COURSE GUIDE

BIO307
EVOLUTION

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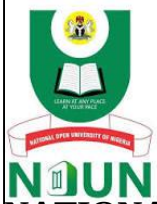

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Introduction

Evolution (BIO 307) is a fundamental course for undergraduate students of Biology that deals with 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.

Course Competencies

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

The Comprehensive Objectives of the Course are to;

- Explain the historical concept of evolution
- Explain the basic of population genetics and gene frequency/equilibrium
- Explain the origin of life and evidence of evolution.
- State the Phyolgeny and geological period and epochs

Working through this 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.

There will be a final examination at the end of the course. The course should take you about 8 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.

Study Units

The course is divided into 3 modules and study units in this course are given below:

BIO 307 EVOLUTION (2 UNITS)

MODULE 1: THEORIES OF EVOLUTION

Unit 1: History and Mechanisms of Evolution
Unit 2: Population Genetics
Unit 3: Gene Frequency/Equilibrium
Unit 4: Hardey-Weinberg Principle
Unit 5: Polymorphism

MODULE 2: GENE VARIATION: TYPES AND CAUSES

Unit 1: Genetic variation
Unit 2: Reshuffling of genes
Unit 3: Mutation, origin and types
Unit 4: Polyploidy
Unit 5: Isolation mechanisms

MODULE 3: EVOLUTION OF LIFE

Unit 1: Origin of Life
Unit 2: Evidence of evolution, adaptation and Speciation
Unit 3: Evolution of the plants
Unit 4: Classification and phylogeny
Unit 5: Geological periods and epoch

References and Further Readings

You would be required to read the recommended references and textbooks in each unit of the course materials.

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 at the stipulated date and time.
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Assessment

There are three aspects to the assessment of this course.
The first one is the in-text questions and the second is self-assessment exercises, while the third is the written examination or the examination to be taken at the end of the course.
Review the exercises or activities in the unit by applying the information and knowledge you acquired during the course.
The work submitted to your tutor for assessment will account for 30% of your total work.
At the end of this course, you will have to sit for a final or end of course examination of about a three hour duration and this will account for 70% of your total course mark.

How to get the Most from the 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 in-text questions and Self-assessment questions 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 Animal Behaviour, a sub-discipline and very interesting field of Biological Science.

Online Facilitation

Eight weeks 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.
The duties of your facilitator is to monitor your progress and provide any necessary assistance you need.
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.

Course Information

Course Code: BIO 307
Course Title: EVOLUTION
Credit Unit: 2
Course Status: Compulsory
Course Blub: This course designed to enable students to understand different theories of evolution postulated and genetic basis of evolution in animals and plant in relation to their development.

Semester: FIRST SEMESTER
Course Duration: 13 WEEKS
Required Hours for Study: 65 HOURS

Ice Breaker

Dr**.** Esenowo, Imeh Kokoete is a Senior Lecturer of Ecology and Environmental Biology in the Department of Animal and Environmental Biology, University of Uyo. Dr. Esenowo moderate and facilitate courses in the National Open University. He has supervised student projects and seminar review in the Department of Biology, Faculty of Science.
Dr. Esenowo research interests are; physico-chemical aspects of freshwater and terrestrial ecosystem, fish biology and environmental toxicology.

Module 1: Theories of Evolution

Module Introduction

In Module One, unit one deals with the historical aspects of ethology, population of genetics, the Hardey-Weinberg Principle and Polymorphism with its different forms.

Unit 1: History and Mechanisms of Evolution
Unit 2: Population Genetics
Unit 3: Gene Frequency/Equilibrium
Unit 4: Hardey-Weinberg Principle
Unit 5: Polymorphism
Glossary

Unit 1: History and Mechanisms of Evolution

Contents

1.1 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.

1.2 Intended Learning Outcomes (ILOs)

At the end 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,
- Understand Darwin's theory of natural selection

1.3 Historical Background of Evolution

i) 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.

ii) 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.

iii) 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.

iv) Immanuel Kant

Immanuel Kant the German philosopher 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.

v) 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.

vi) 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.

vii) 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.

viii) **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.

ix) **Charles Darwin and Alfred Russel Wallace**

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.

Figure 1. Charles Darwin Alfred Wallace
1809-1882 1823-1913

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.

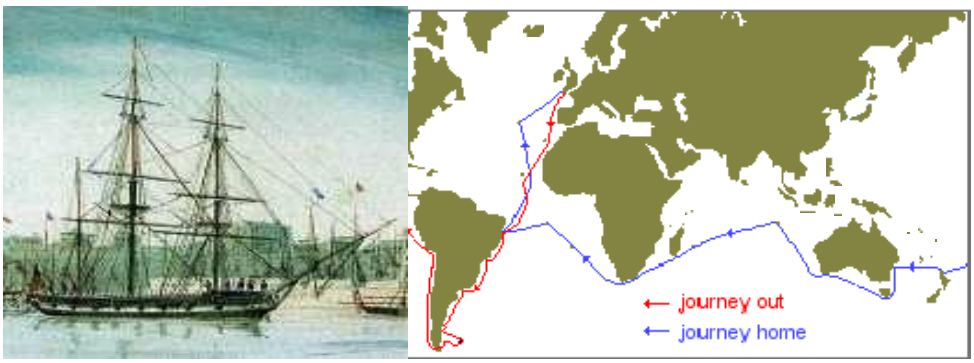

Figure 2: Expedition on board to the Galápagos Islands in the Eastern Pacific Ocean.
Source: www2.palomar.edu/anthro/evolve/evolve_2.html

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.

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.

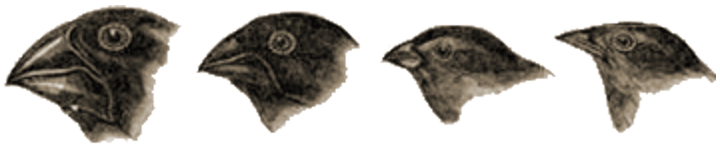

Fig 3: Finches from the Galapagos Islands
Source: www2.palomar.edu/anthro/evolve/evolve_2.htm

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.

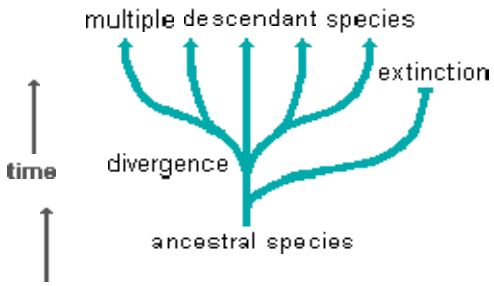

Figure 4: Show the branching evolution in which species become reproductively isolated.

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 human, using paleontology and comparative anatomy to provide strong evidence that human 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.

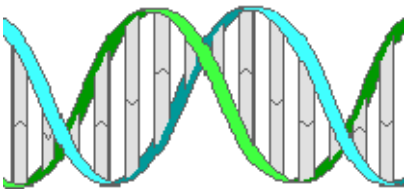

Figure 5: Section of a DNA molecule

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 foundation 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.What is Evolution?
Who is the father of modern taxanomy? Who is Charles Darwin's grandfather?

Self-Assessment Exercises 1

Attempt these exercises to measure what you have learnt so far. This should not take you more than 5 minutes.

1. Thomas Malthus' (1766-1834) theory of population growth inspired who?
2. What is Medieval theories?

1.4 Summary

The concept of evolution has been the center 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 centered 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.

1.5 References/Further Readings/Web Sources

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1.6 Possible Answers to SAEs

Answers to SAEs 1

1. 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.

2. 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

Unit 2: Population Genetics

Contents

2.1 Introduction

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.

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.

2.2 Intended Learning Outcomes (ILOs)

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

- Define Population Genetics
- Describe the History of Population Genetics

2.3 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.
What is Population Genetics? What is Ordinary genetics?

Self-Assessment Exercises 1

Attempt this exercises to measure what you have learnt so far. This should not take you more than 5 minutes.

1. What is Sexual population?
2. Population genetics is based on whose work?

2. 4 Processes of Population Genetics

i) 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.

ii) Genetic Drift

Genetic drift is referred to 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 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 given below;

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

Where t = t generation and, N_e = Log of total number of male and female.

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

iii) 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. 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.

iv) 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.

v) 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.

vi) 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. 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 structure. Genetic structuring can be caused by migration due to historical climate change, species range expansion or current availability of habitat.

vii) 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.
Define Genetic drift? What is Horizontal Gene Transfer?

Self-Assessment Exercises 2

Attempt this exercise to measure what you have learnt so far. This should not take you more than 5 minutes

1. What is Natural Selection?
2. What is the effect of genetic drift?

2.5 Summary

Population genetics is a field of biology that studies the genetic composition of biological population, 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.

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2.7 Possible Answers to SAEs

Answers to SAEs 1

1. A sexual population is a set of organisms in which any pair of members can breed together.

2. Population genetics as a discipline was based on the work of Fisher, Haldane and Wright

Answers to SAEs 2

1. Selection refers to changes in allele frequencies due to the effects of the gene on its host.

2. Genetic drift may cause gene variants to disappear completely, and thereby reduce genetic variability.

Unit 3: Gene Frequency/Equilibrium

Contents

3.1 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 the condition of an allele or genotype in a gene pool (such as a population) where the frequency does not change from generation to generation. 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.

3.2 Intended Learning Outcomes (ILOs)

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

- Define genetic frequency and equilibrium
- Understand the significance of genetic frequency
- Relate genetic frequency and the Hardy-Weinberg Principle

3.3 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: a particular locus on a chromosome and the gene occupying that locus 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) different alleles of the gene exist 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.

i) 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).

ii) The effect of mutation on Genetic frequency

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

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

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,

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

3.3.1 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 mutation 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.
What is genetic equilibrium?

Self-Assessment Exercises 1

Attempt this exercises to measure what you have learnt so far. This should not take you more than 5 minutes.

1. What is Genetic or Allele frequency?

3.4 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.

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3.6 Possible Answers to SAEs

Answers to SAEs 1

1. 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

Unit 4: Hardy-Weinberg Principle

Contents

4.1 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.

4.2 Intended Learning Outcomes (ILOs)

At the end of this topic students should be able to;

- Explain 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

4.3 Hardy-Weinberg Principle

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:

1. mutation is not occurring
2. natural selection is not occurring
3. the population is infinitely large
4. all members of the population breed
5. all mating is totally random
6. everyone produces the same number of offspring
7. 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:

$p = AA + ½Aa$

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).

$q = aa + ½Aa$

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.

i) 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**

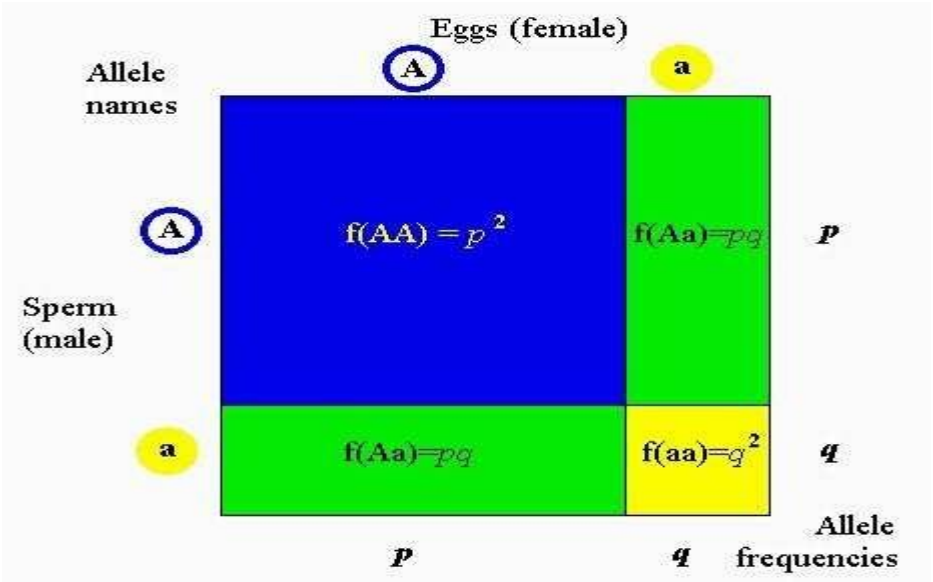

Fig 2: Diagram of Hardy-Weinberg genotype proportions from male (sperm) and female (egg) contributions.

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, 2*pq*, 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.

$AA = p^2 = 0.75 * 0.75 = 0.5625$
$Aa = 2pq = 2 * 0.75 * 0.25 = 0.375$
$aa = q^2 = 0.25 * .025 = 0.0625$ **Eqn. 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**

ii) **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

Table 1: Showing all possible genotype pairing

		Offspring genotype		
Mating	**Frequency**	**A1A1**	**A1A2**	**A2A2**

A1A1 x A1A1	**x^2_{11}**	**1**	**0**	**0**
A1A2	**x11x12**	½	½	**0**
A2A2	**x11x22**	**0**	**1**	**0**
A1A2 x A1A1	**x12x11**	½	½	**0**
A1A2	**x^2_{12}**	¼	½	¼
A2A2	**x12x22**		½	½
A2A2 x A1A1	**x22x11**	**0**	**1**	**0**
A1A2	**x22x12**	**0**	½	½
A2A2	**x^2_{22}**	**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:

2p1 = 2(50)/200 = 0:5, p2 = (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 mating's 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$11 + 1/2x11 x12 + 1/2x12 x11 + 1/4$x^2$12

=$x^2$11+x11x12+1/4$x^2$12

= $(x11 + x12/2)^2 = p^2$

freq. (A1A2 in zygotes) = 2pq

freq. (A2A2 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
What is the first Assumption of the Hardy-Weinberg Principle?

Self-Assessment Exercises 1

Attempt this exercises to measure what you have learnt so far. This should not take you more than 5 minutes.

1. What did Godfrey Hardy and Wilhelm Weinberg develop?

4.5 Summary

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.

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4.7 Possible Answers to SAEs

Answers to SAEs 1

1. 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).

Unit 5: Polymorphism

Contents

5.1 Introduction

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 polymorphism, 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.

5.2 Intended Learning Outcomes (ILOs)

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

- Define polymorphism?
- State and discuss types of polymorphism

5.3 Polymorphism

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).

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).

i) Nomenclature in Polymorphism

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 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).

ii) Ecology and Polymorphism

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.

iii) 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.

vi) 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 males are haploid while the females are diploid. 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. When does Polymorphism occur? What is The Switch?

Self-Assessment Exercises 1

Attempt this exercises to measure what you have learnt so far. This should not take you more than 5 minutes.

1. What is the effect of selection on polymorphism?

5.4 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.

i) Mechanism of Balancing Selection

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

2. 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.

3. 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.

4. 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.

ii) 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.

iii) 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.

iv) 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. Define Genetic polymorphism.

Self-Assessment Exercises 2

Attempt this exercises to measure what you have learnt so far. This should not take you more than 5 minutes.

1. What is Pleiotropism?
2. What is Epistasis?

5.4 Summary

Polymorphism is 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 higher organisms into male and female sexes.

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5.6 Possible Answers to SAEs

Answers to SAEs 1

1. Selection, whether natural or artificial, changes the frequency of morphs within a population; this occurs when morphs reproduce with different degrees of success.

Answers to SAEs 2

1. Pleiotropism occurs when genes have more than one effect on the phenotype of an organism

2. 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

Glossary

AIDs = Acquired immunodeficiency syndrome
ATP =Adenosine triphosphate
°C = degrees Celsius
cm = centimeters
CH4 =Methane
CO = Carbon
CH_4 = methane (CH_4)
DNA =Deoxyribonucleic acid
DSBs = Double strand breaks
°F = degree Fahrenheit
H2 = Hydrogen
ICBN = International Code of Botanical Nomenclature
ICZN = International Code of Zoological Nomenclature
Kgs = kilograms.
km = kilometers
m = meters
mm = millimeters
NH2OH = Hdroxylamine
NH3 = Ammonia
O_2 = Oxygen
Pg = picograms
RNA = Ribonucleic acid

L = length
HIV = Human immunodeficiency virus
% = percentage
g = grams
spp =species
UV = Ultraviolet
< =Less than

End of the Module Questions

1. What did Immanuel Kant develop?
2. List the five condition for equilibrium to remain in effect.
3. List the seven conditions if evolution will not occur in a population
4. What is the 4^{th} Assumption in derivation of the Hardy-Weinberg Principle
5. Who is the founder of niche research?

Module 2: Gene Vairation: Types and Causes

Module Introduction

Unit 1: Genetic variation
Unit 2: Reshuffling of genes
Unit 3: Mutation, origin and types
Unit 4: Polyploidy
Unit 5: Isolation mechanisms
Glossary

Unit 1: Genetic Variation

Contents

1.1 Introduction

Populations evolve and 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 micro-evolutionary 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.

1.2 Intended Learning Outcomes (ILOs)

At the end of lecture, the students should be to:

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

1.3 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.

1.3.1 Examples 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.

i) 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.

ii) 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.

iii) 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 and therefore the order of genes 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 supernumerary 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.

iv) 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.

v) 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 of over 2,300 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. What is variation?
What is the most direct method of studying DNA variation?

Self-Assessment Exercises 1

Attempt these exercises to measure what you have learnt so far. This should not take you more than 5 minutes.

1. What is inversion?
2. What are Restriction enzymes?

1.5 Summary

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?

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1.7 Possible Answers to SAEs

Answers to SAEs 1

1. Inversion occurs when the banding pattern and therefore the order of genes of a region of the chromosome is inverted.

2. Restriction enzymes provide another method of studying DNA variation. Restriction enzymes exist naturally in bacteria and a large number over 2,300 of restriction enzymes are known.

Unit 2: Reshuffling Of Genes

Contents

2.1 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.

2.2 Intended Learning Outcomes (ILOs)

At the end of lecture the students should be able to understand the meaning of

- Genetic recombination
- Chromosomal crossover
- Sexual reproduction

2.3 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 biochemical researchers at 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.

i) 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.

ii) 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.

1. Sexual Reproduction in Plants

Animals typically produce male gametes called sperm, and female 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 develop 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.

2. Sexual Reproduction in 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. Sexual Reproduction in 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.
Define Genetic recombination? Where do recombination occurs?

Self-Assessment Exercises 1

Attempt this exercises to measure what you have learnt so far. This should not take you more than 5 minutes.

1. Explain Chromosomal crossover?
2. What do you understand by Sexual reproduction?

2.4 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.

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2.6 Possible Answers to SAEs

Answers to SAEs 1

1. Chromosomal crossover refers to recombination between the paired chromosomes inherited from each of one's parents, generally occurring during meiosis.

2. Sexual reproduction is the primary method of reproduction for the vast majority of macroscopic organisms, including almost all animals and plants.

Unit 3: Mutation

Contents

3.1 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.

3.2 Intended Learning Outcomes (ILOs)

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

- Define mutation,
- List types of mutation and
- Mention causes mutation.

3.3 Mutation

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 steps 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.

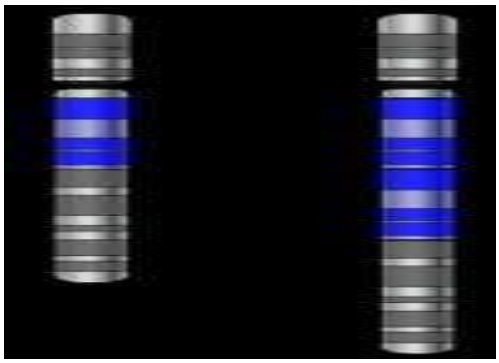

Fig 1: Duplication of part of a chromosome

3.3.1 Classification of mutation

Chromosome mutations are alterations that affect whole chromosomes and whole genes rather than just individual nucleotides. These mutations result from errors in cell division that cause a section of a chromosome to break off, be duplicated or move onto another chromosome.

A) Structural Mutation are divided into the following types depending on the mechanism of the process;

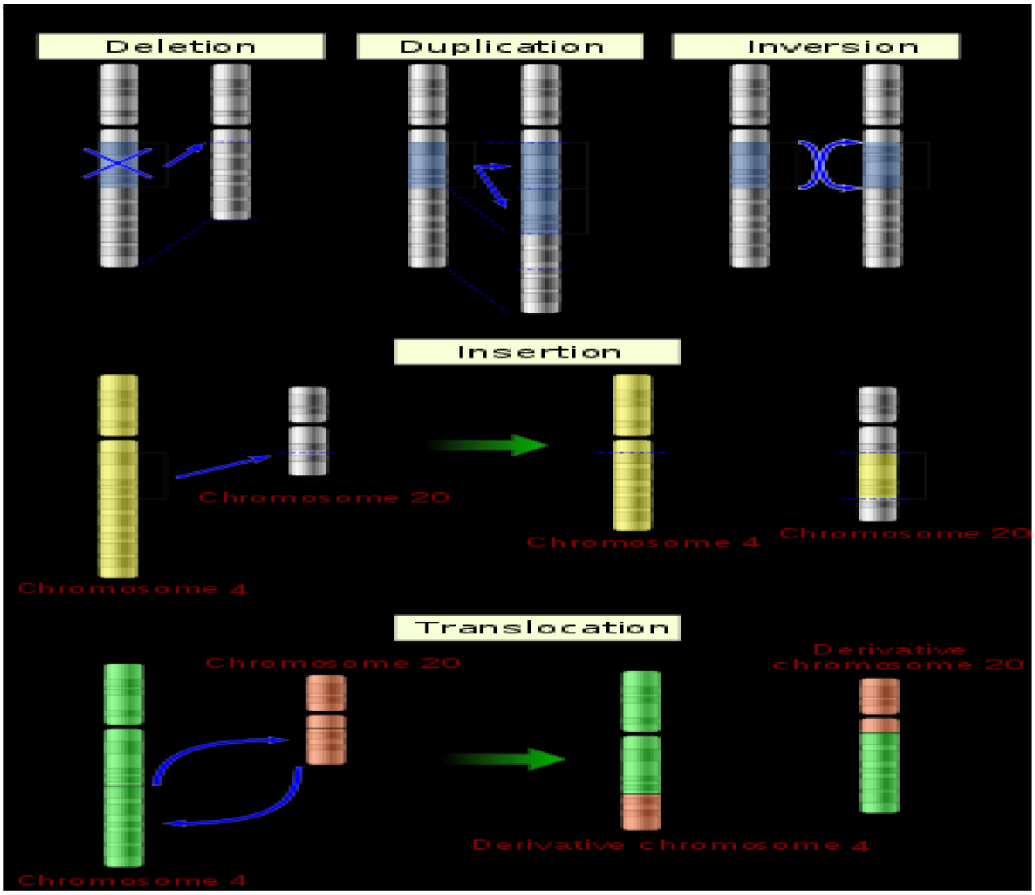

Fig 2: Five types of chromosomal structural mutations.

i) Deletions. This type of mutation occurs when a part of the DNA is not duplicated or is lost during DNA replication. Deletions remove one or more nucleotides from the DNA. Like insertions, these mutations can alter the reading frame of the gene. They are irreversible. Common disorders due to deletion mutation in humans are: Cri du chat, Duchenne muscular dystrophy, Di George's syndrome, etc.

Examples of Deletion

- Deletion of the short arm of chromosome 5 in humans results in a distinctive cat-like cry in babies.
- It is also known as the French name 'cri du chat' syndrome, indicating the cat-like cry.
- These individuals tend to be mentally slow with an IQ below 20 and have different forms of malformation in the larynx, moon faces, saddle noses, and small mandibles.
- The syndrome can be inherited from one generation to another can might even affect the ears and the size of the head.

ii) 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.

iii) Duplication: This type of mutation occurs when an extra copy of a region (or regions) in the DNA is produced. This duplicated region can either be located in its normal location in the chromosome or sometimes be located in other parts of the chromosomes or even in another chromosome. Common disorder due to duplication mutation in humans is: *Charcot-Marie-Tooth disease* type I.

Examples of Duplication

- The duplication of a segment of the X-chromosome, called section 16A, in *Drosophila* is a classic example of duplication.
- Section 16A codes for the bar trait in *Drosophila*, which is characterized by a narrower, oblong, bar-shaped eye with a few facets.
- Each duplicated section 16A intensifies the bar phenotype, which increases the narrowing effect.
- The duplication of the section acts as a genetically dominant factor, and the phenotype intensifies if the duplicated genes occur on the same chromosome.

iv) Inversion: Inversion is a type of structural mutation where a part of chromosomes or a set of genes rotates by 180° on its own axis. During inversion, a portion in the chromosome is reversed and gets inserted back into the chromosome. There is no net loss or gain of genes but simply a rearrangement of the sequence. A part of the chromosome is broken and then rejoined in a different direction. Inversion mutation can be detected cytologically in the meiotic nuclei by the detection of an inversion loop in the paired homologs. The genetic behavior of the changed chromosome depends on the location of the centromere from the site of inversion. Basically, two types of inversion exist: *pericentric* and *paracentric*.

- During a pericentric inversion, the inversion encompasses the **centromere** of the chromosome.
- On the other hand, during a paracentric inversion, it only involves either the short or long arm of the chromosome and the inversion point does not include the centromere.

Examples of inversion

- An example of chromosomal inversion can be observed in the insect, *Coelopa frigid*, where the chromosomal inversion results in the production of differences in phenotype.
- Chromosomal inversion occurs only in the larger species as the smaller species cannot survive the mutation.

- The changes observed as a result of the mutation are a three-fold difference in size in males. The change heterokaryotype has higher viability than the original structure.
- Thus, inversion mutation in the species acts as an evolutionary asset in the species, which increases its fitness in the ecosystem.

Common disorder due to inversion mutation in humans is: *Amniocentensis* during pregnancy.

v) Translocation: Translocation happens when a fragmented chromosome tends to join with a non-homologous chromosome. This newly-formed segment then detaches from the chromosome and moves to a new position on another chromosome. Common disorders due to translocation mutation in humans are: XX male syndrome, Down syndrome, Infertility and Cancer.
Examples of Translocation

- A rare series of reciprocal translocation can be observed in *Oenothera* involving all 7 of the chromosome pairs.
- Multiple translocations exist within the set, which produces different lethal combinations.
- The translocation results in a ring-like structure of the chromosome where the gametes usually do not survive the mutation.
- Viable gametes are formed only when the linkages are alternate disjunction from the ring structure.

B) Chromosomal Number Mutation: Chromosomal mutations II include mutations that are caused by the alterations in the number of chromosomes in a cell. The change in the number of whole chromosomes is called heteroploidy. It produces phenotypic changes, modifications of phenotypic ratios, and alteration of linkage groups. Heteroploidy can be further divided into two different categories depending on the changes in the entire set of chromosomes or in the single whole chromosome.

i) Aneuploidy

- Aneuploidy is a type of mutation that changes parts of a chromosome set, resulting in either the loss of one or more chromosomes or the addition of chromosomes.
- Aneuploidy resulting from the loss of chromosomes is called hypoploidy, whereas that due to the addition of chromosomes is called hyperploidy.
- Hypoploidy usually occurs due to the loss of a single chromosome (monosomy) or due to the loss of a pair of chromosomes (nullisomy).
- Hyperploidy, in turn, might involve the addition of a single chromosome (trisomy) or the addition of a pair of chromosomes (tetrasomy).

- Aneuploids are caused as a result of nondisjunction during mitosis or meiosis.
- If aneuploidy occurs in the gametes of plants, those do not survive, but in animals, some genetic imbalance leading to higher mortality or reduced fertility might occur.

Example of Aneuploidy

- Down's syndrome is an example of aneuploidy which is associated with a trisomic condition for one of the smallest human autosomes (chromosome no. 21).
- Down's syndrome is the most common chromosomal abnormality in live births and exhibits 50 different physical characteristics.
- The characteristics can range from mild and moderate mental retardation to internal epicanthal folds and swollen tongue.
- The main cause of the trisomy is the nondisjunction of chromosome 21 during oogenesis. Down syndrome in children can also occur due to nondisjunction chromosome pairs in spermatogenesis.

ii) Polyploidy

- Polyploidy is a type of euploidy (changes in the entire set of chromosomes) where an organism has more than two sets of genomes (2x).
- Polyploidy includes different combinations like triploid, tetraploid, pentaploid, hexaploid, and octoploid.
- Polyploidy higher than tetraploid is not common in natural environments, but it can be observed in some crops and ornamental flowers.
- Polyploidy can be further divided into three groups; autopolyploids, allopolyploids, and autoallopolyploids.
- Autopolyploids are polyploids that consist of the same basic set of chromosomes but multiplied to form multiple sets.
- Allopolyploids are the polyploids that result from the doubling of chromosome number in a hybrid from two different species.
- The most common morphological effect of polyploidy is gigantism that is commonly seen in large-sized pollen and cells. Besides, it also reduces the fertility of plants to varying degrees.

Example of Polyploidy

- An example of polyploidy can be observed in 'doob' grass (*Cynodon dactylon*) which is mostly cultivated in South Asia.
- It is triploid and sterile but can be propagated vegetatively. Polyploidy results in large-sized plants that have decreased osmotic pressure with increased water content.

- The rate of cell division is low, and thus, the plant growth rate also decreases. These also have reduced auxin content which decreases their rate of respiration

C) Causes of mutation

i) Spontaneous mutation 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.

ii) Induced mutation on the molecular level can be caused by:

- Chemicals; Hydroxylamine NH2OH, 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

D) 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.

E) 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.

In-Text Question(s)

What are Chromosome mutations?
Answer: Chromosome mutations are alterations that affect whole chromosomes and whole genes rather than just individual nucleotides. These mutations result from errors in cell division that cause a section of a chromosome to break off, be duplicated or move onto another chromosome. List the five types of Structural Chromosome mutations?

Self-Assessment Exercises 1

Attempt this exercises to measure what you have learnt so far. This should not take you more than 5 minutes.

1. Define Deletion?
2. What is Translocation?
3. Explain Aneuploidy?

3.4 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 unpleasant, 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.

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3.6 Possible Answers to SAEs

Answers to SAEs 1

1. **Deletions**. This type of mutation occurs when a part of the DNA is not duplicated or is lost during DNA replication. Deletions remove one or more nucleotides from the DNA.

2. **Translocation:** Translocation happens when a fragmented chromosome tends to join with a non-homologous chromosome.

3. Aneuploidy is a type of mutation that changes parts of a chromosome set, resulting in either the loss of one or more chromosomes or the addition of chromosomes.

Unit 4: Polyploidy

Contents

4.1 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.)

4.2 Intended Learning Outcomes (ILOs)

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

- The Importance of Polyploidy
- The different Polyploidy in animals and plants

4.3 POLYPLOIDY

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. The diagram below shows the different types of polyploidy.

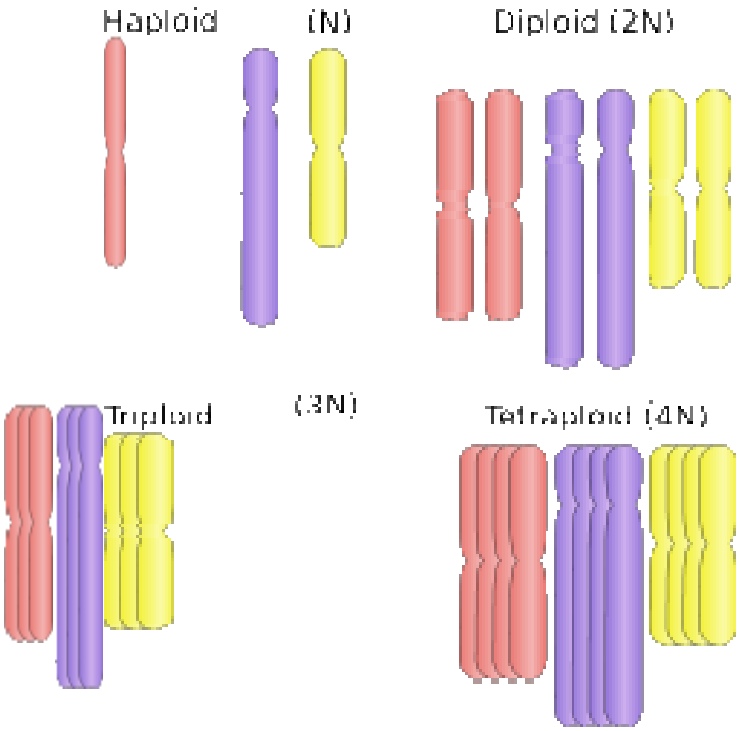

Fig 1: This image shows haploid (single), diploid (double), triploid (triple), and (quadruple) sets of chromosomes.

During meiosis, homologous chromosomes pair 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:

1. Triploid (three sets; 3x), for example seedless watermelons, common in the phylum Tardigrada
2. Tetraploid (four sets; 4x), for example Salmonidae fish
3. Pentaploid (five sets; 5x), for example Kenai Birch (*Betula papyrifera* var. *kenaica*)
4. Hexaploid (six sets; 6x), for example wheat, kiwifruit
5. Octaploid (eight sets; 8x), for example *Acipenser* (genus of sturgeon fish), dahlias decaploid (ten sets; 10x), for example certain strawberries
6. 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 are believed to have undergone a doubling of their genome in the past but now behave as diploids. 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 some of which may lead the organism to a more diploid-like state. In some polyploids, chromosomal re-organization 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.

The phenomenum 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. 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. 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. 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. 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.

Allopolyploidy presents a paradox because it is both a diversifying force and a genetic bottleneck. 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.

4.3.1 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.

1. The gametes may be formed by mitosis instead of meiosis.

2. 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.

1. 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**.

2. 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.

1. *B. oleracea* (cabbage, broccoli, etc.) hybridized with *B. nigra* (black mustard) → *B. carinata* (Abyssinian mustard).
2. *B. oleracea* x *B. campestris* (turnips) → *B. napus* (rutabaga)
3. *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.

i) 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. 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. 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.
Table 1: 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.

ii) Polyploidy in animals

Examples in animals are more common in the 'lower' forms such as flatworms, leeches, brine shrimp goldfish, salmon, and salamanders. 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 23rd 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.

iii) Polyploidy in Man

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 favouring the diploid cells. What is Polyploidy? How can polyploids can acquire variation?

Self-Assessment Exercises 1

Attempt this exercises to measure what you have learnt so far. This should not take you more than 5 minutes.

1. What is the origin of polyploidy?
2. How can polyploidy be induced in the plant –breeding?

4.4 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.

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4.6 Possible Answers to SAEs

Answers to SAEs 1

1. Polyploidy process can begin if **diploid** (**2n**) gametes are formed. These can arise in at least two ways.

i) The gametes may be formed by mitosis instead of meiosis.

ii) 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.

i) 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**.

ii) When rhizobia infect the roots of their legume host, they induce the infected cells to undergo endoreplication producing cells that can become **128n**

2. 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.

Unit 5: Isolation Mechanism

Contents

5.1 Introduction

Any external or internal barrier, which prevents interbreeding between populations, is called isolation. As a result, the population of a species is either separated into smaller units or exchange of genes (gene flow) between them is prevented. Hence, genetic factors such as mutation, recombination, genetic drift, natural selection, etc. occurring in one population will not have any effect on the other population. In the course of time each segregated population forms a separated unit. Thus isolation helps in splitting the population of a species into separate units and their subsequent evolution into distinct species.

5.2 Intended Learning Outcomes (ILOs)

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

- Explain what isolation mechanism?
- State the different types of isolation mechanism.

5.3 Isolation mechanisms

There are a number of processes by which two related populations living in the same area can remain distinct. These have been called Isolating mechanisms by Dobzhansky (1955). Isolation mechanism is the geographical and 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 *geographical* and *reproductive isolation*, i.e. a lack of gene mixture. According to him, the isolating mechanisms are classified into the following types.

5.3.1 Geographical isolation

When the populations are separated by a geographical barrier, such as river, sea, mountain, deserts and for aquatic animals land, they are physically prevented from interbreeding. Such populations are termed as allopatric and are forced to evolve independently and accumulate genetic differences. Geographical isolation may be different for different species.

Large bodies of water are barriers for land-dwelling animals. High Mountain ranges, deserts, dense forests and extremes of temperature serve as affective barriers. Such populations are completely 'out of touch with each other genetically' so that new mutations, genetic drift and the action of natural selection, in one population have no effect on the other population. Thus, a new population may be developed.

5.3.2 Reproductive isolation

It is the property of individuals that prevents interbreeding in populations that are actually sympatric (living in the same area). According to Muller reproductive isolation is due to differences in genes that arise during the origin of sub-species and species in population. According to Dobzhansky, reproductive isolation is the result of natural selection. Hybrids are either sterile or poorly adapted and are, therefore, eliminated by natural selection.

Classification of reproductive isolating mechanism are pre-mating and post-mating mechanisms.

i) Pre-mating mechansims

They prevent interspecific crosses in sympatric populations.

1. Seasonal isolation: Also called temporal isolation, in which potential mates do not come in contact with each other because of differences in breeding seasons of two species, e.g. different flowering seasons in plants. *Bufo americanus* breeds in early rainy season (May), while Bufo fowleri breeds in late rainy season (July) in USA.

2. Habitat isolation: Also called Ecological isolation, in which also potential mates do not meet each other due to differences in habitats, requirements of food, space, climate etc. Potential mates live in different areas and therefore do not come in contact with

one another. For example spawning grounds of riverine fishes are in different tributaries, which prevent interbreeding.

3. Ethological isolation: It is a behavioral isolation, in which potential mates meet but cannot mate, due to differences in courtship displays or other specific signals that are necessary rituals before mating. The signals may be of the following three types, which stimulate the opposite sex for mating.

a) Visual stimuli: Feather displays and dancing in male birds is necessary to attract the female, e.g. peacocks, pheasants and birds of paradise. The colour and shape of the feathers as well as display pattern is so unique for each species that mating between two different species is not possible. Collection of the nest material and construction of the nest as by the weaverbird male is also a very specific display that cannot be imitated by the other species.

b) Auditory stimuli: Songbirds like cuckoos, mynas, nightingales, parakeets etc. use auditory signals to attract the opposite sex. Sometimes the singing goes on for several days before the pair can actually come together for mating. Auditory communication is used by a large number of animals, viz. frogs, toads, cicadas, gibbons, monkeys, jackals etc.

c) Chemical stimuli: This includes odors of the animals that attract the opposite sex for mating. For example scent of musk deer and musthing in elephants attract the females. In insects, particularly Lepidoptera, females produce highly specific pheromones that can be detected by the highly specialized antennae of males from a distance of about 2 kilometers.

4. Mechanical isolation: In this case the above isolating mechanisms are not present and therefore mating is attempted but is not successful due to mechanical problems such as differences in the structure of genitalia. Dufour (1844) described “Lock and key mechanism” in the genitalia of insects. In the species of Drosophila the genitalia are so different that copulation is mechanically not possible.

5. Physiological isolation: This is another type of reproductive isolation in which mating may take place but the gametes are prevented from fertilization due to some physiological factors. For example: F. R. Lillie (1921) has experimentally demonstrated such a physiological disability in the sperms of a particular species of sea urchin in fertilizing the eggs of another species of sea urchin although both were brought together in the same medium. But when the eggs and sperms of the same species brought together, he found that the success of fertilization was nearly 100%.

6. Cytological isolation: In this type fertilization cannot take place due to differences in chromosome number between two species. For example, the bronze frog (*Rana clamitans*) and its close relative the bull frog (*Rana catesbeiana*) do not hybridize mainly because of cytological block to fertilization.

ii) Post-mating mechanisms

These reduce the success of interspecific crosses. In case premating mechanisms fail to prevent mating then several post-mating mechanisms prevent the success of mating and hybridization. There are 4 such mechanisms, which are given below:

1. Gamete mortality: Mating and sperm transfer takes place but egg is not fertilized. In Drosophila vaginal wall swells killing spermatozoa should interspecific crosses take place. If mating takes place between *Bufo fowleri* and *Bufo valliceps*, sperms cannot penetrate the egg membrane of each other, leading to mortality of gametes.

2. Zygote mortality: Egg is fertilized but the zygote dies. Eggs of many species of fishes may be present in the spawning grounds and some may be fertilized by the sperms of different species forming zygote but such zygotes fail to develop due to differences in chromosomes.

3. Zygote inviability: Zygote develops and hybrid is produced but is physically weak and inviable due to physiological disturbances in the body. It fails to survive for long and prematurely dies. Such cases have been recorded in different species of ducks.

4. Hybrid sterility: Hybrid is viable, physically strong and physiologically sound but is sterile due to differences in chromosomes and different gene arrangements. Mule is a cross between male donkey and female horse and Hinny between female donkey and male horse and both are sterile, albeit physically strong.

Sometimes all isolating mechanisms break, leading to fertile hybrids, which are generally not reproductively isolated from the parents and can produce fertile offsprings by Introgression (hybrids backcrossing with parents to produce fertile offsprings). This will be instant speciation.

The following are significance of isolation mechanism

i) Wasteful courtship is avoided. If isolating mechanisms are distinct and specific only individuals of the same species indulge in courtship.
ii) Isolating mechanism protects gene pool of a species and prevents hybridization.
iii) It prevents wastage of gametes and energy.
iv) A weak isolating mechanism leads to production of new species through hybridization.
v) Absence of isolating mechanism leads to production of new species by instant speciation.
vi) Geographical isolation followed by reproductive isolation ultimately leads to production of new species.
vii) Isolating mechanisms protect the identity of a species, which all species fiercely guard.

What is Geographical isolation? What do you understand by Zygote mortality?

Self-Assessment Exercises 1

Attempt this exercises to measure what you have learnt so far. This should not take you more than 5 minutes.

1. What is Reproductive isolation?
2. What is Ethological isolation?
3. What is Cytological isolation?

5.4 Summary

Isolating mechanisms are important for maintaining the integrity of species which is basic biological unit. Their importance was recognized even by Lamarck, Darwin and Wagner. It has been said that without isolation evolution is not possible. There are different types of isolating mechanisms which operate alone or in combination to maintain the integrity of species. Reproductive isolating mechanisms operating between sympatric species/populations play crucial role to prevent interbreeding between populations so that exchange of genes do not occur between them and their integrity is maintained. As a consequence of this, the genetic differences which they have acquired in the long run of time is maintained

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5.6 Possible Answers to SAEs

Answers to SAEs 1

1. Reproductive isolation is the property of individuals that prevents interbreeding in populations that are actually sympatric (living in the same area). According to Muller reproductive isolation is due to differences in genes that arise during the origin of sub-species and species in population. According to Dobzhansky, reproductive isolation is the result of natural selection. Hybrids are either sterile or poorly adapted and are, therefore, eliminated by natural selection.

2. Ethological isolation is a behavioral isolation, in which potential mates meet but cannot mate, due to differences in courtship displays or other specific signals that are necessary rituals before mating. The signals may be of the following three types, which stimulate the opposite sex for mating.

3. Cytological isolation occurs when fertilization cannot take place due to differences in chromosome number between two species. For example, the bronze frog (*Rana clamitans*) and its close relative the bull frog (*Rana catesbeiana*) do not hybridize mainly because of cytological block to fertilization

Glossary

AIDs = Acquired immunodeficiency syndrome
ATP =Adenosine triphosphate
°C = degrees Celsius
cm = centimeters
CH4 =Methane
CO = Carbon
CH_4 = methane (CH_4)
DNA =Deoxyribonucleic acid
DSBs = Double strand breaks
°F = degree Fahrenheit
H2 = Hydrogen

ICBN = International Code of Botanical Nomenclature
ICZN = International Code of Zoological Nomenclature
Kgs = kilograms.
km = kilometers
m = meters
mm = millimeters
NH2OH = Hdroxylamine
NH3 = Ammonia
O_2 = Oxygen
Pg = picograms
RNA = Ribonucleic acid
L = length
HIV = Human immunodeficiency virus
% = percentage
g = grams
spp = species
UV = Ultraviolet
< = Less than

End of the Module Questions

1. What does the *Hardy-Weinberg principle* provides?
2. Mention what is needed for evolution to continue?
3. What causes the inversions of the DNA sequence?
4. Name the enzymes that act as catalyzed in genetic recombination.
5. What is point mutation?

Module 3: Evolution of LIfe

Module Introduction

Unit 1: Origin of Life
Unit 2: Evidence of evolution, adaptation and Speciation
Unit 3: Evolution of the plants
Unit 4: Classification and phylogeny
Unit 5: Geological periods and epoch
Glossary

Unit 1: Origin of Life

Contents

1.1 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.

1.2 Intended Learning Outcomes (ILOs)

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

- Explain the origin of life
- The probable stages in the origin of life

1.3 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.
Origin of life means the appearance of simplest primordial life from non-living matter

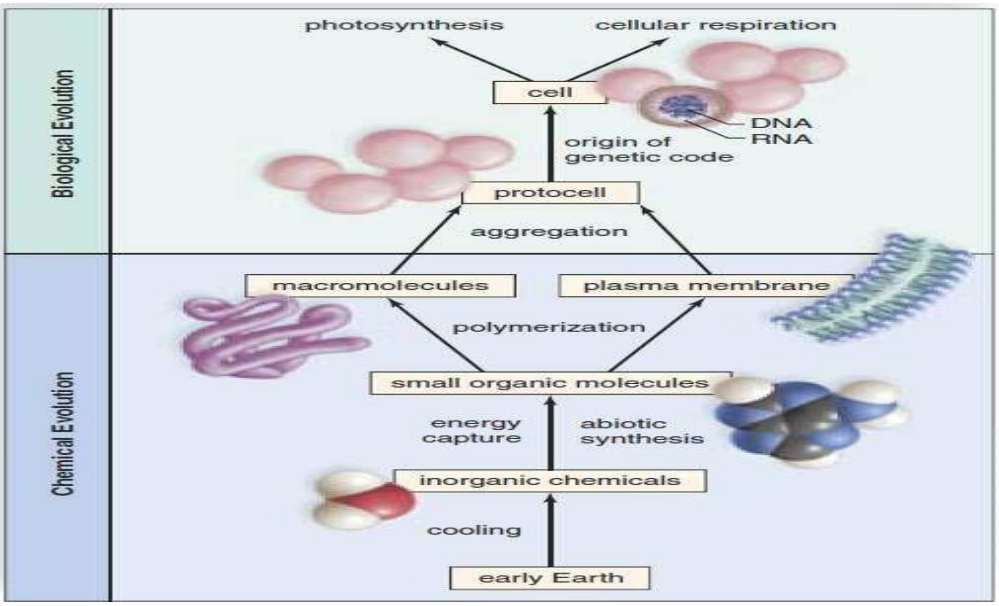

Fig.1: Origin of first cell (s)

Probable stages in the origin of life

ii) 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.

iii) 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 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.

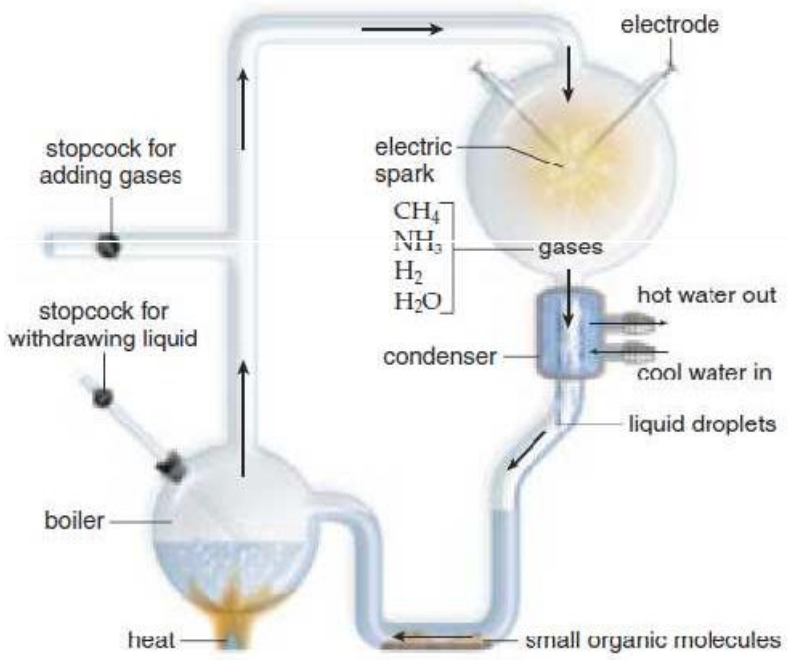

Fig2: Miller and Ureys apparatus and experiment

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

iv) 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.

v) 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), 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.

vi) 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.

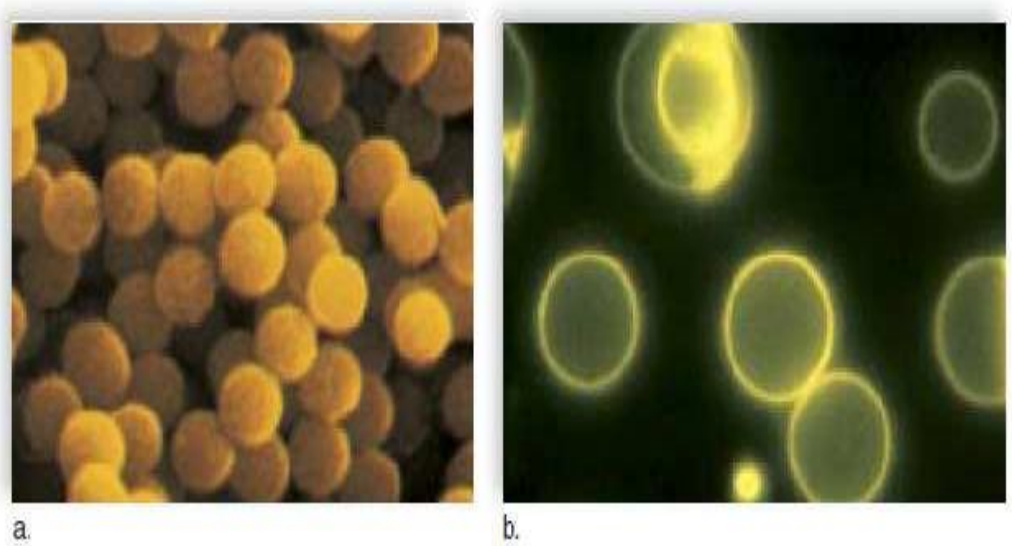

Fig.3. Protocell components

When did the planet come into existence? What was life form in the theorized stage?

Self-Assessment Exercises 1

Attempt these exercises to measure what you have learnt so far. This should not take you more than 5 minutes.

1. When did life evolved on planet?
2. What is origin of life?

1.4 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

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1.6 Possible Answers to SAEs

Answers to SAEs 1

1. Life evolved on planet earth about 3.5 billion years ago. At that time it was extremely hot.

2. Origin of life means the appearance of simplest primordial life from non-living matter

Unit 2: Evidence of evolution, adaptation and Speciation

Contents

2.1 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 behaviour 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.

2.2 Intended Learning Outcomes (ILOs)

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

- Explain the different evidence of evolution
- Explain adaptation and Speciation with the different mechanism involved.

2.3 Evidence of Evolution

The evidences supporting organic evolution are derived from a number of fields of biology are discuss below:

i) 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.1). 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.1: (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 absolute dating methods, we can learn from fossils about the various organisms and environments that existed across the planet during any time period. Fossils allowed scientists to construct the geological timescale that traces the history of life.

ii) 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.

iii) 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 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.

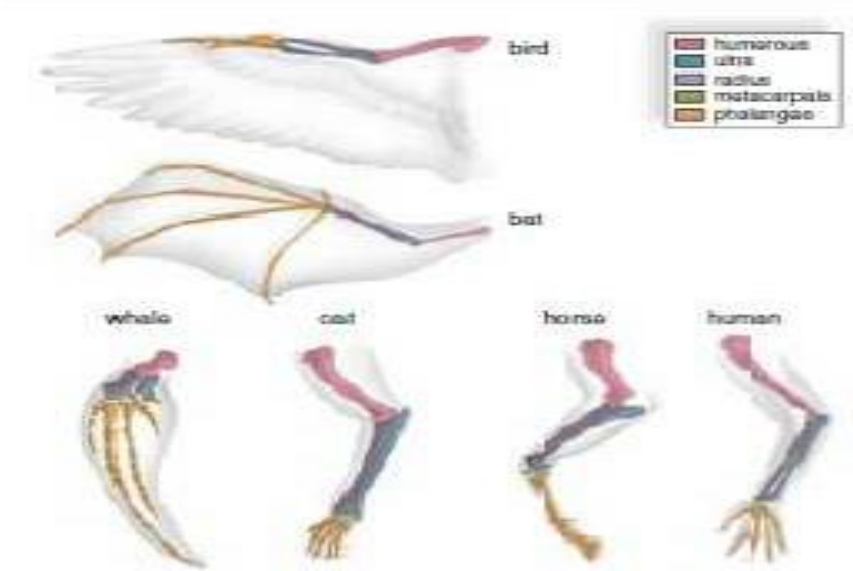

Fig. 2: Significance of homologous structures

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.

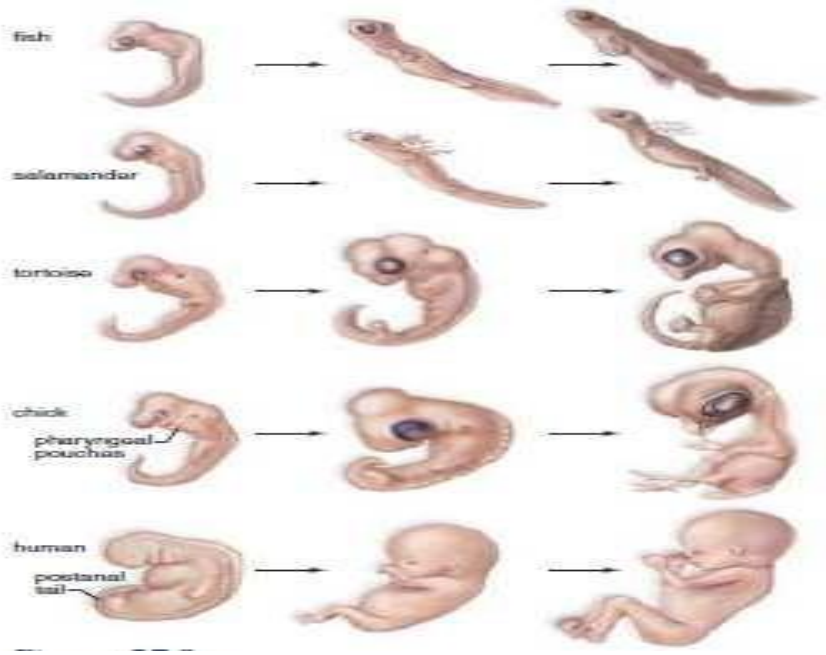

Fig. 3: Significance of developmental similarities

Organisms that share homologous structures are closely related and have a common ancestry. Studies of comparative and embryological development reveal homologous structures.

iv) 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 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 4. 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.

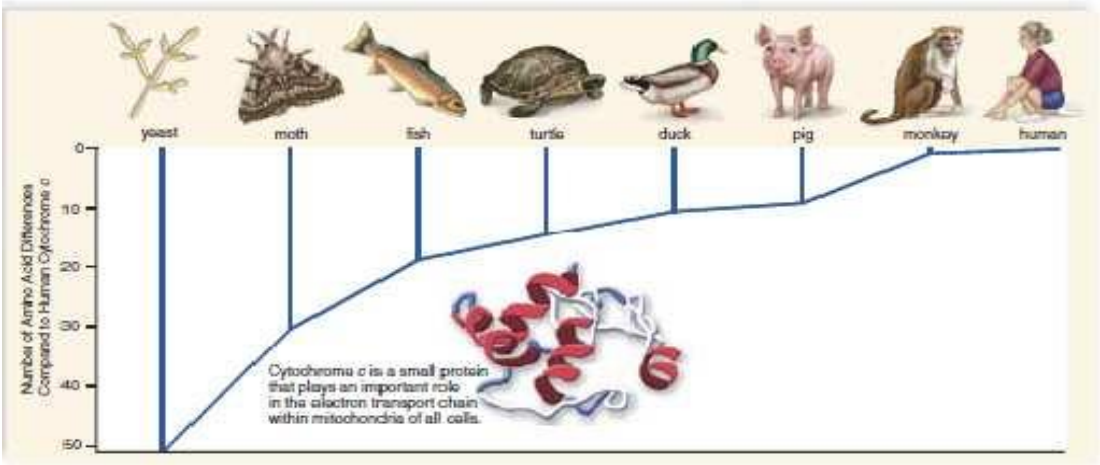

Fig. 4: Significance of biochemical differences
What is Evolution? Explain what you understand by Fossil.
Self-Assessment Exercises 1

Attempt this exercises to measure what you have learnt so far. This should not take you more than 5 minutes.

1. What is Biogeography?
2. What are vestigial structures?

2.4 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. An example is the adaptation of horses' teeth used in grinding of grass as a trait important in its feeding and survival. 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.

"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).

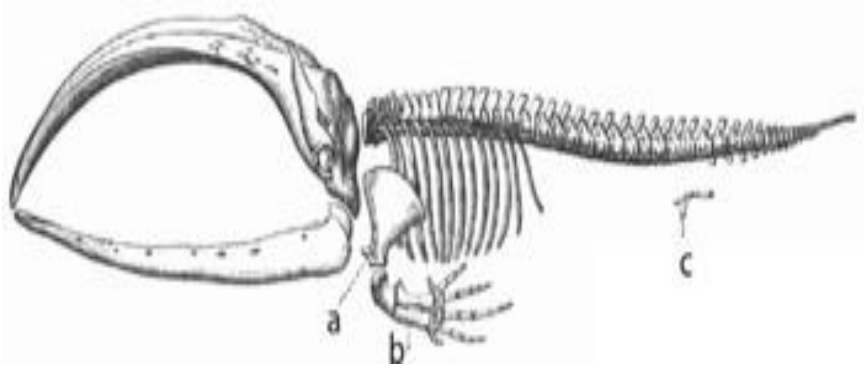

Fig.5 : A baleen whale skeleton, *a* and *b* label flipper bones, which were adapted from front leg bones: while *c* indicates vestigial leg bones, suggesting an adaptation from land to sea.

Adaptation occurs through the gradual modification of existing structure. Consequently, structure 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.

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. Define Adaptation. What is Adaptedness?

Self-Assessment Exercises 2

Attempt this exercises to measure what you have learnt so far. This should not take you more than 5 minutes.

1. What is an adaptive trait?
2. Explain the outcome of Adaptation?

2.5 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 sub-populations 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

ii) 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 population 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.

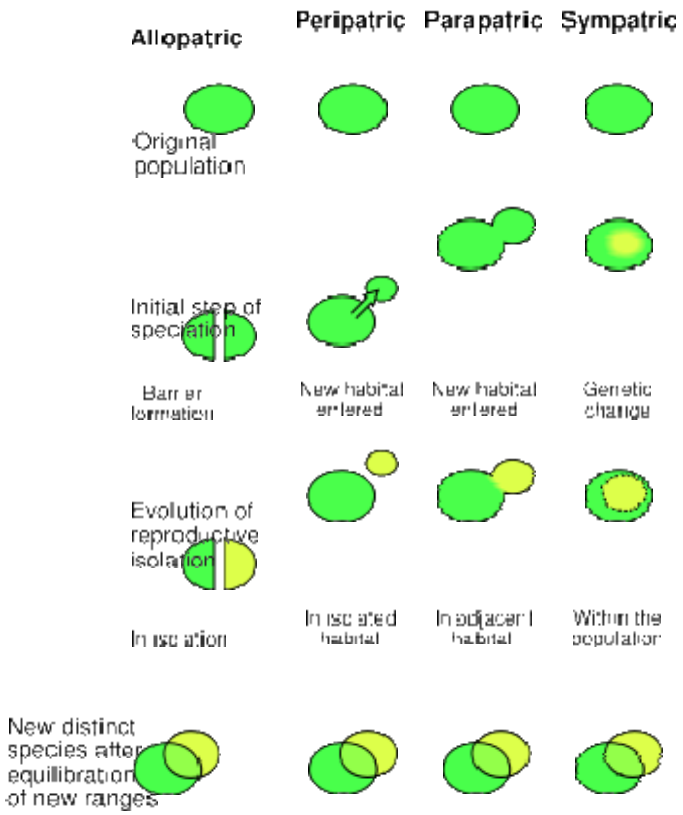

Fig. 6: The four mechanisms

The second mechanism of speciation is **peripatric speciation**, which occurs when small population 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.

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. Define Speciation.
What do you understand by allopatric speciation?

Self-Assessment Exercises 3

Attempt this exercise to measure what you have learnt so far. This should not take you more than 5 minutes

1. What do you understand by sympatric speciation?
2. Explain the third mechanism of speciation?

2.6 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. 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.

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2.8 Possible Answers to SAEs

Answers to SAEs 1

1. 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.

2. 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.

Answers to SAEs 2

1. 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.

2. 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.

Answers to SAEs 3

1. 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.

2. 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 population.

Unit 3: Evolution of Plants

Contents

3.1 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.

3.2 Intended Learning Outcomes (ILOs)

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

- Explain plant evolution
- The theories to explain the appearance of a diplobiontic lifecycle

3.3 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

a. Alternation of generations: Plants developed a more dominant diploid phase of the life cycle.
b. Vascular tissue: Transports water and nutrients throughout the plant body, Thus plants were able to grow larger and in drier conditions.
c. Seeds: Protected the embryo, thus allowing plants to dominate their terrestrial environments.
d. Flowers and fruits

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 are divided 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.

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. 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. Theories to explain the appearance of a diplobiontic life-cycle.

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.

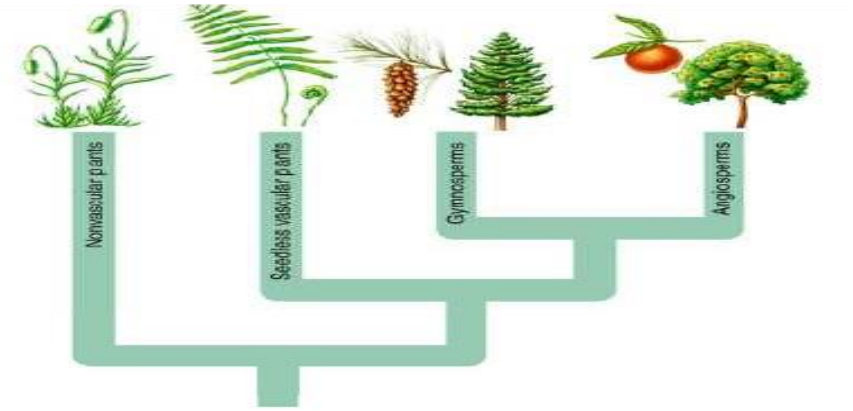

Fig.1: The evolution of plants

4. 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.

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 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. *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 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.

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.

5. Evolution of Seed Plants

In seed plants, the evolutionary trend led to a dominant sporophyte generation, and at the same time, a systematic reduction in the size of the gametophyte: from a conspicuous structure to a microscopic cluster of cells enclosed in the tissues of the sporophyte. Whereas lower vascular plants, such as club mosses and ferns, are mostly homosporous (produce only one type of spore), all seed plants, or **spermatophytes**, are heterosporous. They form two types of spores: megaspores (female) and microspores (male). Megaspores develop into female gametophytes that produce eggs, and microspores mature into male gametophytes that generate sperm. Because the gametophytes mature within the spores, they are not free-living, as are the gametophytes of other seedless vascular plants. Heterosporous seedless plants are seen as the evolutionary forerunners of seed plants.

Seeds and pollen—two critical adaptations to drought, and to reproduction that doesn't require water—distinguish seed plants from other (seedless) vascular plants. Both adaptations were required for the colonization of land begun by the bryophytes and their ancestors. Fossils place the earliest distinct seed plants at about 350 million years ago. The first reliable record of gymnosperms dates their appearance to the Pennsylvanian period, about 319 million years ago. Gymnosperms were preceded by **progymnosperms**, the first naked seed plants, which arose about 380 million years ago. Progymnosperms were a transitional group of plants that superficially resembled conifers (cone bearers) because they produced wood from the secondary growth of the vascular tissues; however, they still reproduced like ferns, releasing spores into the environment. Gymnosperms dominated the landscape in the early (Triassic) and middle (Jurassic) Mesozoic era. Angiosperms surpassed gymnosperms by the middle of the Cretaceous (about 100 million years ago) in the late Mesozoic era, and today are the most abundant plant group in most terrestrial biomes.

Pollen and seed were innovative structures that allowed seed plants to break their dependence on water for reproduction and development of the embryo, and to conquer dry land. The **pollen grains** are the male gametophytes, which contain the sperm (gametes) of the plant. The small haploid (1*n*) cells are encased in a protective coat that prevents desiccation (drying out) and mechanical damage. Pollen grains can travel far from their original sporophyte, spreading the plant's genes. The **seed** offers the embryo protection, nourishment, and a mechanism to maintain dormancy for tens or even thousands of years, ensuring germination can occur when growth conditions are optimal. Seeds therefore allow plants to disperse the next generation through both space

and time. With such evolutionary advantages, seed plants have become the most successful and familiar group of plants, in part because of their size and striking appearance.

6. Evolution of Gymnosperms

The fossil plant *Elkinsia polymorpha*, a "seed fern" from the Devonian period—about 400 million years ago—is considered the earliest seed plant known to date. Seed ferns produced their seeds along their branches without specialized structures. What makes them the first true seed plants is that they developed structures called cupules to enclose and protect the **ovule**—the female gametophyte and associated tissues—which develops into a seed upon fertilization. Seed plants resembling modern tree ferns became more numerous and diverse in the coal swamps of the Carboniferous period.

Fossil records indicate the first gymnosperms (progymnosperms) most likely originated in the Paleozoic era, during the middle Devonian period: about 390 million years ago. Following the wet Mississippian and Pennsylvanian periods, which were dominated by giant fern trees, the Permian period was dry. This gave a reproductive edge to seed plants, which are better adapted to survive dry spells. The Ginkgoales, a group of gymnosperms with only one surviving species—the *Gingko biloba*—were the first gymnosperms to appear during the lower Jurassic. Gymnosperms expanded in the Mesozoic era (about 240 million years ago), supplanting ferns in the landscape, and reaching their greatest diversity during this time. The Jurassic period was as much the age of the cycads (palm-tree-like gymnosperms) as the age of the dinosaurs. Gingkoales and the more familiar conifers also dotted the landscape. Although angiosperms (flowering plants) are the major form of plant life in most biomes, gymnosperms still dominate some ecosystems, such as the taiga (boreal forests) and the alpine forests at higher mountain elevations because of their adaptation to cold and dry growth conditions.

7. Seeds and Pollen as an Evolutionary Adaptation to Dry Land

Unlike bryophyte and fern spores (which are haploid cells dependent on moisture for rapid development of gametophytes), seeds contain a diploid embryo that will germinate into a sporophyte. Storage tissue to sustain growth and a protective coat give seeds their superior evolutionary advantage. Several layers of hardened tissue prevent desiccation, and free reproduction from the need for a constant supply of water. Furthermore, seeds remain in a state of dormancy—induced by desiccation and the hormone abscisic acid—until conditions for growth become favorable. Whether blown by the wind, floating on water, or carried away by animals, seeds are scattered in an expanding geographic range, thus avoiding competition with the parent plant.

Pollen grains are male gametophytes and are carried by wind, water, or a pollinator. The whole structure is protected from desiccation and can reach the female organs without dependence on water. Male gametes reach female gametophyte and the egg cell gamete though a pollen tube:

an extension of a cell within the pollen grain. The sperm of modern gymnosperms lack flagella, but in cycads and the *Gingko*, the sperm still possess flagella that allow them to swim down the **pollen tube** to the female gamete; however, they are enclosed in a pollen grain.

8. Evolution of Angiosperms

Undisputed fossil records place the massive appearance and diversification of angiosperms in the middle to late Mesozoic era. Angiosperms ("seed in a vessel") produce a flower containing male and/or female reproductive structures. Fossil evidence indicates that flowering plants first appeared in the Lower Cretaceous, about 125 million years ago, and were rapidly diversifying by the Middle Cretaceous, about 100 million years ago. Earlier traces of angiosperms are scarce. Fossilized pollen recovered from Jurassic geological material has been attributed to angiosperms. A few early Cretaceous rocks show clear imprints of leaves resembling angiosperm leaves. By the mid-Cretaceous, a staggering number of diverse flowering plants crowd the fossil record. The same geological period is also marked by the appearance of many modern groups of insects, including pollinating insects that played a key role in ecology and the evolution of flowering plants.

Although several hypotheses have been offered to explain this sudden profusion and variety of flowering plants, none have garnered the consensus of paleobotanists (scientists who study ancient plants). New data in comparative genomics and paleobotany have, however, shed some light on the evolution of angiosperms. Rather than being derived from gymnosperms, angiosperms form a sister clade (a species and its descendents) that developed in parallel with the gymnosperms. The two innovative structures of flowers and fruit represent an improved reproductive strategy that served to protect the embryo, while increasing genetic variability and range. Paleobotanists debate whether angiosperms evolved from small woody bushes, or were basal angiosperms related to tropical grasses. Both views draw support from cladistics studies, and the so-called woody magnoliid hypothesis—which proposes that the early ancestors of angiosperms were shrubs—also offers molecular biological evidence.

The most primitive living angiosperm is considered to be *Amborella trichopoda*, a small plant native to the rainforest of New Caledonia, an island in the South Pacific. Analysis of the genome of *A. trichopoda* has shown that it is related to all existing flowering plants and belongs to the oldest confirmed branch of the angiosperm family tree. A few other angiosperm groups called basal angiosperms, are viewed as primitive because they branched off early from the phylogenetic tree. Most modern angiosperms are classified as either monocots or eudicots, based on the structure of their leaves and embryos. Basal angiosperms, such as water lilies, are considered more primitive because they share morphological traits with both monocots and eudicots.

9. Flowers and Fruits as an Evolutionary Adaptation

Angiosperms produce their gametes in separate organs, which are usually housed in a **flower**. Both fertilization and embryo development take place inside an anatomical structure that provides a stable system of sexual reproduction largely sheltered from environmental fluctuations. Flowering plants are the most diverse phylum on Earth after insects; flowers come in a bewildering array of sizes, shapes, colors, smells, and arrangements. Most flowers have a mutualistic pollinator, with the distinctive features of flowers reflecting the nature of the pollination agent. The relationship between pollinator and flower characteristics is one of the great examples of coevolution.

Following fertilization of the egg, the ovule grows into a seed. The surrounding tissues of the ovary thicken, developing into a **fruit** that will protect the seed and often ensure its dispersal over a wide geographic range. Not all fruits develop from an ovary; such structures are "false fruits." Like flowers, fruit can vary tremendously in appearance, size, smell, and taste. Tomatoes, walnut shells and avocados are all examples of fruit. As with pollen and seeds, fruits also act as agents of dispersal. Some may be carried away by the wind. Many attract animals that will eat the fruit and pass the seeds through their digestive systems, then deposit the seeds in another location. Cockleburs are covered with stiff, hooked spines that can hook into fur (or clothing) and hitch a ride on an animal for long distances. The cockleburs that clung to the velvet trousers of an enterprising Swiss hiker, George de Mestral, inspired his invention of the loop and hook fastener he named Velcro. When did evolution of Land plants start? Which plant evolve strands of specialized conduction cells?

Self-Assessment Exercises 1

Attempt this exercises to measure what you have learnt so far. This should not take you more than 5 minutes.

1. What are Pollen and Seed?
2. Which plant is the earliest seed plant known?

3.4 Summary

All seed plants are derived from a single common ancestor which appeared about one million years ago, during the Carboniferous period. The gametophytes of seed plants shrank, while the sporophytes became prominent structures and the diploid stage became the longest phase of the lifecycle. Gymnosperms became the dominant group during the Triassic. In these, pollen grains and seeds protect against desiccation. The seed, unlike a spore, is a diploid embryo surrounded by storage tissue and protective layers. It is equipped to delay germination until growth

conditions are optimal. Angiosperms bear both flowers and fruit. The structures protect the gametes and the embryo during its development. Angiosperms appeared during the Mesozoic era and have become the dominant plant life in terrestrial habitats.

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3.6 Possible Answers to SAEs

Answers to SAEs 1

1. Pollen and seed were innovative structures that allowed seed plants to break their dependence on water for reproduction and development of the embryo, and to conquer dry land. The **pollen grains** are the male gametophytes, which contain the sperm (gametes) of the plant. The small haploid (1*n*) cells are encased in a protective coat that prevents desiccation (drying out) and mechanical damage. Pollen grains can travel far from their original sporophyte, spreading the plant's genes. The **seed** offers the embryo protection, nourishment, and a mechanism to maintain dormancy for tens or even thousands of years, ensuring germination can occur when growth conditions are optimal. Seeds therefore allow plants to disperse the next generation through both space and time.

2. The fossil plant *Elkinsia polymorpha*, a "seed fern" from the Devonian period—about 400 million years ago—is considered the earliest seed plant known to date.

Unit 4: Classification and Phylogeny

Contents

4.1 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.

4.2 Intended Learning Outcomes (ILOs)

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

- Define classification and phylogeny
- List the bases of classification

4.3 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.

There are different types of classification systems and they are all used for different purposes. Organism classification takes place in the form of a taxonomic system. This is called the **Linnean classification**. The Linnean classification involves the usage of the grouping organisms using a hierarchy called taxa. This animal classification system contains taxonomic levels and taxonomic groups that animals are placed in based on their features and even shared ancestry.

The various taxonomic categories are given below:

i) Species: Species classification is the final ranking for the biological classification of living things. A species is defined as a group of organisms with similar characteristics that are able to procreate or interbreed with one another. The offspring they produce must be sustainable and also being able to create a new generation of the species as well. Sometimes, species can form evolve into another, this is known as speciation.

ii) Genus: Genus is the systematic unit in the organism classification that helps determine the species of organisms as the genus groups multiple species together. A genus could also consist of one unusual species whose attributes are so unique it is classified on its own.

iii) Family: When a group of genera with similar characteristics and traits are pulled together it is called a *family***.** The family is the ranking between orders and genus in the Linnean classification. Multiple genera make up a family group. Group of genera (singular-genus) resembling each other. For example, *Felis domestica* (the cat) and *Panthera tigris* (the tiger), both belong to family Felidae.

iv) Order: **Order** is the classification that consists of several families. Orders above the family classification and below the ranking of

classes. An order consists of multiple families that share many characteristics and evolutionary traits.

v) Class: The class falls just between the phylum and order classifications. Just like kingdoms contain multiple phyla, each phylum can contain multiple classes. These generally end with the suffix "*ae*" when they are named. Sometimes, if classes are very large, they may be divided into subclasses.

vi) Phylum: This taxonomic rank, sometimes termed as "*division*" lies after the kingdom and further classifies based on phenetic and phylogenetic. Phenetics is based on the number of shared characteristics using a numerical system and phylogenetics is based on evolution and shared relationships but using a systematic study. Each kingdom is broken down into numerous phyla. This can range from as few as four like in Kingdom Protista to as many as the nine phyla that kingdom Animalia contains

vii) Kingdom: The kingdom classification is the second-highest ranking in the taxonomic groupings of organisms. *How many kingdoms are there?* There are five kingdoms, though some recent studies have claimed six and seven kingdoms. The five kingdoms consist of the animals, plants, fungi, protists, and monerans.
The kingdom Animalia is the most diverse out of the kingdoms and this is because it has evolved the most. This kingdom is generally divided into invertebrate and vertebrate (animals that have a backbone and those that do not).

The kingdom Plantae is made up of all trees, flowers, bushes, and vegetation. It is the oldest of all the kingdoms. Its members are characterized by having limited movement, eukaryotic features, and autotrophic abilities.
Yeasts, toadstools, molds and mushrooms belong to the kingdom Fungi. These organisms are carnivores that have chitin in their cell walls yet reproduce in a sporic manner.
Protista can be considered the *mother* of all eukaryotes as all of them are descendants of this kingdom. It can be a very difficult kingdom to distinguish as it is intertwined with many of the others. The monera kingdom consists of all archaea and bacteria as it deals with all microscopic organisms.
Example of classification system

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* (Fossil men and modern man)
Species: H.*sapiens* (Modern man)
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.

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). The lack of internal membranes in prokaryotes distinguishes them from eukaryotes. The prokaryotic cell membrane is made up of phospholipids and constitutes the cell's primary osmotic barrier.

The cytoplasm contains ribosomes, which carry out protein synthesis, and a double-stranded deoxyribonucleic acid (DNA) chromosome, which is usually circular. Many prokaryotes also contain additional circular DNA molecules called plasmids, with additional dispensable cell functions, such as encoding proteins to inactivate antibiotics. Some prokaryotes have flagella. All bacteria are prokaryotes.

Eukaryote are organism whose cells contain a nucleus and other membrane-organelles. The eukaryotic cell has a nuclear membrane that surrounds the nucleus, in which the well-defined chromosomes (bodies containing the hereditary material) are located. Eukaryotic cells also contain organelles, including mitochondria (cellular energy exchangers), a Golgi apparatus (secretory device), an endoplasmic reticulum (a canal-like system of membranes within the cell), and lysosomes (digestive apparatus within many cell types). There are several exceptions to this, however; for example, the absence of mitochondria and a nucleus in red blood cells and the lack of mitochondria is found in the oxymonad *Monocercomonoides* species. Eukaryotes are thought to have evolved between about 1.7 billion and 1.9 billion years ago. The earliest known microfossils resembling eukaryotic organisms date to approximately 1.8 billion years ago.

Table 1: Differences between Prokaryotes and Eukaryotes

	Prokaryote	**Eukaryote**
Nucleus	Absent	Present
Membrane-bound organelles	Absent	Present
Cell structure	Unicellular	Mostly multicellular; some unicellular
Cell size	Smaller (0.1-5 µm)	Larger (10-100 µm)
Complexity	Simpler	More complex
DNA Form	Circular	Linear
Examples	Bacteria, archaea	Animals, plants, fungi, protists

The five kingdom classification of organisms are;

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

What is Classification? How was organism classified by early taxonomists?

Self-Assessment Exercises 1

Attempt this exercises to measure what you have learnt so far. This should not take you more than 5 minutes.

1. What do you understand by Species?
2. What is Kingdom?
3. Explain the System of naming organism?

4.4 Phylogeny

Phylogenetics is the study of the evolutionary relatedness among groups of organisms. Molecular phylogenetics uses sequence data to infer these relationships for both organisms and the genes they maintain. Life's history is depicted as a branching tree and all forms of life, including many extinct forms that represent dead branches, will connect to this tree somewhere.

Fundamental to phylogeny is the proposition, universally accepted in the scientific community that plants or animals of different species descended from common ancestors. The evidence for such relationships, however, is nearly always incomplete, for the vast majority of species that have ever lived are extinct, and relatively few of their remains have been preserved in the fossil record. Most phylogenies therefore are hypotheses and are based on indirect evidence. Different phylogenies often emerge using the same evidence. Nevertheless, there is universal agreement that the tree of life is the result of organic descent from earlier ancestors and that true phylogenies are discoverable, at least in principle.

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 1 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.

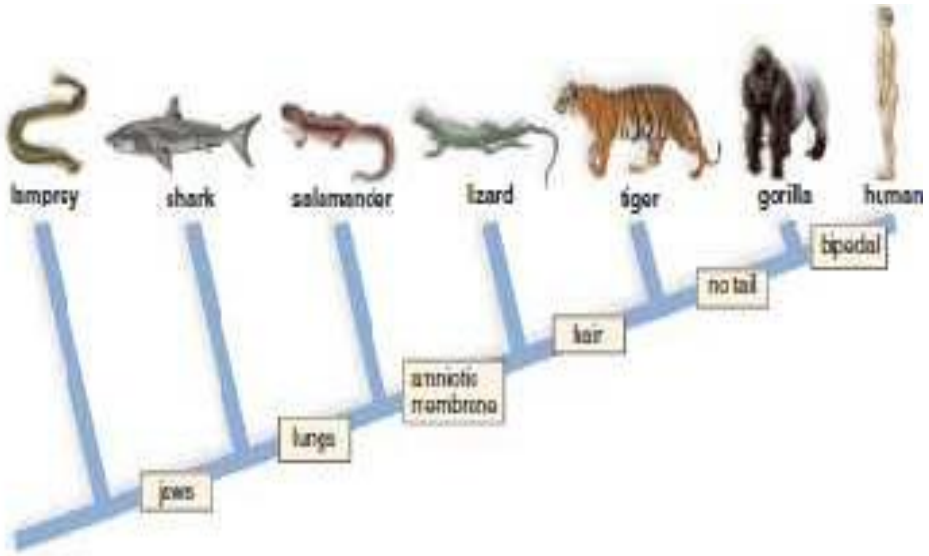

Fig. 1: Cladogram

In-Text Question(s)

What is Molecular phylogenetics?
Answer: Molecular phylogenetics uses sequence data to infer these relationships for both organisms and the genes they maintain.

Self-Assessment Exercises 2

Attempt this exercises to measure what you have learnt so far. This should not take you more than 5 minutes.

1. Describe Phylogeny?
2. What is a clade?

4.5 Summary

Classification involves assigning species to a hierarchy of categories: kingdom, phylum, class, order, family, genus, and species. The five-kingdom system of classification recognizes these kingdoms: Monera (the bacteria), Protista (algae, protozoans), Fungi, Plantae, and Animalia. 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 ancestor.

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4.7 Possible Answers to SAEs

Answers to SAEs 1

1. Species: Species classification is the final ranking for the biological classification of living things. A species is defined as a group of organisms with similar characteristics that are able to procreate or interbreed with one another. The offspring they produce must be sustainable and also being able to create a new generation of the species as well. Sometimes, species can form evolve into another, this is known as speciation.
2. The kingdom classification is the second-highest ranking in the taxonomic groupings of organisms. *How many kingdoms are there?* There are five kingdoms, though some recent studies have claimed six and seven kingdoms. The five kingdoms consist of the animals, plants, fungi, protists, and monerans.
3. 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.

Answers to SAEs 2

1. 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. A clade contains a most recent common ancestor and all its descendant species—the common ancestor is presumed and not identified.

Unit 5: Geological Periods and Epoch

Contents

5.1 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.

5.2 Intended Learning Outcomes (ILOs)

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

- Explain geological time scale and epoch.
- Explain the division of earth's history

5.3 Geological Time Scale

The geological time scale is one of the crowning achievements of science in general and geology in particular. It is a reference and communication system for comparing rocks and fossils from throughout the world and is geology's equivalent of the periodic table of the elements. Most of the boundaries on the geological time scale correspond to the origination or extinction of particular kinds of fossils. Knowing when major groups of fossils first appeared or went extinct is therefore incredibly useful for determining the ages of rocks in the field. For example, if you find a rock with a trilobite fossil upon it, you will immediately know that the rock is Paleozoic in age (541 Ma to 252 Ma) and not older or younger; knowing the species of trilobite allows even greater precision.

As a result of studying strata, scientists have divided Earth's history into eras, and then periods and epochs (Table1). 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 1: Geological time scale

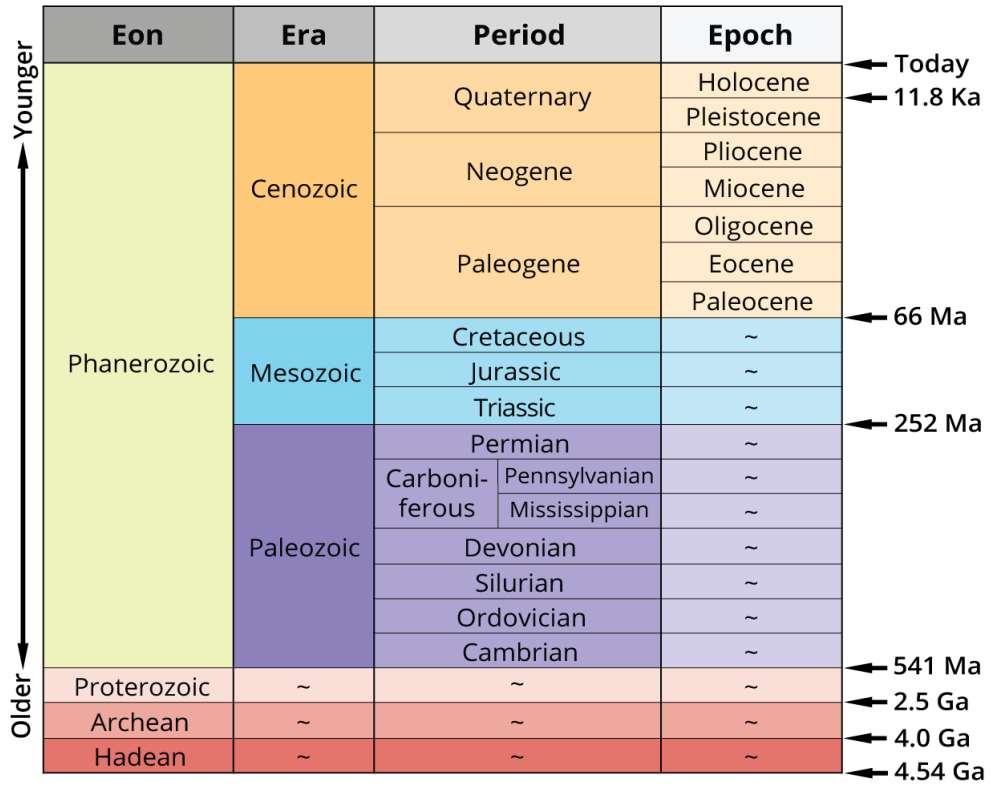
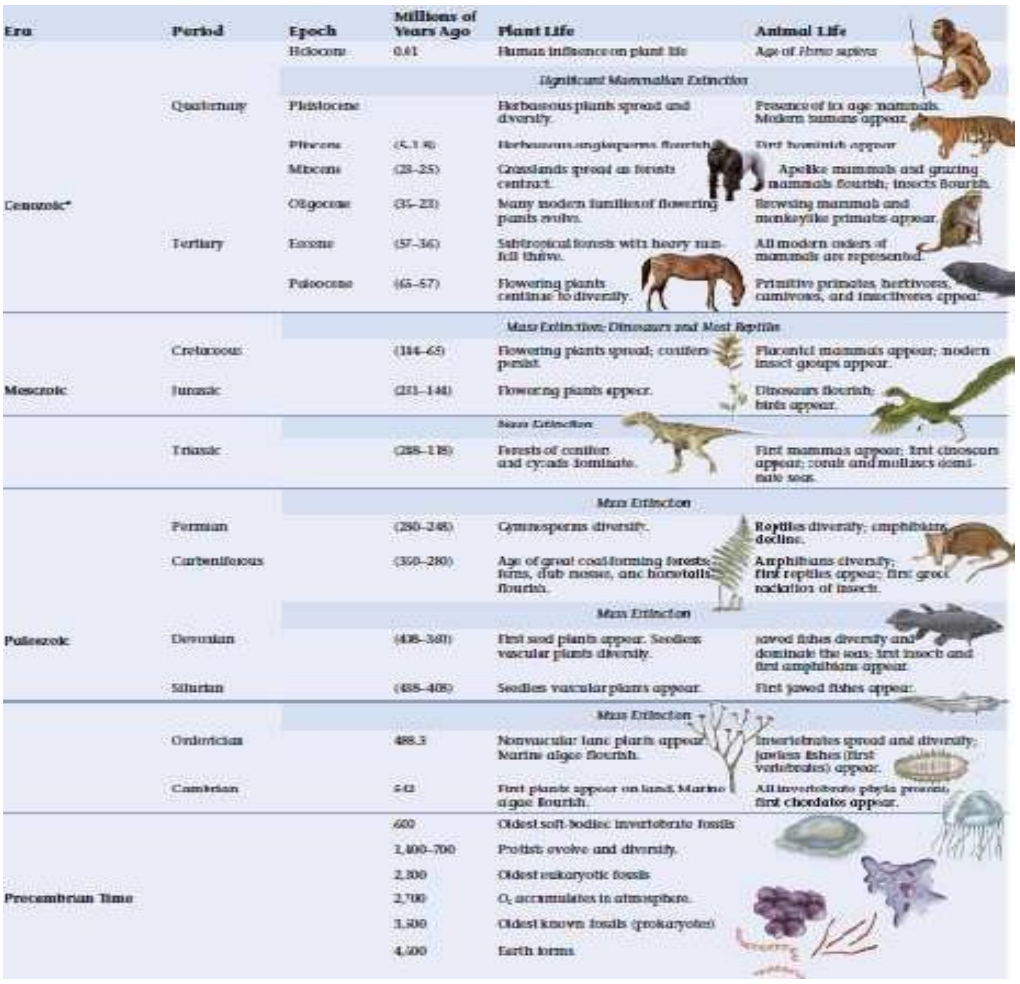


Table 2: Division of earth's history

Eon	Era	Time
Phanerozoic	Cenozoic (New Life)	65 MA to 0 MA
	Mesozoic (Middle Life)	245 MA to 65 MA
	Paleozoic (Old Life)	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
		3.96 Ga to 2.5 Ga
		4.6 Ga to 3.96 Ga

Faunal succession is the principle that different kinds of fossils characterize different intervals of time. This is because evolution and extinction are facts of nature. The principle of faunal succession was developed by an English surveyor named William "Strata" Smith (1769-1839). As he studied layers of rocks to determine where to build canals, he noticed that he found the same ordering of fossil species from place to place; Fossil A was always found below Fossil B, which in turn was always found below Fossil C, and so on. By documenting these sequences of fossils, Smith was able to temporally correlate rock layers (or, strata) from place to place (in other words, to establish that rock layers in two different places are equivalent in age based upon the fact that they include the same types of fossils).

Table 3: Major Divisions of Geological Time and 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	(5.3–1.8)	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		(248–213)	Forests of conifers and cycads dominate.	First mammals appear; first dinosaurs appear; corals and mollusks dominate seas.
				Mass Extinction	
	Permian		(290–248)	Gymnosperms diversify.	Reptiles diversify; amphibians decline.
	Carboniferous		(360–290)	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	

i) Eons

The eon is the broadest category of geological time. Earth's history is characterized by four eons; in order from oldest to youngest, these are the Hadeon, Archean, Proterozoic, and Phanerozoic. Collectively, the Hadean, Archean, and Proterozoic are sometimes informally referred to as the "Precambrian." (The Cambrian period defines the beginning of the

Phanerozoic eon; so, all rocks older than the Cambrian are Precambrian in age.)

We live during the Phanerozoic, which means "visible life." This is the interval of geological time characterized by abundant, complex fossilized remains. Being the youngest eon of time, it is also very well represented by rock at Earth's surface. Because of these two factors, most paleontologists and geologists study fossils and rocks from the Phanerozoic eon.

The Phanerozoic eon began 541 million years ago (or, 0.541 billion years ago) and represents a paltry 12% of Earth's history. Most of Earth's history is represented by the three Precambrian eons. These older eons tell the story of Earth's beginning, life's origin, and the rise of complex life.

The Hadean and Archean are difficult eons to study, however, because they are exposed in very limited places on Earth's surface. (Since they are the oldest eons, rocks that are Hadean and Archean in age are often buried far below younger rocks at Earth's surface.) Proterozoic rocks--which span nearly 2 billion years (42% of Earth's history)--are much more accessible, but, until recently, have received significantly less attention from paleontologists than rocks from the younger, fossil-rich Phanerozoic eon. That is slowly beginning to change, however, as more clues about the origins of complex life begin to be revealed from Proterozoic-aged rocks.

ii) Eras

Eons of geological time are subdivided into eras, which are the second-longest units of geological time. The Phanerozoic eon is divided into three eras: the Paleozoic, Mesozoic, and Cenozoic.

The Paleozoic ("old life") era is characterized by trilobites, the first four-limbed vertebrates, and the origin of land plants. The Mesozoic ("middle life") era represents the "age of dinosaurs," though also is noteworthy for the first appearances of mammals and flowering plants. Finally, the Cenozoic ("new life") era is sometimes called the "age of mammals" and is the era during which we live today.

The older Archean and Proterozoic eons are similarly divided into several eras. For example, the youngest era of the Proterozoic eon is called the Neoproterozoic. For the sake of simplicity, these older eras are not included on the time scale shown at the top of this page; they do, however, exist.

iii) Periods

Just as eons are subdivided into eras, eras are subdivided into units of time called periods. The most well-known of all geological periods is the Jurassic period of the Mesozoic era.

The Paleozoic era is divided into six periods. From oldest to youngest, these are the Cambrian, Ordovician, Silurian, Devonian, Carboniferous, and Permian. Note that in the United States, the Carboniferous is divided into two separate periods: the Mississippian and the Pennsylvanian.

The Mesozoic era is divided into the Triassic, Jurassic, and Cretaceous periods.

Finally, the Cenozoic era is divided into three periods: the Paleogene, Neogene, and Quaternary.

iv) Epochs and Ages

Periods of geological time are subdivided into epochs. In turn, epochs are divided into even narrower units of time called ages. For the sake of simplicity, only the epochs of the Paleogene, Neogene, and Quaternary periods are shown on the time scale at the top of this page. It is important to note, however, that all of the periods of the Phanerozoic era are subdivided into the epochs and ages.

The Paleogene period is divided into--from oldest to youngest--the Paleocene, Eocene, and Oligocene epochs. The Neogene is divided into the Miocene and Pliocene epochs. Finally, the Quaternary is divided into the Pleistocene and Holocene epochs. Some geologists now think that--since humans are having such a notable impact on the Earth and its life--a new, youngest epoch should be added to the Quaternary: the Anthropocene. Define geological time scale. What is Faunal succession?

Self-Assessment Exercises 1

Attempt this exercises to measure what you have learnt so far. This should not take you more than 5 minutes.

1. Write on the Eons.
2. What is period?

5.4 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.

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5.6 Possible Answers to SAEs

Answers to SAEs 1

1. The eon is the broadest category of geological time. Earth's history is characterized by four eons; in order from oldest to youngest, these are the Hadeon, Archean, Proterozoic, and Phanerozoic. Collectively, the Hadean, Archean, and Proterozoic are sometimes informally referred to as the "Precambrian." (The Cambrian period defines the beginning of the Phanerozoic eon; so, all rocks older than the Cambrian are Precambrian in age.)
2. Eras are subdivided into units of time called periods. The most well-known of all geological periods is the Jurassic period of the Mesozoic era. The Paleozoic era is divided into six periods. From oldest to youngest, these are the Cambrian, Ordovician, Silurian, Devonian, Carboniferous, and Permian. Note that in the United States, the Carboniferous is divided into two separate periods: the Mississippian and the Pennsylvanian. The Mesozoic era is divided into the Triassic, Jurassic, and Cretaceous periods. Finally, the Cenozoic era is divided into three periods: the Paleogene, Neogene, and Quaternary.

Glossary

AIDs = Acquired immunodeficiency syndrome
ATP =Adenosine triphosphate
°C = degrees Celsius
cm = centimeters
CH4 =Methane
CO = Carbon
CH_4 = methane (CH_4)
DNA =Deoxyribonucleic acid
DSBs = Double strand breaks
°F = degree Fahrenheit
H2 = Hydrogen
ICBN = International Code of Botanical Nomenclature
ICZN = International Code of Zoological Nomenclature
Kgs = kilograms.
km = kilometers
m = meters
mm = millimeters
NH2OH = Hdroxylamine

NH3 = Ammonia
O_2 = Oxygen
Pg = picograms
RNA = Ribonucleic acid
L = length
HIV = Human immunodeficiency virus
% = percentage
g = grams
spp =species
UV = Ultraviolet
< =Less than

End of the Module Questions

1. What years did the Planet Earth come into existence?
2. Where are the majority of fossils found?
3. What is the benefit of Adaptation in nature?
4. List the four mechanisms of speciation
5. List the four key evolutionary innovation to trace the evolution of the plant kingdom