The Origin of Life on Earth

Recent discoveries of **prebiotic** conditions on other planets and their moons has rekindled interest in the origin of life on primeval Earth. Experiments demonstrate that both peptides and nucleic acids may form polymers naturally in the conditions that are thought to have existed in a primitive terrestrial environment. RNA has also been shown to have enzymatic properties (**ribozymes**) and is capable of self-replication. These discoveries have removed some fundamental obstacles to creating a plausible scientific model for the origin of life from a prebiotic soup. Much research is now underway and space probes have been sent to Mercury, Venus, Mars, Pluto and its moon, Charon. They will search for evidence of prebiotic conditions or primitive microorganisms. The study of life in such regions beyond our planet is called **exobiology**.

Steps Proposed in the Origin of Life

The appearance of life on our planet may be understood as the result of evolutionary processes that involve the following major steps:

1. Formation of the Earth (4600 mya) and its acquisition of volatile organic chemicals by collision with comets and meteorites, which provided the precursors of biochemical molecules.
2. Prebiotic synthesis and accumulation of amino acids, purines, pyrimidines, sugars, lipids, and other organic molecules in the primitive terrestrial environment.
3. Prebiotic condensation reactions involving the synthesis of polymers of peptides (proteins), and nucleic acids (most probably just RNA) with self-replicating and catalytic (enzymatic) abilities.
4. Synthesis of lipids, their self-assembly into double-layered membranes and liposomes, and the 'capturing' of prebiotic (self-replicating and catalytic) molecules within their boundaries.
5. Formation of a **protobiont**; an immediate precursor to the first living systems. Such protobionts would exhibit cooperative interactions between small catalytic peptides, replicative molecules, proto-tRNA, and protoribosomes.

An RNA World

RNA has the ability to act as both genes and enzymes and offers a way around the "chicken-and-egg" problem: genes require enzymes to form; enzymes require genes to form. The first stage of evolution may have proceeded by RNA molecules performing the catalytic activities necessary to assemble themselves from a nucleotide soup. At the next stage, RNA molecules began to synthesize proteins. There is a problem with RNA as a prebiotic molecule because the ribose is unstable. This has led to the idea of a pre-RNA world (PNA).

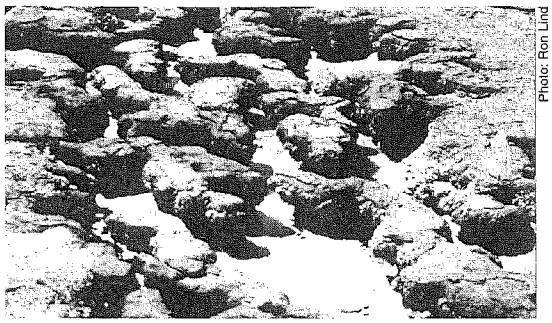
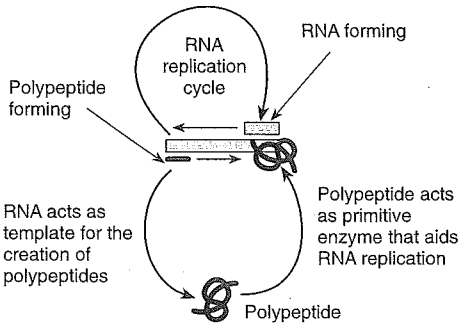


Photo: Ron Lind

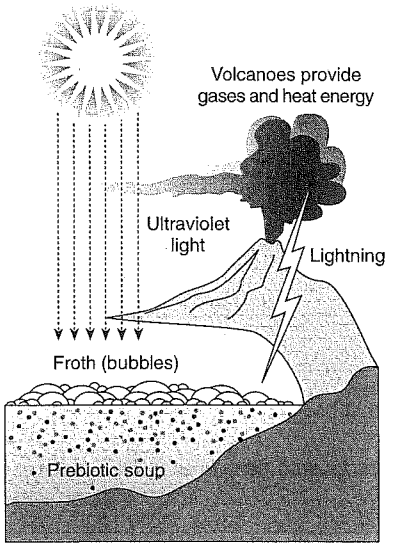
These living **stromatolites** from a beach in Western Australia are created by mats of bacteria. Similar, fossilized stromatolites have been found in rocks dating back to 3500 million years ago.

Dynamics of an RNA world



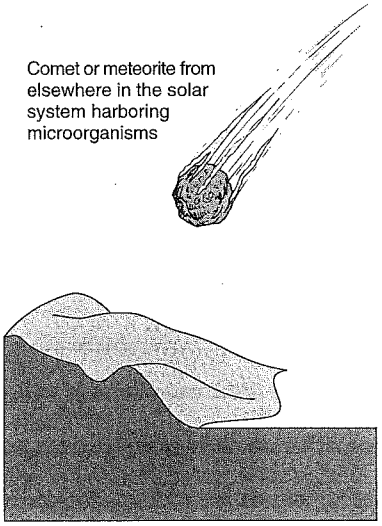
Scenarios for the Origin of Life on Earth

The origin of life remains a matter of scientific speculation. Three alternative views of how the key processes occurred are illustrated below:



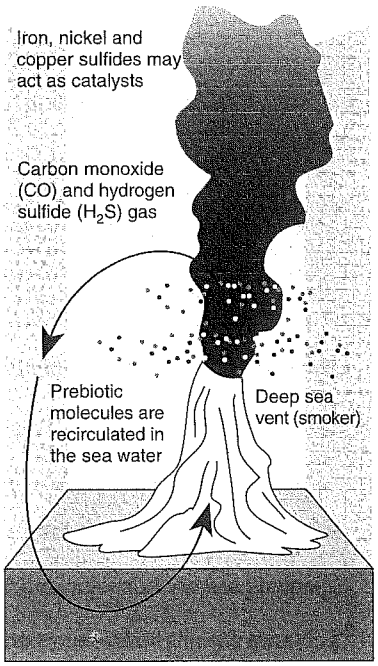
Ocean surface (tidal pools)

This popular theory suggests that life arose in a tidepool, pond or on moist clay on the primeval Earth. Gases from volcanoes would have been energized by UV light or electrical discharges to form the prebiotic molecules in froth.



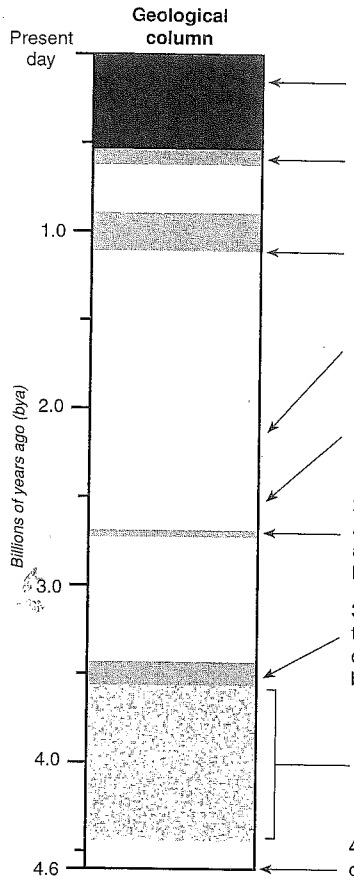
Panspermia

Cosmic ancestry (panspermia) is a serious scientific theory that proposes living organisms were 'seeded' on Earth as 'passengers' aboard comets and meteors. Such incoming organisms would have to survive the heat of re-entry.



Undersea thermal vents

A recently proposed theory suggests that life may have arisen at ancient volcanic vents (called smokers). This environment provides the necessary gases, energy, and a possible source of catalysts (metal sulfides).



Landmarks in the Origin and Evolution of Life

Fossil history of large eukaryotic organisms is well documented.

0.55 bya: Fossils of more complex, multicelled creatures were thought to have first appeared here (but see below). Fossils 600-540 my old, reaching 1 meter across have been found in the Ediacara Hills, Flinders Ranges, South Australia.

1.1 bya: Grooves in sandstone from the Vindhyan Basin, central India, may be the burrows of ancient worm-like creatures. This is 500 million years earlier than any previous evidence for multicellular animals.

2.1 bya: First fossil imprints appear in the geological record that are so large they can only be eukaryotes.

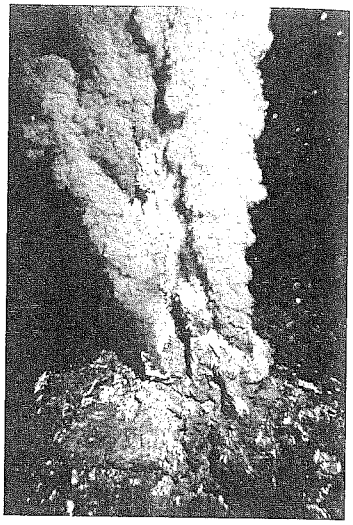
2.5 bya: Molecular fossil of cyanobacteria, 2-methylhopane, is abundant in organic-rich sedimentary rocks from the Mount McRae shale in Western Australia.

2.7 bya: Compounds in the oily residue squeezed out of Australian shale suggest the presence of eukaryotic cells. It appears to push the beginnings of complex life on Earth a billion years earlier than scientists had first thought.

3.5 bya: The oldest microbial community now known is from the Apex chert of northwestern Western Australia. It is a diverse assemblage of cyanobacteria fossils, leaving behind big, layered mounds of fossil bacterial colonies (stromatolites).

A series of prebiotic steps that lead to the formation of the **protobiont**; an immediate precursor to the first living systems.

4.6 bya: Formation of the Earth and its acquisition of volatile organic chemicals by collision with comets and meteorites, which provided the precursors of biochemical molecules.



A black smoker: In 1977, a vent was discovered at the Galapagos spreading center (mid-oceanic ridge), out of which gushed hot water laden with dissolved minerals. Since this discovery, hydrothermal venting has been found to be common along the length of the 55 000 km ridge crest system. Such black smokers are named after the dirty looking, high temperature water (350°C) that gushes from the chimney structures that they form. Such an environment is thought to be a possible site for prebiotic synthesis of life molecules.

1. Summarize the main features of the three most accepted scientific models for the origin of life on Earth:

   (a) Ocean surface: _____

   _____

   _____

   (b) Panspermia: _____

   _____

   _____

   (c) Undersea thermal vents: _____

   _____

   _____

2. Explain how the discovery of ribozymes has assisted in creating a plausible model for the prebiotic origin of life:

   _____

   _____

3. State how old the earliest fossils of microscopic life are known to be: _____

4. Scientists are seriously looking for evidence of life on other planets of our solar system, as well as some of their moons.

   (a) Name a planet or a moon that are pending targets for such spacecraft missions: _____

   (b) Explain how the discovery of life elsewhere in our solar system may affect the explanations for the origin of life:

   _____

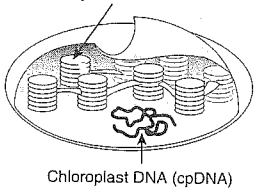
   _____

The Origin of Eukaryotes

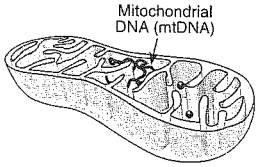
The first firm evidence of eukaryote cells is found in the fossil record at 540-600 mya. It is thought that eukaryote cells evolved from large prokaryote cells that ingested other free-floating prokaryotes. They formed a symbiotic relationship with the cells they engulfed (**endosymbiosis**). The two most important organelles that developed in eukaryote cells were mitochondria, for aerobic respiration, and chloroplasts, for photosynthesis in aerobic conditions. Primitive eukaryotes probably acquired mitochondria by engulfing purple bacteria. Similarly, chloroplasts may have been acquired by engulfing primitive cyanobacteria (which were already capable of photosynthesis). In both instances the organelles produced became dependent on the nucleus of the host cell to direct some of their metabolic processes. The sequence of evolutionary change shown below suggests that the lines leading to animal cells diverged before those leading to plant cells, but the reverse could also be true. Animal cells might have evolved from plant-like cells which subsequently lost their chloroplasts.

The Origin of Eukaryotic Cells

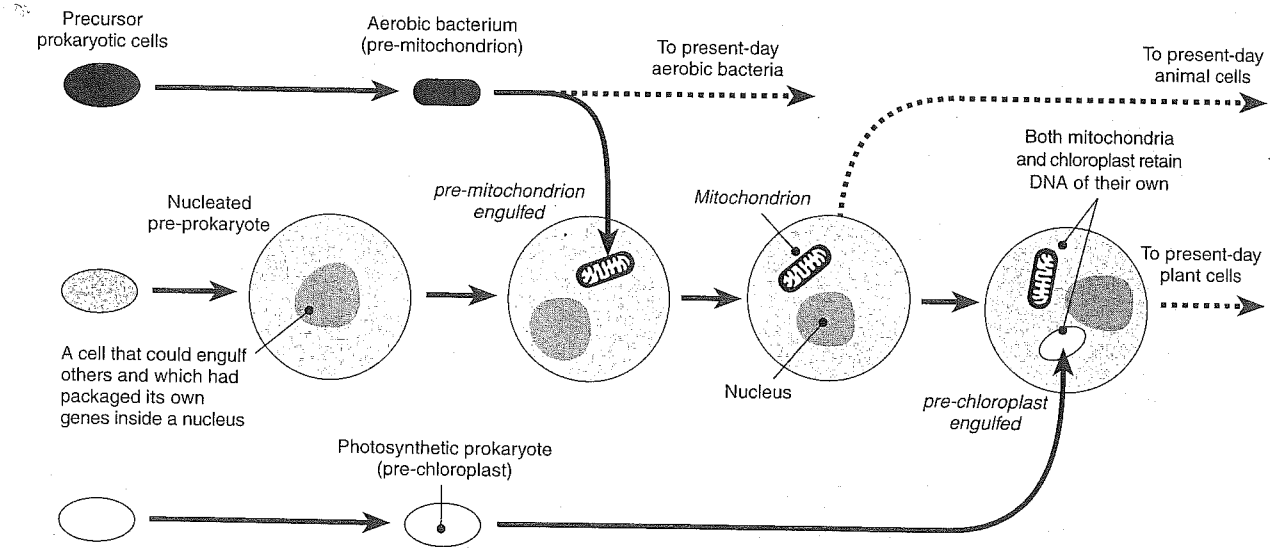
Chloroplasts have an internal membrane structure that is nearly identical to that of modern cyanobacteria.



Chloroplasts possess a self-replicating circular chromosome. It uses a genetic code that contains prokaryotic features, supporting the probable origins of chloroplasts as endosymbionts.



Mitochondria possess a self-replicating circular chromosome and use a genetic code identical to that used by prokaryotes. This supports the probable origins of mitochondria as endosymbionts.



1. Distinguish between the two possible sequences of evolutionary change suggested in the endosymbiosis theory:

2. Explain how the endosymbiosis theory accounts for the origins of the following organelles in eukaryotic cells:

   (a) Mitochondria:

   (b) Chloroplasts:

3. Describe the evidence that is found in modern mitochondria and chloroplasts that supports the endosymbiosis theory:

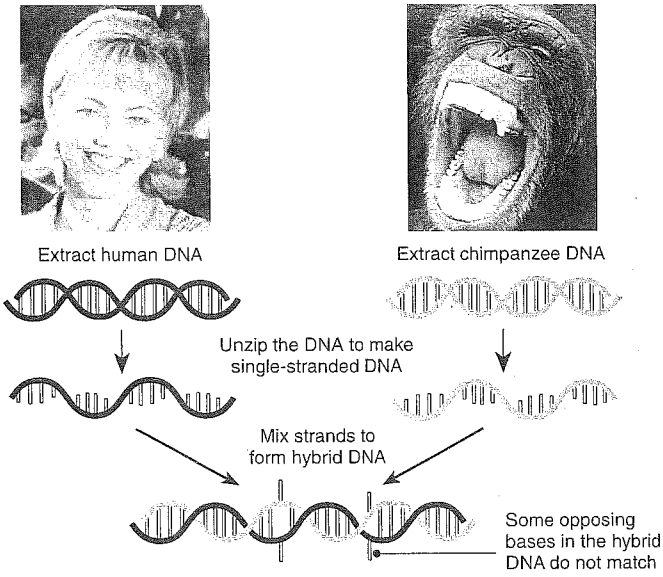
4. Comment on how the fossil evidence of early life supports or contradicts the endosymbiotic theory:

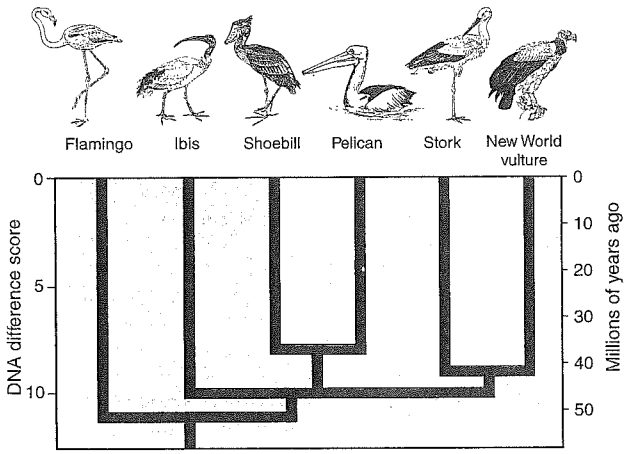
DNA Hybridization

The more closely two species are related, the fewer differences there will be in the exact sequence of bases. This is because there has been less time for the point mutations that will bring about these changes to occur. Modern species can be compared to see how long ago they shared a **common ancestor.** This technique gives a measure of 'relatedness', and can be calibrated as a **molecular clock** against known fossil dates. It is then possible to give approximate dates of common origin to species with no or poor fossil data. This method has been applied to primate DNA samples to help determine the approximate date of human divergence from the apes, which has been estimated to be between 10 and 5 million years ago.

DNA Hybridization

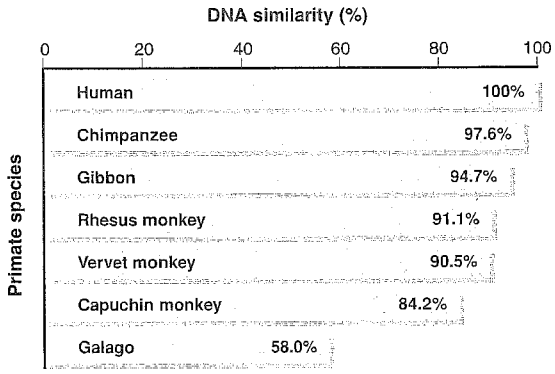
1. Blood samples from each species are taken, from which the DNA is isolated.
2. The DNA from each species is made to unwind into single strands by applying heat (both human and chimpanzee DNA unwinds at 86°C).
3. Enzymes are used to snip the single strands of DNA into smaller pieces (about 500 base pairs long).
4. The segments from human and chimpanzee DNA are combined to see how closely they bind to each other (single strand segments tend to find their complementary segments and rewind into a double helix again).
5. The greater the similarity in DNA base sequence, the stronger the attraction between the two strands and therefore they are harder to separate again. By measuring how hard this hybrid DNA is to separate, a crude measure of DNA 'relatedness' can be achieved.
6. The degree of similarity of the hybrid DNA can be measured by finding the temperature that it unzips into single strands again (in this case it would be 83.6°C).





The relationships among the **New World vultures** and **storks** have been determined using DNA hybridization. It has been possible to estimate how long ago various members of the group shared a common ancestor.

Similarity of human DNA to that of other primates



The genetic relationships among the **primates** has been investigated using DNA hybridization. Human DNA was compared with that of the other primates. It largely confirmed what was suspected from anatomical evidence.

1. Explain how **DNA hybridization** can give a measure of genetic relatedness between species:

_____

_____

_____

2. Study the graph showing the results of a DNA hybridization between human DNA and that of other primates.

   (a) Identify which is the most closely related primate to humans: _____

   (b) Identify which is the most distantly related primate to humans: _____

3. State the DNA difference score for: (a) Shoebills and pelicans: _____ (b) Storks and flamingos: _____

4. On the basis of DNA hybridization, state how long ago the ibises and New World vultures shared a common ancestor:

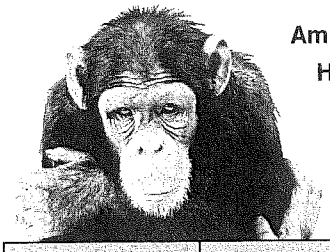
_____

Other Evidence for Evolution

Amino Acid Sequences

Each of our proteins has a specific number of amino acids arranged in a specific order. Any differences in the sequence reflect changes in the DNA sequence. The hemoglobin beta chain has been used as a standard molecule for comparing the precise sequence of amino acids in different species. Hemoglobin is the protein in our red blood cells that is responsible for carrying oxygen around our bodies. The hemoglobin in adults is made up of four polypeptide chains: two alpha chains and two beta chains. Each is coded for by a separate gene.

Example right: When the sequence of human hemoglobin, which is 146 amino acids long, was compared with that of five other primate species it was found that chimpanzees had an identical sequence while those that were already considered less closely related had a greater number of differences. This suggests a very close genetic relationship between humans, chimpanzees and gorillas, but less with the other primates.



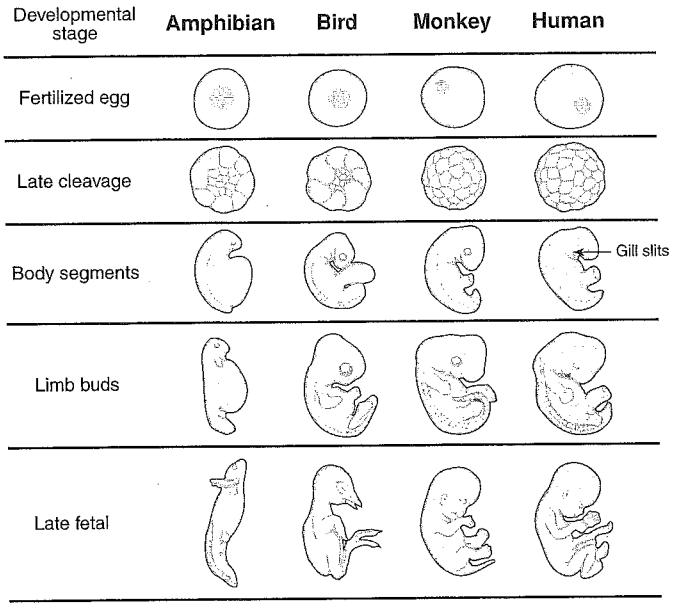
Amino Acid Differences Between Humans and Other Primates

The *'position of changed amino acid'* is the point in the protein, composed of 146 amino acids, at which the **different** amino acids occurs

Primate	No. of amino acids different from humans	Position of changed amino acids
Chimpanzee	Identical	–
Gorilla	1	104
Gibbon	3	80 87 125
Rhesus monkey	8	9 13 33 50 76 87 104 125
Squirrel monkey	9	5 6 9 21 22 56 76 87 125

Comparative Embryology

By comparing the development of embryos from different species, Ernst von Bayer in 1828 noticed that animals are more similar during early stages of their embryological development than later as adults. This later led to Ernst Haeckel (1834-1919) to propose his famous principle: *ontogeny recapitulates phylogeny*. He claimed that the development of an individual (ontogeny) retraces the stages through which the individual species has passed during its evolution (phylogeny). This idea is now known to be an oversimplification and is misleading. Although early developmental sequences between all vertebrates are similar, there are important deviations from the general developmental plan in different species. Notice the gill slits that briefly appear in the human embryo (arrowed). The more closely related forms of the monkey and humans continue to appear similar until a later stage in development, compared to more distantly related species. From the study of fetal development it is possible to find clues as to how evolution generates the diversity of life forms through time, but 'ontogeny does not recapitulate phylogeny'.



1. Study the table of data showing the differences in **amino acid sequences** for selected primates. Explain why chimpanzees and gorillas are considered most closely related to humans, while monkeys are less so:

2. Briefly describe how **comparative embryology** has contributed evidence to support the concept of evolution:

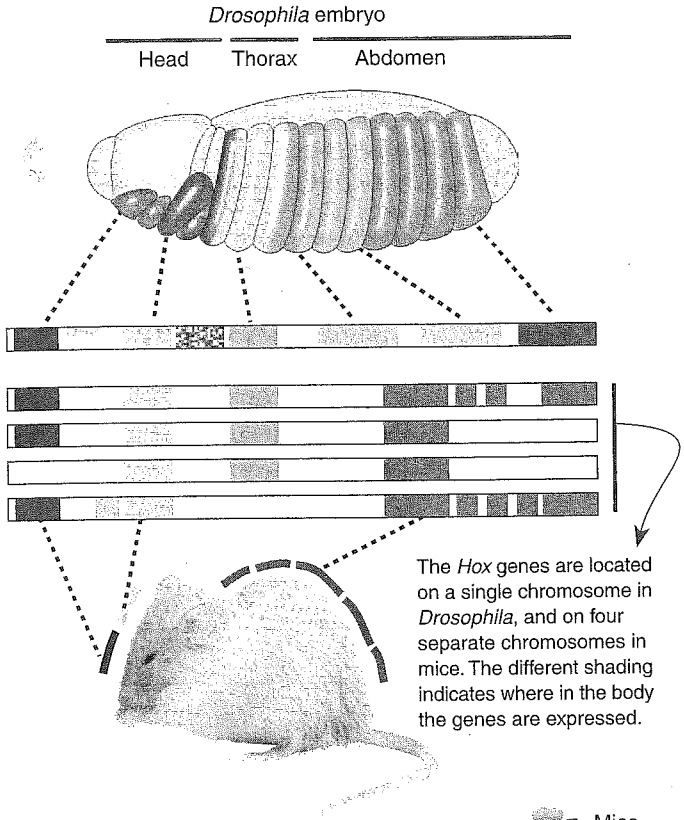
3. Describe a commonly used biochemical method for precisely analyzing the genes in organisms to determine their evolutionary relationships:

The Evolution of Novel Forms

The relatively new field of **evolutionary developmental biology** (or evo-devo) addresses the origin and evolution of embryonic development and looks at how modifications of developmental processes can lead to novel features. Scientists now know that specific genes in animals, including a subgroup of genes called ***Hox* genes**, are part of a basic **'tool kit'** of genes that control animal development. Genomic studies have shown that these genes are **highly conserved** (i.e. they show little change in different lineages). Very disparate organisms share the same **tool kit** of genes, but regulate them differently. The implication of this is that large changes in morphology or function are associated with changes in gene regulation, rather than the evolution of new genes, and natural selection associated with gene switches plays a major role in evolution.

The Role of *Hox* Genes

Hox genes control the development of back and front parts of the body.The same genes (or homologous ones) are present in essentially all animals, including humans.



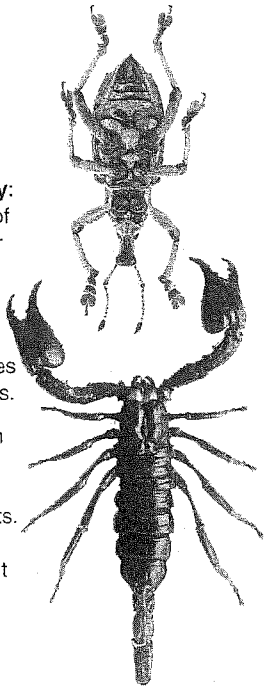
The *Hox* genes are located on a single chromosome in *Drosophila*, and on four separate chromosomes in mice. The different shading indicates where in the body the genes are expressed.

The Evolution of Novel Forms

Even very small changes (mutations) in the *Hox* genes can have a profound effect on morphology. Such changes to the genes controlling development have almost certainly been important in the evolution of novel structures and body plans. Four principles underly the evo-devo thinking regarding the evolution of novel forms:

- **Evolution works with what is already present**: New structures are modifications of pre-existing structures.
- **Multifunctionality** and **redundancy**: Functional redundancy in any part of a multifunctional structure allows for specialisation and division of labour through the development of two separate structures.

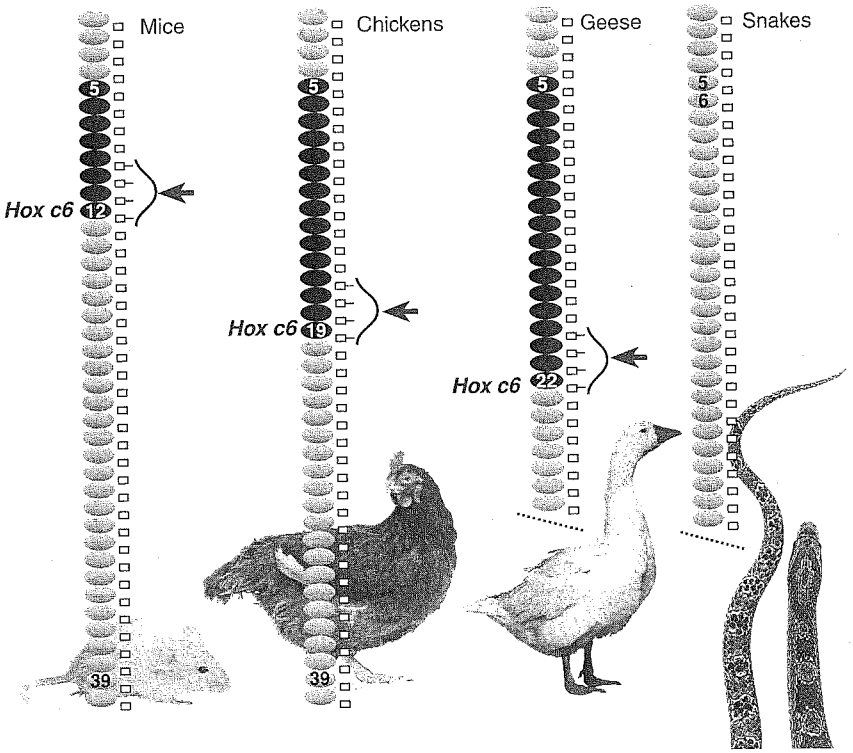
  Example: the diversity of appendages (including mouthparts) in arthropods.
- **Modularity**: Modular architecture in animals (arthropods, vertebrates) allows for the modification and specialisation of individual body parts. Genetic switches allow changes in one part of a structure, independent of other parts.



Shifting *Hox* Expression

Huge diversity in morphology in organisms within and across phyla could have arisen through small changes in the genes controlling development.

Differences in neck length in vertebrates provides a good example of how changes in gene expression can bring about changes in morphology. Different vertebrates have different numbers of neck vertebrae.The boundary between neck and trunk vertebrae is marked by expression of the ***Hox c6* gene** (c6 denotes the sixth cervical or neck vertebra) in all cases, but the position varies in each animal relative to the overall body. The forelimb (arrow) arises at this boundary in all four-legged vertebrates. In snakes, the boundary is shifted forward to the base of the skull and no limbs develop. As a result of these differences in expression, mice have a short neck, geese a long neck, and snakes, no neck at all.



Natural Selection

Natural selection operates on the phenotypes of individuals, produced by their particular combinations of alleles. In natural populations, the allele combinations of some individuals are perpetuated at the expense of other genotypes. This differential survival of some genotypes over others is called **natural selection**. The effect of natural selection can vary; it can act to maintain the genotype of a species or to change it. **Stabilizing selection** maintains the established favorable characteristics and is associated with stable environments. In contrast, **directional selection** favors phenotypes at one extreme of the phenotypic range and is associated with gradually changing environments. **Disruptive selection** is a much rarer form of selection favoring two phenotypic extremes, and is a feature of fluctuating environments.

Stabilizing Selection

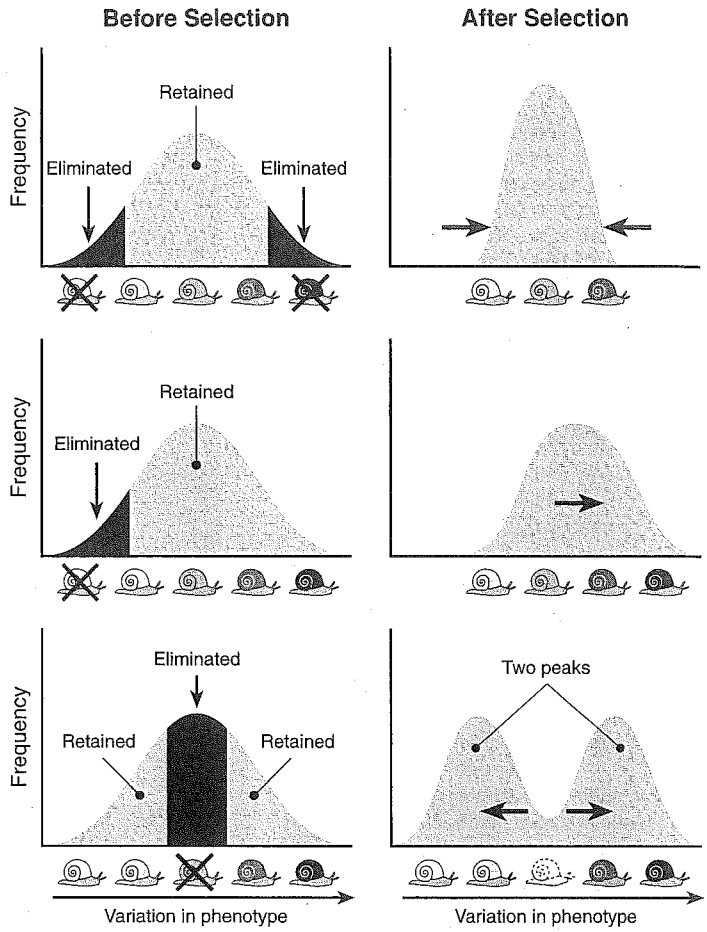
Extreme variations are culled from the population (there is selection against them). Those with the established (middle range) adaptive phenotype are retained in greater numbers. This reduces the variation for the phenotypic character. In the example right, light and dark snails are eliminated, leaving medium colored snails. Stabilizing selection can be seen in the selection pressures on human birth weights.

Directional Selection

Directional selection is associated with gradually changing conditions, where the adaptive phenotype is shifted in one direction and one aspect of a trait becomes emphasized (e.g. coloration). In the example right, light colored snails are eliminated and the population becomes darker. Directional selection was observed in peppered moths in England during the Industrial Revolution. They responded to the air pollution of industrialization by increasing the frequency of darker, melanic forms.

Disruptive or Diversifying Selection

Disruptive selection favors two extremes of a trait at the expense of intermediate forms. It is associated with a fluctuating environment and gives rise to **balanced polymorphism** in the population. In the example right, there is selection against medium colored snails, which are eliminated. There is considerable evidence that predators, such as insectivorous birds, are more likely to find and eat common morphs and ignore rare morphs. This enables the rarer forms to persist in the population.



1. (a) Distinguish between directional selection and disruptive selection, identifying when each is likely to operate:

(b) Identify which of the three types of selection described above will lead to evolution, and explain why:

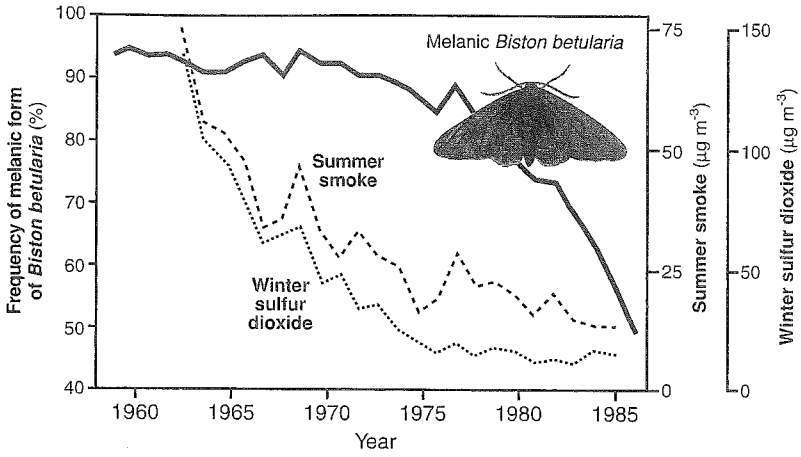
2. Explain how a change in environment may result in selection becoming directional rather than stabilizing:

3. Explain how, in a population of snails, through natural selection, shell color could change from light to dark over time:

Changes in frequency of melanic peppered moths

In the 1940s and 1950s, coal burning was still at intense levels around the industrial centers of Manchester and Liverpool. During this time, the melanic form of the moth was still very dominant. In the rural areas further south and west of these industrial centers, the gray or speckled forms increased dramatically. With the decline of coal burning factories and the Clean Air Acts in cities, the air quality improved between 1960 and 1980. Sulfur dioxide and smoke levels dropped to a fraction of their previous levels. This coincided with a sharp fall in the relative numbers of melanic moths.

Frequency of melanic peppered moth related to reduced air pollution



1. The populations of peppered moth in England have undergone changes in the frequency of an obvious phenotypic character over the last 150 years. Describe the phenotypic character that changed in its frequency:

_____

2. (a) Identify the (proposed) selective agent for phenotypic change in *Biston*: _____

   (b) Describe how the selection pressure on the light colored morph has changed with changing environmental conditions over the last 150 years:

_____

_____

3. The industrial centers for England in 1950 were located around London, Birmingham, Liverpool, Manchester, and Leeds. Glasgow in Scotland also had a large industrial base. Comment on how the relative frequencies of the two forms of peppered moth were affected by the geographic location of industrial regions:

_____

_____

_____

_____

4. The level of pollution dropped around Manchester and Liverpool between 1960 and 1985.

   (a) State how much the pollution dropped by: _____

   (b) Describe how the frequency of the darker melanic form changed during the period of reduced pollution:

_____

_____

5. In the example of the peppered moths, state whether the selection pressure is disruptive, stabilizing, or directional:

_____

6. Outline the key difference between natural and artificial selection: _____

_____

7. Discuss the statement "the environment directs natural selection": _____

_____

_____

_____

Selection for Human Birth Weight

Selection pressures operate on populations in such a way as to reduce mortality. For humans, giving birth is a special, but often traumatic, event. In a study of human birth weights it is possible to observe the effect of selection pressures operating to constrain human birth weight within certain limits. This is a good example of **stabilizing selection**. This activity explores the selection pressures acting on the birth weight of human babies. Carry out the steps below:

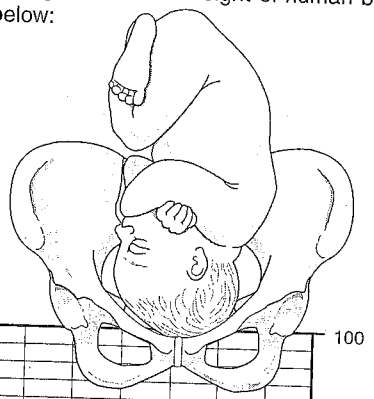
Step 1: Collect the birth weights from 100 birth notices from your local newspaper (or 50 if you are having difficulty getting enough; this should involve looking back through the last 2-3 weeks of birth notices). If you cannot obtain birth weights in your local newspaper, a set of 100 sample birth weights is provided in the Model Answers booklet.

Step 2: Group the weights into each of the 12 weight classes (of 0.5 kg increments). Determine what percentage (of the total sample) fall into each weight class (e.g. 17 babies weigh 2.5-3.0 kg out of the 100 sampled = 17%)

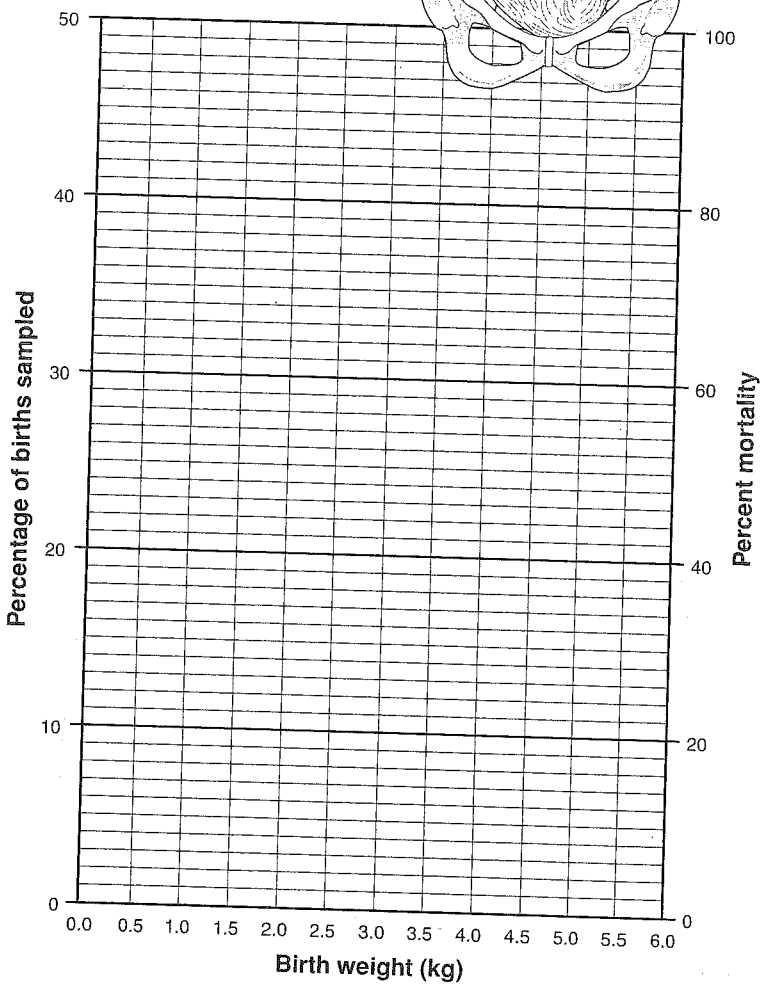
Step 3: Graph these in the form of a histogram for the 12 weight classes (use the graphing grid provided right). Be sure to use the scale provided on the left vertical (y) axis.

Step 4: Create a second graph by plotting percentage mortality of newborn babies in relation to their birth weight. Use the scale on the right y axis and data provided (below).

Step 5: Draw a line of 'best fit' through these points.



The size of the baby and the diameter and shape of the birth canal are the two crucial factors in determining whether a normal delivery is possible.



Mortality of newborn babies related to birth weight

Weight (kg)	Mortality (%)
1.0	80
1.5	30
2.0	12
2.5	4
3.0	3
3.5	2
4.0	3
4.5	7
5.0	15

Source: Biology: The Unity & Diversity of Life (4th ed), by Starr and Taggart

1. Describe the shape of the histogram for birth weights: _____
2. State the optimum birth weight in terms of the lowest newborn mortality: _____
3. Describe the relationship between newborn mortality and birth weight: _____
4. Describe the selection pressures that are operating to control the range of birth weight: _____
5. Describe how medical intervention methods during pregnancy and childbirth may have altered these selection pressures: _____

1. For each of the 2 demes shown on the previous page (treating the mutant in deme 1 as a AA):

   (a) Count up the numbers of **allele types** (**A** and **a**).

   (b) Count up the numbers of **allele combinations** (**AA, Aa, aa**).

2. Calculate the frequencies as percentages (%) for the allele types and combinations:

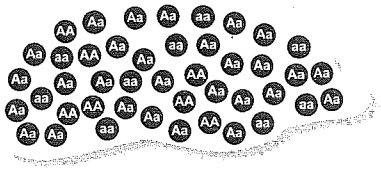
Deme 1		Number counted	%
Allele types	A		
	a		
Allele combinations	AA		
	Aa		
	aa		

Deme 2		Number counted	%
Allele types	A		
	a		
Allele combinations	AA		
	Aa		
	aa		

3. One of the fundamental concepts for population genetics is that of **genetic equilibrium**, stated as: *"For a very large, randomly mating population, the proportion of dominant to recessive alleles remains constant from one generation to the next".* If a gene pool is to remain unchanged, it must satisfy all of the criteria below that favour gene pool stability. Few populations meet all (or any) of these criteria and their genetic makeup must therefore by continually changing. For each of the five factors (a-e) below, state briefly **how** and **why** each would affect the allele frequency in a gene pool:

Factors Favoring Gene Pool Stability	Factors Favoring Gene Pool Change

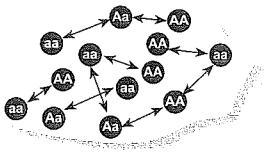
(a) Population size: _____



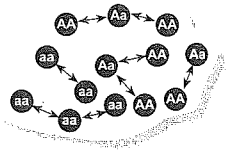
LARGE POPULATION

SMALL POPULATION

(b) Mate selection: _____

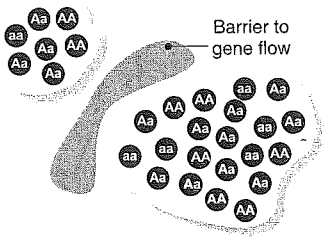


RANDOM MATING

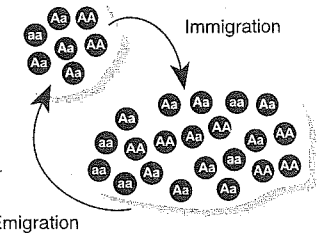


ASSORTATIVE MATING

(c) Gene flow between populations:



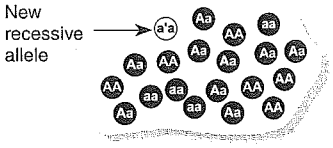
NO GENE FLOW



GENE FLOW

(d) Mutations: _____

NO MUTATION

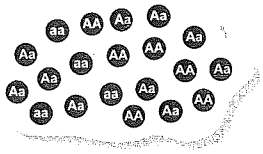


MUTATIONS

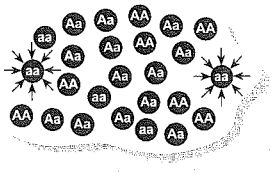
(e) Natural selection: _____

4. Identify the factors that tend to:

   (a) Increase genetic variation in populations:



NO NATURAL SELECTION



NATURAL SELECTION

   (b) Decrease genetic variation in populations:

2. You are working with pea plants and found 36 plants out of 400 were dwarf.
   Data: Frequency of recessive phenotype (36 out of 400 = 9%)

   (a) Calculate the frequency of the tall gene: _____

   (b) Determine the number of heterozygous pea plants:

   _____

   _____

Recessive allele:	q	=
Dominant allele:	p	=
Recessive phenotype:	q^2	=
Homozygous dominant:	p^2	=
Heterozygous:	2pq	=

3. In humans, the ability to taste the chemical phenylthiocarbamide (PTC) is inherited as a simple dominant characteristic. Suppose you found out that 360 out of 1000 college students could not taste the chemical.
   Data: Frequency of recessive phenotype (360 out of 1000).

   (a) State the frequency of the gene for tasting PTC:

   _____

   (b) Determine the number of heterozygous students in this population:

   _____

Recessive allele:	q	=
Dominant allele:	p	=
Recessive phenotype:	q^2	=
Homozygous dominant:	p^2	=
Heterozygous:	2pq	=

4. A type of deformity appears in 4% of a large herd of cattle. Assume the deformity was caused by a recessive gene.
   Data: Frequency of recessive phenotype (4% deformity).

   (a) Calculate the percentage of the herd that are carriers of the gene:

   _____

   (b) Determine the frequency of the dominant gene in this case:

   _____

Recessive allele:	q	=
Dominant allele:	p	=
Recessive phenotype:	q^2	=
Homozygous dominant:	p^2	=
Heterozygous:	2pq	=

5. Assume you placed 50 pure bred black guinea pigs (dominant allele) with 50 albino guinea pigs (recessive allele) and allowed the population to attain genetic equilibrium (several generations have passed).
   Data: Frequency of recessive allele (50%) and dominant allele (50%).

   Determine the proportion (%) of the population that becomes white:

   _____

Recessive allele:	q	=
Dominant allele:	p	=
Recessive phenotype:	q^2	=
Homozygous dominant:	p^2	=
Heterozygous:	2pq	=

6. It is known that 64% of a large population exhibit the recessive trait of a characteristic controlled by two alleles (one is dominant over the other).
   Data: Frequency of recessive phenotype (64%). Determine the following:

   (a) The frequency of the recessive allele: _____

   (b) The percentage that are heterozygous for this trait: _____

   (c) The percentage that exhibit the dominant trait: _____

   (d) The percentage that are homozygous for the dominant trait: _____

   (e) The percentage that has one or more recessive alleles: _____

7. Albinism is recessive to normal pigmentation in humans. The frequency of the albino allele was 10% in a population.
   Data: Frequency of recessive allele (10% albino allele).

   Determine the proportion of people that you would expect to be albino:

   _____

   _____

   _____

Recessive allele:	q	=
Dominant allele:	p	=
Recessive phenotype:	q^2	=
Homozygous dominant:	p^2	=
Heterozygous:	2pq	=

Analysis of a Squirrel Gene Pool

In Olney, Illinois, in the United States, there is a unique population of albino (white) and gray squirrels. Between 1977 and 1990, students at Olney Central College carried out a study of this population. They recorded the frequency of gray and albino squirrels. The albinos displayed a mutant allele expressed as an albino phenotype only in the homozygous recessive condition. The data they collected are provided in the table below. Using the **Hardy-Weinberg equation** for calculating genotype frequencies, it was possible to estimate the frequency of the normal 'wild' allele (G) providing gray fur coloring, and the frequency of the mutant albino allele (g) producing white squirrels. This study provided real, first hand, data that students could use to see how genotype frequencies can change in a real population.

Thanks to **Dr. John Stencel**, Olney Central College, Olney, Illinois, US, for providing the data for this exercise.

Gray squirrel, usual color form

Albino form of gray squirrel

Population of gray and white squirrels in Olney, Illinois (1977-1990)

Year	Gray	White	Total	GG	Gg	gg	Freq. of g	Freq. of G
1977	602	182	784	26.85	49.93	23.21	48.18	51.82
1978	511	172	683	24.82	50.00	25.18	50.18	49.82
1979	482	134	616	28.47	49.77	21.75	46.64	53.36
1980	489	133	622	28.90	49.72	21.38	46.24	53.76
1981	536	163	699	26.74	49.94	23.32	48.29	51.71
1982	618	151	769	31.01	49.35	19.64	44.31	55.69
1983	419	141	560	24.82	50.00	25.18	50.18	49.82
1984	378	106	484	28.30	49.79	21.90	46.80	53.20
1985	448	125	573	28.40	49.78	21.82	46.71	53.29
1986	536	155	691	27.71	49.86	22.43	47.36	52.64
1987	*No data collected this year*							
1988	652	122	774	36.36	47.88	15.76	39.70	60.30
1989	552	146	698	29.45	49.64	20.92	45.74	54.26
1990	603	111	714	36.69	47.76	15.55	39.43	60.57

1. **Graph population changes**: Use the data in the first 3 columns of the table above to plot a line graph. This will show changes in the phenotypes: numbers of gray and white (albino) squirrels, as well as changes in the total population. Plot: **gray**, **white**, and **total** for each year:

   (a) Determine by how much (as a %) total population numbers have fluctuated over the sampling period:

   _____

   (b) Describe the overall trend in total population numbers and any pattern that may exist:

   _____

   _____

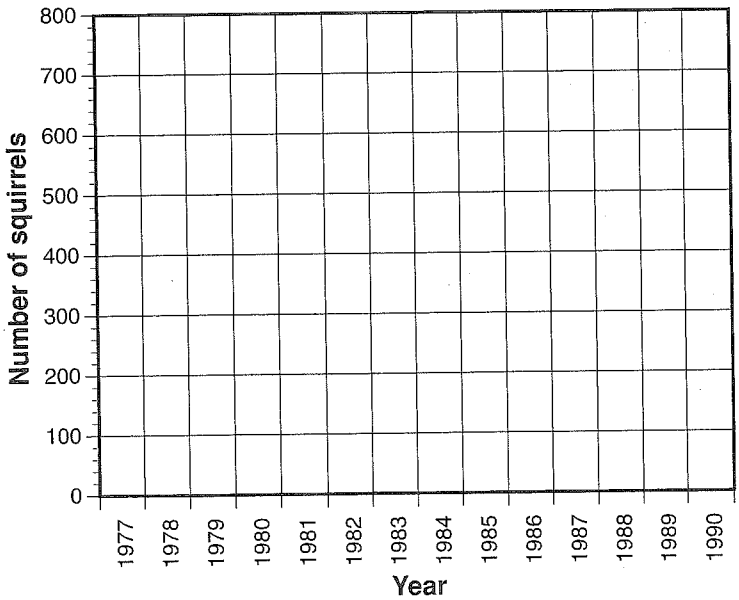
   _____

   _____

   _____

   _____

   _____



2. **Graph genotype changes**: Use the data in the genotype columns of the table on the opposite page to plot a line graph. This will show changes in the allele combinations (**GG**, **Gg**, **gg**). Plot: **GG**, **Gg**, and **gg** for each year:

Describe the overall trend in the frequency of:

(a) Homozygous dominant (**GG**) genotype:

_____

_____

(b) Heterozygous (**Gg**) genotype:

_____

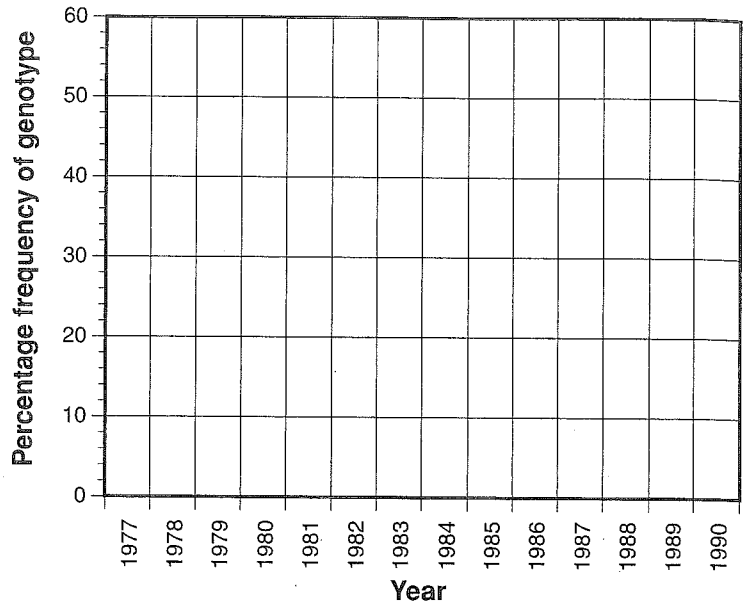
_____

(c) Homozygous recessive (gg) genotype:

_____

_____

_____



3. **Graph allele changes**: Use the data in the last two columns of the table on the previous page to plot a line graph. This will show changes in the *allele frequencies* for each of the dominant (**G**) and recessive (**g**) alleles.
Plot: the frequency of **G** and the frequency of **g**:

(a) Describe the overall trend in the frequency of the dominant allele (**G**):

_____

_____

_____

(b) Describe the overall trend in the frequency of the recessive allele (**g**):

_____

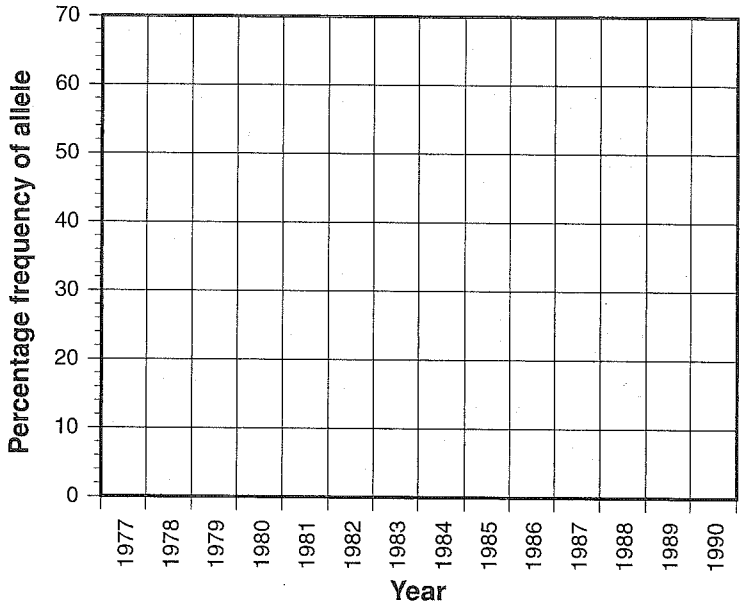
_____

_____

_____

_____

_____



4. (a) State which of the three graphs best indicates that a significant change may be taking place in the gene pool of this population of squirrels:

_____

(b) Give a reason for your answer: _____

_____

_____

5. Describe a possible cause of the changes in allele frequencies over the sampling period: _____

_____

_____

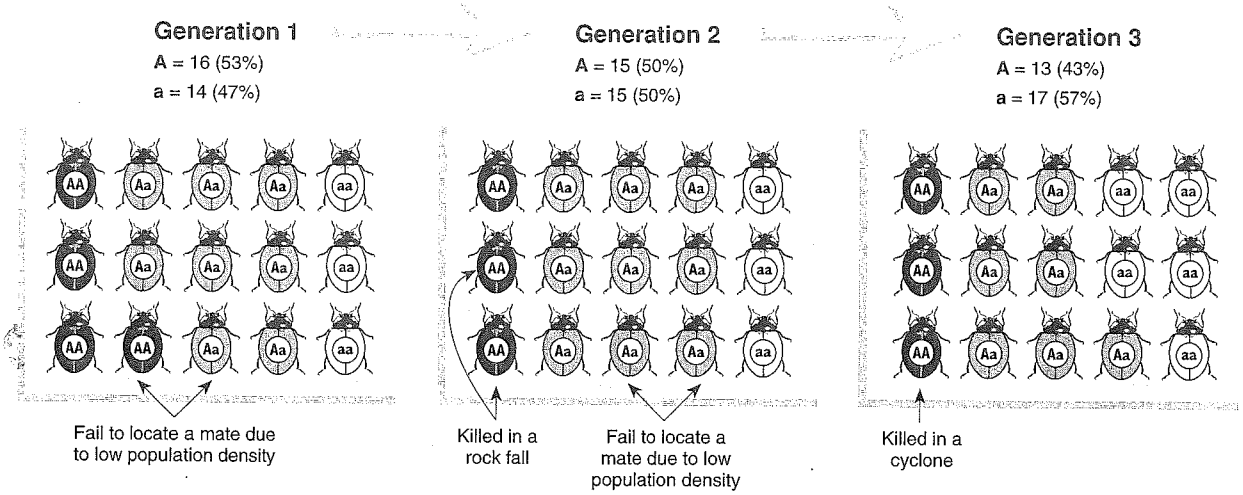
_____

_____

Genetic Drift

Not all individuals, for various reasons, will be able to contribute their genes to the next generation. **Genetic drift** (also known as the Sewell-Wright Effect) refers to the *random changes in allele frequency* that occur in all populations, but are much more pronounced in small populations. In a small population, the effect of a few individuals not contributing their alleles to the next generation can have a great effect on allele frequencies. Alleles may even become **lost** from the gene pool altogether (frequency becomes 0%) or **fixed** as the only allele for the gene present (frequency becomes 100%).

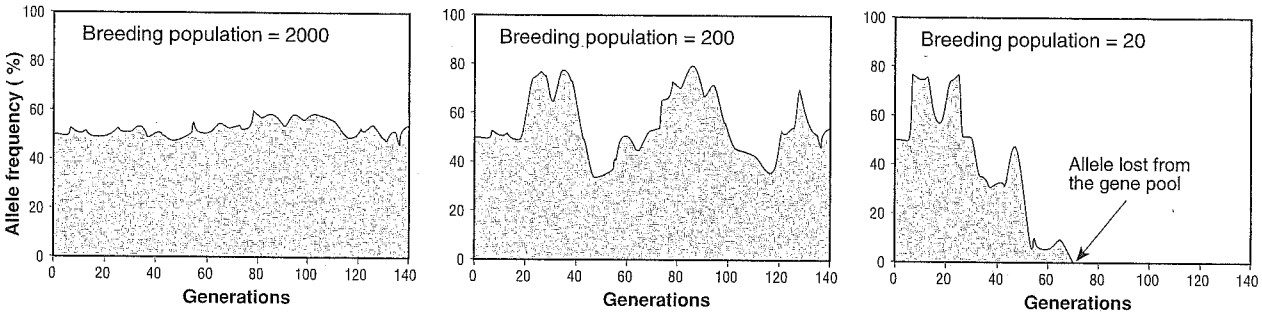
The genetic makeup (allele frequencies) of the population changes randomly over a period of time



This diagram shows the gene pool of a hypothetical small population over three generations. For various reasons, not all individuals contribute alleles to the next generation. With the random loss of the alleles carried by these individuals, the allele frequency changes from one generation to the next. The change in frequency is directionless as there is no selecting force. The allele combinations for each successive generation are determined by how many alleles of each type are passed on from the preceding one.

Computer Simulation of Genetic Drift

Below are displayed the change in allele frequencies in a computer simulation showing random genetic drift. The breeding population progressively gets smaller from left to right. Each simulation was run for 140 generations.



Large breeding population

Fluctuations are minimal in large breeding populations because the large numbers buffer the population against random loss of alleles. On average, losses for each allele type will be similar in frequency and little change occurs.

Small breeding population

Fluctuations are more severe in smaller breeding populations because random changes in a few alleles cause a greater percentage change in allele frequencies.

Very small breeding population

Fluctuations in very small breeding populations are so extreme that the allele can become fixed (frequency of 100%) or lost from the gene pool altogether (frequency of 0%).

1. Explain what is meant by **genetic drift**: _____

2. Describe how genetic drift affects the amount of genetic variation within very small populations: _____

3. Identify a small breeding population of animals or plants in your country in which genetic drift could be occurring:

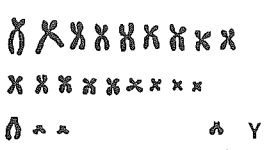
Postzygotic Isolating Mechanisms

Hybrid sterility

Even if two species mate and produce hybrid offspring that are vigorous, the species are still reproductively isolated if the hybrids are sterile (genes cannot flow from one species' gene pool to the other). Such cases are common among the horse family (such as the zebra and donkey shown on the right). One cause of this sterility is the failure of meiosis to produce normal gametes in the hybrid. This can occur if the chromosomes of the two parents are different in number or structure (see the "**zebronkey**" karyotype on the right). The **mule**, a cross between a donkey stallion and a horse mare, is also an example of **hybrid vigor** (they are robust) as well as **hybrid sterility**. Female mules sometimes produce viable eggs but males are infertile.

Zebra stallion (2N = 44) X Donkey jenny (2N = 62)

Karyotype of '**Zebronkey**' offspring (2N = 53)



Y

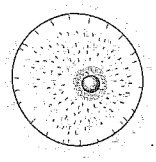
Chromosomes contributed by zebra stallion

X

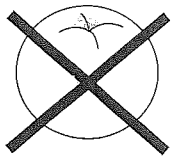
Chromosomes contributed by donkey jenny

Hybrid inviability

Mating between individuals of two different species may sometimes produce a zygote. In such cases, the genetic incompatibility between the two species may stop development of the fertilized egg at some embryonic stage. Fertilized eggs often fail to divide because of unmatched chromosome numbers from each gamete (a kind of aneuploidy between species). Very occasionally, the hybrid zygote will complete embryonic development but will not survive for long.



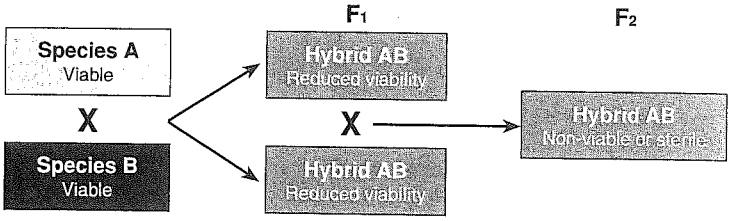
Zygote is formed



Gastrula fails to develop

Hybrid breakdown

First generation (F_1) are fertile, but the second generation (F_2) are infertile or inviable. Conflict between the genes of two species sometimes manifests itself in the second generation.



1. In general terms, explain the role of reproductive isolating mechanisms in maintaining the integrity of a species:

_____

_____

2. In the following examples, classify the reproductive isolating mechanism as either **prezygotic** or **postzygotic** and describe the mechanisms by which the isolation is achieved (e.g. temporal isolation, hybrid sterility etc.):

   (a) Some different cotton species can produce fertile hybrids, but breakdown of the hybrid occurs in the next generation when the offspring of the hybrid die in their seeds or grow into defective plants:

   Prezygotic / postzygotic (delete one) Mechanism of isolation: _____

   (b) Many plants have unique arrangements of their floral parts that stops transfer of pollen between plants:

   Prezygotic / postzygotic (delete one) Mechanism of isolation: _____

   (c) Three species of orchid living in the same rainforest do not hybridize because they flower on different days:

   Prezygotic / postzygotic (delete one) Mechanism of isolation: _____

   (d) Several species of the frog genus *Rana*, live in the same regions and habitats, where they may occasionally hybridize. The hybrids generally do not complete development, and those that do are weak and do not survive long:

   Prezygotic / postzygotic (delete one) Mechanism of isolation: _____

3. Postzygotic isolating mechanisms are said to reinforce prezygotic ones. Explain why this is the case:

_____

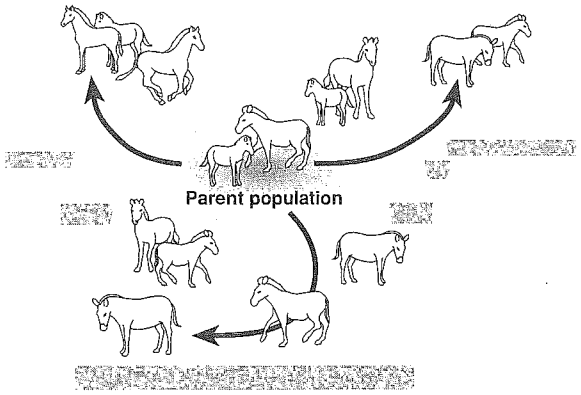
_____

Allopatric Speciation

Allopatric speciation is a process thought to have been responsible for a great many instances of species formation. It has certainly been important in countries which have had a number of cycles of geographical fragmentation. Such cycles can occur as the result of glacial and interglacial periods, where ice expands and then retreats over a land mass. Such events are also accompanied by sea level changes which can isolate populations within relatively small geographical regions.

Stage 1: Moving into new environments

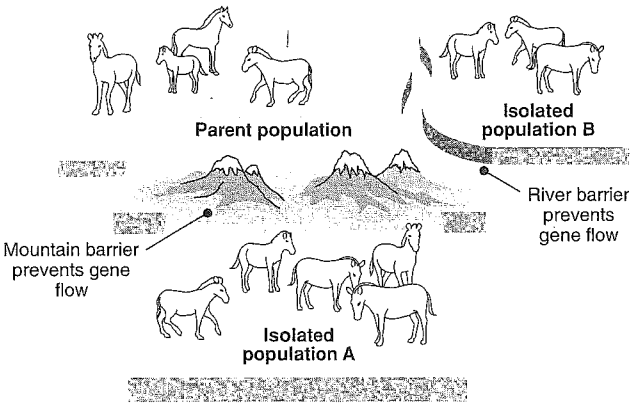
There are times when the range of a species expands for a variety of different reasons. A single population in a relatively homogeneous environment will move into new regions of their environment when they are subjected to intense competition (whether it is interspecific or intraspecific). The most severe form of competition is between members of the same species since they are competing for identical resources in the habitat. In the diagram on the right there is a 'parent population' of a single species with a common gene pool with regular 'gene flow' (theoretically any individual has access to all members of the opposite sex for mating purposes).



Stage 2: Geographical isolation

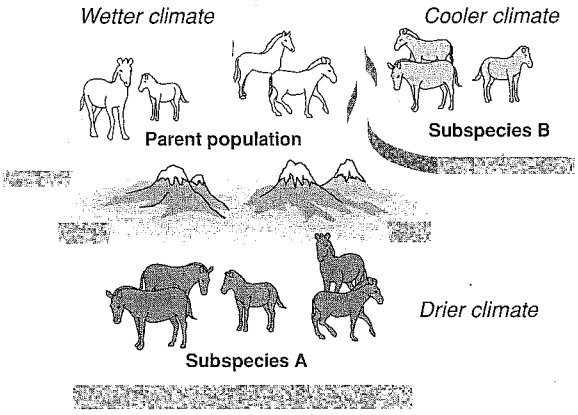
Isolation of parts of the population may occur due to the formation of **physical barriers**. These barriers may cut off those parts of the population that are at the extremes of the species range and gene flow is prevented or rare. The rise and fall of the sea level has been particularly important in functioning as an isolating mechanism. Climatic change can leave 'islands' of habitat separated by large inhospitable zones that the species cannot traverse.

Example: In mountainous regions, alpine species are free to range widely over extensive habitat during cool climatic periods. During warmer periods, however, they may become isolated because their habitat is reduced to 'islands' of high ground surrounded by inhospitable lowland habitat.



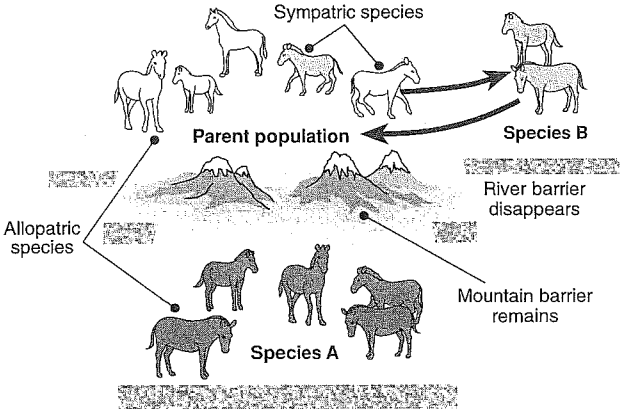
Stage 3: Different selection pressures

The isolated populations (A and B) may be subjected to quite different selection pressures. These will favor individuals with traits that suit each particular environment. For example, population A will be subjected to selection pressures that relate to drier conditions. This will favor those individuals with phenotypes (and therefore genotypes) that are better suited to dry conditions. They may for instance have a better ability to conserve water. This would result in improved health, allowing better disease resistance and greater reproductive performance (i.e. more of their offspring survive). Finally, as allele frequencies for certain genes change, the population takes on the status of a **subspecies**. Reproductive isolation is not yet established but the subspecies are significantly different genetically from other related populations.



Stage 4: Reproductive isolation

The separated populations (isolated subspecies) will often undergo changes in their genetic makeup as well as their behavior patterns. These ensure that the gene pool of each population remains isolated and 'undiluted' by genes from other populations, even if the two populations should be able to remix (due to the removal of the geographical barrier). Gene flow does not occur. The arrows (in the diagram to the right) indicate the zone of overlap between two species after the new Species B has moved back into the range inhabited by the parent population. Closely-related species whose distribution overlaps are said to be **sympatric species**. Those that remain geographically isolated are called **allopatric species**.



Sympatric Speciation

New species may be formed even where there is no separation of the gene pools by physical barriers. Called **sympatric speciation**, it is rarer than allopatric speciation, although not uncommon in plants which form **polyploids**. There are two situations where sympatric speciation is thought to occur. These are described below:

Speciation Through Niche Differentiation

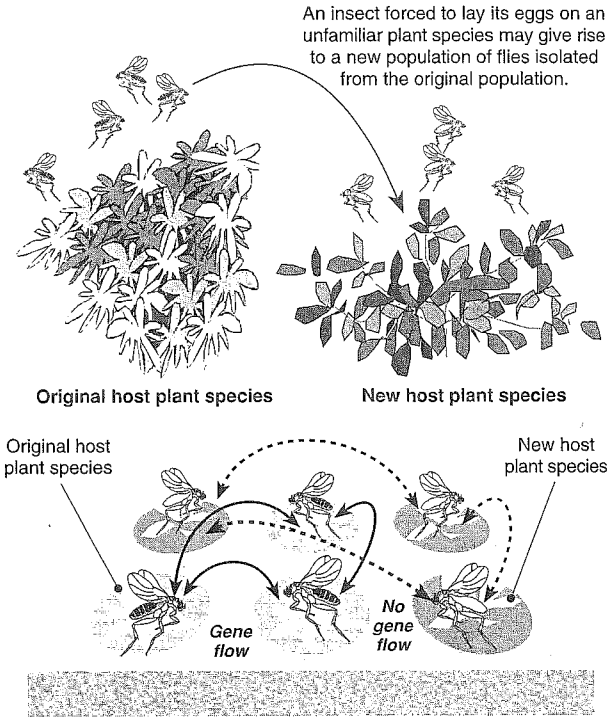
Niche isolation

In a heterogeneous environment (one that is not the same everywhere), a population exists within a diverse collection of **microhabitats**. Some organisms prefer to occupy one particular type of 'microhabitat' most of the time, only rarely coming in contact with fellow organisms that prefer other microhabitats. Some organisms become so dependent on the resources offered by their particular microhabitat that they never meet up with their counterparts in different microhabitats.

Reproductive isolation

Finally, the individual groups have remained genetically isolated for so long because of their microhabitat preferences, that they have become reproductively isolated. They have become new species that have developed subtle differences in behavior, structure, and physiology. Gene flow (via sexual reproduction) is limited to organisms that share a similar microhabitat preference (as shown in the diagram on the right).

Example: When it is time for them to lay eggs, some beetles preferentially locate the same plant species as they grew up on. Individual beetles of the same species have different preferences.



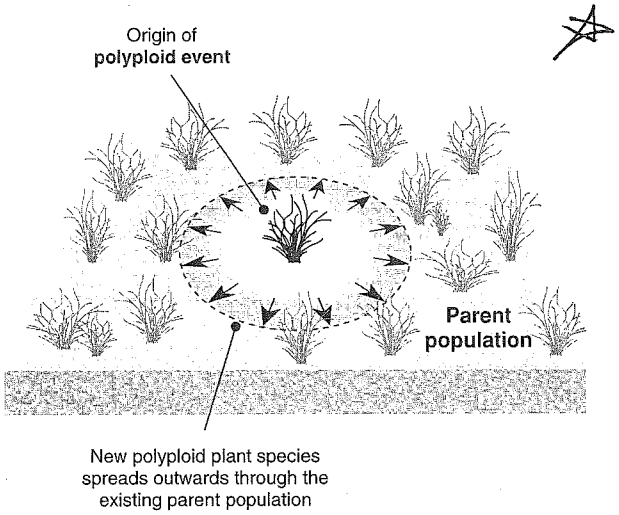
Instant Speciation by Polyploidy

When polyploidy occurs, it is possible to form a completely new species without isolation from the parent species. This type of malfunction during the process of meiosis produces sudden reproductive isolation for the new group. Because the sex-determining mechanism is disturbed, animals are rarely able to achieve new species status this way (they are effectively sterile e.g. tetraploid XXXX). Many plants, on the other hand, are able to reproduce vegetatively, or carry out self pollination. This ability to reproduce on their own enables such polyploid plants to produce a breeding population.

Speciation by allopolyploidy

This type of polyploidy usually arises from the doubling of chromosomes in a hybrid between two different species. The doubling often makes the hybrid fertile.

Examples: Modern wheat. Swedes are polyploid species formed from a hybrid between a type of cabbage and a type of turnip.



1. Explain what is meant by **sympatric speciation** and identify the mechanisms by which it can occur:

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2. Explain briefly how polyploidy may cause the formation of a new species: _____

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3. Identify an example of a species that has been formed by polyploidy: _____

4. Explain how niche differentiation may cause the formation of a new species: _____

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