Cambridge Philosophical Society*, 26-28, Parts I-IX

http://www.uic.edu/classes/bms/bms655/index.html

UNIT 3: HARDY-WEINBERG PRINCIPLE

CONTENTS

Unit 3 Hardy-Weinberg Principle

1.0 Introduction

Evolution is not only the development of new species from older ones, as most people assume. It is also the minor changes within a species from generation to generation over long periods of time that can result in the gradual transition to new species. Evolution has been defined as the sum total of the genetically inherited changes in the individuals who are the members of a population's gene pool. It is clear that the effects of evolution are felt by individuals, but it is the population as a whole that actually evolves.

2.0 Objectives

At the end of this study, students should be able to:

- Discuss the importance of the Hardy-Weinberg Principle
- Apply the Hardy-Weinberg Law in analyzing population genetics for gene frequency, sex linkage, equilibrium, and heterozygote frequency.

3.0 Modern Theories of Evolution

Evolution is simply a change in frequencies of alleles in the gene pool of a population. For instance, let us assume that there is a trait that is determined by the inheritance of a gene with two alleles--B and b. If the parent generation has 92% B and 8% b and their offspring collectively have 90% B and 10% b, evolution has occurred between the generations. The entire population's gene pool has evolved in the direction of a higher frequency of the b allele--it was not just those individuals who inherited the b allele who evolved. This definition of evolution was developed largely as a result of independent work in the early 20th century by **Godfrey Hardy**, an English mathematician, and **Wilhelm Weinberg**, a German physician. Through mathematical modelling based on probability, they concluded in 1908 that gene pool frequencies are inherently stable but that evolution should be expected in all populations virtually all of the time. They resolved this apparent paradox by analyzing the net effects of potential evolutionary mechanisms.

Godfrey Hardy(1877-194)

Wilhelm Weinberg
(1862-1937)

Hardy, Weinberg, and the population geneticists who followed them came to understand that evolution will not occur in a population if seven conditions are met:

- mutation is not occurring
- natural selection is not occurring
- the population is infinitely large
- all members of the population breed
- all mating is totally random
- everyone produces the same number of offspring
- there is no migration in or out of the population

These conditions are the absence of the things that can cause evolution. In other words, if no mechanisms of evolution are acting on a population, evolution will not occur--the gene pool frequencies will remain unchanged. However, since it is highly unlikely that any of these seven conditions, let alone all of them, will happen in the real world, evolution is the inevitable result.

Godfrey Hardy and Wilhelm Weinberg went on to develop a simple equation that can be used to discover the probable genotype frequencies in a population and to track their changes from one generation to another. This has become known as the Hardy-Weinberg equilibrium

equation. In this equation ($p^2 + 2pq + q^2 = 1$), p is defined as the frequency of the dominant allele and q as the frequency of the recessive allele for a trait controlled by a pair of alleles (A and a). In other words, p equals all of the alleles in individuals who are homozygous dominant (AA) and half of the alleles in people who are heterozygous (Aa) for this trait in a population. In mathematical terms, this is:

$$p = AA + \frac{1}{2}Aa$$

Likewise, q equals all of the alleles in individuals who are homozygous recessive (aa) and the other half of the alleles in people who are heterozygous (Aa).

$$q = aa + \frac{1}{2}Aa$$

Because there are only two alleles in this case, the frequency of one plus the frequency of the other must equal 100%, which is to say

$$p + q = 1$$

Since this is logically true, then the following must also be correct:

$$p = 1 - q$$

There were only a few short steps from this knowledge for Hardy and Weinberg to realize that the chances of all possible combinations of alleles occurring randomly is

$$(p + q)^2 = 1$$

or more simply

$$p^2 + 2pq + q^2 = 1$$

In this equation, p^2 is the predicted frequency of homozygous dominant (AA) people in a population, $2pq$ is the predicted frequency of heterozygous (Aa) people, and q^2 is the predicted frequency of homozygous recessive (aa) ones.

From observations of phenotypes, it is usually only possible to know the frequency of homozygous recessive people, or q^2 in the equation, since they will not have the dominant trait. Those who express the trait in their phenotype could be either homozygous dominant (p^2) or heterozygous ($2pq$). The Hardy-Weinberg equation allows us to predict which ones they are. Since $p = 1 - q$ and q is known, it is possible to calculate p as well. Knowing p and q, it is a simple matter to plug these values into the Hardy-Weinberg equation ($p^2 + 2pq + q^2 = 1$). This then provides the predicted frequencies of all three genotypes for the selected trait within the population. By comparing genotype frequencies from the next generation with those of the current generation in a population, one can also learn whether or not evolution has occurred and in what direction and rate for the selected trait. However, the Hardy-Weinberg equation cannot determine which of the various possible causes of evolution were responsible for the changes in gene pool frequencies.

3.1 Examples of the Hardy-Weinberg Principle

The assumptions of the Hardy-Weinberg principle make it easy to calculate the genotype frequencies for a gene with two alleles (*A* and *a*). The frequency of homozygous genotype *AA* is the probability of one allele *A* being in combination with another allele *A*. The expected frequency is simply the product of the separate allele frequencies. We will use the term *p* to refer to the frequency of allele *A*:

Frequency of *AA* = p^2 (Homozygote for *A*) **Eqn 1**

The frequency of heterozygous genotype *Aa* is the probability of allele *A* being in combination with allele *a*. Note that there are two possible ways to get those combinations -- *A* from Dad and *a* from Mom, or vice versa (See the figure below).

Frequency of *Aa* = 2pq (Heterozygote) **Eqn 2**

The frequency of homozygous genotype *aa* is the probability of one allele *a* in combination with another allele *a*.

Frequency of $aa = q^2$ (Homozygote for *a*) **Eqn 3**

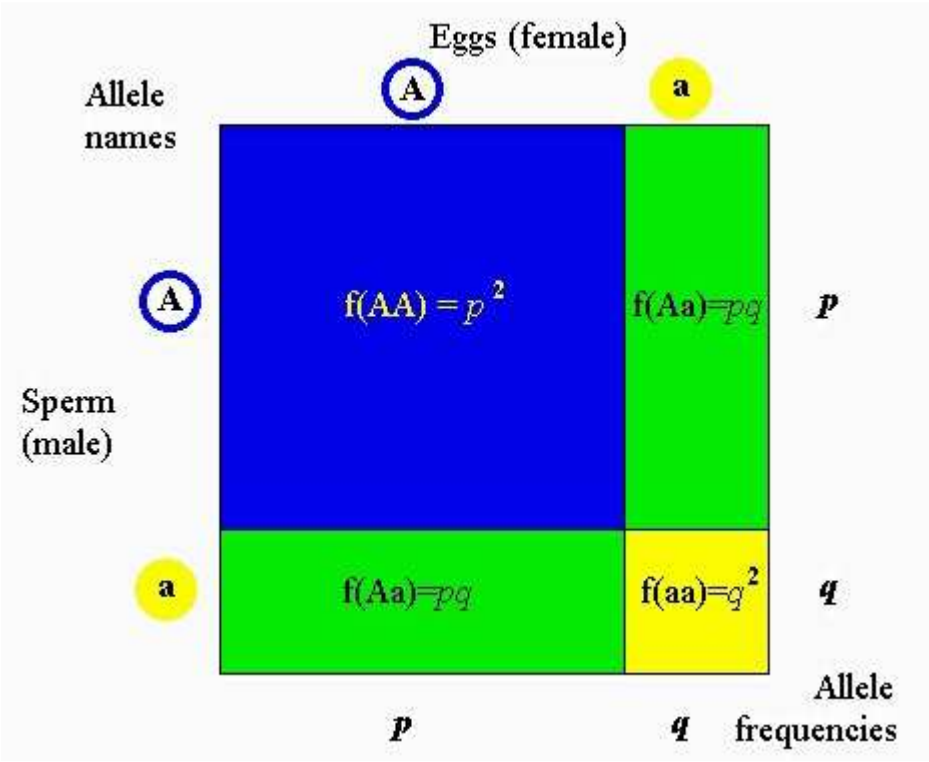


Diagram of Hardy-Weinberg genotype proportions from male (sperm) and female (egg) contributions. Given a locus with two alleles designated *A* and *a* that occur with frequencies *p* and *q*, the chart shows the genotype frequencies (p^2, $2pq$, and q^2) as differently colored areas. Note that the heterozygotes (blue + yellow = green) can be formed in two different ways.

Example 1 -- calculation of expected genotype frequencies from gene (allele) frequencies:

If p = 0.75 and q = 0.25 we can use Eqns 1, 2 and 3 to calculate the expected genotype frequencies.

$AA = p^2 = 0.75 * 0.75 = 0.5625$

$Aa = 2pq = 2 * 0.75 * 0.25 = 0.375$

$aa = q^2 = 0.25 * .025 = 0.0625$ **Eqns 4.**

The values we have just calculated are **EXPECTED** genotype frequencies **IF** the Hardy-Weinberg assumptions hold. We now turn to how we could check that from actual OBSERVED genotypic data (such as microsatellite data for Wyoming black bears). In order to calculate allele frequencies all we need are the observed genotype frequencies.

$p = p^2 + (2pq/2)$ and $q = q^2 + (2pq/2)$ **Eqn 5**

3.2 Derivation of the Hardy-Weinberg Principle

We can describe empirically and algebraically how genotype frequencies in one generation are related to genotype frequencies in the next. Let's explore that a bit further. To do so we're going to use a technique that is broadly useful in population genetics, i.e., we're going to construct a mating table. A mating table consists of three components:

1. A list of all possible genotype pairings.
2. The frequency with which each genotype pairing occurs.
3. The genotypes produced by each pairing.

		Offspring genotype		
Mating	**Frequency**	A_1A_1	A_1A_2	A_2A_2
A_1A_1 x A_1A_1	x_{11}^2	1	0	0
A_1A_2	$x_{11}x_{12}$	½	½	0
A_2A_2	$x_{11}x_{22}$	0	1	0
A_1A_2 x A_1A_1	$x_{12}x_{11}$	½	½	0
A_1A_2	x_{12}^2	¼	½	¼
A_2A_2	$x_{12}x_{22}$		½	1/2
A_2A_2 x A_1A_1	$x_{22}x_{11}$	0	1	0
A_1A_2	$x_{22}x_{12}$	0	½	½
A_2A_2	x_{22}^2	0	0	1

In constructing this table we've already made three assumptions about the transmission of genetic variation from one generation to the next:

$2p_1 = 2(50)/200 = 0:5$, $p_2 = (2(25) + 50)/200 = 0:5$.

Assumption 1 Genotype frequencies are the same in males and females, e.g., x_{11} is the frequency of the A_1A_1 genotype in both males and females.

Assumption 2 Genotypes mate at random with respect to their genotype at this particular locus.

Assumption 3 Meiosis is fair. More specifically, we assume that there is no segregation distortion; no gamete competition; no differences in the developmental ability of eggs, or the fertilization ability of sperm.

Now that we have this table we can use it to calculate the frequency of each genotype in newly formed zygotes in the population,5 provided that we're willing to make three additional assumptions:

Assumption 4 There is no input of new genetic material, i.e., gametes are produced without mutation, and all offspring are produced from the union of gametes within this population, i.e., no migration from outside the population.

Assumption 5 The population is of infinite size so that the actual frequency of mating is equal to their expected frequency and the actual frequency of offspring from each mating is equal to the Mendelian expectations.

Assumption 6 All matings produce the same number of offspring, on average.

Taking these three assumptions together allows us to conclude that the frequency of a particular genotype in the pool of newly formed zygotes is

$\sum$ (frequency of mating) (frequency of genotype produce from mating)

So freq. (A1A1 in zygotes) = $x^2_{11} + 1/2x_{11}\,x_{12} + 1/2x_{12}\,x_{11} + 1/4x^2_{12}$

$$= x^2_{11} + x_{11}x_{12} + 1/4x^2_{12}$$

$= (x_{11} + x_{12}/2)^2$

$= p^2$

freq. (A1A2 in zygotes) = 2pq

freq. (A_2A_2 in zygotes) = q^2

In order to say that these proportions will also be the genotype proportions of adults in the progeny generation, we have to make two more assumptions:

Assumption 7 Generations do not overlap.

Assumption 8 There are no differences among genotypes in the probability of survival.

4.0 Conclusion

In this unit the student learnt:

- An understanding of evolution depends upon knowledge of population genetics.
- that both allele and genotype frequencies in a population remain constant—that is, they are in equilibrium—from generation to generation unless specific disturbing influences are introduced.
- Genetic equilibrium is an ideal state that provides a baseline against which to measure change.
- Static allele frequencies in a population across generations assume: no mutation, no migration or emigration, infinitely large population size, and no selective pressure for or against any genotypes

5.0 Summary

Probably the most important basic concept in population genetics is the Hardy-Weinberg principle. It provides an expectation for genotypic patterns in populations. Deviations from the predicted pattern can provide very important insights into processes of genetic and

evolutionary change. The Hardy-Weinberg principle is a model that relates allele frequencies to genotype frequencies. Like most models, Hardy-Weinberg is a simplification of real world complexities.

6.0Tutor-Marked Assignment

- What is the Hardy-Weinberg Principle?
- What is the significance of this principle?
- What are the deviations from the Hardy-Weinberg Principle?

7.0 Reference Reading

Crow, Jf (Jul 1999). "Hardy, Weinberg and language impediments". *Genetics* **152** (3): 821–5

Hardy, Gh (Jul 1908). "MENDELIAN PROPORTIONS IN A MIXED POPULATION". *Science* **28** (706): 49–50

Modern theories of Evolution : Hardy-Weinberg equilibrium model. http://anthro.palomar.edu/synthetic/Default.htm

R. Ceppellini, M. Siniscalco, and C. A. B. Smith. The estimation of gene frequencies in a random-mating population. Annals of Human Genetics, 20:97{115, 1955.

A. P. Dempster, N. M. Laird, and D. B. Rubin. Maximum likelihood from incomplete data. Journal of the Royal Statistical Society Series B, 39:1-38, 1977.

A. E. Gelfand and A. F. M. Smith. Sampling-based approaches to calculating marginal densities. Journal of the American Statististical Association, 85:398-409, 1990.

UNIT 4: GENE FREQUENCY/EQUILIBRIUM

Contents

Unit 4 Gene Frequency/Equilibrium

1.0 Introduction

Gene frequency measures the frequency in the population of a particular gene relative to other genes at its locus. It is expressed as a proportion (between 0 and 1) or percentage (between 0 and 100%). A genetic equilibrium is at hand for an allele in a gene pool when the frequency of that allele is not changing. For this to be the case, evolutionary forces acting upon the allele must be equal and opposite. The only basic requirement is that the population be large enough that the effects of genetic drift are minimized.

2.0 Objectives

At the end of this course, students should be able to:

- Define genetic frequency
- Significance of genetic frequency
- Relate genetic frequency and the Hardy-Weinberg Principle

3.0 Genetic frequency

Genetic or Allele frequency is the proportion of all copies of a gene that is made up of a particular gene variant. In other words, it is the number of copies of a particular allele divided by the number of copies of all alleles at the genetic place in a population. It can be expressed for example as a percentage. In population genetics, allele frequencies are used to depict the amount of genetic diversity at the individual, population, and species level. It is also the relative proportion of all alleles of a gene that are of a designated type.

Given the following:

1. a particular locus on a chromosome and the gene occupying that locus

2. a population of *N* individuals carrying *n* loci in each of their somatic cells (e.g. two loci in the cells of diploid species, which contain two sets of chromosomes)
3. different alleles of the gene exist
4. one allele exists in *a* copies

then the allele frequency is the fraction or percentage of all the occurrences of that locus that is occupied by a given allele and the frequency of one of the alleles is *a/(n*N)*.For example, if the frequency of an allele is 20% in a given population, then among population members, one in five chromosomes will carry that allele. Four out of five will be occupied by other variant(s) of the gene. Note that for diploid genes the fraction of *individuals* that carry this allele may be nearly *two in five* (36%). The reason for this is that if the allele distributes randomly, then the binomial theorem will apply: 32% of the population will be heterozygous for the allele (i.e. carry one copy of that allele and one copy of another in each somatic cell) and 4% will be homozygous. Together, this means that 36% of diploid individuals would be expected to carry an allele that has a frequency of 20%. However, alleles distribute randomly only under certain assumptions, including the absence of selection. When these conditions apply, a population is said to be in Hardy–Weinberg equilibrium. The frequencies of all the alleles of a given gene often are graphed together as an *allele frequency distribution histogram*, or *allele frequency spectrum*. Population genetics studies the different "forces" that might lead to changes in the distribution and frequencies of alleles—in other words, to evolution. Besides selection, these forces include genetic drift, mutation and migration.

3.1 Calculation of Genetic Frequency

If *f*(*AA*), *f*(*Aa*), and *f*(*aa*) are the frequencies of the three genotypes at a locus with two alleles, then the frequency *p* of the A-allele and the frequency *q* of the a-allele are obtained by counting alleles. Because each homozygote AA consists only of A-alleles, and because half

of the alleles of each heterozygote Aa are A-alleles, the total frequency *p* of A-alleles in the population is calculated as

$$p = f(\mathbf{AA}) + \frac{1}{2}f(\mathbf{Aa}) = \text{frequency of A}$$

Similarly, the frequency *q* of the a allele is given by

$$q = f(\mathbf{aa}) + \frac{1}{2}f(\mathbf{Aa}) = \text{frequency of a}$$

It would be expected that *p* and *q* sum to 1, since they are the frequencies of the only two alleles present. Indeed they do:

$$p + q = f(\mathbf{AA}) + f(\mathbf{aa}) + f(\mathbf{Aa}) = 1$$

and from this we get:

$q = 1 - p$ and $p = 1 - q$

If there are more than two different allelic forms, the frequency for each allele is simply the frequency of its homozygote plus half the sum of the frequencies for all the heterozygotes in which it appears. Allele frequency can always be calculated from genotype frequency, whereas the reverse requires that the Hardy–Weinberg conditions of random mating apply. This is partly due to the *three* genotype frequencies and the *two* allele frequencies. It is easier to reduce from three to two.

In the simplest case, gene frequency is measured by counting the frequencies of each gene in the population. If a genotype contains two genes, then there are a total of 16 genes per locus in a population of eight individuals:

Aa AA aa aa AA Aa AA Aa

In the population above,

Frequency of A = 9/16 = 0.5625

frequency of a = 7/16 = 0.4375.

Algebraically, we can define p as the frequency of A and q as the frequency of a. p and q are always called 'gene' frequencies, but in a strict sense they are allele frequencies: they are the frequencies of the different alleles at one genetic locus.

The gene frequencies can be calculated from the genotype frequencies (P, Q, R):

p = P + 1/2Q

q = R + 1/2Q

(and p + q = 1). The calculation of gene from genotype frequencies is highly important. Although the gene frequencies can be calculated from the genotype frequencies, the opposite is not true: the genotype frequencies cannot be calculated from the gene frequencies (p , q).

3.2 The effect of mutation

Let ú be the mutation rate from allele *A* to some other allele *a* (the probability that a copy of gene *A* will become *a* during the DNA replication preceding meiosis). If p_t is the frequency of the *A* allele in generation *t*, then $q_t = 1 - p_t$ is the frequency of the *a* allele in generation *t*, and if there are no other causes of gene frequency change (no natural selection, for example), then the change in allele frequency in one generation is

$$\Delta p = p_t - p_{t-1} = (p_{t-1} - úp_{t-1}) - p_{t-1} = -úp_{t-1}$$

where p_{t-1} is the frequency of the preceding generation. This tells us that the frequency of *A* decreases (and the frequency of *a* increases) by an amount that is proportional to the mutation rate ú and to the proportion *p* of all the genes that are still available to mutate. Thus Δp gets smaller as the frequency of *p* itself decreases, because there are fewer and fewer *A* alleles to mutate into *a* alleles. We can make an approximation that, after *n* generations of mutation,

$$p_n = p_0 e^{-n\acute{u}}$$

3.3 Population Genetics and the Hardy-Weinberg Law

A **genetic equilibrium** is at hand for an allele in a gene pool when the frequency of that allele is not changing (i.e. when it is not evolving). For this to be the case, evolutionary forces acting upon the allele must be equal and opposite. The only basic requirement is that the population be large enough that the effects of genetic drift are minimized.

The Hardy-Weinberg formulas allow scientists to determine whether evolution has occurred. Any changes in the gene frequencies in the population over time can be detected. The law essentially states that if no evolution is occurring, then an equilibrium of allele frequencies will remain in effect in each succeeding generation of sexually reproducing individuals. In order for equilibrium to remain in effect (i.e. that no evolution is occurring) then the following five conditions must be met:

1. No mutations must occur so that new alleles do not enter the population.
2. No gene flow can occur (i.e. no migration of individuals into, or out of, the population).
3. Random mating must occur (i.e. individuals must pair by chance)
4. The population must be large so that no genetic drift (random chance) can cause the allele frequencies to change.
5. No selection can occur so that certain alleles are not selected for, or against.

Obviously, the Hardy-Weinberg equilibrium cannot exist in real life. Some or all of these types of forces all act on living populations at various times and evolution at some level occurs in all living organisms. The Hardy-Weinberg formulas allow us to detect some allele frequencies that change from generation to generation, thus allowing a simplified method of determining that evolution is occurring. There are two formulas that must be memorized:

$p^2 + 2pq + q^2 = 1$ *and* $p + q = 1$

p = frequency of the dominant allele in the population

q = frequency of the recessive allele in the population

p^2 = percentage of homozygous dominant individuals

q^2 = percentage of homozygous recessive individuals

2pq = percentage of heterozygous individuals

Despite the fact that evolution is a common occurrence in natural populations, allele frequencies will remain unaltered indefinitely unless evolutionary mechanisms such as mutation and natural selection cause them to change. Before Hardy and Weinberg, it was thought that dominant alleles must, over time, inevitably swamp recessive alleles out of existence. This incorrect theory was called "genophagy" (literally "gene eating"). According to this wrong idea, dominant alleles always increase in frequency from generation to generation. Hardy and Weinberg were able to demonstrate with their equation that dominant alleles can just as easily decrease in frequency.

4.0 Conclusion

At the end of this study, the student has learnt:

- Gene frequency measures the frequency in the population of a particular gene relative to other genes at its locus.
- The significance of genetic frequency to the process of evolution
- The relationship between gene frequency and the Hardy-Weinberg Principle

5.0 Summary

A population is a group of individuals of the same species in a given area whose members can interbreed. Because the individuals of a population can interbreed, they share a common group of genes known as the gene pool. Each gene pool contains all the alleles for all the traits of all the population. For evolution to occur in real populations, some of the gene frequencies must change with time. The gene frequency of an allele is the number of times an allele for a particular trait occurs compared to the total number of alleles for that trait. The Hardy-Weinberg principle is a model that relates genetic frequencies to genotype frequencies and the possible range for an allele frequency or genotype frequency therefore lies between zero and one,

Gene frequency = the number of a specific type of allele / the total number of alleles in the gene pool. An important way of discovering why real populations change with time is to construct a model of a population that does not change. This is just what Hardy and Weinberg did. Their principle describes a hypothetical situation in which there is no change in the gene pool (frequencies of alleles), hence no evolution.

6.0 Tutor-Marked Assignment

- What do you understand by the term ‘Genetic Frequency’?
- What is the importance of genetic frequency?

- What is the relationship between genetic frequency and the Hardy-Weinberg Principle?

7.0 Reference Reading

Cheung, KH; Osier MV, Kidd JR, Pakstis AJ, Miller PL, Kidd KK (2000). "ALFRED: an allele frequency database for diverse populations and DNA polymorphisms". *Nucleic Acids Research* **28** (1): 361–3.

Crow, Jf (Jul 1999). "Hardy, Weinberg and language impediments". *Genetics* **152** (3): 821–5

Emigh, T.H. (1980). "A comparison of tests for Hardy–Weinberg equilibrium". *Biometrics* **36** (4): 627–642

Guo, Sw; Thompson, Ea (Jun 1992). "Performing the exact test of Hardy-Weinberg proportion for multiple alleles". *Biometrics* (Biometrics, Vol. 48, No. 2) **48** (2): 361–72

Middleton, D; Menchaca L, Rood H, Komerofsky R (2002). "New allele frequency database: http://www.allelefrequencies.net". *Tissue Antigens* **61** (5): 403–7.

Allele Frequency http://www.ehstrafd.org/

Population Gene Frequencies. http://www.animalgenome.org/edu/genetics/main.html

Evolution. http://www.blackwellpublishing.com/ridley/

MODULE 3: POLYMORPHISM

Unit 1: Ecology

Unit 2: Introduction to Polymorphism

Unit 3: Examples of Polymorphism

Unit 1 Ecology

Contents

3.6.2 Social Ecology

3.6.3 C0evolution

3.6.4 Biogeography

3.6.5 r/K-Selection Theory

3.6.6 Molecular Ecology

3.6.7 Human Ecology

4.0 Conclusion

5.0 Summary

6.0 Tutor-Marked Assignment

7.0 Reference/Further Readings

Unit 1: Ecology

1.0 Introduction

In biology, polymorphism takes place when two or more different phenotypes exist in the same population of a species. This means the occurrence of more than one morph or form. In order to be regarded as such, morphs must occupy the same habitat at the same time and belong to a panmictic population, which refers to a population with random mating.

Polymorphism is related to biodiversity, genetic variation and adaptation and is common in nature. It functions to retain variety of form in a population living in a varied environment.

The most common example is sexual dimorphism, which occurs in many organisms. Other examples are mimetic forms of butterflies, and human haemoglobin and blood types. Polymorphism results from evolutionary processes, as does any aspect of a species. It is heritable and is modified by natural selection.

2.0 Objectives

At the end of this study, students should be able to;

- Define the Ecology and discuss its significance to Evolution
- Differentiate the types of Ecology
- Discuss Ecological Models

3.0 The Science of Ecology

The term “ecology” was coined by the German zoologist, Ernst Haeckel, in 1866 to describe the “economies” of living forms. The theoretical practice of ecology consists, by and large, of the construction of models of the interaction of living systems with their environment that include other living systems. These models are then tested in the laboratory and the field. Theory in ecology consists of the heuristics—or principles—used to construct models. Unlike evolutionary theory, ecology has no generally accepted global principles such as Mendel's and other rules of genetic inheritance. Contemporary ecology consists of a patchwork of sub-disciplines including population ecology, community ecology, conservation ecology, ecosystem ecology, metapopulation ecology, metacommunity ecology, spatial ecology, landscape ecology, physiological ecology, evolutionary ecology, functional ecology, and behavioral ecology. What is common to all these fields is the view that:

(i) different biota interact in ways that can be described with sufficient precision and generality to permit their scientific study;

(ii) (ii) ecological interactions set the stage for evolution to occur primarily because they provide the external component of an entity's fitness. The latter aspect makes ecology a central part of biology. As van Valen once put it: “evolution is the control of development by ecology

3.1 Population Ecology

The golden age of theoretical ecology (1920-1940)—to borrow the title of a book edited by Scudo and Ziegler—consisted primarily of population ecology. The next generation saw a shift of theoretical interest to community ecology. In recent years, interest has reverted to population ecology, sometimes in the form of metapopulation models, which consist of a set of populations with migration between them. Models in population ecology are based on representing an ecological system as the set of populations, which are usually of the same or different species it consists of. Each population, in turn, consists of potentially interacting individuals of a species. Populations may be characterized by their *state* variables - parameters representing properties of the population as a whole, for instance, size, density, growth-rate, *etc*, or by individual variables, that is, the properties of the individuals in them for example, individual fecundity, interactions, *etc*. Classical population ecology was restricted to the study of state-based models primarily because of the requirement that models be tractable, so as to permit predictions and explanations. Classical population ecology is the part of ecology that is theoretically the most developed. The central issue of interest in population ecology is the change in the size of populations over time. Population ecology considers both deterministic and stochastic models. Much of philosophical writing on population ecology has been restricted to deterministic population models and this relatively large body of work will only be very briefly summarized. More attention will be paid to

stochastic models which raise much more interesting philosophical issues that have not been adequately explored.

3.1.1 Deterministic Models

If population sizes are large, they can be studied using deterministic models, that is, fluctuations in populations sizes due to chance factors (such as accidental births and deaths) can be ignored. Usually a model considers members of a single or a very few interacting species, for instance, a few predator and a prey species. A typical result, based on the Lotka-Volterra (coupled differential equations) model, is that predator-prey interactions lead to population cycles, with the predator population cycle temporally tracking the prey population cycle. The explanation of this phenomenon is straightforward: as prey populations increase, the increased availability of resources allows a rise in predator populations a little later in time. But the increase of predators leads to an increase of prey consumption and, consequently, a decrease in prey populations. But, now, the lack of resources leads to a decline of predator populations. As predator populations decline, prey populations increase initiating the cycle once again. See the figure below:

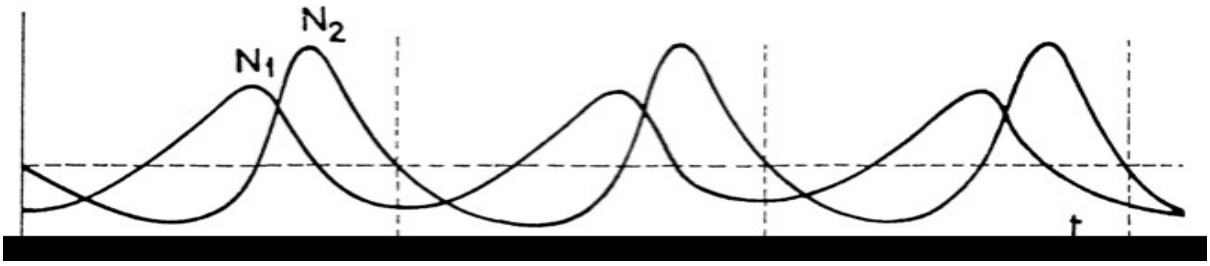


Figure 1: Predator-Prey Population Cycles

The model is due to Volterra 1978. There are two species, a predator species with a population, N_2, which only feeds on a single prey species with population, N_1. The model

incorporates demographic stochasticity which, nevertheless, does not stamp out the basic cyclic pattern. (*t* is a measure of time.)

The Lotka-Volterra model mathematically predicts these cycles. As such, it exemplifies the explanatory ideal of ecology: not only is there a predictively accurate quantitative model, but the mechanisms incorporated in the model have a perspicuous biological interpretation. Unfortunately, in ecology, because of the formalization and interpretation indeterminacy problems, the last condition is rarely satisfied.

For the simpler case of single species, two standard models are that of exponential and logistic growth. The exponential growth model is supposed to capture the behaviour of a population when there is no resource limitation; the logistic growth model is one of the simplest ways to try to capture the self-regulation of population sizes when there is such a limitation. See Examples 1.1 and 1.2.

Example1.1 Exponential Growth Model

Let a population consist of n individuals at time t. Suppose that, in an infinitesimal time interval between t and $t+dt$,

a fraction b/n of individuals give birth and a fraction d/n die.

Let the change in the size of the population be dn.

Then

$$dn/dt = (b - d).$$

Let $r = b - d$. Then the population dynamics is described by the growth equation:

$$dn/dt = m.$$

This is the exponential growth model. It assumes that no resource limitation constrains the "intrinsic growth rate", r. It can be solved to give:

$$n(t) = n_0 e^{rt},$$

where n_0 is the size of the population at t_0.

Example 1.2 Logistic Growth Model

One way to modify the exponential growth model to incorporate resource limitation is to replace the growth equation of the exponential model by that of the logistic growth model:

$$dn/dt = m(1 - n/K),$$

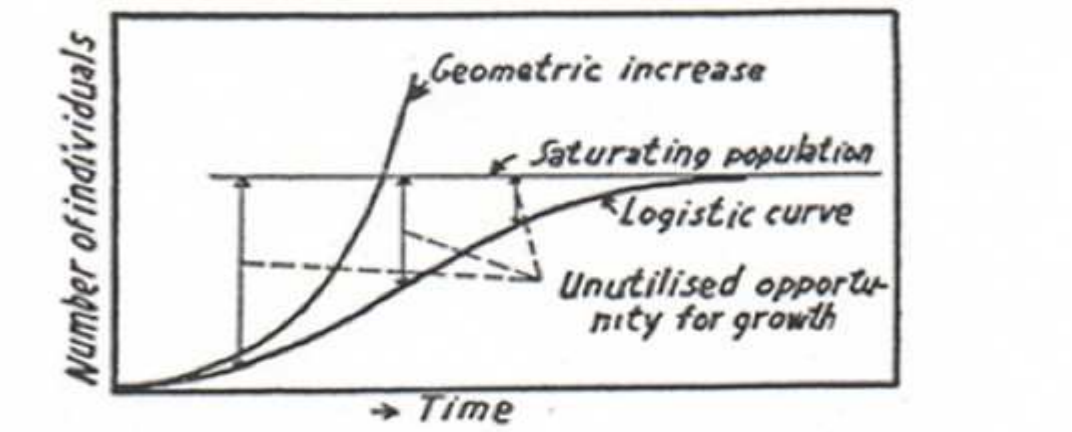
where K is called the "carrying capacity" of the environment; this parameter is supposed to incorporate how resource limitation affects population growth by regulating it.

When $n = K$, the growth rate,

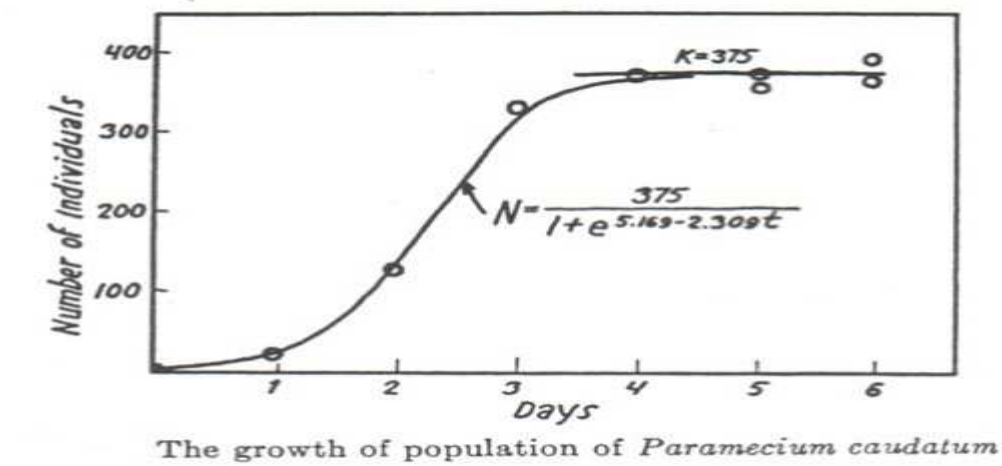
$$dn/dt = 0$$

and the population does not grow any further. Moreover, when there is no resource limitation; that is $K \to \infty$, this model reduces to the exponential growth model. **Figure 2b** shows how a population governed by the logistic equation grows in size. At the level of individual behaviour, this model does not have the kind of justification that the exponential growth model does in the sense that the logistic equation cannot be plausibly derived from the properties of individuals. In this sense it is a purely "phenomenological" model. The exponential growth model appeals to only one essentially ecological parameter, the intrinsic growth rate (r) of a population, interpreted as the rate at which the population would grow if there were no external factor limiting growth; the logistic model also appeals to the carrying

capacity **(*K*),** interpreted as the maximum size of the population that can persist in a given environment. See Figures 2a and 2b:



2a The curve of geometric increase and the logistic curve



2b The growth of population of *Paramecium caudatum*

Figures 2a and 2b: The Logistic Growth Model

The figure on the top shows theoretical curves. “Geometric increase” represents the exponential growth model discussed in the text; “saturating population” refers to the carrying capacity (see example 1.2). The figure on the bottom shows an example of an empirical

growth curve obtained in the laboratory. If the curve is fitted to a logistic curve (to which it shows similarity), then $K = 375$ is the estimated carrying capacity.

In general, biological experience suggests that all populations regulate their sizes, that is, they show self-regulation. Theoretical exploration of models has made it clear that a wide variety of mechanisms can lead to such self-regulation but it is usually unclear which models are more plausible than others thanks to the typical formalization indeterminacy of the field. Moreover, the precise mechanisms that are playing regulative roles in individual cases are often very hard to determine in the field, a classic case of partial observability. Even parameters such as the intrinsic growth rate and carrying capacity are unusually difficult to estimate precisely.

The last mentioned difficulties are perhaps most famously illustrated by the 10-year cycle of snowshoe hares, muskrats and their predators in the North American boreal forests and, especially, the 4-year cycle of lemmings and, possibly, other microtines in the arctic tundra of Eurasia and North America. In spite of almost seventy-five years of continuous research on these well-documented cycles the mechanisms driving them remain unresolved. Models producing such cycles abound, but the structural uncertainty of most of these models, coupled with partial observability of many of the parameters in the field have precluded resolution of the debate.

The models discussed so far are continuous-time models, that is, the temporal or dynamic parameter is assumed to be a continuous variable. However, discrete-time models have also been used to study population processes. A discrete analog of the logistic growth model was one of the first systems in which chaotic dynamic phenomena were discovered. Over the years there has been considerable debate over the question whether ecological systems with chaotic dynamics exist in nature; the current consensus is that they have not yet been found.

3.1.2 Stochastic Models

If population sizes are small, then models should be stochastic: the effects of fluctuations due of population size must be explicitly analyzed. Stochastic models in ecology are among the most mathematically complex models in science. Nevertheless they have begun to be systematically studied because of their relevance to biological conservation—see the entry on conservation biology. They also raise philosophically interesting questions because they underscore the extent to which the nature of randomness and uncertainty remains poorly explored in biological contexts.

What has, by and large, become the standard classification of stochasticity goes back to a 1978 dissertation by Shaffer. The context of that dissertation provides a striking exemplar of the social determination of science. The United States National Forest Management Act of 1976 required the Forest Service to "provide for diversity of plant and animal communities based on the suitability and capability of the specific land area." In 1979 the planning regulations developed to implement this provision required the Forest Service to "maintain viable populations of existing native and desired non-native vertebrate species in the planning area." A viable population was defined as "one which has the estimated numbers and distribution of reproductive individuals to insure its continued existence in the planning area." For large populations, falling within the domain of deterministic models, establishing viability is relatively trivial: all that must be ensured is that, on the average, a population is not declining in size. For small populations, even if it is increasing in size on the average, a chance fluctuation can result in extinction. Stochastic models are necessary to predict parameters such as the probability of extinction within a specified time period or the expected time to extinction.

In his dissertation, Shaffer attempted such an analysis for the grizzly bears (*Ursus arctos*) of Yellowstone which were believed to face the prospect of stochastic extinction. Shaffer distinguished four sources of uncertainty that can contribute to random extinction:

1. *demographic stochasticity* which arises from the chance events in the survival and reproductive success of a finite number of individuals…
2. *environmental stochasticity* due to temporal variation of birth and death rates, carrying capacity, and the population of competitors, predators, parasites, and diseases…
3. *natural catastrophes* such as floods, fires, droughts, etc.…
4. *genetic stochasticity* resulting from changes in gene frequencies due to founder effect, random fixation, or inbreeding."

Shaffer went on to argue that all these factors increase in importance as the population size decreases—a claim that will be questioned below—and, therefore, that their effects are hard to distinguish. Finally, he defined a minimum viable population (MVP): "A minimum viable population for any given species in any given habitat is the smallest population having at least a 95% chance of remaining extant for 100 years despite the foreseeable effects of demographic, environmental, and genetic stochasticity, and natural catastrophes."Both numbers (95% and 100 years) are conventional, and to be determined by social choice, rather than by biological factors, a point that Shaffer explicitly recognized.

In the 1980s, techniques to determine MVPs came to be called "population viability analysis" and the enthusiasm for the new framework was captured in the much-worn slogan: "MVP is the product, PVA the process." By the late 1980s, however, it became clear that the concept of a MVP was at best of very limited use. Leaving aside the conventional elements of the definition given above, even for the same species, populations in marginally different habitat

patches often show highly variable demographic trends resulting in highly variable MVP estimates for them, with each estimate depending critically on the local context. Moreover, as will be illustrated below for stochastic population models in general, the determination of MVPs suffers from irremediable structural uncertainty. This should not come as a surprise: what would have been more surprising is if legislative fiat had identified a scientifically valuable parameter. After the demise of the concept of the MVP, PVA began to be performed largely to estimate other parameters, especially the expected time to extinction of a population the estimation of which does not require any conventional choices—see the entry on conservation biology.

Within the context of PVA, Shaffer's classification—though usually with “random catastrophe” replacing “natural catastrophe”—became canonical within both ecology and conservation biology. However, the philosophical question remains as to whether it makes sense—classification, as many philosophers have pointed out, is not innocent of substantive theoretical assumptions. The first point to note is that genetic stochasticity is not even the same type of mechanism as the other three: its presence makes Shaffer's classification oddly heterogeneous. The reason for this is that genetic stochasticity is a consequence of demographic stochasticity: in small populations, a particular allele may reach fixation purely by chance reproductive events. It is even possible that stochasticity increases the rate at which a beneficial allele may go to fixation in a small population provided that the initial frequency of that allele is already high.

3.2 Community Ecology

Community ecology consists of models of interacting species, forming an ecological “community,” in which each species is treated as a unit. The appropriate definition of “community” has been widely debated among ecologists and philosophers; what is being

given here is an interactive definition. Alternative options include defining community by mere geographical association of species at one extreme, or by requiring a good deal of structure in the interactions at the other, making the community analogous to an organism. The interactive definition given above is attractive for two reasons: (a) mere association leaves little of theoretical or practical interest to study, while requiring some specified elevated levels of interaction introduces an unnecessary arbitrariness in a definition of community; and (b) the former would make any association of species a community whereas the latter would typically introduce so much structure that virtually no association would constitute a community.

Community models can be conveniently represented as loop, diagrams generalized graphs that have each species as a vertex and edges connecting these vertices when the species interact. The edges indicate whether the relevant species benefit or are harmed by the interaction, that is, whether they tend to increase or decrease in abundance, by an interaction. See Figure 3:

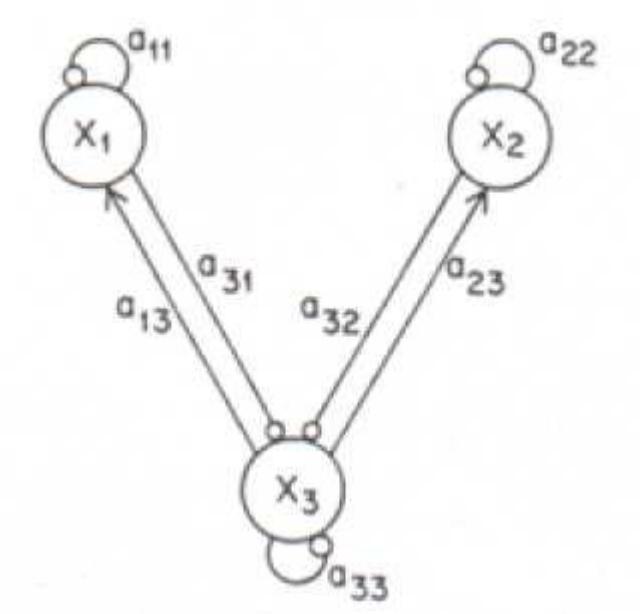


Figure 3: Structure of an Ecological Community

This is the loop diagram of a community of three species (from Diamond [1975a], p. 435). Species X_1 and X_2 both prey upon the resource species X_3. There is resource coupling and and competitive exclusion.

As with population ecology, what is of most interest is are the changes in a community over time. This brings us to one of the most interesting—and one of the most vexed—questions of ecology: the relationship between diversity and stability. A deeply rooted intuition among ecologists has been that diversity begets stability. If this claim is true, it has significant consequences for biodiversity conservation

What confuses this question from the very beginning, is the multiplicity of possible definitions of "diversity" and "stability." There are probably no better instances of formalization indeterminacy in any scientific context. For instance, a reasonable first attempt to define diversity would be to equate the diversity of a community to the number of species in it, that is, its species "richness." The trouble is that there is ample reason to doubt that richness captures all that is relevant about diversity, whether or not we are interested in only its relationship to stability. Consider two communities, the first consisting of 50% species *A* and 50% species *B*, and a second consisting of 99.9 % species *A* and 0.1% species *B*. Both communities have the same richness because they both have two species; however, there is a clear sense in which the first is more diverse—or less homogeneous—than the second. Moreover, the difference is likely to be relevant. If diversity does beget stability in these communities, then that stability must be a result some interaction between the two species. If species *B* comprises only 0.1% of the community, the scope for such interaction is typically much less than if it comprises 50%. Diversity must mean more than richness. There have

been several attempts to define and quantify diversity beyond richness; one of them is described in example 2 below.

Example2. Measures of Diversity

Ecologists often distinguish between three concepts of biodiversity, usually using species as the appropriate unit to measure diversity: (i) "α-diversity", the diversity within a community/place (MacArthur 1965); (ii) "β-diversity", the diversity between communities/places (Whittaker 1972, 1975); and (iii) "γ-diversity", the diversity between regions, that is, β-diversity on a larger spatial scale (Whittaker 1972). Though many measures of α-diversity have been proposed over the years, MacArthur's (1965) proposal to use the Shannon measure of information content in a communication process (Shannon 1948) has remained the most popular (though not universally accepted).

According to this measure, the α-diversity of a community with *n* species is given by

$$\alpha = -\sum_{i=1}^{n} p_i \ln p_i$$

Where p_i is the frequency of the *i*-th species. This is a measure of the diversity of a community in the same way that the Shannon measure of information content is a measure of the variety in a signal.

Turning to the two communities discussed in the text, a simple calculation shows that the diversity of the first is given by α = 0.693, while the diversity of the second is given by α = 0.008, verifying the intuition that the first is more diverse than the second.

3.3 Ecosystem Ecology

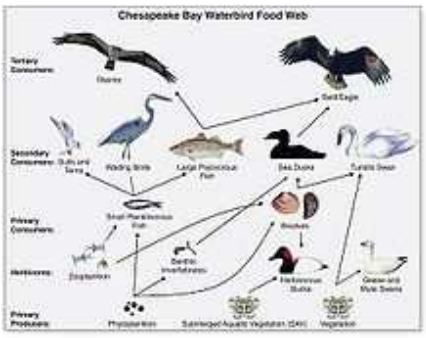
The term “ecosystem” was coined in 1935 by Tansley who defined it as “the whole *system* (in the sense of physics) including not only the organism-complex [that is, the community], but also the whole complex of physical factors forming what we call the environment of the biome—the habitat factors in the widest sense.” Tansley went on to argue that ecosystems “are the basic units of nature on the face of the earth.” For Tansley, using the term “ecosystem” implied a physical description of a community in its habitat. Even though that perspective still illuminates ecosystem studies (see below), it is no longer a necessary or even common connotation of the term “ecosystem.” However, independent of the use of “ecosystem,” ecosystem ecology, in contrast to other ecological sub-disciplines, and in continuity with its history, does retain an emphasis on physical processes. Should ecosystem ecology, then, be regarded as an instance of the unification of the physical and biological sciences? There has been so little philosophical attention to ecology that this question does not appear ever to have been broached.

The introduction and rapidly growing popularity of the term “ecosystem,” especially during the late 1950s and 1960s, was marked by two major cognitive and one sociological shift in the practice of ecology: (a) coming at the end of the so-called golden age of theoretical population ecology of the late 1920s and 1930s, turning to ecosystems helped shift emphasis from populations with interacting individuals to much larger and more inclusive systems. In this sense it was a deliberate anti-“reductionist” move. Ecosystem enthusiasts follow a long holistic tradition in natural history that tends to deify complexity and deny the possibility of explaining wholes in terms of their parts. “Systems thinking” was supposed to replace reductionism, the decomposition of wholes into parts for the sake of analysis; (b) a second cognitive shift is that ecosystem studies involve models based at least partly on non-biological variables. For instance, instead of tracking individuals or even species in communities, models may track energy or matter flow in food webs as a whole; and (c) at the

sociological level, the expansion of ecosystem studies led to what one historian has called the invention of “big biology” in the 1960s, chiefly in the US. These studies, for instance the massive Hubbard Brook Ecosystem Study, required more than just many biologists working together. They also demanded that other specialists, including geochemists and soil scientists, be brought in so that all the relevant physical parameters of ecosystems, besides the biological ones, could be tracked simultaneously. This study constituted the biologists' attempt to engage in publicly-funded Big Science, initiated by the physicists during the Manhattan Project, and subsequently profitably exploited by social scientists since the 1950s.

3.4 Food Webs

A food web is the archetypal ecological network. Plants capture and convert solar energy into the biomolecular bonds of simple sugars during photosynthesis. This food energy is transferred through a series of organisms starting with those that feed on plants and are themselves consumed. The simplified linear feeding pathways that move from a basal trophic species to a top consumer is called the food chain. The larger interlocking pattern of food chains in an ecological community creates a complex food web. Food webs are a type of concept map or a heuristic device that is used illustrate and study pathways of energy and material flows.



Generalized food web of waterbirds from Chesapeake Bay

Food webs are often limited relative to the real world. Complete empirical measurements are generally restricted to a specific habitat, such as a cave or a pond. Principles gleaned from food web microcosm studies are used to extrapolate smaller dynamic concepts to larger systems. Feeding relations require extensive investigations into the gut contents of organisms, which can be very difficult to decipher, or stable isotopes can be used to trace the flow of nutrient diets and energy through a food web. While food webs often give an incomplete measure of ecosystems, they are nonetheless a valuable tool in understanding community ecosystems.

Food-webs exhibit principals of ecological emergence through the nature of trophic entanglement, where some species have many weak feeding links (e.g., omnivores) while some are more specialized with fewer stronger feeding links (e.g., primary predators). Theoretical and empirical studies identify non-random emergent patterns of few strong and many weak linkages that serve to explain how ecological communities remain stable over time. Food-webs have compartments, where the many strong interactions create subgroups among some members in a community and the few weak interactions occur between these subgroups. These compartments increase the stability of food-webs.

As plants grow, they accumulate carbohydrates and are eaten by grazing herbivores. Step by step lines or relations are drawn until a web of life is illustrated.

3.5 Trophic Level

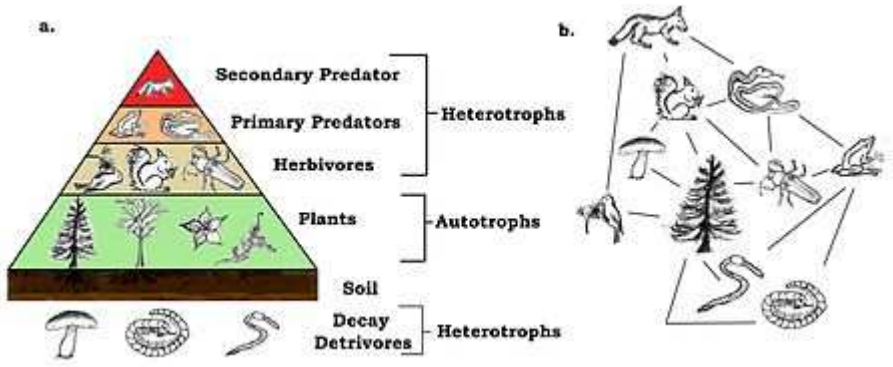
The Greek root of the word *troph*, τροφή, trophē, means food or feeding. Links in food-webs primarily connect feeding relations or trophism among species. Biodiversity within ecosystems can be organized into vertical and horizontal dimensions. The vertical dimension

represents feeding relations that become further removed from the base of the food chain up toward top predators. A trophic level is defined as "a group of organisms acquiring a considerable majority of its energy from the adjacent level nearer the abiotic source." The horizontal dimension represents the abundance or biomass at each level. When the relative abundance or biomass of each functional feeding group is stacked into their respective trophic levels they naturally sort into a 'pyramid of numbers'.

Functional groups are broadly categorized as autotrophs (or primary producers), heterotrophs (or consumers), and detrivores (or decomposers). Autotrophs are organisms that can produce their own food (production is greater than respiration) and are usually plants or cyanobacteria that are capable of photosynthesis but can also be other organisms such as bacteria near ocean vents that are capable of chemosynthesis. Heterotrophs are organisms that must feed on others for nourishment and energy (respiration exceeds production). Heterotrophs can be further sub-divided into different functional groups, including: primary consumers (strict herbivores), secondary consumers (carnivorous predators that feed exclusively on herbivores) and tertiary consumers (predators that feed on a mix of herbivores and predators). Omnivores do not fit neatly into a functional category because they eat both plant and animal tissues. It has been suggested that omnivores have a greater functional influence as predators because relative to herbivores they are comparatively inefficient at grazing.

Trophic levels are part of the holistic or complex systems view of ecosystems. Each trophic level contains unrelated species that grouped together because they share common ecological functions. Grouping functionally similar species into a trophic system gives a macroscopic image of the larger functional design. While the notion of trophic levels provides insight into energy flow and top-down control within food webs, it is troubled by the prevalence of omnivory in real ecosystems. This has lead some ecologists to "reiterate that the notion that

species clearly aggregate into discrete, homogeneous trophic levels is fiction." Nonetheless, recent studies have shown that real trophic levels do exist, but "above the herbivore trophic level, food webs are better characterized as a tangled web of omnivores.



A trophic pyramid (a) and a food-web (b) illustrating ecological relationships among creatures that are typical of a northern Boreal terrestrial ecosystem. The trophic pyramid roughly represents the biomass (usually measured as total dry-weight) at each level. Plants generally have the greatest biomass. Names of trophic categories are shown to the right of the pyramid. Some ecosystems, such as many wetlands, do not organize as a strict pyramid, because aquatic plants are not as productive as long-lived terrestrial plants such as trees. Ecological trophic pyramids are typically one of three kinds: 1) pyramid of numbers, 2) pyramid of biomass, or 3) pyramid of energy

3.6 Ecology and Evolution

Ecology and evolution are considered sister disciplines of the life sciences. Natural selection, life history, development, adaptation, populations, and inheritance are examples of concepts that thread equally into ecological and evolutionary theory. Morphological, behavioral and/or genetic traits, for example, can be mapped onto evolutionary trees to study the historical development of a species in relation to their functions and roles in different ecological circumstances. In this framework, the analytical tools of ecologists and evolutionists overlap as they organize, classify and investigate life through common systematic principals, such as phylogenetics or the Linnaean system of taxonomy. The two disciplines often appear together, such as in the title of the journal *Trends in Ecology and Evolution*. There is no sharp boundary separating ecology from evolution and they differ more in their areas of applied

focus. Both disciplines discover and explain emergent and unique properties and processes operating across different spatial or temporal scales of organization. While the boundary between ecology and evolution is not always clear, it is understood that ecologists study the abiotic and biotic factors that influence the evolutionary process.

3.6.1 Behavioural Ecology

All organisms are motile to some extent. Even plants express complex behavior, including memory and communication. Behavioral ecology is the study of ethology and its ecological and evolutionary implications. Ethology is the study of observable movement or behavior in nature. This could include investigations of motile sperm of plants, mobile phytoplankton, zooplankton swimming toward the female egg, the cultivation of fungi by weevils, the mating dance of a salamander, or social gatherings of amoeba.

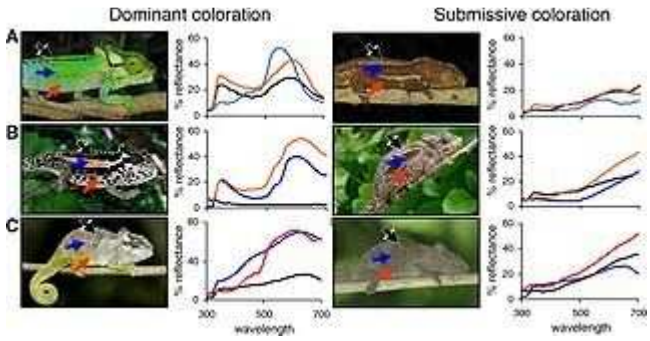
Adaptation is the central unifying concept in behavioral ecology. Behaviors can be recorded as traits and inherited in much the same way that eye and hair color can. Behaviors evolve and become adapted to the ecosystem because they are subject to the forces of natural selection. Hence, behaviors can be adaptive, meaning that they evolve functional utilities that increases reproductive success for the individuals that inherit such traits. This is also the technical definition for fitness in biology, which is a measure of reproductive success over successive generations.

Predator-prey interactions are an introductory concept into food-web studies as well as behavioral ecology. Prey species can exhibit different kinds of behavioral adaptations to predators, such as avoid, flee or defend. Many prey species are faced with multiple predators that differ in the degree of danger posed. To be adapted to their environment and face predatory threats, organisms must balance their energy budgets as they invest in different

aspects of their life history, such as growth, feeding, mating, socializing, or modifying their habitat. Hypotheses posited in behavioral ecology are generally based on adaptive principals of conservation, optimization or efficiency. For example,

"The threat-sensitive predator avoidance hypothesis predicts that prey should assess the degree of threat posed by different predators and match their behavior according to current levels of risk."[

"The optimal flight initiation distance occurs where expected postencounter fitness is maximized, which depends on the prey's initial fitness, benefits obtainable by not fleeing, energetic escape costs, and expected fitness loss due to predation risk."



Social display and color variation in differently adapted species of chameleons (*Bradypodion* spp.). Chameleons change their skin color to match their background as a behavioral defense mechanism and also use color to communicate with other members of their species, such as dominant (left) versus submissive (right) patterns shown in the three species (A-C) above.

Elaborate sexual displays and posturing are encountered in the behavioral ecology of animals. The birds of paradise, for example, display elaborate ornaments and song during courtship. These displays serve a dual purpose of signaling healthy or well-adapted individuals and desirable genes. The elaborate displays are driven by sexual selection as an advertisement of quality of traits among male suitors.

3.6.2 Social Ecology

Social ecological behaviors are notable in the social insects, slime moulds, social spiders, human society, and naked mole rats where eusocialism has evolved. Social behaviors include reciprocally beneficial behaviors among kin and nest mates. Social behaviors evolve from kin and group selection. Kin selection explains altruism through genetic relationships, whereby an altruistic behavior leading to death is rewarded by the survival of genetic copies distributed among surviving relatives. The social insects, including ants, bees and wasps are most famously studied for this type of relationship because the male drones are clones that share the same genetic make-up as every other male in the colony. In contrast, group selectionists find examples of altruism among non-genetic relatives and explain this through selection acting on the group, whereby it becomes selectively advantageous for groups if their members express altruistic behaviors to one another. Groups that are predominantly altruists beat groups that are predominantly selfish.

Symbiosis: Leafhoppers (*Eurymela fenestrata*) are protected by ants (*Iridomyrmex purpureus*) in a symbiotic relationship. The ants protect the leafhoppers from predators and in return the leafhoppers feeding on plants exude honeydew from their anus that provides energy and nutrients to tending ants.

3.6.3 Coevolution

Ecological interactions can be divided into host and associate relationships. A host is any entity that harbors another that is called the associate. Host and associate relationships among species that are mutually or reciprocally beneficial are called mutualisms. If the host and associate are physically connected, the relationship is called symbiosis. Approximately 60% of all plants, for example, have a symbiotic relationship with arbuscular mycorrhizal fungi. Symbiotic plants and fungi exchange carbohydrates for mineral nutrients. Symbiosis differs from indirect mutualisms where the organisms live apart. For example, tropical rainforests regulate the Earth's atmosphere. Trees living in the equatorial regions of the planet supply oxygen into the atmosphere that sustains species living in distant polar regions of the planet. This relationship is called commensalism because many other host species receive the benefits of clean air at no cost or harm to the associate tree species supplying the oxygen. The host and associate relationship is called parasitism if one species benefits while the other suffers. Competition among species or among members of the same species is defined as reciprocal antagonism, such as grasses competing for growth space. Popular ecological study systems for mutualism include, fungus-growing ants employing agricultural symbiosis, bacteria living in the guts of insects and other organisms, the fig wasp and yucca moth pollination complex, lichens with fungi and photosynthetic algae, and corals with photosynthetic algae. Nevertheless, many organisms exploit host rewards without reciprocating and thus have been branded with a myriad of not-very-flattering names such as 'cheaters', 'exploiters', 'robbers', and 'thieves'. Although cheaters impose several host cots (e.g., via damage to their reproductive organs or propagules, denying the services of a beneficial partner), their net effect on host fitness is not necessarily negative and, thus, becomes difficult to forecast.

3.6.4 Biogeography

The word *biogeography* is an amalgamation of *biology* and *geography*. Biogeography is the comparative study of the geographic distribution of organisms and the corresponding evolution of their traits in space and time. The Journal of Biogeography was established in 1974. Biogeography and ecology share many of their disciplinary roots. For example, the theory of island biogeography, published by the mathematician Robert MacArthur and ecologist Edward O. Wilson in 1967is considered one of the fundamentals of ecological theory.

Biogeography has a long history in the natural sciences where questions arise concerning the spatial distribution of plants and animals. Ecology and evolution provide the explanatory context for biogeographical studies. Biogeographical patterns result from ecological processes that influence range distributions, such as migration and dispersal. and from historical processes that split populations or species into different areas. The biogeographic processes that result in the natural splitting of species explains much of the modern distribution of the Earth's biota. The splitting of lineages in a species is called vicariance biogeography and it is a sub-discipline of biogeography. There are also practical applications in the field of biogeography concerning ecological systems and processes. For example, the range and distribution of biodiversity and invasive species responding to climate change is a serious concern and active area of research in context of global warming

3.6.5 r/K-Selection theory

A population ecology concept (introduced in MacArthur and Wilson's (1967) book, *The Theory of Island Biogeography*) is r/K selection theory, one of the first predictive models in ecology used to explain life-history evolution. The premise behind the r/K selection model is that natural selection pressures change according to population density. For example, when an island is first colonized, density of individuals is low. The initial increase in population

size is *not* limited by competition, leaving an abundance of available resources for rapid population growth. These early phases of population growth experience *density-independent* forces of natural selection, which is called *r*-selection. As the population becomes more crowded, it approaches the island's carrying capacity, thus forcing individuals to compete more heavily for fewer available resources. Under crowded conditions the population experiences density-dependent forces of natural selection, called *K*-selection.

In the *r/K*-selection model, the first variable *r* is the intrinsic rate of natural increase in population size and the second variable *K* is the carrying capacity of a population. Different species evolve different life-history strategies spanning a continuum between these two selective forces. An *r*-selected species is one that has high birth rates, low levels of parental investment, and high rates of mortality before individuals reach maturity. Evolution favors high rates of fecundity in *r*-selected species. Many kinds of insects and invasive species exhibit *r*-selected characteristics. In contrast, a *K*-selected species has low rates of fecundity, high levels of parental investment in the young, and low rates of mortality as individuals mature. Humans and elephants are examples of species exhibiting *K*-selected characteristics, including longevity and efficiency in the conversion of more resources into fewer offspring.

3.6.6 Molecular ecology

The important relationship between ecology and genetic inheritance predates modern techniques for molecular analysis. Molecular ecological research became more feasible with the development of rapid and accessible genetic technologies, such as the polymerase chain reaction (PCR). The rise of molecular technologies and influx of research questions into this new ecological field resulted in the publication *Molecular Ecology* in 1992. Molecular ecology uses various analytical techniques to study genes in an evolutionary and ecological context. In 1994, John Avise also played a leading role in this area of science with the

publication of his book, *Molecular Markers, Natural History and Evolution*. Newer technologies opened a wave of genetic analysis into organisms once difficult to study from an ecological or evolutionary standpoint, such as bacteria, fungi and nematodes. Molecular ecology engendered a new research paradigm for investigating ecological questions considered otherwise intractable. Molecular investigations revealed previously obscured details in the tiny intricacies of nature and improved resolution into probing questions about behavioral and biogeographical ecology. For example, molecular ecology revealed promiscuous sexual behavior and multiple male partners in tree swallows previously thought to be socially monogamous. In a biogeographical context, the marriage between genetics, ecology and evolution resulted in a new sub-discipline called phylogeography.

3.6.7 Human Ecology

Human ecology is the interdisciplinary investigation into the ecology of our species. "Human ecology may be defined: (1) from a bio-ecological standpoint as the study of man as the ecological dominant in plant and animal communities and systems; (2) from a bio-ecological standpoint as simply another animal affecting and being affected by his physical environment; and (3) as a human being, somehow different from animal life in general, interacting with physical and modified environments in a distinctive and creative way. A truly interdisciplinary human ecology will most likely address itself to all three." The term human ecology was formally introduced in 1921, but many sociologists, geographers, psychologists, and other disciplines were interested in human relations to natural systems centuries prior, especially in the late 19th century. Some authors have identified a new

unifying science in coupled human and natural systems that builds upon, but moves beyond the field human ecology. Ecology is as much a biological science as it is a human science. "Perhaps the most important implication involves our view of human society. *Homo sapiens* is not an external disturbance, it is a keystone species within the system. In the long term, it may not be the magnitude of extracted goods and services that will determine sustainability. It may well be our disruption of ecological recovery and stability mechanisms that determines system collapse."

4.0 Conclusion

The student has learnt the following

- The concept of ecology
- The significance of ecology to evolution
- Different types of ecology
- Ecological models

5.0 Summary

The science of ecology studies interactions between individual organisms and their environments, including interactions with both conspecifics and members of other species. Though ecology emerged in the 19th century much of its theoretical structure only emerged in the twentieth century. Though ecology includes a wide variety of sub-fields, philosophical analysis of ecology has so far been restricted to population, community, and ecosystem ecology. Central philosophical problems include explication of relevant notions of ecological diversity and stability the relation between diversity and stability. Other debated questions are the nature of laws and theories in ecology, strategies of model-building, and reductionism. Contemporary ecology is undergoing a conceptual upheaval because of increased

computational power. The recent emphasis on individual-based models, which embrace methodological individualism, should be viewed as a return of reductionism in ecology. Other important developments include widespread interest in spatially explicit models and the advent of Geographical Information Systems.

6.0 Tutor-Marked Assignment

- What do you understand by the term ecology
- State the different types of ecology
- Explain one model in ecology

7.0 Reference/Further Reading

Aerts, R. and Chapin III, F. S. 2000. "The Mineral Nutrition of Wild Plants Revisited: A Re-Evaluation of Processes and Patterns." *Advances in Ecological Research* **30**: 1-67.

Beissinger, S. R. 2002. "Population Viability Analysis: Past, Present, Future." In Beissinger, S. R. and McCullough, D. R. Ed. *Population Viability Analysis*. Chicago: University of Chicago Press, pp. 5-17.

Beissinger, S. R. and Westphal, M. I. 1998. "On the Use of Demographic Models of Population Viability in Endangered Species Management." *Journal of Wildlife Management* **62**: 821 –841.

Connor, E. F. and McCoy, E. D. 1979. "The Statistics and Biology of the Species-Area Relationship." *American Naturalist* **113**: 791-833.

Cooper, G. J. 2004. *The Science of the Struggle for Existence: On the Foundations of Ecology*. New York: Cambridge University Press.

Cuddington, K. 2001. “The ‘Balance of Nature’ Metaphor and Equilibrium in Population Ecology.” *Biology and Philosophy* **16**: 463-479.

UNIT 2: INTRODUCTION TO POLYMORPHISM

Content

1.0 Introduction

Polymorphism in biology occurs when two or more clearly different phenotypes exist in the same population of a species — in other words, the occurrence of more than one *form* or *morph*. In order to be classified as such, morphs must occupy the same habitat at the same time and belong to a panmictic population (one with random mating).

Polymorphism is common in nature; it is related to biodiversity, genetic variation and adaptation; it usually functions to retain variety of form in a population living in a varied environment. The most common example is sexual dimorphism, which occurs in many organisms. Other examples are mimetic forms of butterflies (see mimicry), and human haemoglobin and blood types.

Polymorphism results from evolutionary processes, as does any aspect of a species. It is heritable and is modified by natural selection. In polyphenism, an individual's genetic make-up allows for different morphs, and the switch mechanism that determines which morph is shown is environmental. In *genetic polymorphism*, the genetic make-up determines the morph. Ants exhibit both types in a single population.

2.0 Objectives

At the end of this unit, the student should be able to:

- Define polymorphism
- State and discuss types of polymorphism

3.0 Terminology

Although in general use polymorphism is quite a broad term, in biology it has been given a specific meaning.

- The term omits characters showing *continuous variation* (such as weight), even though this has a heritable component. Polymorphism deals with forms in which the variation is discrete (discontinuous) or strongly bimodal or polymodal.
- Morphs must occupy the same habitat at the same time: this excludes geographical races and seasonal forms. The use of the words *morph* or *polymorphism* for what is a visibly different *geographical race or variant* is common, but incorrect. The significance of geographical variation is in that it may lead to allopatric speciation, whereas true polymorphism takes place in panmictic populations.
- The term was first used to describe *visible forms*, but nowadays it has been extended to include *cryptic morphs*, for instance blood types, which can be revealed by a test.
- Rare variations are not classified as polymorphisms; and mutations by themselves do not constitute polymorphisms. To qualify as a polymorphism there has to be some kind of balance between morphs underpinned by inheritance. The criterion is that the frequency of the *least* common morph is too high simply to be the result of new mutations or, as a rough guide, that it is greater than 1 percent (though that is far higher than any normal mutation rate for a single allele)

3.1 Nomenclature

Polymorphism crosses several discipline boundaries, including ecology and genetics, evolution theory, taxonomy, cytology and biochemistry. Different disciplines may give the same concept different names, and different concepts may be given the same name. For

example, there are the terms established in ecological genetics by E.B. Ford (1975) and for classical genetics by John Maynard Smith (1998).The shorter term *morphism* may be more accurate than *polymorphism*, but is not often used. It was the preferred term of the evolutionary biologist Julian Huxley (1955).

Various synonymous terms exist for the various polymorphic forms of an organism. The most common are *morph* and *morpha*, while a more formal term is *morphotype*. *Form* and *phase* are sometimes also used, but are easily confused in zoology with, respectively, "form" in a population of animals, and "phase" as a color or other change in an organism due to environmental conditions (temperature, humidity, etc.). Phenotypic traits and characteristics are also possible descriptions, though that would imply just a limited aspect of the body.

In the taxonomic nomenclature of zoology, the word "morpha" plus a Latin name for the morph can be added to a binomial or trinomial name. However, this invites confusion with geographically-variant ring species or subspecies, especially if polytypic. Morphs have no formal standing in the ICZN. In botanical taxonomy, the concept of morphs is represented with the terms "variety", "subvariety" and "form", which are formally regulated by the ICBN. Horticulturalists sometimes confuse this usage of "variety" both with cultivar ("variety" in viticultural usage, rice agriculture jargon, and informal gardening lingo) and with the legal concept "plant variety" (protection of a cultivar as a form of intellectual property)

3.2 Ecology

Selection, whether natural or artificial, changes the frequency of morphs within a population; this occurs when morphs reproduce with different degrees of success. A genetic (or *balanced*) polymorphism usually persists over many generations, maintained by two or more opposed and powerful selection pressures. Diver (1929) found banding morphs in *Cepaea nemoralis*

could be seen in pre-fossil shells going back to the Mesolithic Holocene. Apes have similar blood groups to humans; this suggests rather strongly that this kind of polymorphism is quite ancient, at least as far back as the last common ancestor of the apes and man, and possibly even further.

The relative proportions of the morphs may vary; the actual values are determined by the effective fitness of the morphs at a particular time and place. The mechanism of heterozygote advantage assures the population of some alternative alleles at the locus or loci involved. Only if competing selection disappears will an allele disappear. However, heterozygote advantage is not the only way a polymorphism can be maintained. Apostatic selection, whereby a predator consumes a common morph whilst overlooking rarer morphs is possible and does occur. This would tend to preserve rarer morphs from extinction.

A polymorphic population does not initiate speciation; nor does it prevent speciation. It has little or nothing to do with species splitting. However, *it has a lot to do with the adaptation of a species to its environment*, which may vary in colour, food supply, predation and in many other ways. Polymorphism is one good way the opportunities get to be used; it has survival value, and the selection of modifier genes may reinforce the polymorphism.

3.3 Polymorphism and Niche Diversity

G. Evelyn Hutchinson, a founder of niche research, commented "It is very likely from an ecological point of view that all species, or at least all common species, consist of populations adapted to more than one niche". He gave as examples sexual size dimorphism and mimicry. In many cases where the male is short-lived and smaller than the female, he does not compete with her during her late pre-adult and adult life. Size difference may permit both sexes to exploit different niches. In elaborate cases of mimicry, such as the African butterfly *Papilio dardanus*, female morphs mimic a range of distasteful models, often in the

same region. The fitness of each type of mimic decreases as it becomes more common, so the polymorphism is maintained by frequency-dependent selection. Thus the efficiency of the mimicry is maintained in a much increased total population.

3.4 The switch

The mechanism which decides which of several morphs an individual displays is called the *switch*. This switch may be genetic, or it may be environmental. Taking sex determination as the example, in humans the determination is genetic, by the XY sex-determination system. In Hymenoptera (ants, bees and wasps), sex determination is by haplo-diploidy: the females are all diploid, the males are haploid. However, in some animals an environmental trigger determines the sex: alligators are a famous case in point. In ants the distinction between workers and guards is environmental, by the feeding of the grubs. Polymorphism with an environmental trigger is called polyphenism.

The polyphenic system does have a degree of environmental flexibility not present in the genetic polymorphism. However, such environmental triggers are the less common of the two methods.

3.5 Investigative Methods

Investigation of polymorphism requires a coming together of field and laboratory technique. In the field:

- detailed survey of occurrence, habits and predation
- selection of an ecological area or areas, with well-defined boundaries
- capture, mark, release, recapture data (see Mark and recapture)
- relative numbers and distribution of morphs

- estimation of population sizes

And in the laboratory:

- genetic data from crosses
- population cages
- chromosome cytology if possible
- use of chromatography or similar techniques if morphs are cryptic (for example, biochemical)

Both types of work are equally important. Without proper field-work the significance of the polymorphism to the species is uncertain; without laboratory breeding the genetic basis is obscure. Even with insects the work may take many years; examples of Batesian mimicry noted in the nineteenth century are still being researched.

3.6 Genetic Polymorphism

Since all polymorphism has a genetic basis, *genetic polymorphism* has a particular meaning:

- Genetic polymorphism is the simultaneous occurrence in the same locality of two or more discontinuous forms in such proportions that the rarest of them cannot be maintained just by recurrent mutation or immigration.

The definition has three parts: a) sympatry: one interbreeding population; b) discrete forms; and c) not maintained just by mutation.

Genetic polymorphism is actively and steadily maintained in populations by natural selection, in contrast to *transient polymorphisms* where a form is progressively replaced by another. By

definition, genetic polymorphism relates to a balance or equilibrium between morphs. The mechanisms that conserve it are types of balancing selection.

3.7 Mechanism of Balancing Selection

- Heterosis (or heterozygote advantage): "Heterosis: the heterozygote at a locus is fitter than either homozygote".

- Frequency dependent selection: The fitness of a particular phenotype is dependent on its frequency relative to other phenotypes in a given population. Example: prey switching, where rare morphs of prey are actually fitter due to predators concentrating on the more frequent morphs.

- Fitness varies in time and space. Fitness of a genotype may vary greatly between larval and adult stages, or between parts of a habitat range.

- Selection acts differently at different levels. The fitness of a genotype may depend on the fitness of other genotypes in the population: this covers many natural situations where the best thing to do (from the point of view of survival and reproduction) depends on what other members of the population are doing at the time.

3.8 Pleiotropism

Most genes have more than one effect on the phenotype of an organism (pleiotropism). Some of these effects may be visible, and others cryptic, so it is often important to look beyond the most obvious effects of a gene to identify other effects. Cases occur where a gene affects an unimportant visible character, yet a change in fitness is recorded. In such cases the gene's other (cryptic or 'physiological') effects may be responsible for the change in fitness.

"If a neutral trait is pleiotropically linked to an advantageous one, it may emerge because of a process of natural selection. It was selected but this doesn't mean it is an adaptation. The reason is that, although it was selected, there was no selection for that trait.

3.9 Epistasis

Epistasis occurs when the expression of one gene is modified by another gene. For example, gene A only shows its effect when allele B1 (at another Locus) is present, but not if it is absent. This is one of the ways in which two or more genes may combine to produce a coordinated change in more than one characteristic (for instance, in mimicry). Unlike the supergene, epistatic genes do not need to be closely linked or even on the same chromosome.

Both pleiotropism and epistasis show that a gene need not relate to a character in the simple manner that was once supposed.

4.0 Origin of supergenes

Although a polymorphism can be controlled by alleles at a single locus (e.g. human ABO blood groups), the more complex forms are controlled by supergenes consisting of several tightly linked genes on a single chromosome. Batesian mimicry in butterflies and heterostyly in angiosperms are good examples. There is a long-standing debate as to how this situation could have arisen, and the question is not yet resolved.

Whereas a gene family (several tighly linked genes performing similar or identical functions) arises by duplication of a single original gene, this is usually not the case with supergenes. In a supergene some of the constituent genes have quite distinct functions, so they must have

come together under selection. This process might involve suppression of crossing-over, translocation of chromosome fragments and possibly occasional cistron duplication. That crossing-over can be suppressed by selection has been known for many years.

Debate has centred round the question of whether the component genes in a super-gene could have started off on separate chromosomes, with subsequent reorganization, or if it is necessary for them to start on the same chromosome. Originally, it was held that chromosome rearrangement would play an important role. This explanation was accepted by E. B. Ford and incorporated into his accounts of ecological genetics.

However, today many believe it more likely that the genes start on the same chromosome. They argue that supergenes arose *in situ*. This is known as Turner's sieve hypothesis. John Maynard Smith agreed with this view in his authoritative textbook but the question is still not definitively settled.

5.0 Conclusion

In this unit the student learnt the following:

- Definition of polymorphism
- Nomenclature associated with polymorphism
- Ecology
- Genetics and polymorphism

6.0 Summary

Polymorphism, in biology, a discontinuous genetic variation resulting in the occurrence of several different forms or types of individuals among the members of a single species. A discontinuous genetic variation divides the individuals of a population into two or more

sharply distinct forms. The most obvious example of this is the separation of most higher organisms into male and female sexes.

7.0 Tutor-Marked Assignment

- Define polymorphism
- Explain polymorphism and niche diversity
- Discuss mechanism of balancing selection
- What is Epistatsis

8.0 Reference/ Further Reading

Ford E.B. 1965. *Genetic polymorphism*. Faber & Faber, London

Dobzhansky, Theodosius. 1970. *Genetics of the Evolutionary Process*

Clark, W. C. (1976). "The Environment and the Genotype in Polymorphism". *Zoological Journal of the Linnaean Society* **58** (3): 255–262.

Ford, E. B. 1975. *Ecological Genetics* (4th ed.). London: Chapman & Hall

Cain, Arthur J. 1971. "Colour and Banding Morphs in Subfossil Samples of the Snail *Cepaea*". In R. Creed (ed.), *Ecological genetics and Evolution: Essays in Honour of E.B. Ford*. Oxford: Blackwell.

Begon, Townsend, Harper. 2006. *Ecology: from individuals to ecosystems*. 4th ed, Blackwell, Oxford. ISBN 978-1405111171

Charlesworth D., Charlesworth B. (1975). "Theoretical Genetics of Batesian Mimicry". "I. Single-locus Models". *Journal of Theoretical Biology*, **55**:283–303. "II. Evolution of

Supergenes". *J. Theoret. Biol.*, **55**:305–324. "III. Evolution of Dominance"". *J. Theoret. Biol.* **55**: 325–337.

UNIT 3: EXAMPLES OF POLYMORPHISM

Contents

1.0 INTRODUCTION

Polymorphism results from evolutionary processes, as does any aspect of a species. It is heritable and is modified by natural selection. In polyphenism, an individual's genetic make-up allows for different morphs, and the switch mechanism that determines which morph is shown is environmental. In *genetic polymorphism*, the genetic make-up determines the morph. Ants exhibit both types in a single population

2.0 Objective

In this unit the student will learn the following:

- Sexual dimorphism
- Human polymorphism
- Polymorphism in lower organisms

3.0 Sexual dimorphism

We meet genetic polymorphism daily, since our species uses sexual reproduction, and of course, the sexes are differentiated. However, even if the sexes were identical in superficial appearance, the division into two sexes is a dimorphism, albeit cryptic. This is because the phenotype of an organism includes its sexual organs and its chromosomes, and all the behaviour associated with reproduction. So research into sexual dimorphism has addressed two issues: first, the advantage of sex in evolutionary terms; second, the role of visible sexual differentiation.

The system is relatively stable and heritable, usually by means of sex chromosomes. Every aspect of this everyday phenomenon bristles with questions for the theoretical biologist. Why is the ratio ~50/50? How could the evolution of sex occur from an original situation of asexual reproduction, which has the advantage that every member of a species could reproduce? Why the visible differences between the sexes? These questions have engaged the attentions of biologists such as Charles Darwin, August Weismann, Ronald Fisher, George C. Williams, John Maynard Smith and W. D. Hamilton, with varied success.

Of the many issues involved, there is widespread agreement on the following: the advantage of sexual and hermaphroditic reproduction over asexual reproduction lies in the way recombination increases the genetic diversity of the ensuing population. The advantage of sexual reproduction over hermaphroditic is not so clear. In forms that have two separate sexes, same sex combinations are excluded from mating which decreases the amount of diversity compared with hermaphrodites by at least twice. So, why are almost all progressive species bi-sexual, considering the asexual process is more efficient and simple, whilst hermaphrodites produce a more diversified progeny? It has been suggested that differentiation into two sexes has evolutionary advantages allowing changes to concentrate in the male part of the population and at the same time preserving the existing genotype

distribution in the females. This enables the population to better meet the challenges of infection, parasitism, predation and other hazards of the varied environment.

3.1 Human polymorphism

3.1.1 Human blood groups

All the common blood types, such as the ABO blood group system, are genetic polymorphisms. Here we see a system where there are more than two morphs: the phenotypes are A, B, AB and O are present in all human populations, but vary in proportion in different parts of the world. The phenotypes are controlled by multiple alleles at one locus. These polymorphisms are seemingly never eliminated by natural selection; the reason came from a study of disease statistics.

Statistical research has shown that the various phenotypes are more, or less, likely to suffer a variety of diseases. For example, an individual's susceptibility to cholera (and other diarrheal infections) is correlated with their blood type: those with type O blood are the most susceptible, while those with type AB are the most resistant. Between these two extremes are the A and B blood types, with type A being more resistant than type B. This suggests that the pleiotropic effects of the genes set up opposing selective forces, thus maintaining a balance. Geographical distribution of blood groups is broadly consistent with the classification of "races" developed by early anthropologists on the basis of visible features.

3.1.2 Sickle-cell anaemia

Such a balance is seen more simply in sickle-cell anaemia, which is found mostly in tropical populations in Africa and India. An individual homozygous for the recessive sickle haemoglobin, HgbS, has a short expectancy of life, whereas the life expectancy of the standard haemoglobin (HgbA) homozygote and also the heterozygote is normal (though

heterozygote individuals will suffer periodic problems). The sickle-cell variant survives in the population because the heterozygote is resistant to malaria and the malaria parasite kills a huge number of people each year. This is balancing selection or genetic polymorphism, balanced between fierce selection against homozygous sickle-cell sufferers, and selection against the standard HgbA homozygotes by malaria. The heterozygote has a permanent advantage (a higher fitness) so long as malaria exists; and it has existed as a human parasite for a long time. Because the heterozygote survives, so does the HgbS allele survive at a rate much higher than the mutation rate.

3.1.3 Duffy system

The Duffy antigen is a protein located on the surface of red blood cells, encoded by the FY (DARC) gene. The protein encoded by this gene is a non-specific receptor for several chemokines, and is the known entry-point for the human malarial parasites Plasmodium vivax and Plasmodium knowlesi. Polymorphisms in this gene are the basis of the Duffy blood group system. In humans, a mutant variant at a single site in the FY cis-regulatory region abolishes all expression of the gene in erythrocyte precursors. As a result, homozygous mutants are strongly protected from infection by P. vivax, and a lower level of protection is conferred on heterozygotes. The variant has apparently arisen twice in geographically distinct human populations, in Africa and Papua New Guinea. It has been driven to high frequencies on at least two haplotypic backgrounds within Africa. Recent work indicates a similar, but not identical, pattern exists in baboons (Papio cynocephalus), which suffer a mosquito-carried malaria-like pathogen, Hepatocystis kochi. Researchers interpret this as a case of convergent evolution.

3.1.4 G6PD

G6PD (Glucose-6-phosphate dehydrogenase) human polymorphism is also implicated in malarial resistance. G6PD alleles with reduced activity are maintained at a high level in endemic malarial regions, despite reduced general viability. Variant A (with 85% activity) reaches 40% in sub-Saharan Africa, but is generally less than 1% outside Africa and the Middle East.

3.1.5 Cystic fibrosis

Cystic fibrosis, a congenital disorder which affects about one in 2000 children, is caused by a mutant form of the CF transmembrane regulator gene, CFTR. The transmission is Mendelian: the normal gene is dominant, so all heterozygotes are healthy, but those who inherit two mutated genes have the condition. The mutated allele is present in about 1:25 of the population (mostly heterozygotes), which is much higher than expected from the rate of mutation alone. Sufferers from this disease have shortened life expectancy (and males are usually sterile if they survive), and the disease was effectively lethal in pre-modern societies. The incidence of the disease varies greatly between ethnic groups, but is highest in Caucasian populations.

Although over 1500 mutations are known in the CFTR gene, by far the most common mutant is DF508. This mutant is being kept at a high level in the population despite the lethal or near-lethal effects of the mutant homozygote. It seems that some kind of heterozygote advantage is operating. Early theories that the heterozygotes might enjoy increased fertility have not been borne out. Present indications are that the bacterium which causes typhoid fever enters cells using CFTR, and experiments with mice suggest that heterozygotes are resistant to the disease. If the same were true in humans, then heterozygotes would have had

an advantage during typhoid epidemics. Cystic fibrosis is a prime target for gene therapy research.

3.1.6 Human taste morphisms

A famous puzzle in human genetics is the genetic ability to taste phenylthiocarbamide (phenylthiourea or PTC), a morphism which was discovered in 1931. This substance, which to some of us is bitter, and to others tasteless, is of no great significance in itself, yet it is a genetic dimorphism. Because of its high frequency (which varies in different ethnic groups) it must be connected to some function of selective value. The ability to taste PTC itself is correlated with the ability to taste other bitter substances, many of which are toxic. Indeed, PTC itself is toxic, though not at the level of tasting it on litmus. Variation in PTC perception may reflect variation in dietary preferences throughout human evolution, and might correlate with susceptibility to diet-related diseases in modern populations. There is a statistical correlation between PTC tasting and liability to thyroid disease.

Fisher, Ford and Huxley tested orangutans and chimpanzees for PTC perception with positive results, thus demonstrating the long-standing existence of this dimorphism.[48] The recently identified PTC gene, which accounts for 85% of the tasting variance, has now been analysed for sequence variation with results which suggest selection is maintaining the morphism.

3.1.7 Lactose tolerance/intolerance

The ability to metabolize lactose, a sugar found in milk and other dairy products, is a prominent dimorphism that has been linked to recent human evolution.

3.1.8 MHC molecules

The genes of the major histocompatibility complex (MHC) are highly polymorphic, and this diversity plays a very important role in resistance to pathogens. This is true for other species as well.

4.0 Polymorphism in lower organisms

4.1 The Cuckoo

Over fifty species in this family of birds practise brood parasitism; the details are best seen in the British or European cuckoo (*Cuculus canorus*). The female lays 15–20 eggs in a season, but only one in each nest of another bird. She removes some or all of the host's clutch of eggs, and lays an egg which closely matches the host eggs. Although, in Britain, the hosts are always smaller than the cuckoo itself, the eggs she lays are small, and coloured to match the host clutch but thick-shelled. This latter is a defence which protects the egg if the host detects the fraud.

The intruded egg develops exceptionally quickly; when the newly-hatched cuckoo is only ten hours old, and still blind, it exhibits an urge to eject the other eggs or nestlings. It rolls them into a special depression on its back and heaves them out of the nest. The cuckoo nestling is apparently able to pressure the host adults for feeding by mimicking the cries of the host nestlings. The diversity of the cuckoo's eggs is extraordinary, the forms resembling those of its most usual hosts. In Britain these are:

- Meadow pipit (*Anthus pratensis*): brown eggs speckled with darker brown.
- European robin (*Erithacus rubecula*): whitish-grey eggs speckled with bright red.
- Reed warbler (*Acrocephalus scirpensis*): light dull green eggs blotched with olive.

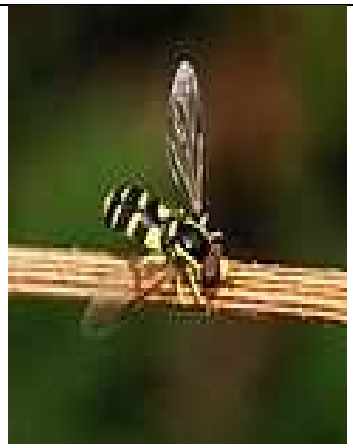
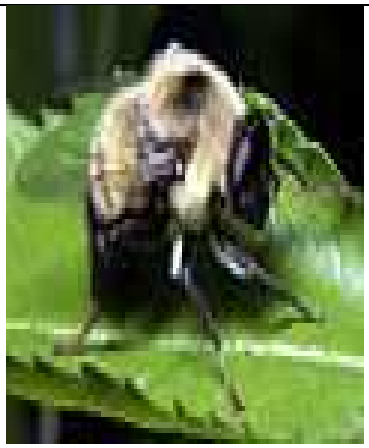
- Redstart (*Phoenicurus phoenicurus*): clear blue eggs.
- Hedge sparrow (*Prunella modularis*): clear blue eggs, unmarked, not mimicked. This bird is an uncritical fosterer; it tolerates in its nest eggs that do not resemble its own.

Each female cuckoo lays one type only; the same type laid by her mother. In this way female cuckoos are divided into groups (known as *gentes*, singular *gens*), each parasitises the host to which it is adapted. The male cuckoo has its own territory, and mates with females from any gens; thus the population (all gentes) is interbreeding.

The standard explanation of how the inheritance of gens works is as follows. The egg colour is inherited by sex chromosome. In birds sex determination is ZZ/ZW, and unlike mammals, the heterogametic sex is the female. The determining gene (or super-gene) for the inheritance of egg colour is believed to be carried on the W chromosome, which is directly transmitted in the female line. The female behaviour in choosing the host species is set by imprinting after birth, a common mechanism in bird behaviour.

Ecologically, the system of multiple hosts protects host species from a critical reduction in numbers, and maximises the egg-laying capacity of the population of cuckoos. It also extends the range of habitats where the cuckoo eggs may be raised successfully. Detailed work on the Cuckoo started with E. Chance in 1922, and continues to the present day; in particular, the inheritance of gens is still a live issue.

4.2 Hover fly polymorphism

		
Xanthogramma pedissiquum, a wasp mimic	*Volucella zonaria*, a large bumblebee mimic	*Mallota* sp., a bumblebee mimic

Hoverfly mimics can be seen in almost any garden in the temperate zone. The Syrphidae are a large (5600+ species) family of flies; their imagos feed on nectar and pollen, and are well known for their mimicry of social hymenoptera. The mimicry is Batesian in nature: hoverflies are palatable but hymenoptera are generally unpalatable and may also be protected by stings and/or armour.

Many social wasp (Vespidae) species exhibit Müllerian mimicry, where a group of unpalatable species benefit from sharing the same kind of warning (aposematic) colouration. Wasps are decidedly noxious: nasty-tasting and with a painful sting. They form a Mullerian 'ring' of similarly coloured models; the wasps are often accompanied by clusters of hover-fly mimics, who tend to arrive at the flowers at a similar time of day, and whose flight pattern is passably similar to wasp flight.

Observers in a garden can see for themselves that hoverfly mimics are quite common, usually many times more common than the models, and are relatively poor mimics, often easy to

distinguish from real wasps. However, it has been established in other cases that imperfect mimicry can confer significant advantage to the mimic, especially if the model is really noxious. Also, not only is polymorphism absent from these mimics, it is absent in the wasps also: these facts are presumably connected.

The situation with bumblebees (*Bombus*) is rather different. They too are unpalatable, in the sense of being difficult to eat: their body is covered with setae and is armoured; they are sometimes described as being 'non-food'. Mostler in 1935 carried out tests of their palatability: with the exception of specialist bee-eaters, adults of 19 species of birds ate only 2% of 646 bumblebees presented to them. After various trials, Mostler attributed their avoidance mainly to mechanical difficulties in handling: one young bird took 18 minutes to subdue, kill and eat a bumblebee.

Bumblebees form Mullerian rings of species, and they do often exhibit polymorphism. The hoverfly species mimicking bumblebees are generally accurate mimics, and many of their species are polymorphic. Many of the polymorphisms are different between the sexes, for example by the mimicry being limited to one sex only.

The question is, how can the differences between social wasp mimics and bumblebee mimics be explained? Evidently if model species are common, and have overlapping distributions, they are less likely to be polymorphic. Their mimics are widespread and develop a kind of rough and ready jack-of-all-trades mimicry. But if model species are less common and have patchy distribution they develop polymorphism; and their mimics match them more exactly and are polymorphic also. The issues are currently being investigated.

4.3 Scarlet tiger moth

The scarlet tiger moth *Callimorpha* (*Panaxia*) *dominula* (family Arctiidae) occurs in continental Europe, western Asia and southern England. It is a day-flying moth, noxious-tasting, with brilliant warning colour in flight, but cryptic at rest. The moth is colonial in habit, and prefers marshy ground or hedgerows. The preferred food of the larvae is the herb Comfrey (*Symphytum officinale*). In England it has one generation per year.

Callimorpha dominula morpha *typica* with spread wings. The red with black rear wings, revealed in flight, warn of its noxious taste. The front wings are cryptic, covering the rear wings at rest. Here the moth is resting but alert, and has jinked the front wings forward to reveal the warning flash.

The moth is known to be polymorphic in its colony at Cothill, about five miles (8 km) from Oxford, with three forms: the typical homozygote; the rare homozygote (*bimacula*) and the heterozygote (*medionigra*). It was studied there by Ford and later by Sheppard and their co-workers over many years. Data is available from 1939 to the present day, got by the usual field method of capture-mark-release-recapture and by genetic analysis from breeding in captivity. The records cover gene frequency and population-size for much of the twentieth century.

In this instance the genetics appears to be simple: two alleles at a single locus, producing the three phenotypes. Total captures over 26 years 1939-64 came to 15,784 homozygous

dominula (i.e. *typica*), 1,221 heterozygous *medionigra* and 28 homozygous *bimacula*. Now, assuming equal viability of the genotypes 1,209 heterozygotes would be expected, so the field results do *not* suggest any heterozygous advantage. It was Sheppard who found that the polymorphism is maintained by selective mating: each genotype preferentially mates with other morphs. This is sufficient to maintain the system despite the fact that in this case the heterozygote has slightly lower viability.

4.4 Peppered moth

The peppered moth, *Biston betularia*, is justly famous as an example of a population responding in a heritable way to a significant change in their ecological circumstances. E.B. Ford described peppered moth evolution as "one of the most striking, though not the most profound, evolutionary changes ever actually witnessed in nature".

Although the moths are cryptically camouflaged and rest during the day in unexposed positions on trees, they are predated by birds hunting by sight. The original camouflage (or crypsis) seems near-perfect against a background of lichen growing on trees. The sudden growth of industrial pollution in the nineteenth century changed the effectiveness of the moths' camouflage: the trees became blackened by soot, and the lichen died off. In 1848 a dark version of this moth was found in the Manchester area. By 1895 98% of the Peppered Moths in this area were black. This was a rapid change for a species that has only one generation a year.

Biston betularia morpha *typica*, the standard light-coloured Peppered Moth.

Biston betularia morpha *carbonaria*, the melanic Peppered Moth.

In Europe, there are three morphs: the typical white morph (*betularia* or *typica*), and *carbonaria*, the melanic black morph. They are controlled by alleles at one locus, with the carbonaria being dominant. There is also an intermediate or semi-melanic morph *insularia*, controlled by other alleles (see Majerus 1998).

A key fact, not realised initially, is the advantage of the heterozygotes, which survive better than either of the homozygotes. This affects the caterpillars as well as the moths, in spite of the caterpillars being monomorphic in appearance (they are twig mimics). In practice heterozygote advantage puts a limit to the effect of selection, since neither homozygote can reach 100% of the population. For this reason, it is likely that the carbonaria allele was in the

population originally, pre-industrialisation, at a low level. With the recent reduction in pollution, the balance between the forms has already shifted back significantly.

Another interesting feature is that the carbonaria had noticeably darkened after about a century. This was seen quite clearly when specimens collected about 1880 were compared with specimens collected more recently: clearly the dark morph has been adjusted by the strong selection acting on the gene complex. This might happen if a more extreme allele was available at the same locus; or genes at other loci might act as modifiers. We do not, of course, know anything about the genetics of the original melanics from the nineteenth century.

This type of industrial melanism has only affected such moths as obtain protection from insect-eating birds by resting on trees where they are concealed by an accurate resemblance to their background (over 100 species of moth in Britain with melanic forms were known by 1980). No species which hide during the day, for instance, among dead leaves, is affected, nor has the melanic change been observed among butterflies.

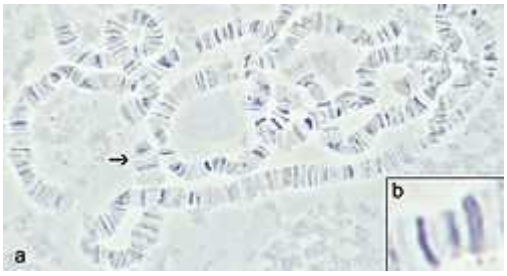
This is, as shown in many textbooks, "evolution in action". Much of the early work was done by Bernard Kettlewell, whose methods came under scrutiny later on. The entomologist Michael Majerus discussed criticisms made of Kettlewell's experimental methods in his 1998 book *Melanism: Evolution in Action*. This book was misrepresented in some reviews, and the story picked up by creationist campaigners. Judith Hooper, in her controversial book *Of Moths and Men* (2002), implied that Kettlewell's work was fraudulent or incompetent. Careful studies of Kettlewell's surviving papers by Rudge (2005) and Young (2004) found that Hooper's accusation of fraud was unjustified, and that "Hooper does not provide one shred of evidence to support this serious allegation". Majerus himself described *Of Moths and Men* as "littered with errors, misrepresentations, misinterpretations and falsehoods".A

suitably restrained 2004 summary of opinion mostly favoured predation as the main selective force. Starting in 2000, Majerus conducted a detailed seven year study of moths, experimenting to assess the various criticisms. He concluded that that differential bird predation was a major factor responsible for the decline in *carbonaria* frequency compared to *typica* in Cambridge during the study period, and described his results as a complete vindication of the peppered moth story. He said, "If the rise and fall of the peppered moth is one of the most visually impacting and easily understood examples of Darwinian evolution in action, it should be taught. It provides after all the proof of evolution."

Current interpretation of the available evidence is that the peppered moth is in fact a valid example of natural selection and adaptation. It illustrates a polymorphic species maintaining adaptation to a varied and sometimes changing environment.

4.5 Chromosome polymorphism in *Drosophila*

In the 1930s Dobzhansky and his co-workers collected *Drosophila pseudoobscura* and *D. persimilis* from wild populations in California and neighbouring states. Using Painter's technique they studied the polytene chromosomes and discovered that the wild populations were polymorphic for chromosomal inversions. All the flies look alike whatever inversions they carry: this is an example of a cryptic polymorphism. Accordingly, Dobzhansky favoured the idea that the morphs became fixed in the population by means of Sewall Wright's drift. However, evidence rapidly accumulated to show that natural selection was responsible:



Drosophila polytene chromosome

1. Values for heterozygote inversions of the third chromosome were often much higher than they should be under the null assumption: if no advantage for any form the number of heterozygotes should conform to N_s (number in sample) = $p^2+2pq+q^2$ where 2pq is the number of heterozygotes (see Hardy-Weinberg equilibrium).

2. Using a method invented by l'Heretier and Teissier, Dobzhansky bred populations in *population cages*, which enabled feeding, breeding and sampling whilst preventing escape. This had the benefit of eliminating migration as a possible explanation of the results. Stocks containing inversions at a known initial frequency can be maintained in controlled conditions. It was found that the various chromosome types do not fluctuate at random, as they would if selectively neutral, but adjust to certain frequencies at which they become stabilised. With *D. persimilis* he found that the caged population followed the values expected on the Hardy-Weinberg equilibrium when conditions were optimal (which disproved any idea of non-random mating), but with a restricted food supply heterozygotes had a distinct advantage.

3. Different proportions of chromosome morphs were found in different areas. There is, for example, a polymorph-ratio cline in *D. robusta* along an 18-mile (29 km) transect near Gatlinburg, TN passing from 1,000 feet (300 m) to 4,000 feet. Also, the same areas sampled at different times of year yielded significant differences in the proportions of forms. This indicates a regular cycle of changes which adjust the population to the seasonal conditions. For these results selection is by far the most likely explanation.

4. Lastly, morphs cannot be maintained at the high levels found simply by mutation, nor is drift a possible explanation when population numbers are high.

By the time Dobzhansky published the third edition of his book in 1951, he was persuaded that the chromosome morphs were being maintained in the population by the selective advantage of the heterozygotes, as with most polymorphisms. Later he made yet another interesting discovery. One of the inversions, known as PP, was quite rare up to 1946, but by 1958 its proportion had risen to 8%. Not only that, but the proportion was similar over an area of some 200,000 square miles (520,000 km^2) in California. This cannot have happened by migration of PP morphs from, say, Mexico (where the inversion is common) because the rate of dispersal (at less than 2 km/year) is of the wrong order. The change therefore reflected a change in prevailing selection whose basis was not yet known.

5.0 Conclusion

In this unit the student learnt the following:

- Sexual polymorphism
- Human polymorphism
- Polymorphism in lower organisms

6.0 Summary

Polymorphism is a condition in which a population possesses more than one allele at a locus. Sometimes it is defined as the condition of having more than one allele with a frequency of over 5% in the population.

There may be several causes of polymorphism:

• Polymorphism can be maintained by a balance between variation created by new mutations and natural selection (see mutational load).

• Genetic variation may be caused by frequency-dependent selection.

• Multiple niche polymorphism exists when different genotypes should have different fitnesses in different niches.

• Heterozygous advantage may maintain alleles which would otherwise be selected against.

• If selection is operating, migration can introduce polymorphism into a population.

These are all sources of polymorphism which make use of the mechanisms of natural selection. Genetic drift is also a possible source of genetic variation.

7.0 Reference/Further Reading

Gillespie J.G. 2004. *Population genetics: a concise guide*. 2nd ed, Johns Hopkins University Press, Baltimore.

Geodakyan V. A. (2000). "Evolutionary chromosomes and evolutionary sex dimorphism". *Biology Bulletin* **27**: 99–113.

Fisher, Ronald. 1930. *The Genetical Theory of Natural Selection*

Hamilton, W. D. 2002. *Narrow Roads of Gene Land, Vol. 2: Evolution of Sex*. Oxford: Oxford U. Pr.

Smith, John Maynard. 1978. *The Evolution of Sex*. Cambridge: Cambridge U. Pr.

Cooke G. S., Hill A. V. S. (2001). "Genetics of susceptibility to human infectious disease". *Nat. Rev. Genet* **2** (12): 967–977. doi:10.1038/35103577. PMID 11733749.

Sykes, B. 1999. *The human inheritance: genes, language and evolution*. Oxford: Oxford U. Pr.

Clarke, Cyril A. 1964. *Genetics for the Clinician*. Oxford: Blackwell

Polymorphism. http://www.blackwellpublishing.com/ridley/

MODULE 4: VARIATION; TYPES AND CAUSES

UNIT 1: EVOLUTION AND VARIATION

1.0 Introduction

Evolution is the cornerstone of modern biology. It unites all the fields of biology under one theoretical umbrella. It is not a difficult concept, but very few people -- the majority of biologists included -- have a satisfactory grasp of it. One common mistake is believing that species can be arranged on an evolutionary ladder from bacteria through "lower" animals, to "higher" animals and, finally, up to man. Mistakes permeate popular science expositions of evolutionary biology. Mistakes even filter into biology journals and texts. For example, Lodish, et. al., in their cell biology text, proclaim, "It was Charles Darwin's great insight that organisms are all related in a great chain of being..." In fact, the idea of a great chain of being, which traces to Linnaeus, was overturned by Darwin's idea of common descent.

Misunderstandings about evolution are damaging to the study of evolution and biology as a whole. People who have a general interest in science are likely to dismiss evolution as a soft science after absorbing the pop science nonsense that abounds. The impression of it being a soft science is reinforced when biologists in unrelated fields speculate publicly about evolution. Evolution is a change in the gene pool of a population over time. A gene is a

hereditary unit that can be passed on unaltered for many generations. The gene pool is the set of all genes in a species or population.

The English moth, *Biston betularia*, is a frequently cited example of observed evolution. [evolution: a change in the gene pool] In this moth there are two color morphs, light and dark. H. B. D. Kettlewell found that dark moths constituted less than 2% of the population prior to 1848. The frequency of the dark morph increased in the years following. By 1898, the 95% of the moths in Manchester and other highly industrialized areas were of the dark type. Their frequency was less in rural areas. The moth population changed from mostly light colored moths to mostly dark colored moths. The moths' color was primarily determined by a single gene. So, the change in frequency of dark colored moths represented a change in the gene pool. This change was, by definition, evolution.

The increase in relative abundance of the dark type was due to natural selection. The late eighteen hundreds was the time of England's industrial revolution. Soot from factories darkened the birch trees the moths landed on. Against a sooty background, birds could see the lighter colored moths better and ate more of them. As a result, more dark moths survived until reproductive age and left offspring. The greater number of offspring left by dark moths is what caused their increase in frequency. This is an example of natural selection.

Populations evolve. In order to understand evolution, it is necessary to view populations as a collection of individuals, each harboring a different set of traits. A single organism is never typical of an entire population unless there is no variation within that population. Individual organisms do not evolve, they retain the same genes throughout their life. When a population is evolving, the ratio of different genetic types is changing -- each individual organism within a population does not change. For example, in the previous example, the frequency of black moths increased; the moths did not turn from light to gray to dark in concert. The process of

evolution can be summarized in three sentences: Genes mutate. Individuals are selected. Populations evolve.

Evolution can be divided into microevolution and macroevolution. The kind of evolution documented above is microevolution. Larger changes, such as when a new species is formed, are called macroevolution. Some biologists feel the mechanisms of macroevolution are different from those of microevolutionary change. Others think the distinction between the two is arbitrary -- macroevolution is cumulative microevolution.

The word evolution has a variety of meanings. The fact that all organisms are linked via descent to a common ancestor is often called evolution. The theory of how the first living organisms appeared is often called evolution. This should be called abiogenesis. And frequently, people use the word evolution when they really mean natural selection -- one of the many mechanisms of evolution.

2.0 0bjectives

At the end of this unit, the students should be able to:

- Understand the basis for variation: Evolution
- Describe variation
- Highlight the importance of natural selection

3.0 Natural Selection

In elephants, as in cod, many individuals die between egg and adult; they both have excess fecundity. This excess fecundity exists because the world does not contain enough resources to support all the eggs that are laid and all the young that are born. The world contains only limited amounts of food and space. A population may expand to some extent, but logically

there will come a point beyond which the food supply must limit its further expansion. As resources are used up, the death rate in the population increases, and when the death rate equals the birth rate the population will stop growing. Organisms, therefore, in an ecological sense compete to survive and reproduce a both directly, for example by defending territories, and indirectly, for example by eating food that could otherwise be eaten by another individual. The actual competitive factors limiting the sizes of real populations make up a major area of ecological study. Various factors have been shown to operate. What matters here, however, is the general point that the members of a population, and members of different species, compete in order to survive. This competition follows from the conditions of limited resources and excess fecundity. Darwin referred to this ecological competition as the "struggle for existence." The expression is metaphorical: it does not imply a physical fight to survive, though fights do sometimes happen. The struggle for existence takes place within a web of ecological relations. Above an organism in the ecological food chain there will be predators and parasites, seeking to feed off it. Below it are the food resources it must in turn consume in order to stay alive. At the same level in the chain are competitors that may be competing for the same limited resources of food, or space.

An organism competes most closely with other members of its own species, because they have the most similar ecological needs to its own. Other species, in decreasing order of ecological similarity, also compete and exert a negative influence on the organism's chance of survival. Only the successful competitors will reproduce themselves. An organism competes most closely with other members of its own species, because they have the most similar ecological needs to its own. Other species, in decreasing order of ecological similarity, also compete and exert a negative influence on the organism's chance of survival. Only the successful competitors will reproduce themselves.

3.1 Variation

An individual organism's phenotype results from both its genotype and the influence from the environment it has lived in. A substantial part of the variation in phenotypes in a population is caused by the differences between their genotypes. The modern evolutionary synthesis defines evolution as the change over time in this genetic variation. The frequency of one particular allele will become more or less prevalent relative to other forms of that gene. Variation disappears when a new allele reaches the point of fixation — when it either disappears from the population or replaces the ancestral allele entirely.

Natural selection will only cause evolution if there is enough genetic variation in a population. Before the discovery of Mendelian genetics, one common hypothesis was blending inheritance. But with blending inheritance, genetic variance would be rapidly lost, making evolution by natural selection implausible. The *Hardy-Weinberg principle* provides the solution to how variation is maintained in a population with Mendelian inheritance. The frequencies of alleles (variations in a gene) will remain constant in the absence of selection, mutation, migration and genetic drift.

Variation comes from mutations in genetic material, reshuffling of genes through sexual reproduction and migration between populations (gene flow). Despite the constant introduction of new variation through mutation and gene flow, most of the genome of a species is identical in all individuals of that species. However, even relatively small differences in genotype can lead to dramatic differences in phenotype: for example, chimpanzees and humans differ in only about 5% of their genomes.

3.2 Types of variation

How much, and with respect to what characters, do natural populations show variation and, in particular, variation in fitness? Let us consider biological variation through a series of levels of organization, beginning with the organism's morphology, and working down to more microscopic levels. The purpose of this section is to give examples of variation, to show how variation can be seen in almost all the properties of living things, and to introduce some of the methods (particularly molecular methods) that we shall meet again and that are used to study variation.

3.2.1 Genetic Variation

Evolution requires genetic variation. If there were no dark moths, the population could not have evolved from mostly light to mostly dark. In order for continuing evolution there must be mechanisms to increase or create genetic variation and mechanisms to decrease it. Mutation is a change in a gene. These changes are the source of new genetic variation. Natural selection operates on this variation.

3.2.2 Morphological Level

At the morphological level, the individuals of a natural population will be found to vary for almost any character we may measure. In some characters, like body size, every individual differs from every other individual; this is called continuous variation. Other morphological characters show discrete variation as they fall into a limited number of categories. Sex, or gender, is an obvious example, with some individuals of a population being female, others male. This kind of categorical variation is found in other characters too. A population that contains more than one recognizable form is *polymorphic* (the condition is called polymorphism). There can be any number of forms in real cases, and they can have any set of relative frequencies. With sex, there are usually two forms. In the peppered moth (*Biston betularia*), two main color forms are often distinguished, though real populations may contain

three or more As the number of forms in the population increases, the polymorphic, categorical kind of variation blurs into the continuous kind of variation.

3.2.3 Cellular Level

Variation is not confined to morphological characters. If we descend to a cellular character, such as the number and structure of the chromosomes, we again find variation. In the fruitfly *Drosophila melanogaster,* the chromosomes exist in giant forms in the larval salivary glands and they can be studied with a light microscope. They turn out to have characteristic banding patterns, and chromosomes from different individuals in a population have subtly varying banding patterns. One type of variant is called an *inversion,* in which the banding pattern a and therefore the order of genes a of a region of the chromosome is inverted. A population of fruitflies may be polymorphic for a number of different inversions. Chromosomal variation is less easy to study in species that lack giant chromosomal forms, but it is still known to exist. Populations of the Australian grasshopper *Keyacris scurra,* for example, may contain two (normal and inverted) forms for each of two chromosomes; that makes nine kinds of grasshopper in all because an individual may be homozygous or heterozygous for any of the four chromosomal types. The nine differ in size and viability Chromosomes can vary in other respects too. Individuals may vary in their number of chromosomes, for example. In many species, some individuals have one or more extra chromosomes, in addition to the normal number for the species. These “supernumerary”chromosomes, which are often called B chromosomes, have been particularly studied in maize and in grasshoppers. In the grasshopper *Atractomorpha australis,* normal individuals have 18 autosomes, but individuals have been found with from one to six supernumary chromosomes. The population is polymorphic with respect to chromosome number. Inversions and B chromosomes are just two kinds of chromosomal variation. There are other kinds too; but these are enough to make the point that individuals vary at the subcellular, as well as the morphological level.

3.2.4 Biochemical Level

The story is the same at the biochemical level, such as for proteins. Proteins are molecules made up of sequences of amino acid units. A particular protein, like human hemoglobin, has a particular characteristic sequence, which in turn determines the molecule's shape and properties. But do all humans have exactly the same sequence for hemoglobin, or any other protein? In theory, we could find out by taking the protein from several individuals and then working out the sequence in each of them; but it would be excessively laborious to do so. *Gel electrophoresis* is a much faster method. Gel electrophoresis works because different amino acids carry different electric charges. Different proteins and different variants of the same protein a have different net electric charges, because they have different amino acid compositions. If we place a sample of proteins (with the same molecular weight) in an electric field, those with the largest electric charges will move fastest. For the student of biological variation, the importance of the method is that it can reveal different variants of a particular type of protein. A good example is provided by a less well known protein than hemoglobin a the enzyme called alcohol dehydrogenase, in the fruitfly. Fruitflies, as their name suggests, lay their eggs in, and feed on, decaying fruit. They are attracted to rotting fruit because of the yeast it contains. Fruitflies can be collected almost anywhere in the world by leaving out rotting fruit as a lure; and drowned fruitflies are usually found in a glass of wine left out overnight after a garden party in the late summer. As fruit rots, it forms a number of chemicals, including alcohol, which is both a poison and a potential energy source. Fruitflies cope with alcohol by means of an enzyme called alcohol dehydrogenase. The enzyme is crucial. If the alcohol dehydrogenase gene is deleted from fruitflies, and those flies are then fed on mere 5% alcohol, "they have difficulty flying and walking, and finally, cannot stay on their feet" (quoted in Ashburner 1998). Gel electrophoresis reveals that, in most populations of the fruitfly *Drosophila melanogaster*, alcohol dehydrogenase comes in two main forms.

The two forms show up as different bands on the gel after the sample has been put on it, an electric current put across it for a few hours, and the position of the enzyme has been exposed by a specific stain. The two variants are called slow (*Adh-s*) or fast (*Adh-f*) according to how far they have moved in the time. The multiple bands show that the protein is polymorphic. The enzyme called alcohol dehydrogenase is actually a class of two polypeptides with slightly different amino acid sequences. Gel electrophoresis has been applied to a large number of proteins in a large number of species and different proteins show different degrees of variability. But the point for now is that many of these proteins have been found to be variable a extensive variation exists in proteins in natural

populations.

3.2.5 DNA Level

If variation is found in every organ, at every level, among the individuals of a population, variation will almost inevitably also be found at the DNA level too. The inversion polymorphisms of chromosomes that we met above, for example, are due to inversions of the DNA sequence. However, the most direct method of studying DNA variation is to sequence the DNA itself. Let us stay with alcohol dehydrogenase in the fruitfly. Kreitman (1983) isolated the DNA encoding alcohol dehydrogenase from 11 independent lines of *D. melanogaster* and individually sequenced them all. Some of the 11 had *Adh-f*, others *Adh-s*, and the difference between *Adh-f* and *Adh-s* was always due to a single amino acid difference (Thr or Lys at codon 192). The amino acid difference appears as a base difference in the DNA, but this was not the only source of variation at the DNA level. The DNA is even more variable than the protein study suggests. At the protein level, only the two main variants were found in the sample of 11 genes, but at the DNA level there were 11 different sequences with 43 different variable sites. The amount of variation that we find is therefore highest at the DNA level. At the level of gross morphology, a *Drosophila* with two *Adh-f* genes is

indistinguishable from one with two *Adh-s* genes; gel electrophoresis resolves two classes of fly; but at the DNA level, the two classes decompose into innumerable individual variants. Restriction enzymes provide another method of studying DNA variation. Restriction enzymes exist naturally in bacteria, and a large number a over 2,300 a of restriction enzymes are known. Any one restriction enzyme cuts a DNA strand wherever it has a particular sequence, usually of about 4–8 base pairs. The restriction enzyme called *EcoR1,* for instance, which is found in the bacterium *Escherichia coli,* recognizes the base sequence ...GAATTC... and cuts it between the initial G and the first A. In the bacterium, the enzymes help to protect against viral invasion by cleaving foreign DNA, but the enzymes can be isolated in the laboratory and used to investigate DNA sequences. Suppose the DNA of two individuals differs, and that one has the sequence GAATTC at a certain site whereas the other individual has another sequence such as GTATT. If the DNA of each individual is put with *EcoR1,* only that of the first individual will be cleaved. The difference can be detected in the length of the DNA fragments: the pattern of fragment lengths will differ for the two individuals. The variation is called *restriction fragment length polymorphism* and has been found in all populations that have been studied.

4.0 Conclusion

The student has learnt the following:

- Background history on the importance of evolution
- Variation
- Types of variation

5.0 Summary

Evolution has been called the cornerstone of biology, and for good reasons. It is possible to do research in biology with little or no knowledge of evolution. Most biologists do. But,

without evolution biology becomes a disparate set of fields. Evolutionary explanations pervade all fields in biology and brings them together under one theoretical umbrella.

We know that natural selection should optimize the existing genetic variation in a population to maximize reproductive success. This provides a framework for interpreting a variety of biological traits and their relative importance. For example, a signal intended to attract a mate could be intercepted by predators. Natural selection has caused a trade- off between attracting mates and getting preyed upon. If you assume something other than reproductive success is optimized, many things in biology would make little sense. Without the theory of evolution, life history strategies would be poorly understood.

Organisms are modified over time by cumulative natural selection. The numerous examples of jury- rigged design in nature are a direct result of this. The distribution of genetically based traits across groups is explained by splitting of lineages and the continued production of new traits by mutation. The traits are restricted to the lineages they arise in.

Details of the past also hold explanatory power in biology. Plants obtain their carbon by joining carbon dioxide gas to an organic molecule within their cells. This is called carbon fixation. The enzyme that fixes carbon is RuBP carboxlyase. Plants using C3 photosynthesis lose 1/3 to 1/2 of the carbon dioxide they originally fix. RuBP carboxlyase works well in the absence of oxygen, but poorly in its presence. This is because photosynthesis evolved when there was little gaseous oxygen present. Later, when oxygen became more abundant, the efficiency of photosynthesis decreased. Photosynthetic organisms compensated by making more of the enzyme. RuBP carboxylase is the most abundant protein on the planet partially because it is one of the least efficient.

Ecosystems, species, organisms and their genes all have long histories. A complete explanation of any biological trait must have two components. First, a proximal explanation -- how does it work? And second, an ultimate explanation -- what was it modified from? For centuries humans have asked, "Why are we here?" The answer to that question lies outside the realm of science. Biologists, however, can provide an elegant answer to the question, "How did we get here?"

6.0 Tutor-Marked Assignment

- What do you know about evolution?
- What is variation?
- Discuss natural selection.

7.0 Reference/ Further Readings

Futuyma, Douglas J. (1997). *Evolutionary Biology*. Sunderland, Mass.: Sinauer Associates.

Ridley, Mark. (2003). *Evolution*. Boston: Blackwell Scientific.

Hartl, Daniel L. & Andrew G. Clark. (1997). *Principles of Population Genetics*. Sunderland, Mass.: Sinauer Associates.

Crow, James F. & Motoo Kimura. (1970). *Introduction to Population Genetics Theory*. Edina, Minn.: Burgess Publishing Company.

Graur, Dan & Wen-Hsiung Li. (2000). *Fundamentals of Molecular Evolution*. Sunderland, Mass.: Sinauer Associates.

Lewontin, Richard C. (1974). *The Genetic Basis of Evolutionary Change*. New York: Columbia Univ. Press.

Gillespie, John H. (1997). *The Causes of Molecular Evolution*. New York: Oxford Univ. Press.

Golding, Brian, ed. (1994). *Non-Neutral Evolution*. Boston: Chapman and Hall.

Kimura, Motoo. (1983). *The Neutral Theory of Molecular Evolution*. Cambridge, U.K.: Cambridge Univ. Press.

Endler, John A. (1986). *Natural Selection in the Wild*. Princeton, N.J.: Princeton Univ. Press.

UNIT 2: GENETIC VARIATION

1.0 Introduction

In order for continuing evolution there must be mechanisms to increase or create genetic variation and mechanisms to decrease it. The mechanisms of evolution are mutation, natural selection, genetic drift, recombination and gene flow. I have grouped them into two classes -- those that decrease genetic variation and those that increase it.

2.0 Objective

At the end of this unit, the student should be able to:

- Discuss on mechanisms that affect genetic variation
- Discuss on conditions for natural selection
- Have an understanding of Mutation, alleles, recombination and Gene flow.

3.0 Mechanisms that Decrease Genetic Variation

3.1.1 Genetic variation has two components: allelic diversity and non- random associations of alleles. Alleles are different versions of the same gene. For example, humans can have A, B or O alleles that determine one aspect of their blood type. Most animals, including humans, are diploid -- they contain two alleles for every gene at every locus, one inherited from their

mother and one inherited from their father. Locus is the location of a gene on a chromosome. Humans can be AA, AB, AO, BB, BO or OO at the blood group locus. If the two alleles at a locus are the same type (for instance two A alleles) the individual would be called homozygous. An individual with two different alleles at a locus (for example, an AB individual) is called heterozygous. At any locus there can be many different alleles in a population, more alleles than any single organism can possess. For example, no single human can have an A, B and an O allele.

Considerable variation is present in natural populations. At 45 percent of loci in plants there is more than one allele in the gene pool. Any given plant is likely to be heterozygous at about 15 percent of its loci. Levels of genetic variation in animals range from roughly 15% of loci having more than one allele (polymorphic) in birds, to over 50% of loci being polymorphic in insects. Mammals and reptiles are polymorphic at about 20% of their loci - - amphibians and fish are polymorphic at around 30% of their loci. In most populations, there are enough loci and enough different alleles that every individual, identical twins excepted, has a unique combination of alleles.

Linkage disequilibrium is a measure of association between alleles of two different genes. If two alleles were found together in organisms more often than would be expected, the alleles are in linkage disequilibrium. If there two loci in an organism (A and B) and two alleles at each of these loci (A1, A2, B1 and B2) linkage disequilibrium (D) is calculated as D = f(A1B1) * f(A2B2) - f(A1B2) * f(A2B1) (where f(X) is the frequency of X in the population). [Loci (plural of locus): location of a gene on a chromosome] D varies between -1/4 and 1/4; the greater the deviation from zero, the greater the linkage. The sign is simply a consequence of how the alleles are numbered. Linkage disequilibrium can be the result of

physical proximity of the genes. Or, it can be maintained by natural selection if some combinations of alleles work better as a team.

Natural selection maintains the linkage disequilibrium between color and pattern alleles in *Papilio memnon*. [linkage disequilibrium: association between alleles at different loci] In this moth species, there is a gene that determines wing morphology. One allele at this locus leads to a moth that has a tail; the other allele codes for a untailed moth. There is another gene that determines if the wing is brightly or darkly colored. There are thus four possible types of moths: brightly colored moths with and without tails, and dark moths with and without tails. All four can be produced when moths are brought into the lab and bred. However, only two of these types of moths are found in the wild: brightly colored moths with tails and darkly colored moths without tails. The non-random association is maintained by natural selection. Bright, tailed moths mimic the pattern of an unpalatable species. The dark morph is cryptic. The other two combinations are neither mimetic nor cryptic and are quickly eaten by birds.

Assortative mating causes a non-random distribution of alleles at a single locus. [locus: location of a gene on a chromosome] If there are two alleles (A and a) at a locus with frequencies p and q, the frequency of the three possible genotypes (AA, Aa and aa) will be p^2, 2pq and q^2, respectively. For example, if the frequency of A is 0.9 and the frequency of a is 0.1, the frequencies of AA, Aa and aa individuals are: 0.81, 0.18 and 0.01. This distribution is called the Hardy-Weinberg equilibrium.

Non-random mating results in a deviation from the Hardy-Weinberg distribution. Humans mate assortatively according to race; we are more likely to mate with someone of own race than another. In populations that mate this way, fewer heterozygotes are found than would be predicted under random mating. A decrease in heterozygotes can be the result of mate choice,

or simply the result of population subdivision. Most organisms have a limited dispersal capability, so their mate will be chosen from the local population.

3.1.2 Conditions in Natural Selection

The excess fecundity, and consequent competition to survive in every species, provide the preconditions for the process Darwin called natural selection. Natural selection is easiest to understand, in the abstract, as a logical argument, leading from premises to conclusion. The argument, in its most general form, requires four conditions:

1. Reproduction. Entities must reproduce to form a new generation.
2. Heredity. The offspring must tend to resemble their parents: roughly speaking, "like must produce like."
3. Variation in individual characters among the members of the population. If we are studying natural selection on body size, then different individuals in the population must have different body sizes.
4. Variation in the *fitness* of organisms according to the state they have for a heritable character. In evolutionary theory, fitness is a technical term, meaning the average number of offspring left by an individual relative to the number of offspring left by an average member of the population. This condition therefore means that individuals in the population with some characters must be more likely to reproduce (i.e., have higher fitness) than others. (The evolutionary meaning of the term fitness differs from its athletic meaning.)

If these conditions are met for any property of a species, natural selection automatically results. And if any are not, it does not. Thus entities, like planets, that do not reproduce,

cannot evolve by natural selection. Entities that reproduce but in which parental characters are not inherited by their offspring also cannot evolve by natural selection. But when the four conditions apply, the entities with the property conferring higher fitness will leave more offspring, and the frequency of that type of entity will increase in the population. The evolution of drug resistance in HIV illustrates the process The usual form of HIV has a reverse transcriptase that binds to drugs called nucleoside inhibitors as well as the proper constituents of DNA (A, C, G, and T). In particular, one nucleoside inhibitor called 3TC is a molecular analog of C. When reverse transcriptase places a 3TC molecule, instead of a C, in a replicating DNA chain, chain elongation is stopped and the reproduction of HIV is also stopped. In the presence of the drug 3TC, the HIV population in a human body evolves a discriminating form of reverse transcriptase a a form that does not bind 3TCbut does bind C. The HIV has then evolved drug resistance. The frequency of the drug-resistant HIV increases from an undetectably low frequency at the time the drug is first given to the patient up to 100% about 3 weeks later. The increase in the frequency of drug-resistant HIV is almost certainly driven by natural selection. The virus satisfies all four conditions for natural selection to operate. The virus reproduces; the ability to resist drugs is inherited (because the ability is due to a genetic change in the virus); the viral population within one human body shows genetic variation in drug-resistance ability; and the different forms of HIV have different fitnesses. In a human AIDS patient who is being treated with a drug such as 3TC, the HIV with the right change of amino acid in their reverse transcriptase will reproduce better, produce more offspring virus like themselves, and increase in frequency. Natural selection favors them.

3.1.3 Natural selection explains both evolution and adaptation

When the environment of HIV changes, such that the host cell contains nucleoside inhibitors such as 3TC as well as valuable resources such as C, the population of HIV changes over time. In other words, the HIV population evolves. Natural selection produces evolution when the environment changes; it will also produce evolutionary change in a constant environment if a new form arises that survives better than the current form of the species. The process that operates in any AIDS patient on drug treatment has been operating in all life for 4,000 million years since life originated, and has driven much larger evolutionary changes over those long periods of time. Natural selection can not only produce evolutionary change, it can also cause a population to stay constant. If the environment is constant and no superior form arises in the population, natural selection will keep the population the way it is. Natural selection can explain both evolutionary change and the absence of change. Natural selection also explains adaptation. The drug resistance of HIV is an example of an adaptation The discriminatory reverse transcriptase enzyme enables HIV to reproduce in an environment containing nucleoside inhibitors. The new adaptation was needed because of the change in the environment. In the drug treated AIDS patient, a fast but undiscriminating reverse transcriptase was no longer adaptive. The action of natural selection to increase the frequency of the gene coding for a discriminating reverse transcriptase resulted in the HIV becoming adapted to its environment. Over time, natural selection generates adaptation. The theory of natural selection therefore passes the key test set by Darwin for a satisfactory theory of evolution.

3.1.4 Sexual Selection

In many species, males develop prominent secondary sexual characteristics. A few oft cited examples are the peacock's tail, coloring and patterns in male birds in general, voice calls in

frogs and flashes in fireflies. Many of these traits are a liability from the standpoint of survival. Any ostentatious trait or noisy, attention getting behavior will alert predators as well as potential mates. How then could natural selection favor these traits?

Natural selection can be broken down into many components, of which survival is only one. Sexual attractiveness is a very important component of selection, so much so that biologists use the term sexual selection when they talk about this subset of natural selection.

Sexual selection is natural selection operating on factors that contribute to an organism's mating success. Traits that are a liability to survival can evolve when the sexual attractiveness of a trait outweighs the liability incurred for survival. A male who lives a short time, but produces many offspring is much more successful than a long lived one that produces few. The former's genes will eventually dominate the gene pool of his species. In many species, especially polygynous species where only a few males monopolize all the females, sexual selection has caused pronounced sexual dimorphism. In these species males compete against other males for mates. The competition can be either direct or mediated by female choice. In species where females choose, males compete by displaying striking phenotypic characteristics and/or performing elaborate courtship behaviors. The females then mate with the males that most interest them, usually the ones with the most outlandish displays. There are many competing theories as to why females are attracted to these displays.

The good genes model states that the display indicates some component of male fitness. A good genes advocate would say that bright coloring in male birds indicates a lack of parasites. The females are cueing on some signal that is correlated with some other component of viability.

Selection for good genes can be seen in sticklebacks. In these fish, males have red coloration on their sides. Milinski and Bakker showed that intensity of color was correlated to both parasite load and sexual attractiveness. Females preferred redder males. The redness indicated that he was carrying fewer parasites.

Evolution can get stuck in a positive feedback loop. Another model to explain secondary sexual characteristics is called the runaway sexual selection model. R. A. Fisher proposed that females may have an innate preference for some male trait before it appears in a population. Females would then mate with male carriers when the trait appears. The offspring of these matings have the genes for both the trait and the preference for the trait. As a result, the process snowballs until natural selection brings it into check. Suppose that female birds prefer males with longer than average tail feathers. Mutant males with longer than average feathers will produce more offspring than the short feathered males. In the next generation, average tail length will increase. As the generations progress, feather length will increase because females do not prefer a specific length tail, but a longer than average tail. Eventually tail length will increase to the point were the liability to survival is matched by the sexual attractiveness of the trait and an equilibrium will be established. Note that in many exotic birds male plumage is often very showy and many species do in fact have males with greatly elongated feathers. In some cases these feathers are shed after the breeding season.

None of the above models are mutually exclusive. There are millions of sexually dimorphic species on this planet and the forms of sexual selection probably vary amongst them.

3.1.5 Genetic Drift

Allele frequencies can change due to chance alone. This is called genetic drift. Drift is a binomial sampling error of the gene pool. What this means is, the alleles that form the next

generation's gene pool are a sample of the alleles from the current generation. When sampled from a population, the frequency of alleles differs slightly due to chance alone.

Alleles can increase or decrease in frequency due to drift. The average expected change in allele frequency is zero, since increasing or decreasing in frequency is equally probable. A small percentage of alleles may continually change frequency in a single direction for several generations just as flipping a fair coin may, on occasion, result in a string of heads or tails. A very few new mutant alleles can drift to fixation in this manner.

In small populations, the variance in the rate of change of allele frequencies is greater than in large populations. However, the overall rate of genetic drift (measured in substitutions per generation) is independent of population size. If the mutation rate is constant, large and small populations lose alleles to drift at the same rate. This is because large populations will have more alleles in the gene pool, but they will lose them more slowly. Smaller populations will have fewer alleles, but these will quickly cycle through. This assumes that mutation is constantly adding new alleles to the gene pool and selection is not operating on any of these alleles.

Sharp drops in population size can change allele frequencies substantially. When a population crashes, the alleles in the surviving sample may not be representative of the precrash gene pool. This change in the gene pool is called the founder effect, because small populations of organisms that invade a new territory (founders) are subject to this. Many biologists feel the genetic changes brought about by founder effects may contribute to isolated populations developing reproductive isolation from their parent populations. In sufficiently small populations, genetic drift can counteract selection. Mildly deleterious alleles may drift to fixation.

Wright and Fisher disagreed on the importance of drift. Fisher thought populations were sufficiently large that drift could be neglected. Wright argued that populations were often divided into smaller subpopulations. Drift could cause allele frequency differences between subpopulations if gene flow was small enough. If a subpopulation was small enough, the population could even drift through fitness valleys in the adaptive landscape. Then, the subpopulation could climb a larger fitness hill. Gene flow out of this subpopulation could contribute to the population as a whole adapting. This is Wright's Shifting Balance theory of evolution.

Both natural selection and genetic drift decrease genetic variation. If they were the only mechanisms of evolution, populations would eventually become homogeneous and further evolution would be impossible.

3.2 Mechanisms that Increase Genetic Variation

Mutation is any change occurring in the message that a gene carries. Mutations mainly arise as copy errors when DNA is replicated at mitosis and meiosis. Darwinian evolution requires a constant supply of variation: much of it is supplied by mutation, and a mutation-selection balance can maintain a genetic polymorphism. The first major geneticist to study mutation

was H.J. Muller, who demonstrated it can be induced by X-rays. He also recognized that the rate of mutation in nature is extremely low, and that they are almost always deleterious to the fitness of the organism. The accumulation of deleterious mutations places a mutational load on the population. Mutations can occur at single base level or at chromosomal level. The effects of mutation can occasionally be very dramatic: some of these fruitflies have suffered mutations which alter the number of wings that develop.

Mutations are changes in the DNA sequence of a cell's genome. When mutations occur, they can either have no effect, alter the product of a gene, or prevent the gene from functioning. Based on studies in the fly *Drosophila melanogaster*, it has been suggested that if a mutation changes a protein produced by a gene, this will probably be harmful, with about 70% of these mutations having damaging effects, and the remainder being either neutral or weakly beneficial.

Mutations can involve large sections of a chromosome becoming duplicated (usually by genetic recombination), which can introduce extra copies of a gene into a genome. Extra copies of genes are a major source of the raw material needed for new genes to evolve. This is important because most new genes evolve within gene families from pre-existing genes that share common ancestors. For example, the human eye uses four genes to make structures that sense light: three for colour vision and one for night vision; all four are descended from a single ancestral gene.

New genes can be generated from an ancestral gene when a duplicate copy mutates and acquires a new function. This process is easier once a gene has been duplicated because it increases the redundancy of the system; one gene in the pair can acquire a new function while the other copy continues to perform its original function. Other types of mutations can even generate entirely new genes from previously noncoding DNA.

The generation of new genes can also involve small parts of several genes being duplicated, with these fragments then recombining to form new combinations with new functions. When new genes are assembled from shuffling pre-existing parts, domains act as modules with simple independent functions, which can be mixed together to produce new combinations with new and complex functions. For example, polyketide synthases are large enzymes that make antibiotics; they contain up to one hundred independent domains that each catalyze one step in the overall process, like a step in an assembly line. Mutation limits the rate of evolution. The rate of evolution can be expressed in terms of nucleotide substitutions in a lineage per generation. Substitution is the replacement of an allele by another in a population. This is a two step process: First a mutation occurs in an individual, creating a new allele. This allele subsequently increases in frequency to fixation in the population. The rate of evolution is $k = 2Nvu$ (in diploids) where k is nucleotide substitutions, N is the effective population size, v is the rate of mutation and u is the proportion of mutants that eventually fix in the population.

Mutation need not be limiting over short time spans. The rate of evolution expressed above is given as a steady state equation; it assumes the system is at equilibrium. Given the time frames for a single mutant to fix, it is unclear if populations are ever at equilibrium. A change in environment can cause previously neutral alleles to have selective values; in the short term evolution can run on "stored" variation and thus is independent of mutation rate. Other mechanisms can also contribute selectable variation. Recombination creates new combinations of alleles (or new alleles) by joining sequences with separate microevolutionary histories within a population. Gene flow can also supply the gene pool with variants. Of course, the ultimate source of these variants is mutation.

3.2.1 The Fate of Mutant Alleles

Mutation creates new alleles. Each new allele enters the gene pool as a single copy amongst many. Most are lost from the gene pool, the organism carrying them fails to reproduce, or reproduces but does not pass on that particular allele. A mutant's fate is shared with the genetic background it appears in. A new allele will initially be linked to other loci in its genetic background, even loci on other chromosomes. If the allele increases in frequency in the population, initially it will be paired with other alleles at that locus -- the new allele will primarily be carried in individuals heterozygous for that locus. The chance of it being paired with itself is low until it reaches intermediate frequency. If the allele is recessive, its effect won't be seen in any individual until a homozygote is formed. The eventual fate of the allele depends on whether it is neutral, deleterious or beneficial.

3.2.2 Neutral alleles

Most neutral alleles are lost soon after they appear. The average time (in generations) until loss of a neutral allele is 2(Ne/N) ln(2N) where N is the effective population size (the number of individuals contributing to the next generation's gene pool) and N is the total population size. Only a small percentage of alleles fix. Fixation is the process of an allele increasing to a frequency at or near one. The probability of a neutral allele fixing in a population is equal to its frequency. For a new mutant in a diploid population, this frequency is 1/2N.

If mutations are neutral with respect to fitness, the rate of substitution (k) is equal to the rate of mutation(v). This does not mean every new mutant eventually reaches fixation. Alleles are added to the gene pool by mutation at the same rate they are lost to drift. For neutral alleles that do fix, it takes an average of 4N generations to do so. However, at equilibrium there are multiple alleles segregating in the population. In small populations, few mutations appear

each generation. The ones that fix do so quickly relative to large populations. In large populations, more mutants appear over the generations. But, the ones that fix take much longer to do so. Thus, the rate of neutral evolution (in substitutions per generation) is independent of population size.

The rate of mutation determines the level of heterozygosity at a locus according to the neutral theory. Heterozygosity is simply the proportion of the population that is heterozygous. Equilibrium heterozygosity is given as $H = 4Nv/[4Nv+1]$ (for diploid populations). H can vary from a very small number to almost one. In small populations, H is small (because the equation is approximately a very small number divided by one). In (biologically unrealistically) large populations, heterozygosity approaches one (because the equation is approximately a large number divided by itself). Directly testing this model is difficult because N and v can only be estimated for most natural populations. But, heterozygosities are believed to be too low to be described by a strictly neutral model. Solutions offered by neutralists for this discrepancy include hypothesizing that natural populations may not be at equilibrium.

At equilibrium there should be a few alleles at intermediate frequency and many at very low frequencies. This is the Ewens- Watterson distribution. New alleles enter a population every generation, most remain at low frequency until they are lost. A few drift to intermediate frequencies, a very few drift all the way to fixation. In Drosophila pseudoobscura, the protein Xanthine dehydrogenase (Xdh) has many variants. In a single population, Keith, et. al., found that 59 of 96 proteins were of one type, two others were represented ten and nine times and nine other types were present singly or in low numbers.

3.2.3 Deleterious alleles

Deleterious mutants are selected against but remain at low frequency in the gene pool. In diploids, a deleterious recessive mutant may increase in frequency due to drift. Selection cannot see it when it is masked by a dominant allele. Many disease causing alleles remain at low frequency for this reason. People who are carriers do not suffer the negative effect of the allele. Unless they mate with another carrier, the allele may simply continue to be passed on. Deleterious alleles also remain in populations at a low frequency due to a balance between recurrent mutation and selection. This is called the mutation load.

3.2.4 Beneficial alleles

Most new mutants are lost, even beneficial ones. Wright calculated that the probability of fixation of a beneficial allele is 2s. (This assumes a large population size, a small fitness benefit, and that heterozygotes have an intermediate fitness. A benefit of 2s yields an overall rate of evolution: k=4Nvs where v is the mutation rate to beneficial alleles) An allele that conferred a one percent increase in fitness only has a two percent chance of fixing. The probability of fixation of beneficial type of mutant is boosted by recurrent mutation. The beneficial mutant may be lost several times, but eventually it will arise and stick in a population. (Recall that even deleterious mutants recur in a population.)

Directional selection depletes genetic variation at the selected locus as the fitter allele sweeps to fixation. Sequences linked to the selected allele also increase in frequency due to hitchhiking. The lower the rate of recombination, the larger the window of sequence that hitchhikes. Begun and Aquadro compared the level of nucleotide polymorphism within and between species with the rate of recombination at a locus. Low levels of nucleotide polymorphism within species coincided with low rates of recombination. This could be

explained by molecular mechanisms if recombination itself was mutagenic. In this case, recombination with also be correlated with nucleotide divergence between species. But, the level of sequence divergence did not correlate with the rate of recombination. Thus, they inferred that selection was the cause. The correlation between recombination and nucleotide polymorphism leaves the conclusion that selective sweeps occur often enough to leave an imprint on the level of genetic variation in natural populations.

One example of a beneficial mutation comes from the mosquito Culex pipiens. In this organism, a gene that was involved with breaking down organophosphates - common insecticide ingredients -became duplicated. Progeny of the organism with this mutation quickly swept across the worldwide mosquito population. There are numerous examples of insects developing resistance to chemicals, especially DDT which was once heavily used in this country. And, most importantly, even though "good" mutations happen much less frequently than "bad" ones, organisms with "good" mutations thrive while organisms with "bad" ones die out.

If beneficial mutants arise infrequently, the only fitness differences in a population will be due to new deleterious mutants and the deleterious recessives. Selection will simply be weeding out unfit variants. Only occasionally will a beneficial allele be sweeping through a population. The general lack of large fitness differences segregating in natural populations argues that beneficial mutants do indeed arise infrequently. However, the impact of a beneficial mutant on the level of variation at a locus can be large and lasting. It takes many generations for a locus to regain appreciable levels of heterozygosity following a selective sweep.

3.2.5 Recombination

Each chromosome in our sperm or egg cells is a mixture of genes from our mother and our father. Recombination can be thought of as gene shuffling. Most organisms have linear chromosomes and their genes lie at specific location (loci) along them. Bacteria have circular chromosomes. In most sexually reproducing organisms, there are two of each chromosome type in every cell. For instance in humans, every chromosome is paired, one inherited from the mother, the other inherited from the father. When an organism produces gametes, the gametes end up with only one of each chromosome per cell. Haploid gametes are produced from diploid cells by a process called meiosis.

In meiosis, homologous chromosomes line up. The DNA of the chromosome is broken on both chromosomes in several places and rejoined with the other strand. Later, the two homologous chromosomes are split into two separate cells that divide and become gametes. But, because of recombination, both of the chromosomes are a mix of alleles from the mother and father.

Recombination creates new combinations of alleles. Alleles that arose at different times and different places can be brought together. Recombination can occur not only between genes, but within genes as well. Recombination within a gene can form a new allele. Recombination is a mechanism of evolution because it adds new alleles and combinations of alleles to the gene pool.

3.2.6 Gene Flow

New organisms may enter a population by migration from another population. If they mate within the population, they can bring new alleles to the local gene pool. This is called gene

flow. In some closely related species, fertile hybrids can result from interspecific matings. These hybrids can vector genes from species to species.

Gene flow between more distantly related species occurs infrequently. This is called horizontal transfer. One interesting case of this involves genetic elements called P elements. Margaret Kidwell found that P elements were transferred from some species in the Drosophila willistoni group to Drosophila melanogaster. These two species of fruit flies are distantly related and hybrids do not form. Their ranges do, however, overlap. The P elements were vectored into D. melanogaster via a parasitic mite that targets both these species. This mite punctures the exoskeleton of the flies and feeds on the "juices". Material, including DNA, from one fly can be transferred to another when the mite feeds. Since P elements actively move in the genome (they are themselves parasites of DNA), one incorporated itself into the genome of a melanogaster fly and subsequently spread through the species. Laboratory stocks of melanogaster caught prior to the 1940's lack of P elements. All natural populations today harbor them.

4.0 Conclusion

In this unit the student learnt the following:

- Genetic variation
- Mechanisms that decrease genetic variation
- Natural selection
- Conditions for natural selection
- Sexual selection
- Genetic drift
- Mechanisms that increase genetic variation

- Mutation
- Alleles
- Recombination
- Gene flow

5.0 Summary

Evolution is a change in the gene pool of a population over time; it can occur due to several factors. Three mechanisms add new alleles to the gene pool: mutation, recombination and gene flow. Two mechanisms remove alleles, genetic drift and natural selection. Drift removes alleles randomly from the gene pool. Selection removes deleterious alleles from the gene pool. The amount of genetic variation found in a population is the balance between the actions of these mechanisms.

Natural selection can also increase the frequency of an allele. Selection that weeds out harmful alleles is called negative selection. Selection that increases the frequency of helpful alleles is called positive, or sometimes positive Darwinian, selection. A new allele can also drift to high frequency. But, since the change in frequency of an allele each generation is random, nobody speaks of positive or negative drift.

Except in rare cases of high gene flow, new alleles enter the gene pool as a single copy. Most new alleles added to the gene pool are lost almost immediately due to drift or selection; only a small percent ever reach a high frequency in the population. Even most moderately beneficial alleles are lost due to drift when they appear. But, a mutation can reappear numerous times.

The fate of any new allele depends a great deal on the organism it appears in. This allele will be linked to the other alleles near it for many generations. A mutant allele can increase in

frequency simply because it is linked to a beneficial allele at a nearby locus. This can occur even if the mutant allele is deleterious, although it must not be so deleterious as to offset the benefit of the other allele. Likewise a potentially beneficial new allele can be eliminated from the gene pool because it was linked to deleterious alleles when it first arose. An allele "riding on the coat tails" of a beneficial allele is called a hitchhiker. Eventually, recombination will bring the two loci to linkage equilibrium. But, the more closely linked two alleles are, the longer the hitchhiking will last.

The effects of selection and drift are coupled. Drift is intensified as selection pressures increase. This is because increased selection (i.e. a greater difference in reproductive success among organisms in a population) reduces the effective population size, the number of individuals contributing alleles to the next generation.

6.0 Tutor-Marked Assignment

- What is genetic variation?
- What are the mechanisms that affect genetic variation?

7.0 Reference/Further Reading

Eldredge, Niles. (1989). *Macroevolutionary Dynamics*. New York: McGraw-Hill.

Cowen, Richard. (2004). *History of Life*. Boston: Blackwell Scientific.

Dawkins, Richard. (1987). *The Blind Watchmaker*. New York: W.W. Norton.

Kitcher, Philip. (1982). *Abusing Science*. Cambridge, Mass.: MIT Press.

Wilson, Edward O. (1992). *The Diversity of Life*. Cambridge, Mass.: Harvard Belknap.

Darwin, Charles. (1859). *On the Origin of Species*.

Darwin, Charles. (1871). *The Descent of Man*.

Haldane, J.B.S. (1932). *The Causes of Evolution*. Princeton, N.J.: Princeton Univ. Press

(reprinted 1990).

Simpson, George G. (1944). *Tempo and Mode in Evolution*. New York: Columbia Univ. Press.

Mayr, Ernst E. (1982). *The Growth of Biological Thought*. Cambridge, Mass: Harvard Belknap.

Provine, William B. (2001). *The Origins of Theoretical Population Genetics*. Chicago: Univ. of

Chicago Press.

UNIT 3: RESHUFFLING OF GENES

1.0 Introduction

Genetic recombination is a process by which a molecule of nucleic acid (usually DNA, but can also be RNA) is broken and then joined to a different one. Recombination can occur between similar molecules of DNA, as in homologous recombination, or dissimilar molecules, as in non-homologous end joining. Recombination is a common method of DNA repair in both bacteria and eukaryotes.

2.0 Objectives

At the end of this unit, students should learn the following

- Genetic recombination
- Chromosomal crossover
- Sexual reproduction

3.0 Genetic Recombination

The shuffling of genes brought about by genetic recombination is thought to have many advantages, as it is a major engine of genetic variation and also allows sexually reproducing organisms to avoid Muller's ratchet, in which the genomes of an asexual population accumulate deleterious mutations in an irreversible manner.

In genetic engineering, recombination can also refer to artificial and deliberate recombination of disparate pieces of DNA, often from different organisms, creating what is called recombinant DNA. A prime example of such a use of genetic recombination is gene targeting, which can be used to add, delete or otherwise change an organism's genes. This

technique is important to biomedical researchers as it allows them to study the effects of specific genes. Techniques based on genetic recombination are also applied in protein engineering to develop new proteins of biological interest.

Genetic recombination is catalyzed by many different enzymes, called *recombinases*. RecA, the chief recombinase found in *Escherichia coli*, is responsible for the repair of DNA double strand breaks (DSBs). In yeast and other eukaryotic organisms there are two recombinases required for repairing DSBs. The RAD51 protein is required for mitotic and meiotic recombination, whereas the DMC1 protein is specific to meiotic recombination.

3.1 Chromosomal Crossover

Chromosomal crossover refers to recombination between the paired chromosomes inherited from each of one's parents, generally occurring during meiosis. During prophase I the four available chromatids are in tight formation with one another. While in this formation, homologous sites on two chromatids can mesh with one another, and may exchange genetic information.

Because recombination can occur with small probability at any location along chromosome, the frequency of recombination between two locations depends on their distance. Therefore, for genes sufficiently distant on the same chromosome the amount of crossover is high enough to destroy the correlation between alleles.

Tracking the movement of genes during crossovers has proven quite useful to geneticists. Because two genes that are close together are less likely to become separated than genes that are farther apart, geneticists can deduce roughly how far apart two genes are on a chromosome if they know the frequency of the crossovers. Geneticists can also use this

method to infer the presence of certain genes. Genes that typically stay together during recombination are said to be linked. One gene in a linked pair can sometimes be used as a marker to deduce the presence of another gene. This is typically used in order to detect the presence of a disease-causing gene.

3.2 Sexual Reproduction

In asexual organisms, genes are inherited together, or *linked*, as they cannot mix with genes of other organisms during reproduction. In contrast, the offspring of sexual organisms contain random mixtures of their parents' chromosomes that are produced through independent assortment. In a related process called homologous recombination, sexual organisms exchange DNA between two matching chromosomes. Recombination and reassortment do not alter allele frequencies, but instead change which alleles are associated with each other, producing offspring with new combinations of alleles. Sex usually increases genetic variation and may increase the rate of evolution.

Sexual reproduction is the creation of a new organism by combining the genetic material of two organisms. There are two main processes during sexual reproduction; they are: meiosis, involving the halving of the number of chromosomes; and fertilization, involving the fusion of two gametes and the restoration of the original number of chromosomes. During meiosis, the chromosomes of each pair usually cross over to achieve homologous recombination.

The evolution of sexual reproduction is a major puzzle. The first fossilized evidence of sexually reproducing organisms is from eukaryotes of the Stenian period, about 1 to 1.2 billion years ago. Sexual reproduction is the primary method of reproduction for the vast majority of macroscopic organisms, including almost all animals and plants. Bacterial

conjugation, the transfer of DNA between two bacteria, is often mistakenly confused with sexual reproduction, because the mechanics are similar.

Evolutionary thought proposes several explanations for why sexual reproduction developed out of former asexual reproduction. It may be due to selection pressure on the clade itself—the ability for a population to radiate more rapidly in response to a changing environment through sexual recombination than parthenogenesis allows. Also, sexual reproduction allows for the "ratcheting" of evolutionary speed as one clade competes with another for a limited resource

3.3 Plants

Animals typically produce male gametes called sperm, and emale gametes called eggs and ova, following immediately after meiosis, with the gametes produced directly by meiosis. Plants on the other hand have mitosis occurring in spores, which are produced by meiosis. The spores germinate into the gametophyte phase. The gametophytes of different groups of plants vary in size; angiosperms have as few as three cells in pollen, and mosses and other so called primitive plants may have several million cells. Plants have an alternation of generations where the sporophyte phase is succeeded by the gametophyte phase. The sporophyte phase produces spores within the sporangium by meiosis.

Flowering plants are the dominant plant form on land and they reproduce by sexual and asexual means. Often their most distinguishing feature is their reproductive organs, commonly called flowers. The anther produces male gametophytes, the sperm is produced in pollen grains, which attach to the stigma on top of a carpel, in which the female gametophytes (inside ovules) are located. After the pollen tube grows through the carpel's style, the sex cell nuclei from the pollen grain migrate into the ovule to fertilize the egg cell and endosperm nuclei within the female gametophyte in a process termed double fertilization.

The resulting zygote develops into an embryo, while the triploid endosperm (one sperm cell plus two female cells) and female tissues of the ovule give rise to the surrounding tissues in the developing seed. The ovary, which produced the female gametophyte(s), then grows into a fruit, which surrounds the seed(s). Plants may either self-pollinate or cross-pollinate. Nonflowering plants like ferns, moss and liverworts use other means of sexual reproduction.

Bryophytes, which include liverworts, hornworts and mosses, reproduce both sexually and vegetatively. They are small plants found growing in moist locations and like ferns, have motile sperm with flagella and need water to facilitate sexual reproduction. These plants start as a haploid spore that grows into the dominate form, which is a multicellular haploid body with leaf-like structures that photosynthesize. Haploid gametes are produced in antherida and archegonia by mitosis. The sperm released from the antherida respond to chemicals released by ripe archegonia and swim to them in a film of water and fertilize the egg cells thus producing a zygote. The zygote divides by mitotic division and grows into a sporophyte that is diploid. The multicellular diploid sporophyte produces structures called spore capsules, which are connected by seta to the archegonia. The spore capsules produce spores by meiosis, when ripe the capsules burst open and the spores are released. Bryophytes show considerable variation in their breeding structures and the above is a basic outline. Also in some species each plant is one sex while other species produce both sexes on the same plant.

3.4 Insects

Insect species make up more than two-thirds of all extant animal species, and most insect species use sex for reproduction, though some species are facultatively parthenogenetic. Many species have sexual dimorphism, while in others the sexes look nearly identical. Typically they have two sexes with males producing spermatozoa and females ova. The ova develop into eggs that have a covering called the chorion, which forms before internal

fertilization. Insects have very diverse mating and reproductive strategies most often resulting in the male depositing spermatophore within the female, which stores the sperm until she is ready for egg fertilization. After fertilization, and the formation of a zygote, and varying degrees of development; the eggs are deposited outside the female in many species, or in some, they develop further within the female and live born offspring are produced.

3.5 Mammals

There are three extant kinds of mammals: Monotremes, Placentals and Marsupials, all with internal fertilization. In placental mammals, offspring are born as juveniles: complete animals with the sex organs present although not reproductively functional. After several months or years, the sex organs develop further to maturity and the animal becomes sexually mature. Most female mammals are only fertile during certain periods during their estrous cycle, at which point they are ready to mate. Individual male and female mammals meet and carry out copulation. For most mammals, males and females exchange sexual partners throughout their adult lives.

Male

The male reproductive system contains two main divisions: the penis, and the testicles, the latter of which is where sperm are produced. In humans, both of these organs are outside the abdominal cavity, but they can be primarily housed within the abdomen in other animals (for instance, in dogs, the penis is internal except when mating). Having the testicles outside the abdomen best facilitates temperature regulation of the sperm, which require specific temperatures to survive. Sperm are the smaller of the two gametes and are generally very short-lived, requiring males to produce them continuously from the time of sexual maturity until death. Prior to ejaculation the produced sperm are stored in the epididymis. The sperm

cells are motile and they swim using tail-like flagella to propel themselves towards the ovum. The sperm follows temperature gradients (thermotaxis) and chemical gradients (chemotaxis) to locate the ovum.

Female

The female reproductive system likewise contains two main divisions: the vagina and uterus, which act as the receptacle for the sperm, and the ovaries, which produce the female's ova. All of these parts are always internal. The vagina is attached to the uterus through the cervix, while the uterus is attached to the ovaries via the Fallopian tubes. At certain intervals, the ovaries release an ovum, which passes through the fallopian tube into the uterus.

If, in this transit, it meets with sperm, the egg selects sperm with which to merge; this is termed fertilization. The fertilization usually occurs in the oviducts, but can happen in the uterus itself. The zygote then implants itself in the wall of the uterus, where it begins the processes of embryogenesis and morphogenesis. When developed enough to survive outside the womb, the cervix dilates and contractions of the uterus propel the fetus through the birth canal, which is the vagina.

The ova, which are the female sex cells, are much larger than the sperm and are normally formed within the ovaries of the fetus before its birth. They are mostly fixed in location within the ovary until their transit to the uterus, and contain nutrients for the later zygote and embryo. Over a regular interval, in response to hormonal signals, a process of oogenesis matures one ovum which is released and sent down the Fallopian tube. If not fertilized, this egg is released through menstruation in humans and other great apes, and reabsorbed in other mammals in the estrus cycle.

4.0 Conclusion

In this unit, students have learnt the following:

- Genetic recombination
- Chromosomal crossover
- Sexual reproduction

5.0 Summary

The process of recombination takes place when germ cells are produced, when large segments of DNA are exchanged between each pair of chromosomes. This kind of genetic shuffling means that any chromosome inherited is in fact a mosaic of chromosomes she inherited or transferred. This reshuffling increases the possible number of combinations of genetic variants, which in turn ensures greater variability of characteristics among individuals.

6.0 Tutor-Marked Assignment

- What is genetic recombination?
- What processes determine gene reshuffling?
- What are the possible outcomes of gene reshuffling?

7.0 Refernce/ Further reading

Radding C (1982). "Homologous pairing and strand exchange in genetic recombination". *Annu. Rev. Genet.* **16** (1): 405–37. doi:10.1146/annurev.ge.16.120182.002201. PMID 6297377.

Agrawal AF (2006). "Evolution of sex: why do organisms shuffle their genotypes?". *Curr. Biol.* **16** (17): R696. doi:10.1016/j.cub.2006.07.063. PMID 16950096.

Peters AD, Otto SP (2003). "Liberating genetic variance through sex". *Bioessays* **25** (6): 533–7. doi:10.1002/bies.10291. PMID 12766942.

Goddard MR, Godfray HC, Burt A (2005). "Sex increases the efficacy of natural selection in experimental yeast populations". *Nature* **434** (7033): 636–40. Bibcode 2005Natur.434..636G. doi:10.1038/nature03405. PMID 15800622.

N.J. Buttefield (2000). "Bangiomorpha pubescens n. gen., n. sp.: implications for the evolution of sex, multicellularity, and the Mesoproterozoic/Neoproterozoic radiation of eukaryotes". *Paleobiology* **26** (3): 386–404. doi:10.1666/0094-8373(2000)026<0386:BPNGNS>2.0.CO;2.

UNIT 4: POLYPLOIDY

Unit 3 Polyploidy

1.0 Introduction

Polyploidy is a term used to describe cells and organisms containing more than two paired (homologous) sets of chromosomes. Most eukaryotic species are diploid, meaning they have

two sets of chromosomes — one set inherited from each parent. However polyploidy is found in some organisms and is especially common in plants. In addition, polyploidy also occurs in some tissues of animals who are otherwise diploid, such as human muscle tissues. This is known as endopolyploidy. (Monoploid organisms also occur; a monoploid has only one set of chromosomes. These include the vast majority of prokaryotes.)

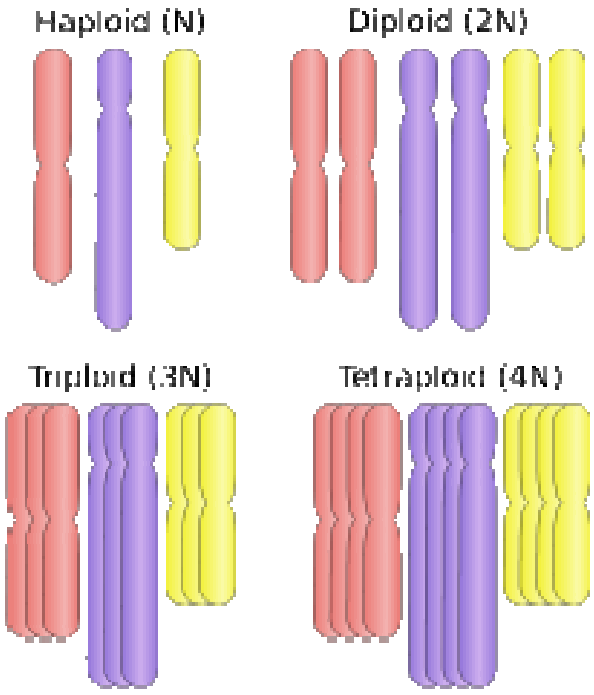
Polyploidy refers to a numerical change in a whole set of chromosomes. Organisms in which a particular chromosome, or chromosome segment, is under- or overrepresented are said to be aneuploid (from the Greek words meaning "not," "good," and "fold"). Therefore the distinction between aneuploidy and polyploidy is that aneuploidy refers to a numerical change in part of the chromosome set, whereas polyploidy refers to a numerical change in the whole set of chromosomes

2.0 Objective

At the end of this unit, students should be able to explain the following:

- Polyploidy
- Importance of Polyploidy
- Polyploidy in animals and plants

3.0 Types of polyploidy



This image shows haploid (single), diploid (double), triploid (triple), and tetraploid (quadruple) sets of chromosomes. Triploid and tetraploid chromosomes are examples of polyploidy.

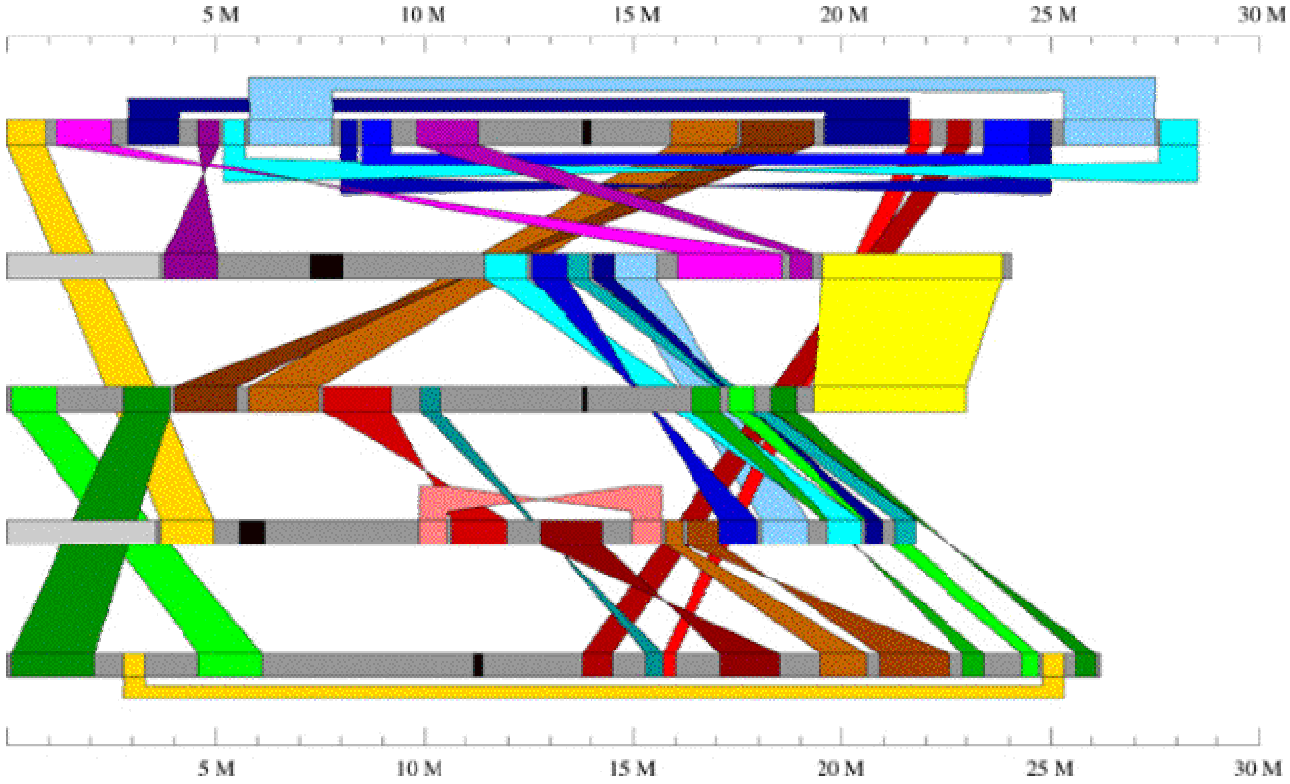
Two main modes of origin of the polyploid condition are recognized somatic doubling in mitosis, and nonreduction in meiosis (Heilborn, 1934; Grant, 1971). The mechanism of somatic doubling is exemplified by polyploid Primula kewensis, and nonreduction was the mode of origin seen in polyploid Rhaphobrassica. It used to be thought most that polyploids formed by hybridization followed by chromosome doubling. However, Harlan and deWet (1975) argued that unreduced gametes played an important role. While agronomy researchers took notice of this (e.g. Peloquin, 19XX), textbooks did not change. Recently, a lot of theoretical modeling (Rodriguez, 1996; Ramsey and Schemske, 1998, 2002) and fieldwork (Husband 1999, 2000) has contributed to the view that unreduced gametes and triploid bridges are a major source of polyploid formation. This is also a mechanism for how diploid and polyploid genomes can interact (thus, the new polyploid species are not strictly sealed off from its diploid progenitors).

During meiosis, homologous chromosomes pair and undergo crossing over resulting in the exchange of parts of their chromosomes. In diploid hybrids derived from crosses of two species, chromosomes from the two species may differ or one of the chromosomes may be absent. This can cause irregularities during meiosis and may result in cell cycle arrest and subsequent embryo abortion. However, if the chromosome number is doubled in the hybrid, allotetraploids are formed, which have four sets of chromosomes. This can occur by crossing autotetraploids of the two species, or more likely in nature, by the fussion of unreduced gametes. Allotetraploids generally will have pairing and crossing over only within the two chromosomes of each original parent (the homologous chromosomes AA) and only rarely between chromosomes from the two original parents (the homeologous chromosomes AA'). This meiotic behavior assures proper pairing of the chromosomes and the correct assortment into gametes

Polyploid types are labeled according to the number of chromosome sets in the nucleus:

- triploid (three sets; 3x), for example seedless watermelons, common in the phylum Tardigrada
- tetraploid (four sets; 4x), for example Salmonidae fish
- pentaploid (five sets; 5x), for example Kenai Birch (*Betula papyrifera* var. *kenaica*)
- hexaploid (six sets; 6x), for example wheat, kiwifruit
- octaploid (eight sets; 8x), for example *Acipenser* (genus of sturgeon fish), dahlias
- decaploid (ten sets; 10x), for example certain strawberries
- dodecaploid (twelve sets; 12x), for example the plant *Celosia argentea* and the amphibian *Xenopus ruwenzoriensis*
- To recognize polyploids, biologists have traditionally counted chromosomes and guess if taxa were diploid or polyploid with rules of thumb. For example, anything

with over a certain number of chromosomes, especially if it were multiple of the number, was assumed to be polyploid (refs/examples). Often, an increase in organ size (e.g., stomates) was used as a substitute measure for comparing genome size (Masterson, 1994). Today, biologists use genomic tools (molecular cytogenetics, genetic maps) to observe a continuum of diploids and polyploids in various states of genome evolution. Many species, such as Arabidopsis, Zea mays, and yeast (TAGI, 2000; Blanc et al., 2003; Langkjaer et al., 2003) are believed to have undergone a doubling of their genome in the past but now behave as diploids (Wendel, 2000), . These ancient polyploids (paleopolyploids) have undergone a multitude of genomic changes, such as deletions of large fragments of chromosomes, silencing of duplicate genes, and recombining of homoeologous chromosome segments (Wolfe, 2001; Levy and Feldman, 2002) some of which may lead the organism to a more diploid-like state. In some polyploids, chromosomal reorganization is so extensive that the genome is no longer structured as an allopolyploid. Paradoxically, the more extensive this process of diploidization, the more difficult it is to discern. For example, “diploid” Brassica can be considered ancient polyploids when compared to Arabidopsis, and Arabidopsis itself shows ancient duplications. Since plant genomes show cycles of polyploidization and diploidization, it can be difficult to determine whether gene duplication arose by polyploidy or some other mechanism.



According to Grant (1971, 1981), the phenomena of polyploidy was discovered during the exploratory phase of plant cytogenetics in the early years of the twentieth century. Winkler (1916) introduced the term polyploidy, and Winge (1917) proposed that polyploidy occurred by somatic doubling in species hybrids. Early polyploid studies included those in in Nicotiana (Clausen and Goodspeed 1925), Raphanus-Brassica (Karpechenko 1927) and Galeopsis (Muntzing 1930, 1932). The distinctions between autopolyploidy and allopolyploidy were made by Kihara and Ono (1926) and later elaborated on by Clausen, Keck, and Hiesey (1945) and Stebbins (1950,1971). Stebbins also proposed the category of segmental allopolyploids , which are essentially intermediate forms between auto- and allo- polyploids (Stebbins 1950). Harlan and deWet (1975) suggested that most polyploids arose through the production of unreduced gametes as oppossed to somatic doubling. Ramsey and Schemske (2002) review the controversies surrounding the confusion over whether to classify polyploids by mode of

origin criteria or by cytological criteria (Ramsey and Schemske, 2002). Using cytological criteria, allopolyploids display mostly bivalent chromosome pairing (aggregates containing two chromosomes) while autopolyploids can have higher frequencies of multivalent chromosome configurations. Here we follow Ramsey and Schemske (2002) and adopt mode of origin criteria: if the chromosomes of one genome within an organism or species are simply duplicated, the resulting polyploid is an autopolyploid. However, if genome duplication occurs during a cross of two different species, the resulting organism is referred to as an allopolyploid.

Polyploids can acquire variation both through mechanisms of population genetics (gene flow with diploids and multiple origins of polyploids), and through mechanisms that generate “de novo variation” such as chromosomal rearrangements and epigenetic phenomena.

Polyploidy has long been considered an important example of instant or sympatric speciation, since polyploid species are mostly reproductively isolated from their diploid progenitors (Stebbins 1950, 1971; Levin 1983). An interesting aspect related to allopolyploidization or hybridization of different species is the question of the “species barrier” when using a biological species concept. Members of the same biological species are commonly defined as related individuals of a population that can interbreed and whose offspring are fertile. Thus, the horse and a donkey are considered separate species because their hybrid offspring are viable but infertile. In plants, hybridization of different species is quite common and many of the well-known crop plants are allopolyploids resulting from inter-species hybrids. Such allopolyploids pose a challenge to phylogenetic species concepts, which define species on strict monophyletic criteria. Over the last decade this challenge has taken on additional relevance as “polyploid species” have been found to form repeatedly in close proximity to one another (Soltis and Soltis, 1993; 1999; 2000). The polyphyly of “polyploid species” calls

into question the very definition of “species.” Allopolyploids – like other organisms with reticulate evolutionary histories (e.g., eukaryotes, lichens) – give biologists important examples when theorizing about evolutionary entities. Aside from philosophical considerations about species definitions, there are many implications for the multiplicity of origins for polyploids. Multiple origins of polyploid species have been reported for mosses, ferns, and many angiosperms (reviewed in Vogel et al., 1999; Soltis and Soltis, 2000).

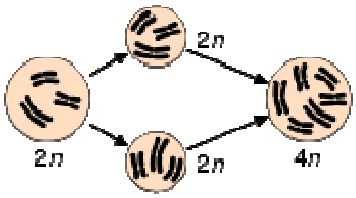
Allopolyploidy presents a paradox because it is both a diversifying force and a genetic bottleneck (Stebbins, 1971). However, the genetic bottleneck problem may be solved by the fact that population-level genetic studies of polyploid plants and animals indicate that polyploidy is not a rare event leading to unique and uniform genotypes. Rather, the multiple independent formations of polyploid species from heterozygous diploid progenitors may provide a significant source of genetic variation (reviewed in Soltis and Soltis, 1993; 1999; 2000).

Many new polyploids also are genetically unstable, and the next section describes mechanisms that can lead to novel variation.

3.1 Polyploidy in animals and plants

Polyploidy occurs in some animals, such as goldfish, salmon, and salamanders. However, polyploidy is especially common among ferns and flowering plants, including both wild and cultivated species. Wheat, for example, after millennia of hybridization and modification by humans, has strains that are *diploid* (two sets of chromosomes); *tetraploid* (four sets of chromosomes), with the common name of durum or macaroni wheat; and *hexaploid* (six sets of chromosomes), with the common name of bread wheat.

Many agriculturally important plants of the genus *Brassica* are also tetraploids. This genus, known as cabbages or mustards, includes turnips, brussels sprouts, cabbage, cauliflower, broccoli, mustard seed and other important crops. The *Triangle of U* is a theory, developed by a Woo Jang-choon, a Korean botanist who was working in Japan, that says the genomes of three ancestral species of Brassica combined to create the three common tetraploid species *Brassica juncea* (Indian mustard), *Brassica napus* (Rapeseed, rutabaga), and *Brassica carinata* (Ethiopian mustard).



Speciation via polyploidy: A diploid cell undergoes failed meiosis, producing diploid gametes, which self-fertilize to produce a tetraploid zygote.

Examples in animals are more common in the lower forms such as flatworms, leeches, and brine shrimp. Polyploid animals are often sterile, so they often reproduce by parthenogenesis, a form of asexual reproduction. Polyploid salamanders and lizards are also quite common and parthenogenetic. While mammalian liver cells are polyploid, rare instances of polyploid mammals are known, but most often result in prenatal death.

The only known exception to this rule is an octodontid rodent of Argentina's harsh desert regions, known as the Red Viscacha-Rat *(Tympanoctomys barrerae),* discovered by Milton Gallardo Narcisi. This rodent is not a rat, but kin to guinea pigs and chinchillas. Its "new" diploid [2n] number is 102 and so its cells are roughly twice normal size. Its closest living relation is *Octomys mimax,* the Andean Viscacha-Rat of the same family, whose 2n=56. It is surmised that an *Octomys*-like ancestor produced tetraploid (i.e., 4n=112) offspring that were,

by virtue of their doubled chromosomes, reproductively isolated from their parents; but that these likely survived the ordinarily catastrophic effects of polyploidy in mammals by shedding (via chromosomal translocation or some similar mechanism) the "extra" set of sex chromosomes gained at this doubling.

Polyploidy can be induced in cell culture by some chemicals: the best known is colchicine, which can result in chromosome doubling, though its use may have other less obvious consequences as well.

3.2 Polyploidy in Plants

Polyploidy is very common in plants, especially in angiosperms. From 30% to 70% of today's angiosperms are thought to be polyploid. Species of coffee plant with 22, 44, 66, and 88 chromosomes are known. This suggests that the ancestral condition was a plant with a haploid (**n**) number of 11 and a diploid (**2n**) number of 22, from which evolved the different polyploid descendants.

In fact, the chromosome content of most plant groups suggests that the basic angiosperm genome consists of the genes on 7–11 chromosomes. Domestic wheat, with its 42 chromosomes, is probably hexaploid (**6n**), where **n** (the ancestral haploid number) was 7.

Some other examples:

Plant	**Probable ancestral haploid number**	**Chromosome number**	**Ploidy level**
Domestic oat	7	42	**6n**
Peanut	10	40	**4n**
Sugar cane	10	80	**8n**
Banana	11	22,33	**2n, 3n**
White potato	12	48	**4n**
Tobacco	12	48	**4n**
Cotton	13	52	**4n**
Apple	17	34,51	**2n, 3n**

Polyploid plants not only have larger cells but the plants themselves are often larger. This has led to the deliberate creation of polyploid varieties of such plants as watermelons, marigolds, and snapdragons.

3.3 Origin of Polyploidy

Polyploidy has occurred often in the evolution of plants.

The process can begin if **diploid** (**2n**) gametes are formed. These can arise in at least two ways.

- The gametes may be formed by mitosis instead of meiosis.
- Plants, in contrast to animals, form germ cells (sperm and eggs) from somatic tissues. If the chromosome content of a precursor somatic cell has accidentally doubled (e.g., as a result of passing through S phase of the cell cycle without following up with mitosis and cytokinesis), then gametes containing **2n** chromosomes are formed

Polyploidy also occurs naturally in certain plant **tissues**.

- As the endosperm (**3n**) develops in corn (maize) kernels (Zea mays), its cells undergo successive rounds (as many as 5) of endoreplication producing nuclei that range as high as **96n**.
- When rhizobia infect the roots of their legume host, they induce the infected cells to undergo endoreplication producing cells that can become **128n**

Polyploidy can also be induced in the plant-breeding laboratory by treating dividing cells with colchicine. This drug disrupts microtubules and thus prevents the formation of a spindle. Consequently, the duplicated chromosomes fail to separate in mitosis. Onion cells exposed to colchicine for several days may have over 1000 chromosomes inside.

When a newly-arisen tetraploid (**4n**) plant tries to breed with its ancestral species (a **backcross**), triploid offspring are formed. These are sterile because they cannot form gametes with a balanced assortment of chromosomes.

However, the tetraploid plants can breed with each other. So in one generation, a new species has been formed.

Polyploidy even allows the formation of new species derived from different ancestors.

In 1928, the Russian plant geneticist Karpechenko produced a new species by crossing a cabbage with a radish. Although belonging to different genera (**Brassica** and **Raphanus** respectively), both parents have a diploid number of 18. Fusion of their respective gametes (**n**=9) produced mostly infertile hybrids.

However, a few fertile plants were formed, probably by the spontaneous doubling of the chromosome number in somatic cells that went on to form gametes (by meiosis). Thus these contained 18 chromosomes — a complete set of both cabbage (**n**=9) and radish (**n**=9) chromosomes.

Fusion of these gametes produced vigorous, fully-fertile, polyploid plants with 36 chromosomes. (They had the roots of the cabbage and the leaves of the radish.)

These plants could breed with each other but not with either the cabbage or radish ancestors, so Karpechenko had produced a new species.

The process also occurs in nature. Three species in the mustard family (Brassicaceae) appear to have arisen by hybridization and polyploidy from three other ancestral species:

- **B. oleracea** (cabbage, broccoli, etc.) hybridized with **B. nigra** (black mustard) → **B. carinata** (Abyssinian mustard).
- **B. oleracea** x **B. campestris** (turnips) → **B. napus** (rutabaga)
- **B. nigra** x **B. campestris** → **B. juncea** (leaf mustard)

Modern wheat and perhaps some of the other plants listed in the table above have probably evolved in a similar way.

3.4 Polyploidy in animals

Examples in animals are more common in the 'lower' forms such as flatworms, leeches, and brine shrimp. Polyploid animals are often sterile, so they often reproduce by parthenogenesis. Polyploid lizards are also quite common and parthenogenetic. Polyploid mole salamanders (mostly triploids) are all female and reproduce by kleptogenesis, "stealing" spermatophores from diploid males of related species to trigger egg development but not incorporating the males' DNA into the offspring. While mammalian liver cells are polyploid, rare instances of polyploid mammals are known, but most often result in prenatal death.

An octodontid rodent of Argentina's harsh desert regions, known as the Plains Viscacha-Rat (*Tympanoctomys barrerae*) has been reported as an exception to this 'rule'. However, careful analysis using chromosome paints shows that there are only two copies of each chromosome in *T. barrerae* not the four expected if it were truly a tetraploid. The rodent is not a rat, but kin to guinea pigs and chinchillas. Its "new" diploid [2n] number is 102 and so its cells are roughly twice normal size. Its closest living relation is *Octomys mimax*, the Andean Viscacha-Rat of the same family, whose 2n = 56. It was therefore surmised that an *Octomys*-like ancestor produced tetraploid (i.e., 4n = 112) offspring that were, by virtue of their doubled chromosomes, reproductively isolated from their parents.

Polyploidy is much rarer in animals. It is found in some insects, fishes, amphibians, and reptiles. Until recently, no polyploid **mammal** was known. However, the 23 September 1999 issue of **Nature** reported that a polyploid (tetraploid; **4n** = 102) rat has been found in Argentina.

Polyploid cells are larger than diploid ones; not surprising in view of the increased amount of DNA in their nucleus. The liver cells of the Argentinian rat are larger than those of its diploid relatives, and its sperm are huge in comparison. Normal mammalian sperm heads contain some 3.3 picograms (10^{-12} g) of DNA; the sperm of the rat contains 9.2 pg.

Although only one mammal is known to have all its cells polyploid, many mammals have polyploid cells in certain of their organs, e.g, the liver.

3.5 Polyploidy in Man

Polyploidy occurs in some animals, such as goldfish, salmon, and salamanders, but is especially common among ferns and flowering plants (see *Hibiscus rosa-sinensis*), including both wild and cultivated species. Wheat, for example, after millennia of hybridization and modification by humans, has strains that are **diploid** (two sets of chromosomes), **tetraploid** (four sets of chromosomes) with the common name of durum or macaroni wheat, and **hexaploid** (six sets of chromosomes) with the common name of bread wheat. Many agriculturally important plants of the genus *Brassica* are also tetraploids. Polyploidization is a mechanism of sympatric speciation because polyploids are usually unable to interbreed with their diploid ancestors.

Polyploidy can be induced in plants and cell cultures by some chemicals: the best known is colchicine, which can result in chromosome doubling, though its use may have other less obvious consequences as well. Oryzalin also will double the existing chromosome content.

True polyploidy rarely occurs in humans, although it occurs in some tissues (especially in the liver). Aneuploidy is more common.

Polyploidy occurs in humans in the form of triploidy, with 69 chromosomes (sometimes called 69,XXX), and tetraploidy with 92 chromosomes (sometimes called 92,XXXX). Triploidy, usually due to polyspermy, occurs in about 2–3% of all human pregnancies and 15% of miscarriages. The vast majority of triploid conceptions end as miscarriage and those that do survive to term typically die shortly after birth. In some cases survival past birth may occur longer if there is mixoploidy with both a diploid and a triploid cell population present.

Triploidy may be the result of either digyny (the extra haploid set is from the mother) or diandry (the extra haploid set is from the father). Diandry is mostly caused by reduplication of the paternal haploid set from a single sperm, but may also be the consequence of dispermic (two sperm) fertilization of the egg. Digyny is most commonly caused by either failure of one meiotic division during oogenesis leading to a diploid oocyte or failure to extrude one polar body from the oocyte. Diandry appears to predominate among early miscarriages while digyny predominates among triploidy that survives into the fetal period. However, among early miscarriages, digyny is also more common in those cases <8.5 weeks gestational age or those in which an embryo is present. There are also two distinct phenotypes in triploid placentas and fetuses that are dependent on the origin of the extra haploid set. In digyny there is typically an asymmetric poorly grown fetus, with marked adrenal hypoplasia and a very small placenta. In diandry, a partial hydatidiform mole develops. These parent-of-origin effects reflect the effects of genomic imprinting.

Complete tetraploidy is more rarely diagnosed than triploidy, but is observed in 1–2% of early miscarriages. However, some tetraploid cells are commonly found in chromosome analysis at prenatal diagnosis and these are generally considered 'harmless'. It is not clear

whether these tetraploid cells simply tend to arise during *in vitro* cell culture or whether they are also present in placental cells *in vivo*. There are, at any rate, very few clinical reports of fetuses/infants diagnosed with tetraploidy mosaicism.

Mixoploidy is quite commonly observed in human preimplantation embryos and includes haploid/diploid as well as diploid/tetraploid mixed cell populations. It is unknown whether these embryos fail to implant and are therefore rarely detected in ongoing pregnancies or if there is simply a selective process favoring the diploid cells.

4.0 Conclusion

At the end of this unit, the student learnt the following:

- Definition of polyploidy
- Polyploidy in animals and plants
- Polyploidy in man

5.0 Summary.

Polyploidy is the condition whereby a biological cell or organism has more than two homologous sets of chromosomes, with each set essentially coding for all the biological traits of the organism. A haploid (n) only has one set of chromosomes. A diploid cell (2n) has two sets of chromosomes. Polyploidy involves three or more times the haploid number of chromosomes. *Polyploid* types are termed according to the number of chromosome sets in the nucleus: *triploid* (three sets; 3n), *tetraploid* (four sets; 4n), *pentaploid* (five sets; 5n), *hexaploid* (six sets; 6n), and so on.

6.0 Tutor-Marked Assignment.

- What is polyploidy?
- Describe polyploidy in animals and plants

7.0 Reference/Further Readings

Parmacek MS, Epstein JA (July 2009). "Cardiomyocyte renewal". *N. Engl. J. Med.* **361** (1): 86–8. doi:10.1056/NEJMcibr0903347. PMID 19571289.

Griffiths, Anthony J. F. (1999). *An Introduction to genetic analysis*. San Francisco: W.H. Freeman. ISBN 0-7167-3520-2.

Ohno S, Muramoto J, Christian L, Atkin NB (1967). "Diploid-tetraploid relationship among old-world members of the fish family *Cyprinidae*". *Chromosoma* **23** (1): 1–9. doi:10.1007/BF00293307.

Bertolani R (2001). "Evolution of the reproductive mechanisms in Tardigrades: a review". *Zoologischer Anzeiger* **240** (3-4): 247–252. doi:10.1078/0044-5231-00032.

The genetic origin of kiwifruit. http://www.actahort.org/books/297/297_5.htm

Bogart JP, Bi K, Fu J, Noble DW, Niedzwiecki J (February 2007). "Unisexual salamanders (genus Ambystoma) present a new reproductive mode for eukaryotes". *Genome* **50** (2): 119–36. doi:10.1139/g06-152. PMID 17546077.

Svartman M, Stone G, Stanyon R (Aprilt 2005). "Molecular cytogenetics discards polyploidy in mammals". *Genomics* **85** (4): 425–30. doi:10.1016/j.ygeno.2004.12.004. PMID 15780745.

MODULE 5: EVOLUTION OF LIFE

Unit 1: Origin of Life

Unit 2: Evidence of evolution, adaptation and Speciation

Unit 3: Classification and phylogeny

Unit 4: Geological periods and epoch

Unit 5: Evolution of the plants

UNIT 1: ORIGIN OF LIFE

CONTENTS

1.0 INTRODUCTION

It is important to understand how life began. The origin of the first cell is an event of low probability, because a complete series of events would have had to occur- but this length of time is long enough for an event of low probability to have occurred. Today we do not believe that life arises spontaneously from nonlife and we say that ‘a life comes from a life’. However, the very first living thing had to have come from non living chemicals.

2.0 OBJECTIVES

At the end of this unit, student should be able to:

- explain the origin of life
- understand the probable stages in the origin of life
- explain what is organic evolution

3.0 MAIN CONTENT

3.1 Origin of life

The planet earth came into existence 4 and 5 billion years ago. Life evolved on planet earth about 3.5 billion years ago. At that time it was extremely hot. The existence of life in any form at that high temperature was not possible.

So, life two questions arise pertaining to

1. How did life originated on earth?
2. How did primitive organisms evolve into new forms resulting in the evolution of a variety of organism on earth?

Origin of life means the appearance of simplest primordial life from non living matter (Fig. 3.1).

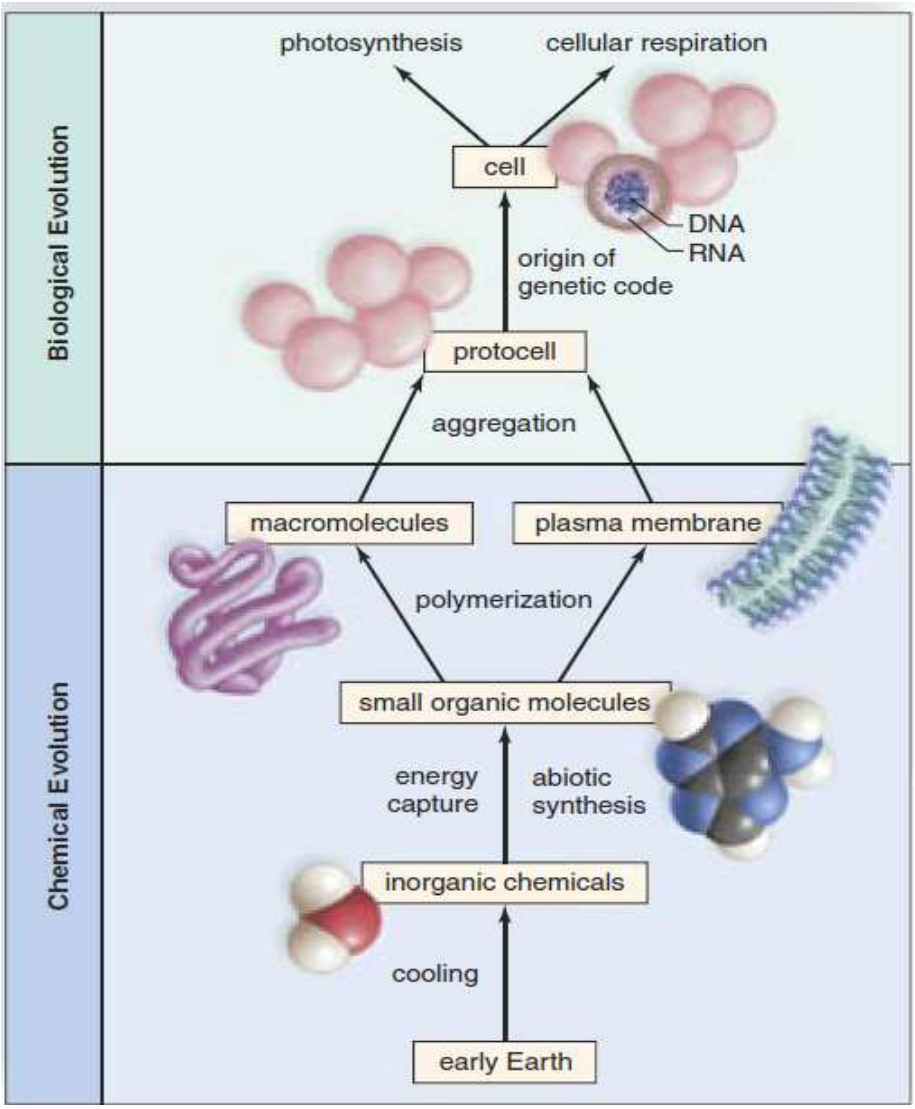


Fig. 3.1: Origin of first cell (s)

3.2 Probable stages in the origin of life

3.2.1 First stage (the primitive earth)

Theorized early primitive atmosphere consisted mostly of water vapour, nitrogen and carbon dioxide with small amounts of hydrogen and carbon monoxide with little if any, of free oxygen.

3.2.2 Second stage (evolution of small organic molecules)

A particular mix of inorganic chemicals could have reacted with one another to produce small organic molecules (or compound) such as glucose, amino acids and nucleotides. Most chemical reactions take place in water and the first proto cell undoubtedly arose in the ocean. In 1953, Stanley Miller and Harold Urey performed an experiment known as the (Miller-Urey experiment) that supports the hypothesis that small organic molecules were formed at the ocean's surface (Fig. 3.2). In the early earth, volcanoes erupted constantly and the first atmospheric gas would have consequently contained methane (CH_4), ammonia (NH_3) and hydrogen (H_2). These gases could have been washed into the ocean by the first rains, fierce lightning and unabated ultraviolent radiation would have allowed them to react and produce the first organic molecules.

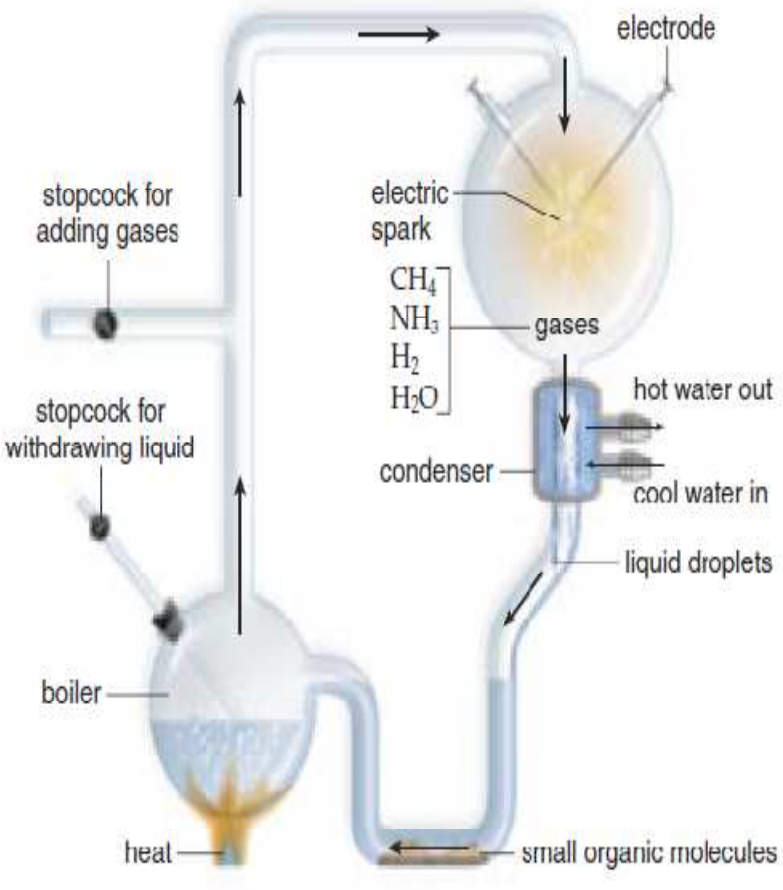


Fig 3.2: Miller and Ureys apparatus and experiment.

To test the hypothesis of chemical evolution, Miller placed the organic chemicals mentioned in a closed system, heated the mixture and circulated it past an electric spark (simulating lightning). After a week, the solution contained a variety amino acids and organic acids. This and other similar experiments support the hypothesis that inorganic chemicals in the absence of oxygen (O2) and in the presence of a strong energy source can result in organic molecules.

The formation of small organic molecules is thought to be the first step toward the origin

3.2.3 Third stage (macromolecules)

Once formed, the first small organic molecules gave rise to still larger molecules. Then these would have polymerized into macromolecules. There are three primary hypotheses concerning this stage in the origin of life.

RNA first- hypothesis: this hypothesis suggests that only the macromolecule RNA (ribonucleic acid) was needed at this time to progress toward formation of the first cell or cell(s). Such RNA molecules are called ribozymes since we know that ribozymes exist.

Protein-first hypothesis: this hypothesis suggests that amino acids collected in shallow puddles along the rocky shore and the heat of the sun caused them to form protenoids, small polypeptides that have some catalytic properties. When protenoids are returned to water, they form the properties of a cell. Some of these proteins could have had enzymatic properties.

The third hypothesis is put forth by Graham Cairns-Smith. He believes that clay was especially helpful in causing the polymerization of both proteins and nucleic acids at the same time. Clay attracts small organic molecules and contains iron and zinc, which may have served as inorganic catalysts for polypeptide formation. In addition, clay tends to collect energy from radioactive decay and then discharge it when the temperature or humidity changes, possibly providing a source of energy for polymerization. Cairns-Smith suggests that RNA nucleotides and amino acids became associated in such a way that polypeptides were ordered by, and helped synthesize, RNA.

Chemical reactions likely produced the macromolecules we associate with living things.

3.2.4 Fourth stage (the protocell)

After macromolecules formed, something akin to a modern plasma membrane was needed to separate them from the environment. Thus before the first true cell arose, there would likely have been a protocell (fig. 3.3), which could carry on metabolism but not reproduce, formed when lipids and microspheres formed a lipid – protein membrane. It has been suggested that the protocell likely was a heterotrophy, an organism that takes in preformed food.

The Heterotroph Hypothesis has been suggested that the protocell likely was a heterotroph, an organism that takes in performed food. During the early evolution of life, the ocean contained abundant nutrition in the form of small organic molecules. This suggests that heterotrophs preceded autotrophs, organisms that make their own food.

Once the protocell was capable of reproduction, it became a true cell, and biological evolution began

3.2.5 Fifth stage (The true cell)

A true cell is a membrane-bounded structure that can carry on protein synthesis to produce the enzymes that allow DNA to replicate. The central concept of genetics states that DNA directs protein synthesis and that information flows from DNA to RNA to protein. It is possible that this sequence developed in stages. Once the protocells acquired genes that could replicate, they became cells capable of reproducing, and biological evolution began.

The hypothesis that the origin of life followed a transition from small organic molecules to macromolecules to protocells to true cells is currently widely favoured by scientists.

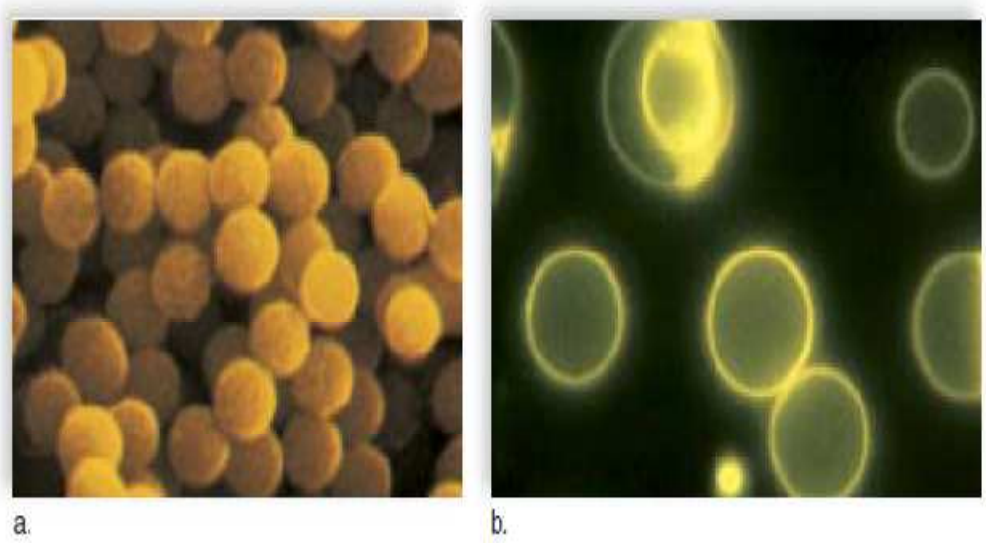


Fig.3.3: Protocell components

4.0 CONCLUSION

In this unit you have leant about the meaning of origin of life and the five probable stages involved in the origin of life. This knowledge will facilitate the understanding of the subsequent study units in this module.

5.0 SUMMARY

Chemical reactions are believed to have led to the formation of the first true cell(s). Inorganic chemicals, probably derived from the primitive atmosphere, reacted to form small organic molecules. These reactions occurred in the ocean, either on the surface or in the region of hydrothermal vents deep within.

After small organic molecules such as glucose, amino acids, and nucleotides arose, they polymerised to form the macromolecules. Amino acids joined to form proteins, and nucleotides joined to form nucleic acids. Perhaps RNA was the first nucleic acid. The RNA-first hypothesis is supported by the

discovery of ribozymes, RNA enzymes. The protein-first hypothesis is supported by the observation that amino acids polymerize abiotically when exposed to dry heat.

Once a plasma membrane developed, the protocell came into being. Eventually, the DNA→RNA→protein system evolved, and a true cell came into being.

6.0 TUTOR – MARKED ASSIGMENT

1. Explain the origin of life

2. List 5 probable stages involved in the origin of life

3. The RNA- first hypothesis for the origin of cells is supported by the discovery of

---

7.0 REFERENCES/FURTHER READINGS

Magurran, A. E., and R. M. May. 1999. Evolution of biological diversity. Oxford, U.K., Oxford University Press. An edited volume covering recent issues in the study of specification, with contributions from many active evolutionary biologists.

www.mhhe.com/zoology

www.mcgraw.com

UNIT 2: EVIDENCE OF EVOLUTION, ADAPTION AND SPECIATION

CONTENTS

1.0 INTRODUCTION

Evolution is defined as “common descent”. Because of descent with modification, all living things share the same fundamental characteristics: they are made of cells, take chemicals and energy from the environment, respond to external stimuli, and reproduce. Living things are diverse because individual organisms exist in the many environments throughout the Earth, and the features that enable them to survive in those environments are quite diverse. Many fields of biology provide evidence that evolution through descent with modification occurred in the past and is still occurring. Let us look at the various types of evidence for evolution.

Evolution influences every aspect of the form and behavior of organisms. Most prominent are the specific behavioural and physical adaptation that are the outcome of natural selection. These adaptations increase fitness by aiding activities such as finding food, avoiding predators or attracting mates.

2.0 OBJECTIVES

After the completing this lesson, you will be able to:

- give the fossils, anatomy, biochemical and biogeographical evidence

- explain adaptation

- explain speciation and four mechanisms involved

3.0 MAIN CONTENT

3.1 EVIDENCES OF EVOLUTION

The evidences supporting organic evolution are derived from a number of fields of biology. Those discuss here are:

1. Fossil evidence 2. Anatomy evidence 3. Biochemical evidence 4. Biogeographical evidence

3.1.1 Fossil Evidence

Fossils are the remains and traces of past life or any other direct evidence of past life. Most fossils consist only of hard parts of organisms, such as shells, bones, or teeth, because these are usually preserved after death. The soft parts of a dead organism are often consumed by scavengers or decomposed by bacteria. Occasionally, however, an organism is buried quickly and in such a way that decomposition is never completed or is completed so slowly that the soft parts leave an imprint of their structure. Traces include trails, footprints, burrows, worm casts, or even preserved droppings.

The great majority of fossils are found embedded in sedimentary rock. Sedimentation, a process that has been going on since Earth formed, can take place on land or in bodies of water. The weathering and erosion of rocks produces particles that vary in size and are called sediment. As such particles accumulate, sediment becomes a stratum (pl., strata), a recognizable layer of rock. Any given stratum is older than the one above it and younger than the one immediately below it, so that the relative age of fossils can be determined based on their depth.

Palaeontologists are biologists who study the fossils record and from it draw conclusions about the history of life. Particularly interesting are the fossils that serve as transitional links between groups. For example, the famous fossils of *Archaeopteryx* are intermediate between reptiles and birds (Fig.3.4). The dinosaur-like skeleton of this fossil has reptilian features, including jaws with teeth and a long, jointed tail, but *Archaeopteryx* also had feathers and wings, all suggesting that reptiles evolved from birds. Other transitional links among fossil vertebrates suggest that fishes evolved before amphibians, which evolved before both birds and mammals in the history of life. As a result of studying strata, scientists have divided Earth's history into eras, and then periods and epochs. The fossil record has helped determine the dates given in the table. There are two ways to date fossils. The

relative dating method determines the relative order of fossils and strata depending on the layer of rock in which they were found, but it does not determine the actual date they were formed.

a.

b.

Fig. 3.4 (a&b): Transitional fossils

The absolute dating method relies on radioactive dating techniques to assign an actual date to a fossil. All radioactive isotopes have a particular half-life, the length of time it takes for half of the radioactive isotope to change into another stable element. Carbon 14 (^{14}C) is the only radioactive isotope in organic matter. Using both relative and absolutedating methods, we can learn from fossils about the various organisms and environments that existed across the planet during any time period. Fossils

Table 3.1: The Geological Time Scale: Major Divisions of Geological Time and Same of the Major Evolutionary Events of Each Time Period

Era	Period	Epoch	Millions of Years Ago	Plant Life	Animal Life
		Holocene	0.01	Human influence on plant life	Age of *Homo sapiens*
				Significant Mammalian Extinction	
	Quaternary	Pleistocene		Herbaceous plants spread and diversify.	Presence of ice age mammals. Modern humans appear.
		Pliocene	(5.1–1.8)	Herbaceous angiosperms flourish.	First hominids appear.
		Miocene	(23–25)	Grasslands spread as forests contract.	Apelike mammals and grazing mammals flourish; insects flourish.
Cenozoic*		Oligocene	(35–23)	Many modern families of flowering plants evolve.	Browsing mammals and monkeylike primates appear.
	Tertiary	Eocene	(57–36)	Subtropical forests with heavy rainfall thrive.	All modern orders of mammals are represented.
		Paleocene	(65–57)	Flowering plants continue to diversify.	Primitive primates, herbivores, carnivores, and insectivores appear.
				Mass Extinction: Dinosaurs and Most Reptiles	
	Cretaceous		(144–65)	Flowering plants spread; conifers persist.	Placental mammals appear; modern insect groups appear.
Mesozoic	Jurassic		(213–144)	Flowering plants appear.	Dinosaurs flourish; birds appear.
				Mass Extinction	
	Triassic		(248–138)	Forests of conifers and cycads dominate.	First mammals appear; first dinosaurs appear; corals and mollusks dominate seas.
				Mass Extinction	
	Permian		(280–248)	Gymnosperms diversify.	Reptiles diversify; amphibians decline.
	Carboniferous		(360–280)	Age of great coal-forming forests: ferns, club mosses, and horsetails flourish.	Amphibians diversify; first reptiles appear; first great radiation of insects.
				Mass Extinction	
Paleozoic	Devonian		(408–360)	First seed plants appear. Seedless vascular plants diversify.	Jawed fishes diversify and dominate the seas; first insects and first amphibians appear.
	Silurian		(438–408)	Seedless vascular plants appear.	First jawed fishes appear.
				Mass Extinction	
	Ordovician		488.3	Nonvascular land plants appear. Marine algae flourish.	Invertebrates spread and diversify; jawless fishes (first vertebrates) appear.
	Cambrian		543	First plants appear on land. Marine algae flourish.	All invertebrate phyla present; first chordates appear.
			600	Oldest soft-bodied invertebrate fossils	
			1,400–700	Protists evolve and diversify.	
			2,300	Oldest eukaryotic fossils	
Precambrian Time			2,700	O_2 accumulates in atmosphere.	
			3,500	Oldest known fossils (prokaryotes)	
			4,600	Earth forms	

allowed scientists to construct the geological timescale that traces the history of life.

3.1.2 Biogeographical evidence

Another type of evidence that supports evolution through descent with modification is found in the field of biogeography, the study of the distribution of species throughout the world.

Different mammals and flowering plants evolved separately in each biogeographical region, and barriers such as mountain ranges and oceans prevented them from migrating to other regions.

Many of these barriers arose through a process called continental drift. That is, the continents have never been fixed; rather, their positions and the positions of the oceans have changed over time.

The distribution of many organisms on earth is explainable by knowing when they evolved, either before or after the continents moved apart

3.1 .3 Anatomical evidence

The fact that anatomical similarities exist among organisms provides further support for evolution via descent with modification. Vertebrate forelimbs are used for flight (birds and bat), orientation during swimming (whales and seals). Running (horses), climbing (arboreal lizard), or swinging from tree branches (monkey). Yet all vertebrate forelimbs contain the same set of bones organized in the same ways, despite the dissimilar functions. The most plausible explanation for this unity is that the basic forelimb plan belongs to a common ancestor, and then the plan was modified in the succeeding groups as each continued along its own evolutionary pathway. Structures that are anatomically similar because they are inherited from a common ancestor called homologous structure structures. In contrast, analogous structures serve the same function, but are not constructed similarly nor do they share a common ancestry. The wings of birds and insect and the eyes of octopi and humans are analogous structure and are similar due to a common ancestry. The presence of homology, analogy, is evidence that organisms are related

Vestigial structures are anatomical features that are fully developed in one group of organisms but that are reduced and may have no functions in similar groups. Most birds, for example , have well-developed wings for flight. However some species (e.g., ostrich) have greatly reduced wings and do not fly. Similarly, snakes have no use for hind limbs, and yet some have remnant of hind limbs in a pelvic girdle and legs. The presence of vestigial structures occur because organisms inherit their anatomy from their ancestors: they are traces of an organism’s evolutionary history.

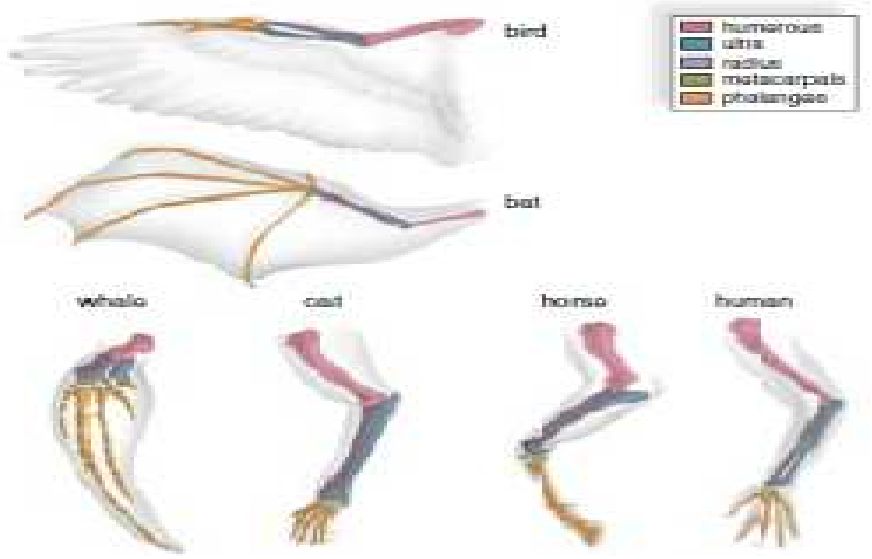


Fig. 3.5: Significance of homologous structures

The homology shared by vertebrates extends to their embryology development. At some time during development, all vertebrates have a post anal tail and exhibit paired pharyngeal pouches. In fishes and amphibian larvae these pouches develop into functioning gills. In humans, the first pair of pouches becomes the cavity of the middle ear and the auditory tube. The second pair becomes the tonsils, while the third and fourth pairs become the thymus and parathyroid gland. Why should terrestrial vertebrates develop and then modify structures like pharyngeal pouches that have lost their original function? The most likely explanation is that fishes are ancestral to other vertebrate groups.

Fig. 3.6: Significance of developmental similarities

Organisms that share homologous structures are closely related and have a common ancestry. Studies of comparative and embryological development reveal homologous structures.

3.1.4 Biochemical evidence

Almost all living organism use the same basic biochemical molecules, including DNA, ATP (adenosine triphosphate), and many identical or nearly identical enzymes. Further, organisms use the same DNA triplet code for the 20 amino acid in their proteins. Since the sequences of DNA bases in

genomes of many organisms are now known, it has become clear that humans share a large number of genes with much

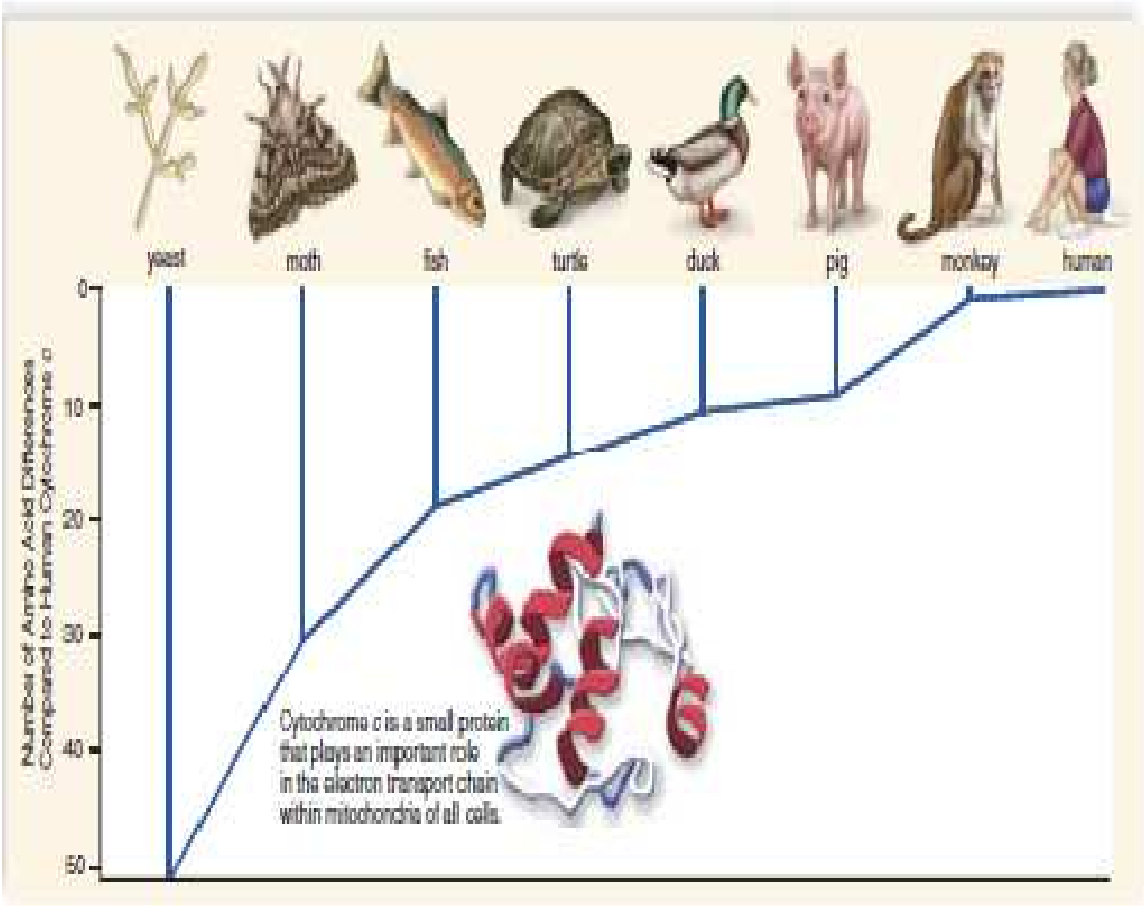


Fig. 3.7: Significance of biochemical differences

simpler organisms. It appears that life's vast diversity has come about by only a slight difference in many of same genes. The result has been widely divergent types of bodies. When the degree of similarity in DNA nucleotide sequences or the degree of similarity in amino acid sequences of proteins is examined, the more similar the DNA sequences are, generally the more closely related the organisms are. For example, humans and chimpanzees' are about 99% similar! Cytochrome c is a molecule that is used in used in electron transport chain of all the organisms appearing in figure 3.7. data regarding differences in the amino acid sequence of cytochrome c show that the sequence in a

human differs from that in a yeast by 51 amino acids. These data are consistent with other data regarding the anatomical similarities of these organisms and, therefore, how closely they are related.

Evolution in no longer considered a hypothesis. It is one of the great unifying theories of biology. In science the word theory is reserved for those conceptual schemes that are supported by a large number of observation and scientific experiments. The theory of evolution has the same status in biology that the germ theory of disease has in medicine.

Many line of evidence support the theory of evolution by descent with modification. Recently biochemical evidence has also been found to support evolution. A hypothesis is strengthened when it is supported by many different lines of evidence.

3.2 Adaptation

Adaptation is the process that makes organisms better suited to their habitat. Also, the term adaptation may refer to a trait that is important for an organism's survival. For example, the adaptation of horses' teeth to the grinding of grass. By using the term *adaptation* for the evolutionary process and *adaptive trait* for the product (the bodily part or function), the two senses of the word may be distinguished. Adaptations are produced by natural selection. The following definitions are due to Theodosius Dobzhansky.

> *Adaptation* is the evolutionary process whereby an organism becomes better able to live in its habitat or habitats.
>
> *Adaptedness* is the state of being adapted: the degree to which an organism is able to live and reproduce in a given set of habitats.
>
> *An adaptive trait is an aspect of the developmental pattern of the organism which enables or enhances the probability of that organism surviving and reproducing.*

Adaptation may cause either the gain of a new feature, or the loss of an ancestral feature. An example that shows both types of change is bacterial adaptation to antibiotic selection, with genetic changes causing antibiotic resistance by both modifying the target of the drug, and increasing the activity of transporters that pump the drug out of the cell. Other striking examples are the bacteria *Escherichia coli* evolving the ability to use citric acid as a nutrient in a long-term laboratory experiment, *Flavobacterium* evolving a novel enzyme that allows these bacteria to grow on the by-products of nylon manufacturing, and the soil bacterium *Sphingobium* evolving an entirely new metabolic pathway that degrades the synthetic pesticide pentachlorophenol. An interesting but still controversial idea is that some adaptations might increase the ability of

organisms to generate genetic diversity and adapt by natural selection (increasing organisms' evolvability).

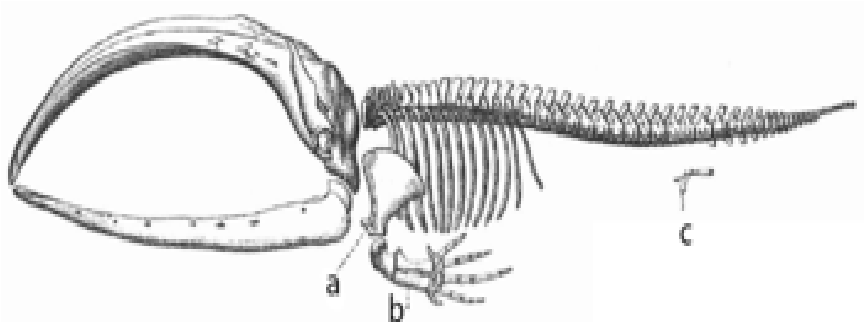


Fig. 3.8: A baleen whale skeleton, *a* and *b* label flipper bones, which were adapted from front leg bones: while *c* indicates vestigial leg bones, suggesting an adaptation from land to sea.

Adaptation occurs through the gradual modification of existing structures. Consequently, structures with similar internal organization may have different functions in related organisms. This is the result of a single ancestral structure being adapted to function in different ways. The bones within bat wings, for example, are very similar to those in mice feet and primate hands, due to the descent of all these structures from a common mammalian ancestor. However, since all living organisms are related to some extent, even organs that appear to have little or no structural similarity, such as arthropod, squid and vertebrate eyes, or the limbs and wings of arthropods and vertebrates, can depend on a common set of homologous genes that control their assembly and function; this is called deep homology.

During evolution, some structures may lose their original function and become vestigial structures. Such structures may have little or no function in a current species, yet have a clear function in ancestral species, or other closely related species. Examples include pseudogenes, the non-functional remains of eyes in blind cave-dwelling fish, wings in flightless birds, and the presence of hip bones in whales and snakes. Examples of vestigial structures in humans include wisdom teeth, the coccyx, the vermiform appendix, and other behavioural vestiges such as goose bumps and primitive reflexes.

However, many traits that appear to be simple adaptations are in fact exaptations: structures originally adapted for one function, but which coincidentally became somewhat useful for some

other function in the process. One example is the African lizard *Holaspis guentheri*, which developed an extremely flat head for hiding in crevices, as can be seen by looking at its near relatives. However, in this species, the head has become so flattened that it assists in gliding from tree to tree—an exaptation. Within cells, molecular machines such as the bacterial flagella and protein sorting machinery evolved by the recruitment of several pre-existing proteins that previously had different functions. Another example is the recruitment of enzymes from glycolysis and xenobiotic metabolism to serve as structural proteins called crystallins within the lenses of organisms' eyes.

A critical principle of ecology is that of competitive exclusion: no two species can occupy the same niche in the same environment for a long time. Consequently, natural selection will tend to force species to adapt to different ecological niches. This may mean that, for example, two species of cichlid fish adapt to live in different habitats, which will minimise the competition between them for food.

An area of current investigation in evolutionary developmental biology is the developmental basis of adaptations and exaptations. This research addresses the origin and evolution of embryonic development and how modifications of development and developmental processes produce novel features. These studies have shown that evolution can alter development to produce new structures, such as embryonic bone structures that develop into the jaw in other animals instead forming part of the middle ear in mammals. It is also possible for structures that have been lost in evolution to reappear due to changes in developmental genes, such as a mutation in chickens causing embryos to grow teeth similar to those of crocodiles. It is now becoming clear that most alterations in the form of organisms are due to changes in a small set of conserved genes.

3.3 Speciation

Speciation is the origin or evolution of new species. Speciation has occurred when one species gives rise to two species, each of which continues on its own evolutionary pathway. Species is a latin word meaning "kind" or "appearance" Species is defined as a group of subpopulations that are capable of inter breeding and are isolated reproductively form other species. The subpopulations of the same species can exchange genes, but different species do not exchange genes. Species have traditionally been described on the basis of their physical form or morphology

Speciation occurs in the following ways and is termed accordingly.

How can we recognize speciation?

Whenever reproductive isolation develops between two formerly interbreeding groups of populations, speciation has occurred.

Allopatric speciation takes place when a part of the population becomes

geographically separated (geographical isolation) from the parental population.

For example a group of birds lives at the base of the mountain, some members

fly up and get geographically isolated. Variation and natural selection act differently

on the two because the environment in which the two live is different. Gradually

genetic changes render them to be reproductively isolated.

Sympatric speciation

Sometimes a genetic barrier (reproductive barrier) prevents reproduction between

a section of a population of a species with other members. Such a section

of population usually arises in plants because of polyploidy. Polyploidy is a

mutation in which the normal diploid number of chromosomes become doubled

or trebled (2n becomes 3n, 4n, 5n etc) in a section of the population of a

species due to certain irregularities during cell division. The polyploid section

of the population is then unable to interbreed (mate and reproduce) with the

other and becomes a new species.

3.3.1 Two isolation mechanisms

Prezygotic isolation mechanisms are in place before fertilization, and thus reproduction is never attempted. Postzygotic isolation mechanisms are in place after fertilization, so reproduction may take place, but it does not produce fertile offspring (Table 3.2).

Reproductive Isolating Mechanisms

Isolation Mechanism	Example
Prezygotic	
Habitat isolation	Species at same locale occupy different habitats
Temporal isolation	Species reproduce at different seasons or different times of day.
Behavioural isolation	In animals, courtship behaviour differs, or they respond to different songs, calls, pheromones, or other signals
Mechanical isolation	Genitalia unsuitable for one another
Postzygotic	
Gamete isolation	Sperm cannot reach or fertilize egg
Zygote mortality	Fertilization occurs, but zygote does not survive
Hybrid sterility	Hybrid survives but is sterile and cannot reproduce
F_2 fitness	Hybrid is sterile, but F_2 hybrid has reduced fitness

3.3.2 Models of speciation

There are two accepted models of speciation that have given rise to the biodiversity

1. Phyletic Gradualism model

Two species from common ancestor gradually become more and more structurally different acquiring adaptations unique to each (Fig. 1.11b(i)). Darwin also believed that evolution is a slow and gradual process.

2. Punctuated equilibrium

A new species arises through major changes in the beginning and then remain constant for long periods before changing again. (Fig. 3.9b (ii)) This model was suggested by paleontologists (scientists who study fossils), Niles Eldredge and Stephen Jay Gould.

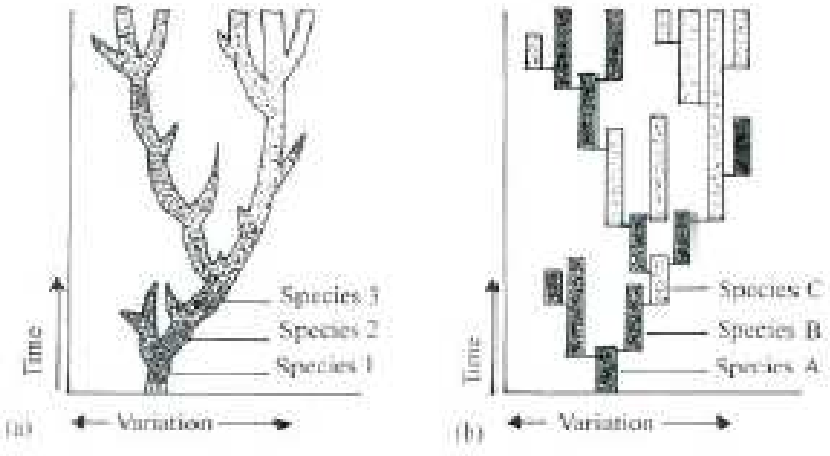


Fig. 3.9: Models of speciation (a) Phyletic gradualism (b) Punctuated equilibrum

3.3.3 The four mechanisms of speciation.

Speciation is the process where a species diverges into two or more descendant species. Barriers to reproduction between two diverging sexual populations are required for the populations to become new species. Gene flow may slow this process by spreading the new genetic variants also to the other populations. Depending on how far two species have diverged since their most recent common ancestor, it may still be possible for them to produce offspring, as with horses and donkeys mating to produce mules. Such hybrids are generally infertile. In this case, closely related species may regularly interbreed, but hybrids will be selected against and the species will remain distinct. However, viable hybrids are occasionally formed and these new species can either have properties intermediate between their parent species, or possess a totally new phenotype. The importance of hybridisation in producing new species of animals is unclear, although cases have been seen in many types of animals, with the gray tree frog being a particularly well-studied example.

Speciation has been observed multiple times under both controlled laboratory conditions and in nature. In sexually reproducing organisms, speciation results from reproductive isolation followed by genealogical divergence. There are four mechanisms for speciation.

The most common in animals is allopatric speciation, which occurs in populations initially isolated geographically, such as by habitat fragmentation or migration. Selection under these conditions can produce very rapid changes in the appearance and behaviour of organisms. As selection and drift act independently on populations isolated from the rest of their species, separation may eventually produce organisms that cannot interbreed.

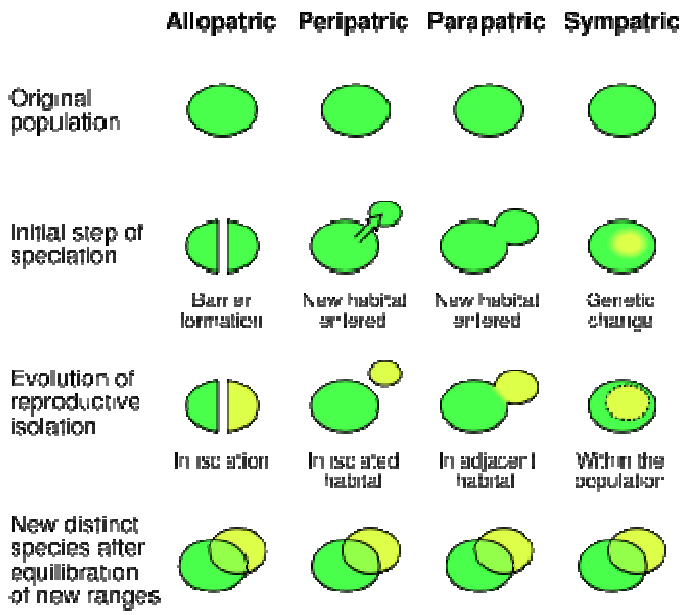


Fig. 3.10: The four mechanisms

The second mechanism of speciation is peripatric speciation, which occurs when small populations of organisms become isolated in a new environment. This differs from allopatric speciation in that the isolated populations are numerically much smaller than the parental population. Here, the founder effect causes rapid speciation after an increase in inbreeding increases selection on homozygotes, leading to rapid genetic change.

The third mechanism of speciation is parapatric speciation. This is similar to peripatric speciation in that a small population enters a new habitat, but differs in that there is no physical separation between these two populations. Instead, speciation results from the evolution of mechanisms that reduce gene flow between the two populations.

Generally this occurs when there has been a drastic change in the environment within the parental species' habitat. One example is the grass *Anthoxanthum odoratum*, which can undergo parapatric speciation in response to localised metal pollution from mines. Here, plants evolve that have resistance to high levels of metals in the soil. Selection against interbreeding with the metal-sensitive parental population produced a gradual change in the flowering time of the metal-resistant plants, which eventually produced complete reproductive isolation. Selection against hybrids between the two populations may cause *reinforcement*, which is the evolution of traits that promote mating within a species, as well as character displacement, which is when two species become more distinct in appearance.

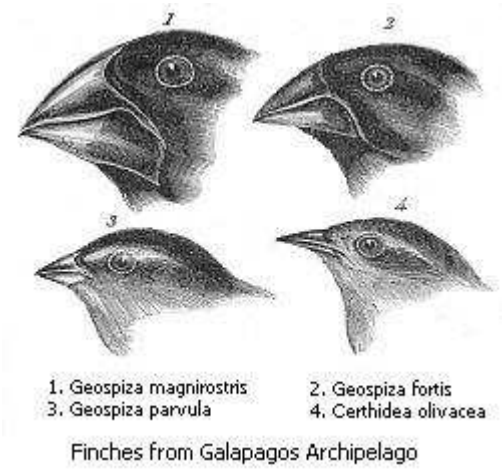


Fig. 3.11: Finches from Galapagos Archipelago

Geographical isolation of finches on the Galápagos Islands produced over a dozen new species.

Finally, in sympatric speciation species diverge without geographic isolation or changes in habitat. This form is rare since even a small amount of gene flow may remove genetic differences between parts of a population. Generally, sympatric speciation in animals requires the evolution of both genetic differences and non-random mating, to allow reproductive isolation to evolve.

4.0 Conclusion

In this unit you have learnt about evidence of evolution, adaptation and speciation, this knowledge will facilitate the understanding of the subsequent study units in this module.

5.0 Summary

The fossil record and biogeography, as well as studies of comparative anatomy, development, and biochemistry, all provide evidence of evolution. The fossil record gives clues about the history of life in general and allows us to trace the descent of a particular group. Biogeography shows that the distribution of organisms on Earth can be influenced by a combination of evolutionary and geological processes. Comparing the anatomy and the development of organisms reveals homologous structures among those that share common ancestry. All organisms have certain biochemical molecules in common, and these chemical similarities indicate the degree of relatedness. Adaptation is the evolutionary process whereby an organism becomes better able to live in its habitat or habitats. Speciation is the origin of new species. This usually requires geographic isolation, followed by reproductive isolation. The evolution of several species of finches on the Galápagos Islands is an example of speciation caused by adaptive radiation because each one has a different way of life. Currently, there are two hypotheses about the pace of speciation. Traditionalists support phyletic gradualism—slow, steady change leading to speciation. In contrast, a more recent model, called punctuated equilibrium, proposes that long periods of stasis are interrupted by rapid speciation.

6.0 TUTOR-MARKED ASSIGNMENT

1. List and explain 4 evidences of evolution
2. What is adaptation?
3. Explain two models of speciation

7.0 REFERENCES/FURTHER READINGS

Raff, R. A. 1996. The ahape of life: genes, development and the evolution of animal form. Chicago, University of Chicago Press. A provocative discussion of the genetic and developmental processes underlying evolution of animal diversity.

www.mhhe.com/zoology

www.mcgraw.com

UNIT 3 : CLASSIFICATION AND PHYLOGENY

CONTENTS

1.0 INTRODUCTION

Classification means identifying similarities and differences between different kinds of organism and then placing similar organisms in one group and different kinds of organisms in different groups. Taxonomy may thus be defined as the science of classification of organisms into categories, maintaining certain rules. Early taxonomists classified organisms according to morphological features only. Once the concept of organic evolution was accepted, taxonomists began to draw evolutionary relationships between different kinds of organisms. This was termed systematic. Today taxonomy and systematic are treated as synonymous since for classification, both morphological and biochemical resemblances and even those of between molecules such as DNA and RNA are studied to establish evolutionary relationships. Phylogeny is the evolutionary relationship among organisms. Ideally, classifications reflect phylogeny in that it tells how organisms are related through evolution and common ancestry. Species in the same genus are more closely related than species in separate genera and so forth as we proceed from genus to domain.

2.0 OBJECTIVES

At the end of this unit, students should be able to:

- Define classification
- Justify the need for classification of organisms
- List the bases of classification
- Explain taxonomy and phylogeny

3.0 MAIN CONTENT

3.1 Classification

Classification involves assigning species to a hierarchy of categories: species, genus, family, order, class, phylum and kingdom. While classifying an organism, it is assigned to categories which show its evolutionary relationship with other groups of organisms. Each level or category is termed taxon (plural-taxa). The lowermost category is species. Other categories are arranged above species so that there is a hierarchy of categories.

3.1.1 Taxonomic categories

The various taxonomic categories are given below:

Species: Group of individuals of one kind which can interbreed to produce fertile offspring.

Genus: Group of species resembling each other in several features indicating common ancestry.

Family: Group of genera (singular-genus) resembling each other. e.g. *Felis domestica* (the cat) and *Panthera tigris* (the tiger), both belong to family Felidae.

Order: Includes families showing similar characteristics.

Class: Includes related orders.

Phylum: Includes related classes.

The various phyla belong to their respective kingdoms. There are five kingdoms

Example.

Kingdom : Animalae (Animals)

Phylum : Chordata (Animals with notochord/ backbone)

Class : Mammalia (Animals that suckle their young ones.)

Order : Primates (Mammals with larger brains and binocular vision)

Family : Hominidae (Humans and human like ancestors)

Genus : *Homo* (Fossilmen and modern man)

Species : H.*sapiens* (Modern man)

3.1.2 Scientific naming of organisms

Different plants and animals have different common names. A cat is called ‘billi’ in Hindi, ‘biral’ in Bengali, ‘punai’ in Tamil and manjar in Marathi. There are different words for cat in French or German. There arose the need to give organisms names which could be understood throughout the world. So, the scientific names were given to organisms. Scientific names of organisms are understood all over the world.

A simplified system of naming organisms called binomial nomenclature has been the standard for more than two centuries now. It was proposed by the Swedish biologist, Carolus Linnaeus (1707-1778). Binomial nomenclature simply means two-name system of naming. The name of every kind of organism has two parts, that of the genus followed by that of species. The generic name is written with a capital letter and the specific name with a small letter. e.g. *Homo sapiens* is the scientific name of modem man, *Mangifera indica* is the biological name of mango.

Three main features of biological naming are as follows:

1. A scientific name, by convention, is printed in italics or underlined when hand written.

2. Scientific naming is according to a set of scientific rules of nomenclature.

3. Scientific names are mostly in *Greek* and *Latin*. They are understood all over the world and have made communication about organisms easier.

3.1.3 Prokaryotes and Eukaryotes

The organisms that are most primitive or the first to evolve on earth are the bacteria.

They do not possess a nuclear membrane around their single chromosome. Absence of a well defined nucleus or in other words a primitive nucleus terms them prokaryotes (pro = primitive, karyon = nucleus). All bacteria are prokaryotes.

As a constrast, organisms other than bacteria possessing a well defined nucleus are eukaryotes (eu = true; karyon = nucleus). There are other differences between them are given in the table 3.3

3.1.4 The Five Kingdoms of Organisms

Till recently there were only two kingdoms for classification - Plantae and Animalae. Such a two kingdom classification had several drawbacks, e.g. bacteria and fungi were kept along with plants although they are very different.

R.H. Whittaker in 1969 suggested the five kingdom classification which is based on 3 criteria:

(i) The presence or absence of a well-defined nucleus.

(ii) Unicellular or multicellular

(iii)Mode of nutrition

Table 3.2 Differences between Prokaryotes and Eukaryotes

Characteristics	Prokaryotes	Eukaryotes
1. Size	0.1-10 μm	10-100 μm (greater volume)
2. Genetic material	Circular DNA, no linear DNA, histones associated with DNA, nucleoid form, no nuclear membrane	Histones present on which DNA molecule wrapped, well defined chromosomes, nuclear membrane present
3. Site of Nuclear material	DNA in cytoplasm	DNA inside distinct nucleus
4. Organelles	Non membrane bound organelles	Mitochondria, golgi body, lysosome present in the cell
5. Cell wall	Always present, contains	None {animals} or made of

6. Respiration 7. Reproduction	peptidoglycan By mesosomes Mostly asexual e.g bacterial, cyanobacterial and blue green algae	cellulose, chitin {plant and fungi} By mitochondria Asexual and sexual e.g protoctista, fungi, plants animal

The five kingdom classification of organisms

Name of Nature of Whether Kind of Kingdom nucleus unicells or nutrition multicells

1. MONERA Prokaryotic Unicellular Diverse type of (Blue green algae nutrition and bacteria)

2. PROTOCTISTA Eukaryotic Unicellular Diverse kind of (Algae and nutrition Protozoa)

3. FUNGI Eukaryotic Multicellular Saprophytic (Moulds, etc.) (Feed on dead, decaying matter)

4. PLANTAE Eukaryotic Multicellular Autotrophic (All green plants) (Synthesize food by photosynthesis)

5. ANIMALAE Eukaryotic Multicellular Heterotrophic (Animals) (Depend on others for food

3.2 Phylogeny

Life's history is depicted as a branching tree, called a phylogeny. All forms of life, including many extinct forms that represent dead branches, will connect to this tree somewhere.

3.2.1 Phylogenetics

Phylogenetics is the modern way in which organisms are classified and arranged in evolutionary trees. Phylogeneticists arrange species and higher classification categories into clades. Clades may be represented on a diagram called a cladogram. A clade contains a most recent common ancestor and all its descendant species—the common ancestor is presumed and not identified. Figure 3.12 depicts a cladogram for seven groups of vertebrates. Only the lamprey, the so-called "outgroup," lacks jaws, but the other six groups of vertebrates are in the same clade because they all have jaws, a derived characteristic relative to their ancestors. On the other hand, the vertebrates beyond the shark are all in the same clade because they have lungs, and so forth.

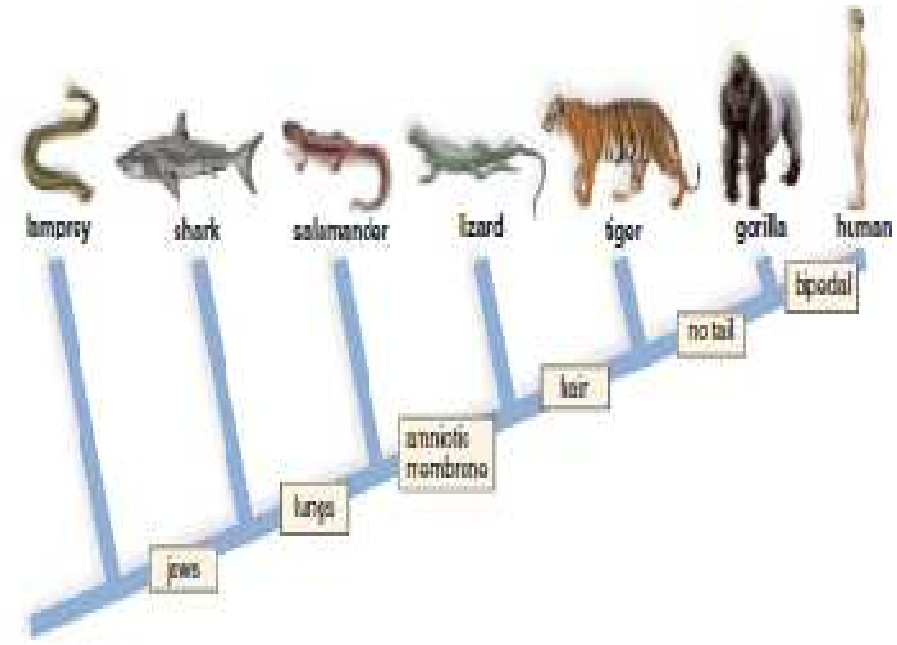


Fig. 3.12: Cladogram

4.0 Conclusion

In this unit you have leant about the classification of organisms, taxonomy, phylogeny and its importance. This knowledge will facilitate the understanding of the subsequent study units in this module.

5.0 Summary

Classification involves assigning species to a hierarchy of categories: kingdom, phylum, class, order, family, genus, and species, and in this text, domain. The five-kingdom system of classification recognizes these kingdoms: Monera (the bacteria), Protista (algae, protozoans), Fungi, Plantae, and Animalia. The more recent which is three-domain system (Bacteria, Archaea, and Eukarya), based on molecular data, is currently preferred. Both bacteria and archaea are prokaryotes. Members of the kingdoms Protista, Fungi, Plantae, and Animalia are eukaryotes.

Phylogeneticists classify and diagram the evolutionary relationships among organisms. They use as many characteristics as possible to put species in clades, which are represented on portions of a diagram called a cladogram. A clade contains a most recent common ancestor and all its descendant species, which share the same derived characteristics relative to their ancestors.

6.0 Tutor – Marked Assignment

(1) Name the Scientists who proposed:

(a) Binomial nomenclature

(b) Five kingdom classification

(2) Name the categories above order level in a correct sequence.

(3) What is cladogram?

7.0 References/Further Readings

Buss L.W. 1987. The evolution of individuality. Princeton, New Jersey, Princeton University Press. An original and provocative thesis on the relationship between development and evolution, with examples drawn from many different animal phyla.

Freeman, S., and J.C. Herron. 1998. Evolutionary analysis. Upper saddle River, New Jersey, Prentice-Hall. An introductory textbook on evolutionary biology designed for undergraduate biology majors.

Gould, S.J. 1989. Wonderful life: the Burgess Shale and the nature of history. New York, W.W. Norton & Company. An insightful discussion of what fossils tell us about the nature of life's evolutionary history.

Hartl, D. L., and A.G. Clark. 1997. Principles of population genetics. Sunderland, Massachusetts, Sinateur Associates. A current textbook on population genetics.

www.mhhe.com/zoology

www.mcgraw.com

http://www.ucmp.berkeley.edu/help/taxaform.html

http://www.ucmp.berkeley.edu/phyla/metazoamm.html

http://www.ucmp.berkeley.edu/plants/plantaemm.html

http://www.ucmp.berkeley.edu/chromista/chromista.html

http://www.ucmp.berkeley.edu/alllife/eukaryotasy.html

http://www.ucmp.berkeley.edu/fungi/fungi.html

http://www.ucmp.berkeley.edu/alllife/virus.html

http://www.ucmp.berkeley.edu/archaea/archaea.html

http://www.ucmp.berkeley.edu/bacteria/bacteria.html

http://www.mansfield.ohio-state.edu/~sabedon/biol3008.htm

UNIT 4: GEOLOGICAL TIME AND EPOCH

CONTENTS

1.0 Introduction

Long before the earth's age was known, geologists divided its history into a table of succeeding events based on the ordered layers of sedimentary rock. Geologists found that they needed some scheme by which to classify time (geological variety). In the 19th Century, fossils were becoming increasing important for correlating rock units, but they were also useful for determining time. Organisms evolved and died off. If you could determine their relative ages, you could start to age (relatively) the rocks that contained them.

2.0 Objectives

At the end of this unit, students should be able to:

- Explain geological time scale
- Explain epoch

3.0 Main content

3.1 Geological time scale

As a result of studying strata, scientists have divided Earth's history into eras, and then periods and epochs (Table 3.1). The largest divisions are Eons. To 19th Century geologists, the rocks could be broken up into 2 divisions 1) those containing visible signs of life (e.g., fossils) and those devoid of life (e.g., pre-fossils).; the Phanerozoic ("visible life; 544 Million years to the present) and the Proterozic (before life; up to 544 Million years). Some Proterozoic rocks did in fact contain primitive life (bacteria and later on soft-tissued organisms), but not beasties that produced skeletal remains. The Proterozoic is now defined as a specific interval of time (2.5 billion years to 544 million years). Two additional Eras have been added to the Geological Time Scale; the Archean (4.0 to 2.5 billion years) and the Hadean (4.5 to 4.0 billion years).

Table 3. 5: Division of earth's history

Eon	Era	Time
Phanerozoic	Cenozoic	65 MA to 0 MA
	Mesozoic	245 MA to 65 MA
	Paleozoic	550 MA to 245 MA
Proterozoic	Neoproterozoic	900 MA to 550 Ma
	Mesoproterozoic	1.6 GA to 900 MA
	Paleoproterozoic	2.5 GA to 1.6 GA

Archean		3.96 Ga to 2.5 Ga
Hadean		4.6 Ga to 3.96 Ga

Table 3.6: The Geological Time Scale

Era	Period	Epoch
Cenozoic (0 to 65 Ma)	Quaternary (0 to 2 Ma)	Holocene (0 to 10 Ka)
		Pleistocene (10 Ka to 2 Ma)
	Tertiary (2 MA to 65 Ma)	Pliocene (2 Ma to 5 Ma)
		Miocene (5 Ma to 24 Ma
		Oligocene (24 Ma – 37Ma)
		Eocene (37 Ma to 58 Ma)
		Paleocene (58 to 65 Ma)
Mesozoic (65 to 245 Ma)	Cretaceous (65 to 144 Ma)	Lots of Epochs in
	Jurassic (144 to 208 Ma)	All Periods

	Triassic (208 to 245 Ma)	
Paleozoic (245 to 544 Ma)	Permian (245 to 286 Ma)	
	Pennsylvanian (286 to 320 Ma)	
	Mississippian (320 to 362 Ma)	
	Devonian (362 to 418 Ma)	
	Silurian (418 to 441 Ma)	
	Ordovician (441 to 505 Ma)	
	Cambrian	

	(505 to 550 Ma)	
Neoproterozoic	Ediacarin (600 to 550 Ma)	
	Cryogenian (850 to 600 Ma)	

The use of fossils for dating rocks was of course limited to those rocks which contained fossils (i.e., mostly the Phanerozoic). Since there were lots of different fossils that came and went, it seemed logical to break the Phanerozoic up into smaller divisions called Eras. Three Paleozoic Eras are recognized:

Cenozoic (0 to 65 million years BP)

Mesozoic (65 to 245 million years BP)

Paleozoic (245 to 544 million years BP)

The Eras are further subdivided into smaller Periods and the Periods can be further subdivided into Epochs. The best way of illustrating this is to show you how these subdivisions work for the Phanerozoic and the latest portion of the Proterozoic (neoproterozoic). That occurs on the next page. The names used (e.g., Devonian) come from specific localities (usually in Europe) where especially well exposed rocks of that particular division are well exposed. You will also note that there are dates assigned to each division. These are absolute dates based upon radiometric dating of materials within

certain rocks. The dates are regularly modified if better radiometric analyses are developed. But the divisions themselves are fixed. They represent major changes in the fossil record. For example, the boundary between the Mesozoic and Cenozoic Eras (65 Ma) represents a major extinction event (the dinosaurs all died off). The boundary between the Paleozoic and Mesozoic (245 Ma) represents an even bigger extinction. Radiometric dating puts an absolute date on the geological divisions, but the divisions are largely paleontological in nature.

4.0 Conclusion

In this unit you have leant about the geological time scale. This knowledge will facilitate the understanding of the subsequent study units in this module.

5.0 Summary

Geological time is unlike normal time, it is broken up into manageable bits. The largest divisions are Eons. The use of fossils for dating rocks was of course limited to those rocks which contained fossils (i.e., mostly the Phanerozoic). Since there were lots of different fossils that came and went, it seemed logical to break the Phanerozoic* up into smaller divisions called Eras. The Eras are further subdivided into smaller Periods and the Periods can be further subdivided into Epochs.

6.0 TUTOR – MARKED ASSIGMENT

1. What is Eon?
2. List all the division of Era

7.0 REFERENCES/FURTHER READINGS

Buss L.W. 1987. The evolution of individuality. Princeton, New Jersey, Princeton University Press. An original and provocative thesis on the relationship between development and evolution, with examples drawn from many different animal phyla.

Freeman, S., and J.C. Herron. 1998. Evolutionary analysis. Upper saddle River, New Jersey, Prentice-Hall. An introductory textbook on evolutionary biology designed for undergraduate biology majors.

Gould, S.J. 1989. Wonderful life: the Burgess Shale and the nature of history. New York, W.W. Norton & Company. An insightful discussion of what fossils tell us about the nature of life's evolutionary history.

www.mhhe.com/zoology

www.mcgraw.com

UNIT 5: EVOLUTION OF THE PLANTS

CONTENTS

1.0 Introduction

In the history of life on Earth, one of the most revolutionary events was the colonization of land, first by plants, then by animals. Fossil and biochemical evidence indicates plants are descended from multicellular green algae. Algae dominated the oceans of the precambrian time over 700 million years ago. The evolution of plants has resulted in increasing levels of complexity, from the earliest algal mats, through bryophytes, lycopods, ferns to the complex gymnosperms and angiosperms of today. While the groups which appeared earlier continue to thrive, especially in the environments in which they evolved, each new grade of organization has eventually become more "successful" than its predecessors by most measures. Between 500 and 400 million years ago, some algae made the transition to land, becoming plants by developing a series of adaptations to help them survive out of the water.

2.0 Objectives

At the end of this unit, student should be able to:

- Explain plant evolution , multicellular development, rise of angiosperms and role of oxygen

3.0 Main Content

3.1 Plant Evolution

Land plants evolved from chlorophyte algae, perhaps as early as 510 million years ago; some molecular estimates place their origin even earlier, as much as 630 million years ago. Their closest living relatives are the charophytes, specifically Charales; assuming that the Charales' habit has changed little since the divergence of lineages, this means that the land plants evolved from a branched, filamentous, alga.

Plants were not the first photosynthesisers on land, though: consideration of weathering rates suggests that organisms were already living on the land 1,200 million years ago.

The first evidence of plants on land comes from spores of Mid-Ordovician age (early Llanvirn, ~470 million years ago). These spores, known as cryptospores, were produced either singly (monads), in pairs (diads) or groups of four (tetrads), and their microstructure resembles that of modern liverwort spores, suggesting they share an equivalent grade of organisation.

Four key evolutionary innovations serve to trace the evolution of the plant kingdom

1. Alternation of generations

Plants developed a more dominant diploid phase of the life cycle

2. Vascular tissue

Transports water and nutrients throughout the plant body, Thus plants were able to grow larger and in drier conditions

3. Seeds

Protected the embryo, thus allowing plants to dominate their terrestrial environments

4. Flowers and fruits

3.2 Multicellular plant

All seed plants are derived from a single common ancestor. The plant kingdom contains multicellular phototrophs that usually live on land. The earliest plant fossils are from terrestrial deposits, although some plants have since returned to the water. All plant cells have a cell wall containing the carbohydrate cellulose, and often have plastids in their cytoplasm. The plant life cycle has an alternation between haploid (gametophyte) and diploid (sporophyte) generations. There are more than 300,000 living species of plants known, as well as an extensive fossil record.

Plants divide into two groups: plants lacking lignin-impregnated conducting cells (the nonvascular plants) and those containing lignin-impregnated conducting cells (the vascular plants). Living groups of nonvascular plants include the bryophytes: liverworts, hornworts, and mosses.

There are five living phyla.

Four are gymnosperms, ovules not completely enclosed by sporophyte at time of pollination.

Fifth is angiosperms, ovules completely enclosed by a vessel of

sporophyte tissue, the carpel, at time of pollination

All multicellular plants have a life cycle comprising two generations or phases. One is termed the gametophyte, has a single set of chromosomes (denoted 1N), and produces gametes (sperm and eggs). The other is termed the sporophyte, has paired chromosomes (denoted 2N), and produces spores. The gametophyte and sporophyte may appear identical – homomorphy – or may be very different – heteromorphy.

The pattern in plant evolution has been a shift from homomorphy to heteromorphy. All land plants (i.e. embryophytes) are diplobiontic – that is, both the haploid and diploid stages are multicellular. Two trends are apparent: bryophytes (liverworts, mosses and hornworts) have developed the gametophyte, with the sporophyte becoming almost entirely dependent on it; vascular plants have developed the sporophyte, with the gametophyte being particularly reduced in the seed plants.

There are two competing theories to explain the appearance of a diplobiontic lifecycle.

The interpolation theory (also known as the antithetic or intercalary theory). This theory implies that the first sporophytes bore a very different morphology that the gametophyte they depended on. This seems to fit well with what we know of the bryophytes, in which a vegetative thalloid gametophyte is parasitised by simple sporophytes, which often comprise no more than a sporangium on a stalk. Increasing complexity of the ancestrally simple sporophyte, including the eventual acquisition of photosynthetic cells, would free it from its dependence on a gametophyte, as we see in some hornworts (*Anthoceros*), and eventually result in the sporophyte developing organs and vascular tissue, and becoming the dominant phase, as in the tracheophytes (vascular plants). This theory may be supported by observations that smaller *Cooksonia* individuals must have been supported by a gametophyte generation. The observed appearance of larger axial sizes, with room for photosynthetic tissue and thus self-sustainability, provides a possible route for the development of a self-sufficient sporophyte phase.

The alternative hypothesis is termed the transformation theory (or homologous theory). This explains the behaviour of some algae, which produce alternating phases of identical sporophytes and gametophytes. Subsequent adaption to the desiccating land environment, which makes sexual reproduction difficult, would result in the simplification of the sexually active gametophyte, and elaboration of the sporophyte phase to better disperse the waterproof spores. The tissue of sporophytes and gametophytes preserved in the Rhynie chert is of similar complexity, which is taken to support this hypothesis.

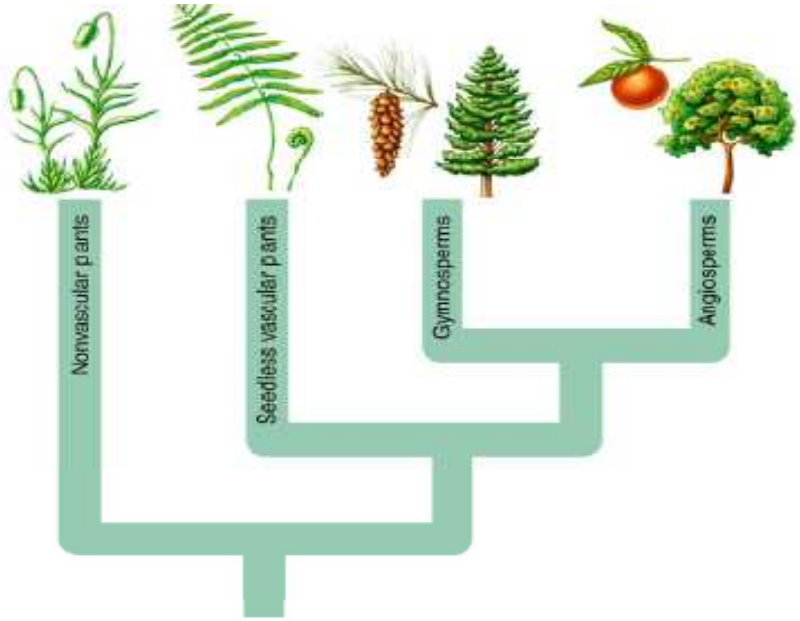


Fig.3.13: The evolution of plants

3.2.1 Nonvascular Plants

Only two phyla of living plants lack a vascular system

Liverworts (Phylum Hepaticophyta) (The simplest of all living plants)

Hornworts (Phylum Anthocerophyta)

Mosses (Phylum Bryophyta) were the first plants to evolve strands of specialized conduction cells. The conducting cells do not have specialized wall thickenings. Thus, a primitive vascular system, at the most

Fig 3.14: Hair-cup moss, Polytrichum

Bryophytes

Bryophytes are small, nonvascular plants that first evolved approximately 500 million years ago. The earliest land plants were most likely bryophytes. Bryophytes lack vascular tissue and have life cycles dominated by the gametophyte phase. The lack of conducting cells limits the size of the plants, generally keeping them under 5 inches high. Roots are absent in bryophytes, instead there are root-like structures known as rhizoids. Bryophytes include the hornworts, liverworts, and mosses.

3.2.2 The Evolution of Vascular plant

The first vascular plant appeared approximately 430 million years ago (mya), Early plants became successful colonizers of land through the development of vascular tissue, efficient water-and food-conducting system. Vascular plants are the more common plants like pines, ferns, corn, and oaks.

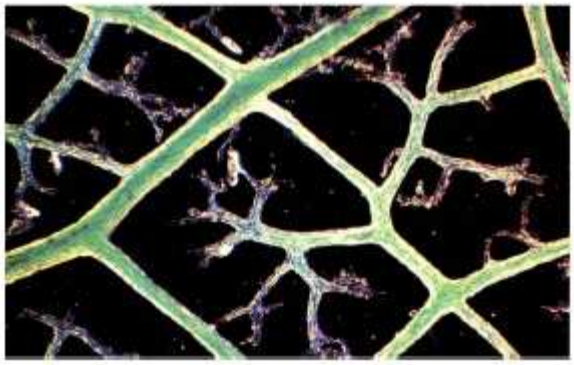


Fig. 3.15: The vascular system of a leaf

Vascular plants first developed during the Silurian Period, about 400 million years ago. The earliest vascular plants had no roots, leaves, fruits, or flowers, and reproduced by producing spores.

Cooksonia, is a typical early vascular plant. It was less than 15 cm tall, with stems that dichotomously branched. Dichotomous branching (where the stem divides into two ewqual branches) appears a primitive or ancestral trait in vascular plants. Some branches terminated in sporangia that produced a single size of spore.

Many scientists now consider "*Cooksonia*" an evolutionary grade rather than a true monophyletic taxon. Their main argument is that not all stems of *Cooksonia*-type plants have vascular tissue. The evolutionary situation of a grade would have some members of the group having the trait, others not. The shapes of sporangia on various specimens of *Cooksonia* also vary considerably.

Rhynia, is another early vascular plant. Like *Cooksonia*, it lacked leaves and roots. One of the species formerly assigned to this genus, *R. major*, has since been reclassified as *Aglaophyton major*. Some paleobotanists consider *A. major* a bryophyte, however, it does have a separate free-living sporophyte that is more prominent than the sporophyte, but appears to lack lignified conducting cells. The remaining species, *R. gwynne-vaughanii* is an undoubted vascular plant.

Devonian plant lines included the trimerophytes and zosterophyllophytes, which have been interpreted as related to ferns and lycophytes.

The Psilophytes

The Division Psilophyta consists of *Psilotum nudum*, a living plant that resembling what paleobotanists believe *Cooksonia* to have been: a naked, photosynthetic stem bearing sporangia. Also in the group is *Tmesipteris*, which resembles Psilotum except for its possession of smallo vascularized leaves arising on opposite sides of the stem. However, most paleobotanists doubt that *Psilotum* is a direct descendant of *Cooksonia*. Molecular studies suggest an affiliation with ferns for *Psilotum. Psilotum* also has three fused sporangia,

termed a synangium, located on the sides of the stems (instead of the tips of stems as in *Cooksonia*).

The Lycophytes

The next group, the Division Lycophyta, have their sporangia organized into strobili (singular: strobilus). A strobilus is a series of sporangia and modified leaves closely grouped on a stem tip. The leaves in strobili are soft and fleshy as opposed to the hard, modified leaves in cones.

Leaves that contained vascular tissue are another major advance for this group.The leaves in lycophytes, both living and fossil forms, are known as microphylls. This term does not imply any size constraint, but rather refers to the absence of a leaf gap in the vascular supply of the stem at the point where the leaf vascular trace departs. Ferns and other plants have megaphylls, leaves that produce this leaf gap.

Today there are fewer genera of lycophytes than during the group's heyday, the Paleozoic Era. Major living lycophytes include *Lycopodium* (commonly called the club moss, although it is NOT a moss), *Isoetes*, and *Selaginella* (the so-called resurrection plant). *Lycopodium* produces isospores that germinate in the soil and produce a bisexual gametophyte. These spores are all approximately the same size. *Selaginella* and *Isoetes* are heterosporous, and thus produce two sizes of spores: small spores (termed microspores) that germinate to produce the male gametophyte; and larger spores (megaspores) that germinate to produce the female gametophyte. The production of two sizes of spores, and also making separate unisexual gametophytes, is thought an important step toward the seed. Modern lycophytes are small, herbaceous plants. Many of the prominent fossil members of this group produced large amounts of wood and were significant trees in the Carboniferous-aged coal swamps.

Selaginella is a heterosporous member of the lycophytes. Some species of this genus are able to withstand drying out by going dormant until they are rehydrated. For this reason these forms of the genus are commonly called resurrection plants.

Fossil Lycophytes: Baragwanathia *and* Drepanophycus

Baragwanathia, is an undoubted lycophyte from the middle Silurian deposits of Australia. It has microphyllous leaves spirally attached to the stem, and sporangia clustered in some areas of the plant, although not in terminal strobili as in modern lycophytes.

Drepanophycus is a middle Devonian lycophyte from the Northern Hemisphere. Its features are very similar to modern lycophytes.

Lepidodendron *and* Sigillaria

The Lycophytes became significant elements of the world's flora during the Carboniferous time (the Mississippian and Pennsylvanian are terms used for this time span in the United States). These non-seed plants evolved into trees placed in the fossil genera *Lepidodendron* and *Sigillaria,* with heights reaching up to 40 meters and 20-30 meters respectively. *Lepidodendron* stems are composed of less wood (secondary xylem) that usually is found in gymnosperm and angiosperm trees.

We know much about the anatomy of these coal-age lycopods because of an odd type of preservation known as a coal ball. Coal balls can be peeled and the plants that are anatomically preserved within them laboriously studied to learn the details of cell structure of these coal age plants. Additionally, we have some exceptional petrifactions and compressions that reveal different layers of the plants' structure. Estimates place the bulk, up to 70%, of coal material as being derived from lycophytes.

Lepidodendron, was a heterosporous lycophyte tree common in coal swamps of the Carboniferous time. As with many large plant fossils, one rarely if ever finds the entire tree preserved intact. Consequently there are a number of fossil plant genera that are "organ taxa" and represent only the leaves (such as *Lepidophylloides*), reproductive structures (*Lepidostrobus*), stem (*Lepidodendron*), spores (*Lycospora*), and roots (*Stigmaria*). *Lepidodendron* had leaves borne spirally on branches that dichotomously forked, with roots also arising spirally from the stigmarian axes, and both small (microspores) and large (megaspores) formed in strobili (a loose type of soft cone). *Lepidodendron* may have attained heigths of nearly 40 meters, with trunks nearly 2 meters in diameter. The trees branched

extensively and produced a large number of leaves. When these leaves fell from the branches, they left behind them the leaf scars characteristic of the genus.

Sigillaria was another arborescent lycopod, and is also common in coal-age deposits. In contrast to the spirally borne leaves of *Lepidodendron, Sigillaria* had leaved arranged in vertical rows along the stem.

The Sphenophyta

The Division Sphenophyta contains once dominant plants (both arborescent as well as herbaceous) in Paleozoic forests, equisetophytes are today relegated to minor roles as herbaceous plants. Today only a single genus, *Equisetum*, survives. The group is defined by their jointed stems, with many leaves being produced at a node, production of isospores in cones borne at the tips of stems, and spores bearing elaters (devices to aid in spore dispersal). The gametophyte is small, bisexual, photosynthetic, and free-living. Silica concentrated in the stems give this group one of their common names: scouring rushes. These plants were reportedly used by American pioneers to scour the pots and pans. The fossil members of this group are often encountered in coal deposits of Carboniferous age in North America and Europe.

The Ferns

Ferns reproduce by spores from which the free-living bisexual gametophyte generation develops. There are 12,000 species of ferns today, placed in the Division Pteridophyta. The fossil history of ferns shows them to have been a dominant plant group during the Paleozoic Era. Most ferns have pinnate leaves, exhibiting small leaflets on a frond, Ferns have megaphyllous leaves, which cause a leaf gap in the vascular cylinder of the stem/rhizome. The first ferns also appear by the end of the Devonian. Some anatomical similarities suggest that ferns and sphenophytes may have shared a common ancestor within the trimerophytes.

3.2.3 Tracheophytes: The Vascular Plants

The vascular plants have specialized transporting cells xylem (for transporting water and mineral nutrients) and phloem (for transporting sugars from leaves to the rest of the plant).

When we think of plants we invariably picture vascular plants. Vascular plants tend to be larger and more complex than bryophytes, and have a life cycle where the sporophyte is more prominent than the gametophyte. Vascular plants also demonstrate increased levels of organization by having organs and organ systems. The novel features oif the vascular plants are summarized in Table 3.7

Table 3.7: Major evolutionary advances of the vascular plants

Advance	Green Algae	Bryophytes	Tracheophytes
Development of the root-stem-leaf vascular system	nonvascularized body (thallus) that may be variously shaped no levaes, shoots, or roots	no vascular system leaflike structures are present, but lack any vascular tissue	early vascular plants are naked, rootless vascularized stems later vascular plants develop vascularized leaves, then roots
Reduction in the size of the gametophyte generation	wide range of life cycles, some gametophyte dominant, others sporophyte dominant	sporophyte generation dependant on gametophyte generation for food; gametophyte is free-living and photosynthetic	progressive reduction in size and complexity of the gametoiphyte generation, leading to its complete dependance on the sporophyte for food in angiosperms, 3 celled male gametophyte and a (usually) 8 celled female gametophyte
Development of seeds in some vascular plants	no seeds	no seeds	seed plants retain the female gametophyte on the sporophyte
Spores/Pollen	spores for	Spores that germinate	Spores that germinate into the

	resisting environmental degradation	into the gametophyte generation	gametophyte generation or spores that have the gametophyte generation develop within themselves

3.2.4 Seedless Vascular Plants

Two phyla of modern-day vascular plants lack seeds

Ferns (Phylum Pterophyta)

Club mosses (Phylum Lycophyta)

Both have free-swimming sperm that require free water for fertilization By far, the largest group are ferns

~ 12,000 living species

3.2.5 Evolution of Seed Plants

Seeds are embryo covers that protect the embryonic plant at its most vulnerable stage.

Seed plants produce two kinds of gametophytes

1. Male gametophytes called pollen grains arise from microspores
2. Female gametophytes contains the egg develops from a megaspore produced within an ovule

Pollination is the transfer of pollen by insects, winds Thus; there is no need for free water for fertilization.

A seed has three parts

1. A sporophyte plant embryo

2. A source of food for the embryo called endosperm

3. A drought-resistant protective cover

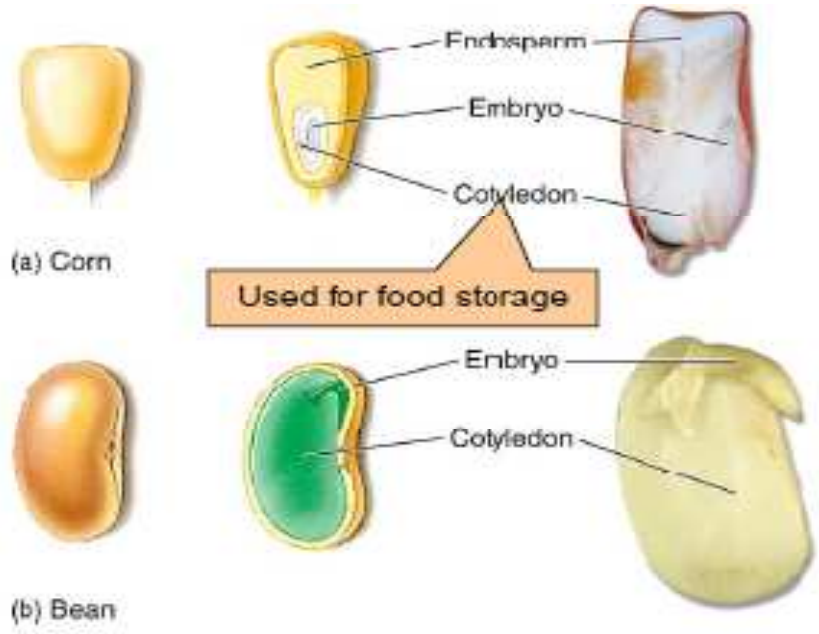


Fig. 3.16: (a) Corn and (b) Bean

Seeds have greatly improved the adaptations of plants to living on land

1. Dispersal: Facilitate migration dispersal

2. Dormancy: Postpone development until conditions are favourable

3. Germination: Permit embryonic development to be synchronized with habitat

4. Nourishment : Offer an energy source of young plants

Fig. 3.17: Cycad

3.2.6 Gymnosperms

Gymnosperms are non flowering seed plants, they include four phyla

Conifers (Coniferophyta)

Cycads (Cycadophyta)

Gnetophytes (Gnetophyta)

Ginkgo (Ginkgophyta)

3.2.6 .1 Conifers

The most common (and familiar) of the gymnosperms include Pine, spruce, cedar, redwood and fir trees, conifers are trees that produce their seeds in cones Seeds (ovules) develop on scales within cones and are exposed at the time of pollination.

Fig. 3. 18: Conifers

3.2.6.2 Cycads

Have short stems and palmlike leaves, The predominant land plant in the Jurassic Period

Fig. 3.19: Cycad

3.2.6.3 Gnetophytes

The most closely related to angiosperms, only three types of plants; all unusual, Have flagellated sperm

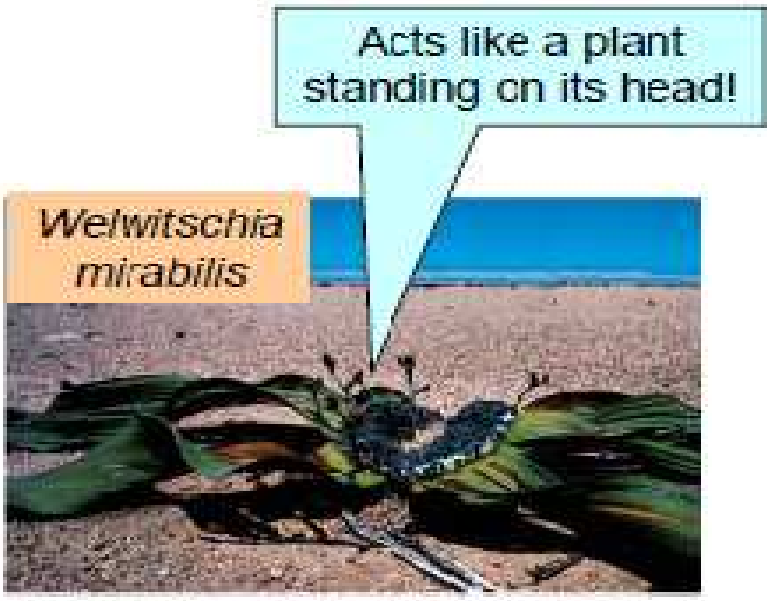


Fig. 3.20: Gnetophytes

3.2.6.4 Ginkgo

Only one living species exists; The maidenhair tree, Ginkgo biloba

Resistant to air pollution, reproductive structures found on different trees and have flagellated sperm.

Fig. 3.20: Ginkgo

3.2.7 Rise of the Angiosperms

Angiosperms comprise 90% of all living plants > 300,000 species, virtually all our food is derived, directly or indirectly from them

In gymnosperm reproduction, pollen grains are carried passively by the wind

Angiosperms have evolved more direct way of transferring pollen

Induce animals to carry it for them

Flowers are the reproductive organs of angiosperms , A flower employs bright colors to attract insects and nectar, to induce the insects to enter the flower, there they are coated with pollen grains, which they carry with them to other flowers

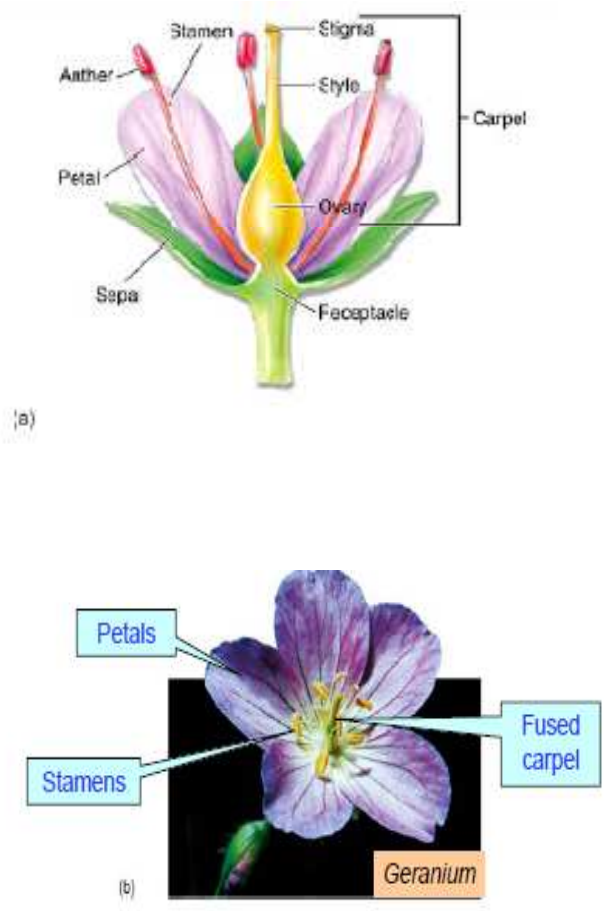


Fig. 3.22: flower (a and b)

3.2.7.1 Flower

A flower consists of four concentric circles, or whorls, connected to a base called the receptacle, outermost whorl (Sepals), protects flower from physical damage, Second whorl (Petals) attracts pollinators, Third whorl (Stamens) produces pollen grains in the anther, Innermost whorl (Carpel)

produces eggs in the ovary, Rising from the ovary, is a slender stalk, the style, with a sticky tip, the stigma

3.2.7.2 Fruits

A fruit is a mature ripened ovary containing fertilized seeds, fruits aid in the dispersal of seeds to new habitats

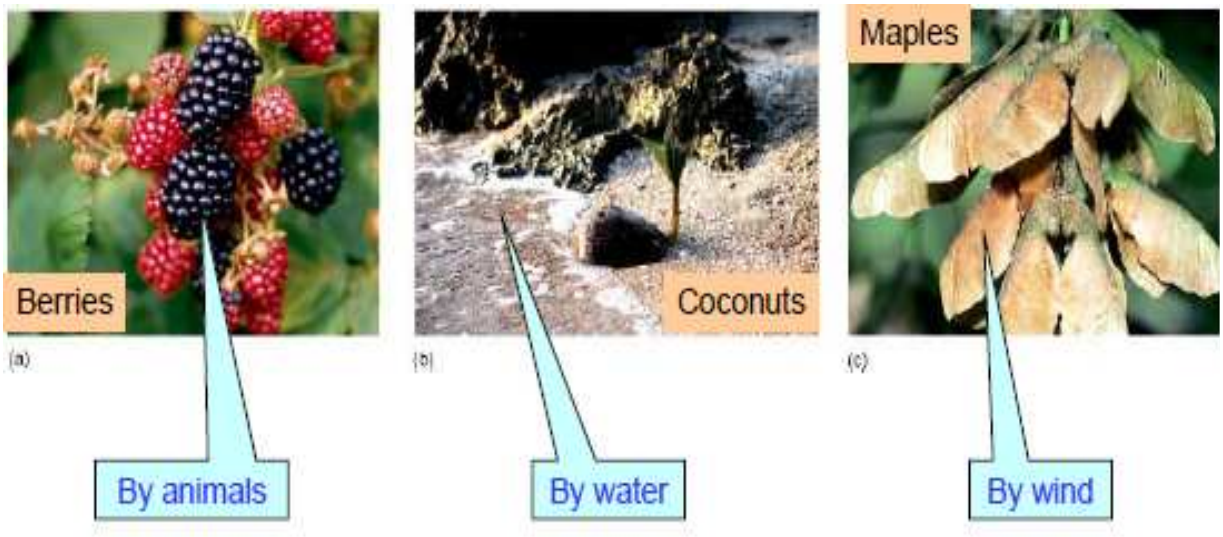


Fig. 3.23: Different ways of dispersing fruit

Flowers protected the egg and improved the odds of its fertilization

Fruits surrounded the seeds and aided in their dispersal

3.3 Plant Adaptations to Life on Land

Organisms in water do not face many of the challenges that terrestrial creatures do. Water supports the organism; the moist surface of the creature is a superb surface for gas exchange, etc. Approximately 288,700 species of plants are now in existence and most of them are terrestrial

However, green algae, the likely ancestors of plants, are aquatic and not well adapted to living on land

Three major challenges had to be overcome

1. Mineral absorption

2. Water conservation

3. Reproduction on land

3.3.1 Mineral Absorption

Plants require relatively large amounts of six inorganic minerals; Nitrogen, potassium, calcium, phosphorus, magnesium and sulphur. Plants absorb these materials through their roots

The first plants developed symbiotic associations with fungi, and these mycorrhizae enabled plants to extract minerals from rocky soil

3.3.2 Water Conservation

To avoid drying out, plants have a watertight outer covering, termed the cuticle Stomata (singular, stoma) are pores in the cuticle that allow gas and vapor exchange

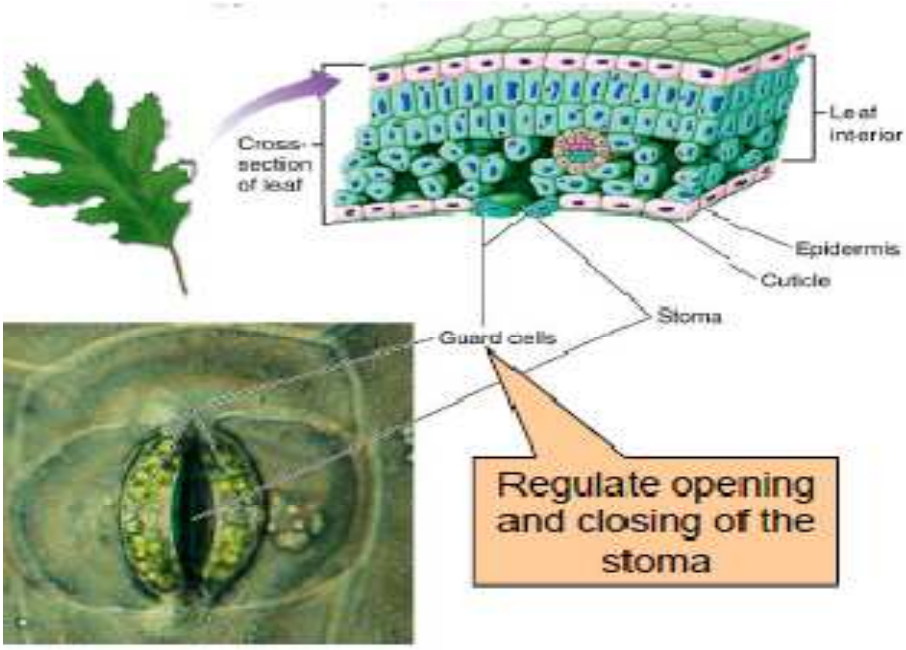


Fig 3.24: stoma and cuticle

Regulate opening and closing of the stoma

3.3.3 Reproduction on Land

Spores developed as a means to protect gametes from drying out on land in a plant life cycle, there is alternation of generations Diploid with haploid

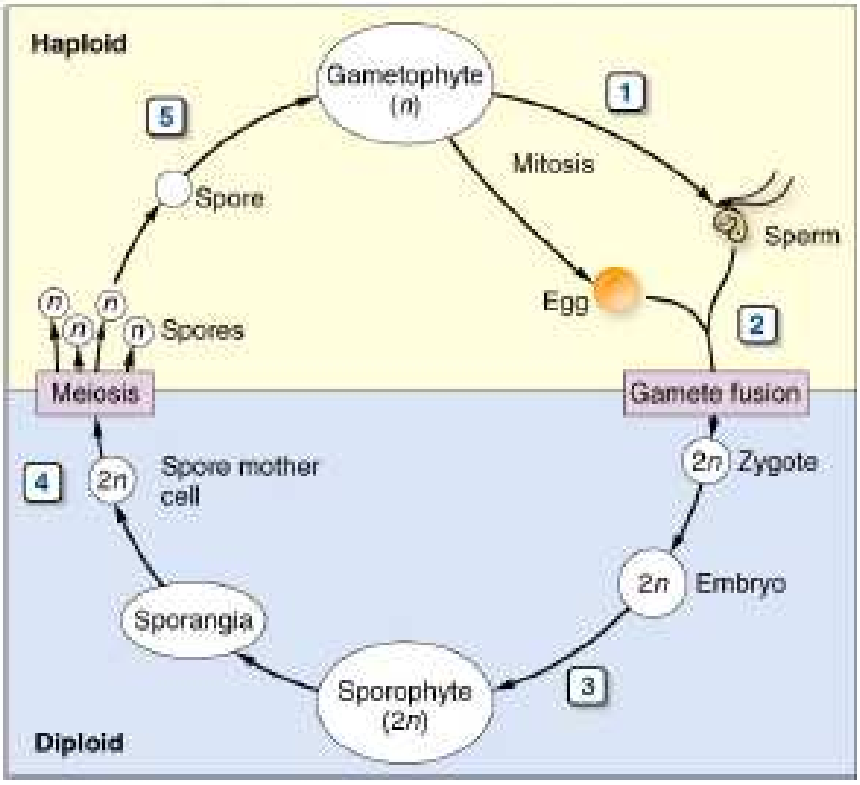


Fig. 3.25 Generalized plant life cycle

The diploid generation is called the sporophyte and the haploid generation is called the gametophyte. As plants evolved, the sporophyte tissue dominated

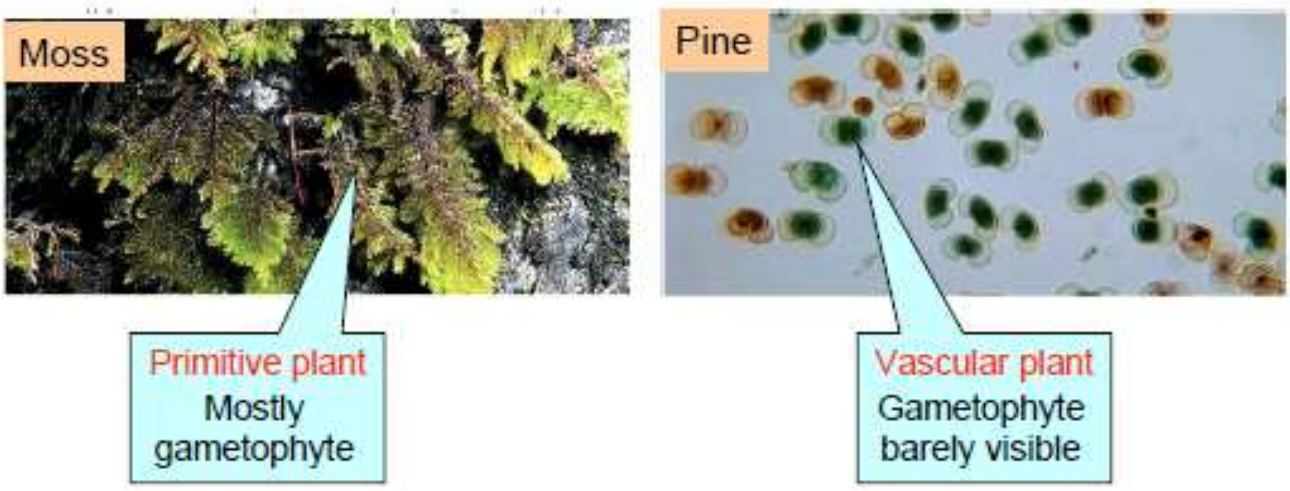


Fig. 3.26: Two types of gametophytes

3.4 Role of Oxygen

As we all know that oxygen is very important not only to animal but also plant. Oxygen plays many important roles but in this course we are going to look at the role it play in the respiration of plant.

Two most important prerequisites of life are continuous supply of materials for growth of body and energy for carrying out various life processes. All systems, from cell to ecosystem, require energy to work. However, the energy in the food has to be made available to the cells in a usable form. This is the role of respiration. Respiration is the process by which energy in organic molecules is released by oxidation. This energy is made available to the living cells in the form of ATP (Adenosine Tri Phosphate). The O_2 required for respiration is obtained from the atmosphere. ATP is the energy currency of the cell.

Respiration: Respiration is the stepwise oxidation of complex organic molecules and release of energy as ATP for various cellular metabolic activities. Respiration involves exchange of gases between the organism and the external environment. The plants obtain oxygen from their environment and return carbon dioxide and water vapour into it.

The biochemical process, which occurs within cells and oxides food to obtain energy, is known as cellular respiration. The processes by which cells obtain energy from complex food molecules depend upon whether or not oxygen is present in their environment and utilised. Respiration is termed aerobic when oxygen is utilised. In aerobic respiration the respiration are followed by an oxygen requiring process that releases much larger quantity of energy in the form of ATP. This occurs in the mitochondria of the eukaryotes and in the plasma membrane of the prokaryotes.

The rate of respiration increases with rise in oxygen concentration. As O_2 concentration increases from Zero, the rate of respiration increases. However, beyond a limit the rate of increase falls.

In plants, the atmospheric air moves in an out by simple diffusion that takes place through

a) The general body surface of the (stains, roots, fruits and seeds);
b) Lenticels (openings in the bark of the tree trunk)
c) Stomata present in the leaves and young stems.

From the atmosphere gases enter the intercellular spaces inside the plants. As o_2 is utilized, more of it diffuses into the plant. Since co_2 is being continuously formed, its concentration result in tissue spaces becomes higher than in the surrounding air. As a result, it diffuses out of the plant, especially when it is being used for photosynthesis.

In plants, o_2 released during photosynthesis in day time is made available for respiration. However, rate of photosynthesis is greater than that of respiration. Thus, plants give out excess o_2 in the daytime.

Oxygen that is absorbed is used to oxidize the nutrient, viz; glucose, amino acids and fatty acids completely producing co_2, water and energy. It occurs within the cells and tissues. Further oxidation of pyruvic acid requires o_2. It then enters mitochondria for aerobic respiration.

4.0 Conclusion

In this unit you learnt:

1. About evolution of the plants
2. About nonvascular and vascular plant
3. About multicellular development
4. Role of oxygen

5.0 Summary

All seed plants are derived from a single common ancestor. The plant kingdom contains multicellular phototrophs that usually live on land and all plant cells have a cell wall containing the carbohydrate cellulose, and often have plastids in their cytoplasm. The plant life cycle has an alternation between haploid (gametophyte) and diploid (sporophyte) generations. There are more than 300,000 living species of plants known, as well as an extensive fossil record.

Plants divide into two groups: plants lacking lignin-impregnated conducting cells (the nonvascular plants) and those containing lignin-impregnated conducting cells (the vascular plants). Living groups of nonvascular plants include the bryophytes: liverworts, hornworts, and mosses. Water supports the organism; the moist surface of the creature is a superb surface for gas exchange, etc.

6.0 TUTOR-MARKED ASSIGNMENT

1. List four key evolutionary innovations serve to trace the evolution of the plant kingdom
2. List four phyla of non flowering seed plant
3. What are the important of xylem and phloem

7.0 REFERENCES/FURTHER READINGS

Labandeira, C. (2007). "The origin of herbivory on land: Initial patterns of plant tissue consumption by arthropods". *Insect Science* 14 (4): 259–275. doi:10.1111/j.1744-7917.2007.00152.x.

"The oldest fossils reveal evolution of non-vascular plants by the middle to late Ordovician Period (~450-440 m.y.a.) on the basis of fossil spores" Transition of plants to land

Haines, Tim. 2000. *Walking with Dinosaurs: A Natural History*, (New York: Dorling Kindersley Publishing, Inc.) ISBN 0-563-38449-2. Page 65.

Behrensmeyer, Anna K., Damuth, J.D., DiMichele, W.A., Potts, R., Sues, H.D. & Wing, S.L. (eds.). 1992. *Terrestrial Ecosystems through Time: the Evolutionary Paleoecology of Terrestrial Plants and Animals*, (Chicago & London: University of Chicago Press), ISBN 0-226-04154-9 (cloth), ISBN 0-226-04155-7 (paper). Page 349.

Butterfield, N. J. (Jan 2009). "Oxygen, animals and oceanic ventilation: an alternative view". *Geobiology* **7** (1): 1–7. doi:10.1111/j.1472-4669.2009.00188.x. ISSN 1472-4677. PMID 19200141. edit

Steemans, P.; Hérissé, L.; Melvin, J.; Miller, A.; Paris, F.; Verniers, J.; Wellman, H. (Apr 2009). "Origin and Radiation of the Earliest Vascular Land Plants". *Science* **324** (5925): 353–353. Bibcode 2009Sci...324..353S. doi:10.1126/science.1169659. ISSN 0036-8075. PMID 19372423. edit

omescu, A. M. F.; Rothwell, G. W. (2006). "Wetlands before tracheophytes: Thalloid terrestrial communities of the Early Silurian Passage Creek biota (Virginia)". *Wetlands Through Time*. doi:10.1130/2006.2399(02). ISBN 9780813723990. http://www.humboldt.edu/biosci/docs/faculty/TomescuRothwell2006.pdf. Retrieved 2011-02-22. edit

Tomescu, A. M. F.; Honegger, R.; Rothwell, G. W. (2008). "Earliest fossil record of bacterial–cyanobacterial mat consortia: the early Silurian Passage Creek biota (440 Ma, Virginia, USA)". *Geobiology* **6** (2): 120. doi:10.1111/j.1472-4669.2007.00143.x. PMID 18380874. edit

Scott, C.; Glasspool, J. (Jul 2006). "The diversification of Paleozoic fire systems and fluctuations in atmospheric oxygen concentration" (Free full text). *Proceedings of the National Academy of Sciences of the United States of America* **103** (29): 10861–10865. Bibcode 2006PNAS..10310861S. doi:10.1073/pnas.0604090103. ISSN 0027-8424. PMC 1544139. PMID 16832054. http://www.pnas.org/cgi/pmidlookup?view=long&pmid=16832054. edit