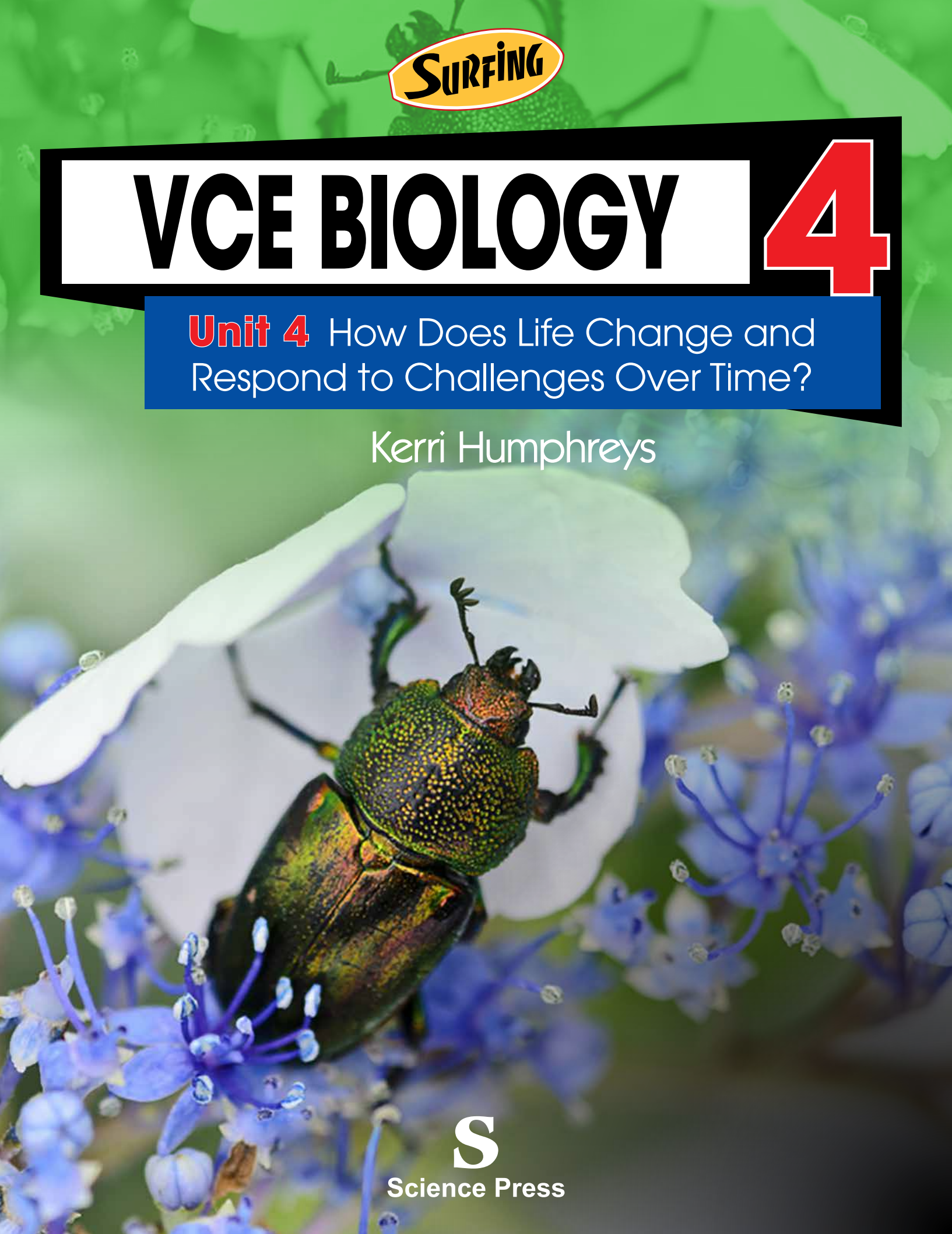
SURFING

VCE BIOLOGY 4

Unit 4 How Does Life Change and Respond to Challenges Over Time?

Kerri Humphreys



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Contents

Introduction

This book covers the Biology content specified in the Victorian Certificate of Education Biology Study Design. Sample data has been included for suggested experiments to give you practice to reinforce practical work in class.

Each book in the *Surfing* series contains a summary, with occasional more detailed sections, of all the mandatory parts of the syllabus, along with questions and answers.

All types of questions – multiple choice, short response, structured response and free response – are provided. Questions are written in exam style so that you will become familiar with the concepts of the topic and answering questions in the required way.

Answers to all questions are included.

A topic test at the end of the book contains an extensive set of summary questions. These cover every aspect of the topic, and are useful for revision and exam practice.

Words To Watch

account, account for State reasons for, report on, give an account of, narrate a series of events or transactions.

analyse Interpret data to reach conclusions.

annotate Add brief notes to a diagram or graph.

apply Put to use in a particular situation.

assess Make a judgement about the value of something.

calculate Find a numerical answer.

clarify Make clear or plain.

classify Arrange into classes, groups or categories.

comment Give a judgement based on a given statement or result of a calculation.

compare Estimate, measure or note how things are similar or different.

construct Represent or develop in graphical form.

contrast Show how things are different or opposite.

create Originate or bring into existence.

deduce Reach a conclusion from given information.

define Give the precise meaning of a word, phrase or physical quantity.

demonstrate Show by example.

derive Manipulate a mathematical relationship(s) to give a new equation or relationship.

describe Give a detailed account.

design Produce a plan, simulation or model.

determine Find the only possible answer.

discuss Talk or write about a topic, taking into account different issues or ideas.

distinguish Give differences between two or more different items.

draw Represent by means of pencil lines.

estimate Find an approximate value for an unknown quantity.

evaluate Assess the implications and limitations.

examine Inquire into.

explain Make something clear or easy to understand.

extract Choose relevant and/or appropriate details.

extrapolate Infer from what is known.

hypothesise Suggest an explanation for a group of facts or phenomena.

identify Recognise and name.

interpret Draw meaning from.

investigate Plan, inquire into and draw conclusions about.

justify Support an argument or conclusion.

label Add labels to a diagram.

list Give a sequence of names or other brief answers.

measure Find a value for a quantity.

outline Give a brief account or summary.

plan Use strategies to develop a series of steps or processes.

predict Give an expected result.

propose Put forward a plan or suggestion for consideration or action.

recall Present remembered ideas, facts or experiences.

relate Tell or report about happenings, events or circumstances.

represent Use words, images or symbols to convey meaning.

select Choose in preference to another or others.

sequence Arrange in order.

show Give the steps in a calculation or derivation.

sketch Make a quick, rough drawing of something.

solve Work out the answer to a problem.

state Give a specific name, value or other brief answer.

suggest Put forward an idea for consideration.

summarise Give a brief statement of the main points.

synthesise Combine various elements to make a whole.

SURFING

VCE BIOLOGY 4

Area of Study 1

How Are Species Related?

1 Assumed Knowledge

1. The diagram shows a chromosome.

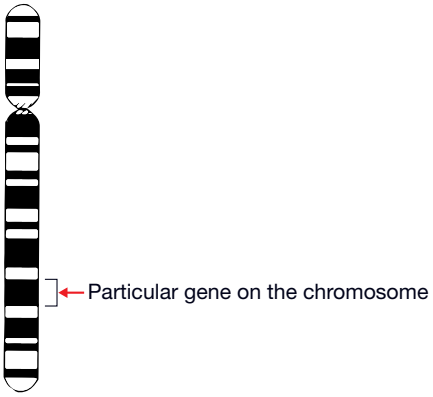


Figure 1.1 Chromosome.

   (a) Define a chromosome.
   (b) What is the relationship between gene and chromosome?
2. Distinguish between a gene and an allele.
3. Distinguish between autosomes and sex chromosomes.
4. The diagram shows the karyogram for person A.

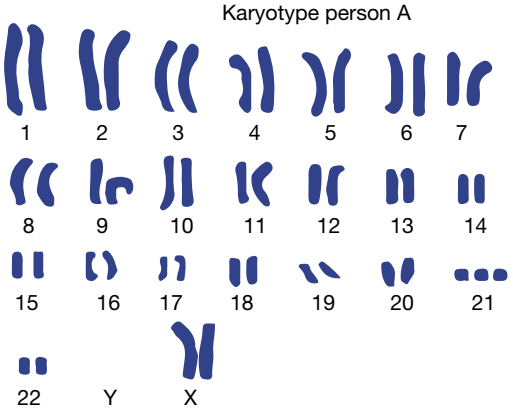


Figure 1.2 Karyogram.

   (a) Is this person male or female?
   (b) Define trisomy.
   (c) Person A has a trisomy disorder. Name this disorder.
5. What is meant by the genome sequence?
6. Define a gene pool.
7. What is meant by allele frequency?
8. Define polyploidy.
9. What is a mutation?
10. Distinguish between genotype and phenotype.
11. What features classify humans as primates?
12. What is comparative genomics?
13. Define biodiversity.
14. What is a fossil?
15. Define a species.
16. Where is most DNA in a eukaryote cell?
17. The diagram shows a biological process.

Figure 1.3 Biological process.

   Name this process.
18. What is an index fossil?
19. State the law of superposition.
20. Distinguish between absolute dating and relative dating.
21. The diagram shows when fossil animals called trilobites existed on Earth.

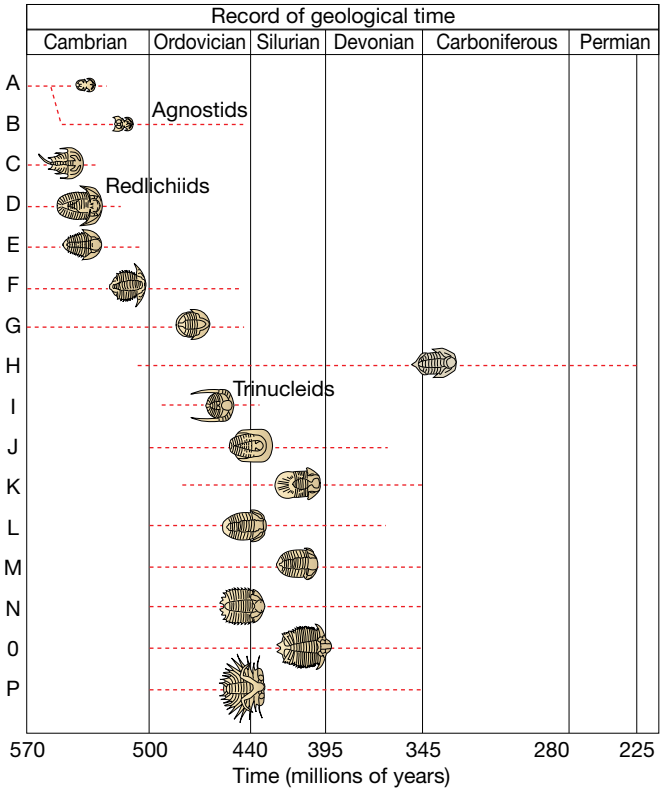


Figure 1.4 When trilobites lived.

   Explain why trilobite D is useful as an index fossil but trilobite H is not particularly suitable to be used as an index fossil.
22. Define natural selection.
23. Use an example to show how present-day organisms have developed from different organisms in the distant past.
24. When does evolution occur?
25. List the major stages in the evolution of living things from organic molecules to multicellular organisms.

2 Allele Frequency

An **allele** is an alternative for a particular trait, e.g. there are two alleles for height in pea plants – tall (T) or short (t). Alleles are the alternative forms of a gene and occupy a particular locus on a chromosome. The **frequency** of an allele in a population refers to the proportion of the population that have that allele.

The Hardy-Weinberg principle

Population genetics studies how populations change genetically over time and the Hardy-Weinberg principle proposes that frequencies of alleles and genotypes remain constant from generation to generation provided that only Mendelian segregation and recombination of alleles occur. This is known as the **Hardy-Weinberg equilibrium**. This means that there is no evolution occurring in the population.

The Hardy-Weinberg principle gives the mathematical formula to show how the frequencies of two alleles (A and a) represented by symbols *p* and *q* maintains constant gene frequencies if the following conditions exist. 1. The population is large. 2. There is no mutation or the rate of mutation of a specific allele equals the reverse mutation. 3. There is no selective mating and reproduction is purely random. 4. There is no migration of individuals into or out of the population. The Hardy-Weinberg equation is as follows.

$$p^2 + 2pq + q^2 = 1 \text{ or } 100\%$$

This means that there is 100% probability that the offspring are AA (p^2), Aa ($2pq$), aa (q^2).

Gene pool

A **gene pool** is the total aggregate of genes in a population at any one time. When studying a gene pool it is important to first define the population. A **population** is a group of organisms which can freely interbreed to produce fertile offspring living in a particular area at a particular time.

The gene pool of a localised population living in a remote area, e.g. Inuit in Greenland will have a different gene pool to the people living in a distant area, e.g. people living in Arnhem Land, Northern Territory. Allele frequencies of particular genes will differ if these two populations are compared.

Genome

A **genome** is the complete complement of an organism's genes and the **genome sequence** gives the order of the As, Ts, Cs and Gs in the DNA code. **Genomics** is the study of whole sets of genes of their interactions.

Genes are coded DNA which is transcribed into RNA that is used in protein formation. Controlling proteins means that when differentiation occurs the final cell structure and functioning is controlled by which genes are switched on or off in gene expression, e.g. the switching on of genes that control specific proteins such as keratin and collagen and the switching on of particular enzymes that regulate specific reactions will determine the shape, composition and functioning ability of the cell. Allele frequency of particular traits will then show the frequency of particular phenotypes in a population.

Phenotype is the observable physical and physiological traits of an organism and the outward appearance of an organism. Phenotype depends not only on which alleles are present but also on environmental influences, e.g. temperature during development can affect gene expression in some organisms.

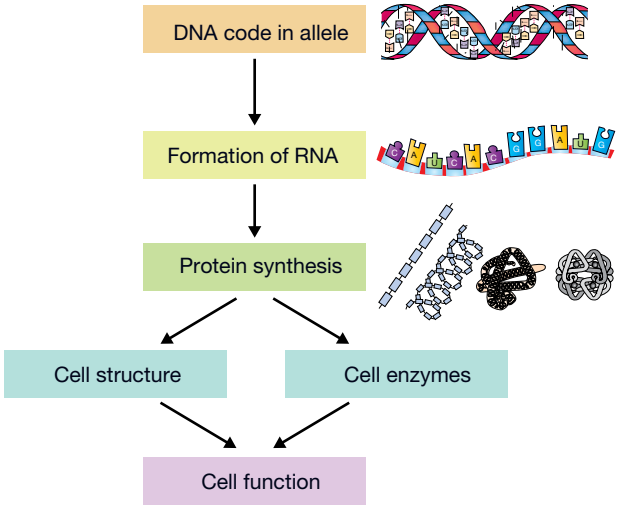


Figure 2.1 Alleles of a gene control cell functions.

QUESTIONS

1. Define an allele.
2. What is meant by allele frequency?
3. How is the Hardy-Weinberg equilibrium used in population genetics?
4. Define a gene pool.
5. What is a population?
6. Define a genome and genomics.
7. Explain how genes determine the structure and functioning of a cell.
8. How does allele frequency affect phenotype in a population?
9. What determines phenotype?
10. What is allele frequency?
    (A) The number of people with a certain trait.
    (B) The number with an alternate trait.
    (C) The proportion of a population with that allele.
    (D) The ratio of different alleles in different groups.

3 Point Mutations

A mutation is a permanent change in the genetic information and is a cause in genetic diversity. A gene mutation is a permanent change in the genetic information in a gene. The mutation can involve one or more base pairs and can be anywhere in the gene.

A point mutation is a change in one base in a single nucleotide in a gene. If the point mutation occurs in a gamete or zygote the change will affect every cell in the developing organism and will be passed to future generations. If the point mutation occurs in the developing embryo or foetus the change will affect tissues and cells that descend from this cell and may be passed to future offspring depending on the location of the mutation, e.g. in reproductive organs. If the point mutation occurs in an adult somatic cell the change will not be inherited by future generations and the effect on the person will depend on the specific mutation and how the body detects and responds to the error.

There are four basic types of point mutation – base pair substitution, insertion, deletion and inversion.

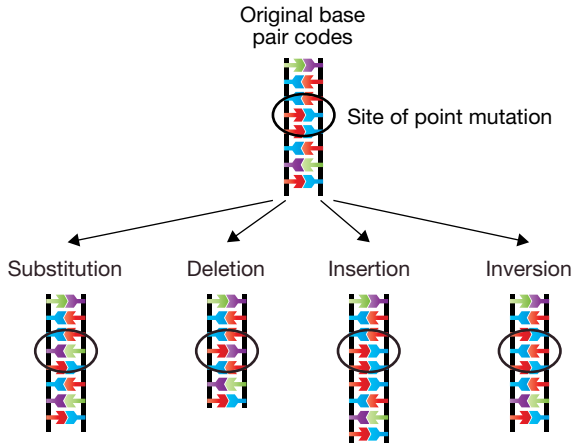


Figure 3.1 Point mutations.

Base pair substitution

In a base pair substitution one nucleotide and its complementary partner replace another pair of nucleotides. Sometimes this change is a **silent mutation** as the change has no effect on the protein being produced. For example, using the mRNA codes from Table 3.1 a base pair substitution from CUU to CUC will be a silent mutation as both code for the amino acid leucine. Other silent mutations may change the amino acid but have little effect on the protein. This can occur when the new amino acid has properties similar to the amino acid it replaced or the substitution has occurred in an area of the protein that is not an important determinant of the shape of the protein and its function.

Silent mutations occur fairly frequently but are hard to detect as they are not easily observed. Occasionally the change can be beneficial and can increase variation.

If the base pair substitution affects the shape of the protein or is at the active site of an enzyme, the change can seriously affect the functioning of the protein. For example, disulfide bridges are formed when two cysteine amino acids are brought close together with their sulfhydryl groups (–SH) on their side chains in close proximity due to the folding of the protein and a change in the code from UGC (cysteine a polar amino) to UUC (phenylalanine a non-polar amino acid) can affect the structure and functioning of the protein. Disulfide bridges are important in maintaining the structure of immunoglobulins (antibodies) and the antigen receptor sites on lymphocytes.

Table 3.1 mRNA codes.

UUU Phe	UCU Ser	UAU Tyr	UGU Cys
UUC Phe	UCC Ser	UAC Tyr	UGC Cys
UUA Leu	UCA Ser	UAA stop	UGA stop
UUG Leu	UCG Ser	UAG stop	UGG Trp
CUU Leu	CCU Pro	CAU His	CGU Arg
CUC Leu	CCC Pro	CAC His	CGC Arg
CUA Leu	CCA Pro	CAA Gln	CGA Arg
CUG Leu	CCG Pro	CAG Gln	CGG Arg
AUU He	ACU Thr	AAU Asn	AGU Ser
AUC He	ACC Thr	AAC Asn	AGC Ser
AUA He	ACA Thr	AAA Lys	AGA Arg
AUG Met	ACG Thr	AAG Lys	AGG Arg
GUU Val	GCU Ala	GAU Asp	GGU Gly
GUC Val	GCC Ala	GAC Asp	GGC Gly
GUA Val	GCA Ala	GAA Glu	GGA Gly
GUG Val	GCG Ala	GAG Glu	GGG Gly

U – uracil (thymine)	Cys – cysteine	Met – methionine
C – cytosine	Gln – glutamine	Phe – phenylalanine
A – adenine	Glu – glutamine	Pro – proline
G – guanine	Gly – glycine	Ser – serine
Ala – alanine	His – histidine	Thr – threonine
Arg – arginine	He – isoleucine	Trp – tryptophan
Asn – asparagine	Leu – leucine	Tyr – tyrosine
Asp – aspartic acid	Lys – lysine	Val – valine

Most base pair substitution mutations cause a missense mutation. A **missense mutation** is a changed codon that codes for an amino acid but does not necessarily make the correct sense. A **nonsense mutation** is a point mutation that changes a codon for an amino acid into a stop codon. The stop codon causes translation to stop shortening the polypeptide chain that is being synthesised. In most cases a nonsense mutation creates a non-functional protein. For example, a change in the code UCA for serine to UAA will create a stop at this point in translation and process of protein synthesis.

Deletion

Deletion is a loss of nucleotide pairs in a gene sequence. This causes a **frameshift mutation**. The nucleotide sequence is read in multiples of three (the code for each amino acid) so the deletion of one nucleotide will change the reading sequence. A frameshift mutation will not occur if the change was an insertion or deletion of three nucleotides. The example in Figure 3.2 shows how the reading frame is changed with a deletion.

The old dog sat on a hot day on a red mat.

As a triplet code this would be read

The/ old/ dog/ sat/ on a/ hot/ day/ on a/ red/ mat/

If the 'e' from 'The' is deleted, the code is read

Th o/ ldd/ ogs/ ato/ nah/ otd/ ayo/ nar/ edm/ at

Figure 3.2 Frameshift mutation due to deletion.

Insertion

Insertion is a point mutation where a nucleotide is added into the code. This will also cause a frameshift mutation. A frameshift mutation can be a nonsense mutation. Depending on the location of the insertion a number of different effects can occur.

Point mutations can occur spontaneously, e.g. during replication or can be induced by mutagens including ionising radiation, e.g. X-rays, cosmic rays, non-ionising radiation, e.g. UV light, various chemicals, e.g. benzene and some biological agents.

Inversion

In an inversion point mutation a nucleotide pair reverse positions with the next pair of nucleotides.

QUESTIONS

1. Define a mutation.
2. What is a gene mutation?
3. Define a point mutation.
4. Name the four basic types of point mutation.
5. What is a silent mutation?
6. Use Table 3.1 with the mRNA codes for amino acids to determine which of the following would be silent mutations.
   (a) GUU to GUG (b) ACG to AAG
   (c) GGU to GAU (d) CGG to UGG
   (e) GCU to GCC
7. Distinguish between a missense mutation and a nonsense mutation.
8. What is a frameshift mutation?
9. Use an example to show how a point mutation can have serious consequences.
10. What are the mRNA stop codes?
11. The diagram shows a section of DNA of a gene which undergoes a point mutation.

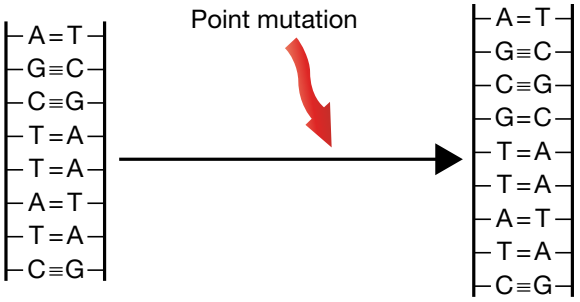


Figure 3.3 Point mutation in a gene.

Copy the diagram and label the location of the point mutation and which type of point mutation has occurred.

Use the diagram which shows a deletion of two nucleotides for the next THREE questions.

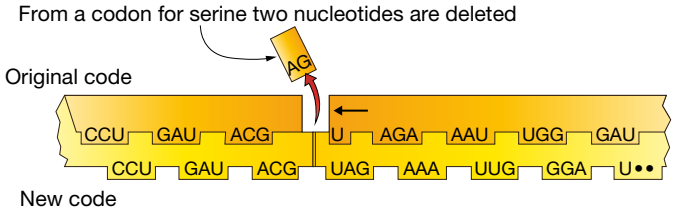


Figure 3.4 Deletion of two nucleotides.

12. What is the best description of this mutation?
    (A) Frameshift.
    (B) Chromosomal.
    (C) Insertion.
    (D) Base pair substitution.
13. Why is this mutation a nonsense mutation?
    (A) The new code does not read correctly.
    (B) The right proteins will not be produced.
    (C) There is no codon with only one nucleotide.
    (D) The new code is UAG which is a stop codon.
14. What was the code for serine that was involved in the mutation?
    (A) AGC (B) AGU (C) UGA (D) UAG
15. What is the best definition of a point mutation?
    (A) A permanent change in the genetic information.
    (B) An agent that interacts with DNA to cause a change.
    (C) A change in one base in a single nucleotide in a gene.
    (D) A change in an amino acid that alters protein structure.

4 Block Mutations

A block mutation is a permanent change to a segment of a chromosome that rearranges, deletes or disrupts many loci. Many block mutations are harmful changing the structure of the chromosome, though some are neutral especially if genes remain intact and sometimes the change can be beneficial, e.g. the change links genes that together now produce a positive effect.

Block mutations can be caused by **transposons** (transposable genetic elements) which are DNA segments that can move from one position to another in the chromosome. Transposons are sometimes called 'jumping genes' which is not how they move as they do not 'jump'. When the folding of the DNA molecule brings segments near each other transposons follow a 'cut and paste' mechanism to move to a new location or follow a 'copy and paste' mechanism replicating a section of DNA and adding it to another area. Transposons were discovered by Barbara McClintock when studying corn in the 1940s and she was awarded the Nobel Prize in Physiology or Medicine in 1983 for her discovery of 'mobile genetic elements'. She was the first female to receive this prize unshared. It is estimated that about 44% of the human genome is **repetitive DNA** which is multiple copies of DNA sections.

Types of block mutations include duplications, inversions, deletions, insertions and translocations.

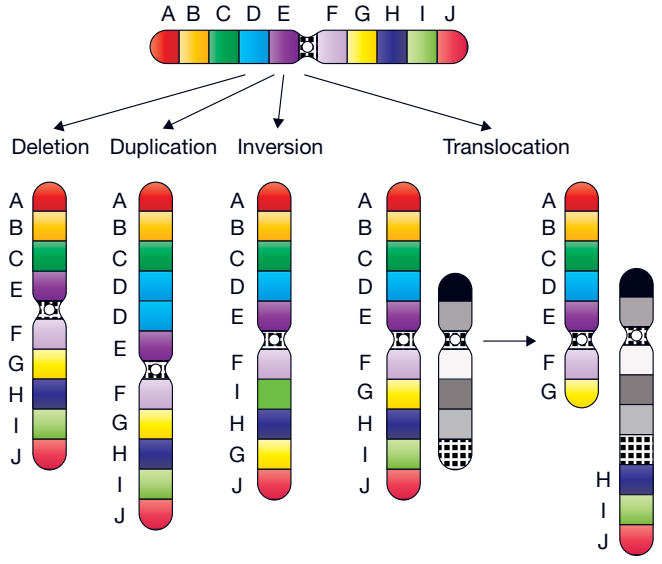


Figure 4.1 Types of block mutations.

Some sections of DNA are called **hotspots** as they are places that are more likely to undergo mutation than other places with an observable higher mutation frequency. Hotspots can be single nucleotides or short stretches of repeated nucleotides that have some basic instability or chemical tendency for nucleotide substitution.

Deletions

Deletions occur when a section of DNA is removed from a chromosome. The effect of a deletion depends on the size and location of the removed block sequence. Large deletions can involve several genes and have greater effect on phenotype and the health of the individual. Cri du chat syndrome is a deletion disorder that is caused by a deletion on the short arm of chromosome 5.

Duplications

Duplications occur when sections of DNA are replicated making the chromosome longer. Repetitive DNA can occur during DNA replication or recombination in crossing over and segregation during mitosis or meiosis. If the copies of a repeat sequence lie adjacent to each other they are called **tandem repeats**. Tandem repeats can vary in length with the large repeat units called satellites. Satellite DNA was first discovered when DNA was centrifuged and the repetitive units appeared as a distinct band in the tube. **Satellite DNA** (also now called simple sequence DNA) is a section of tandem, non-coding DNA that can be thousands of base pairs long. Microsatellite DNA is a short region of repeats that are used as genetic markers in DNA fingerprinting. In humans some microsatellites have 20 or more alleles which provides the variation to assist in identifying particular individuals by their DNA. **Trinucleotide disorders** occur when there are too many trinucleotide repeats in a gene, e.g. Huntington's disease occurs when there are more than 35 CAG repeats on the gene coding for the protein HTT. A genetic disorder due to duplication is Charcot-Marie-Tooth disease type 1 with duplication of 17p12 – a large section on the short arm of chromosome 17.

Inversions

Inversions occur when a section of DNA breaks and is reattached in the reverse orientation and order. This changes chromosome structure. Inversion is a cause for haemophilia A with an inversion in the factor VIII gene on X chromosome. The inversion within this gene stops protein production which means that testing for this disorder often involves measuring protein activity rather than a genetic test for the inversion.

Translocation

In translocation a section of one chromosome moves to a non-homologous chromosome. In a **reciprocal translocation** the non-homologous chromosomes exchange segments. Myeloproliferative syndrome is a genetic disorder caused by translocation of genetic material from chromosome 8 to other chromosomes, e.g. t(8;13)(p11;q12) which involves translocation of chromosomes 8 and 13 in lymphoma cells.

QUESTIONS

1. Define a mutation.
2. What is a transposon?
3. Explain why the term 'jumping gene' is not an accurate description of transposons.
4. What is repetitive DNA?
5. Outline Barbara McClintock's contribution to our understanding of genetics.
6. List the main types of block mutations.
7. Describe hotspots.
8. The diagram shows a type of block mutation.

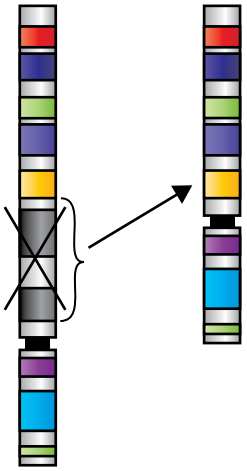


Figure 4.2 Type of block mutation.

Identify this type of block mutation and explain what has happened,

9. Construct a table to summarise the main types of block mutation – deletion, duplication, inversion and translocation explaining what is happening and giving a genetic example.
10. What are tandem repeats?
11. What is satellite DNA and how was it discovered?
12. Outline a use for microsatellite DNA.
13. What is a trinucleotide disorder and give an example.
14. The diagram shows an example of translocation.

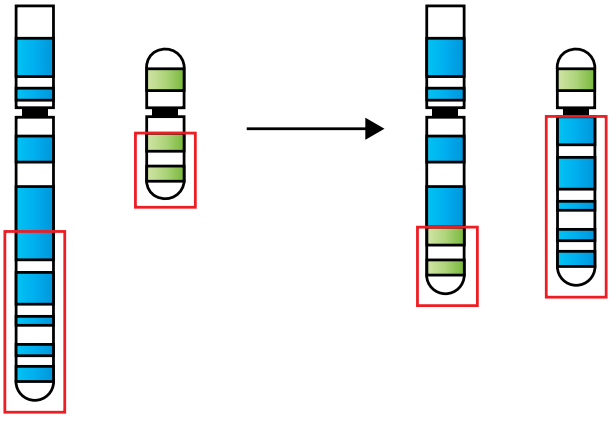


Figure 4.3 Translocation.

Explain why this is an example of reciprocal translocation.

15. If DNA duplications occur that involve one or more genes to make a gene pair and both copies stay in the genome and are inherited by future generations then a multigene family can be created.
    (a) Explain why the genes in a multigene family code for proteins with similar sequences.
    (b) Explain why the genes in a multigene are usually involved in the same body functioning.
    (c) Discuss why comparative genomics is interested in multigene families such as the globin genes for haemoglobin.

Use the following diagram for the next TWO questions.

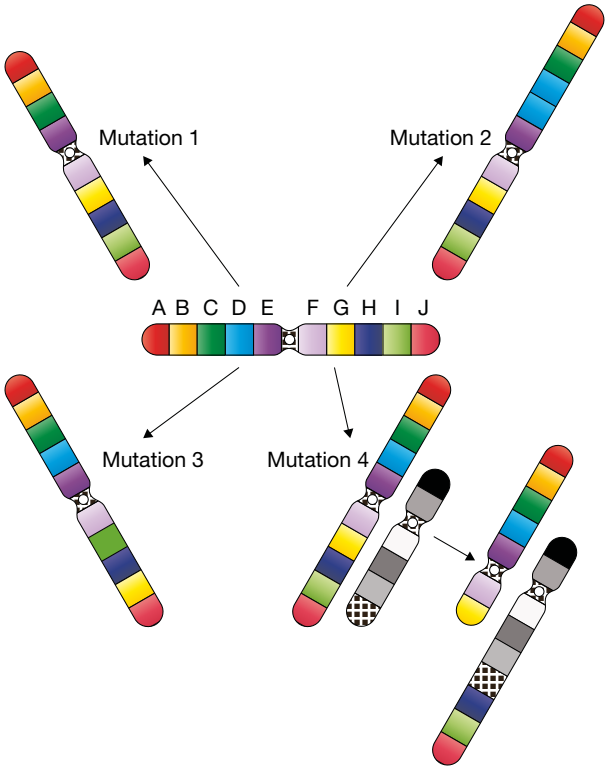


Figure 4.4 Mutation types.

16. Which mutation type shows inversion?
    (A) Mutation 1 (B) Mutation 2
    (C) Mutation 3 (D) Mutation 4
17. Which type of mutation would be involved in the creation of repetitive DNA?
    (A) Mutation 1 (B) Mutation 2
    (C) Mutation 3 (D) Mutation 4
18. The following code shows a section of a simple sequence DNA showing only one strand of the DNA.

    ..GCTTAGCTTAGCTTAGCTTAGCTTAGCTTA..

    How many nucleotides are in this repeat unit?
    (A) 3 (B) 5 (C) 15 (D) 30
19. Which disease can be caused by translocation?
    (A) Trisomy 21. (B) Measles.
    (C) Cri du chat. (D) Malaria.

5 Transposable Genetic Elements

Transposable genetic elements or transposons are DNA segments that can move from one position to another in chromosomes. The movement is called **transposition**. Transposons have been found in both prokaryotes and eukaryotes and were first found in plants. They can cause mutations and can increase/decrease the amount of DNA in the genome. In humans, jumping genes have been linked with antibody production by the immune system and some of the rearrangements caused by jumping genes are believed to be the cause of some forms of cancer. Some people call transposons 'junk' DNA or 'selfish' DNA as they do not seem to benefit the host and often just make copies of themselves.

When the enzyme transposase is activated a 'jumping gene' can move from one locus on one chromosome to another chromosome in a 'cut and paste' action. Retrotransposons also use a 'cut and paste' method but they copy RNA rather than DNA. About 40% of the human genome consists of retrotransposons. Some transposons use a 'copy and paste' method.

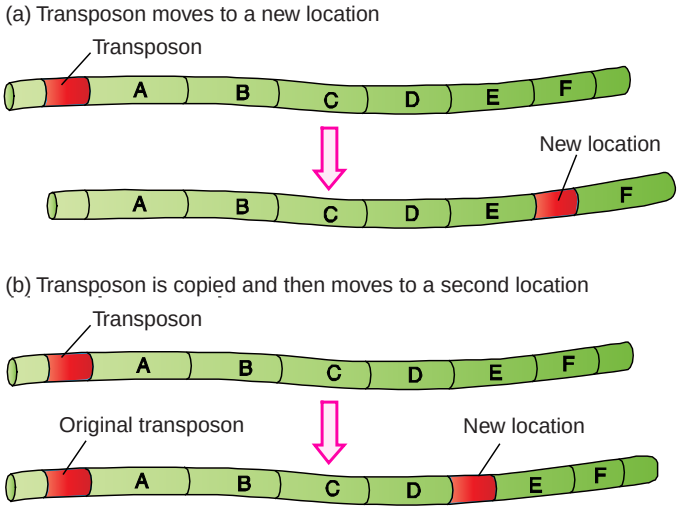


Figure 5.1 Transposons.

Transposons have been used to study gene function and to cause mutagenesis in *Drosophila melanogaster*. In the fruit fly the transposons are called P elements and they seem to have only appeared 50 years ago. In bacteria some transposons not only carry the gene for transposase but also one or more genes for proteins that give antibiotic resistance. The transposons can jump from the DNA chromosome to the plasmid and back again. It is believed these transposons have caused the rapid spread of multidrug antibiotic resistance in bacteria.

Transposons and mutation

Transposons are mutagens. If a transposon is inserted into a functional gene, it will most likely stop the function of that gene, e.g. if an L1 retrotransposon moves to a specific location on the X chromosome it causes the most common form of haemophilia. If the gap caused by the removal of a transposon is not correctly repaired, that site becomes a mutation. And if there are a string of inserts on a chromosome, identical homologous pairing becomes difficult in meiosis. Human diseases caused by transposons include haemophilia A and B, Duchenne muscular dystrophy, SCID and porphyria.

QUESTIONS

1. Define transposable genetic elements.
2. What is transposition?
3. Distinguish between a prokaryote and a eukaryote.
4. In which organisms have transposons been found?
5. In the 1940s Barbara McClintock discovered the first transposons in maize, *Zea mays* (called corn in USA). She found transposons were responsible for gene mutations caused by insertions, deletions and translocations. Transposons cause the different colours, e.g. purple, yellow, white and streaking on 'Indian corn'.

Figure 5.2 Indian corn.

Barbara McClintock received the Nobel Prize in Physiology or Medicine in 1983 for her work on corn genetics. Suggest one reason why her work was not recognised until 1983.

6. Outline how jumping genes affect humans.
7. HIV-1 is a retrovirus that causes AIDS. It acts like a retrotransposon. What is a retrotransposon?
8. Explain why transposons have been called by some people 'junk' DNA and 'selfish' DNA.
9. Transposons have been found in all branches of life. This has led to an interest in when transposons evolved and their effect on the evolution of genomes. Suggest a possible evolutionary history for transposons.
10. Construct a table to compare the effects of transposable genetic elements and germ line mutation on an organism.
11. What is a transposon?
    (A) DNA sequence that changes its position within the genome.
    (B) A permanent change in the genetic information.
    (C) The movement of information from one cell to another.
    (D) DNA sequence that is always expressed.

6 Chromosome Abnormalities

There are several types of chromosome abnormalities. Some individuals can have extra or missing chromosomes and some individuals have a different number of whole sets of chromosomes.

Polyploidy is the possession of more than two sets of chromosomes per nucleus, e.g. hexaploid (6*n*).

Monoploidy is the loss of an entire set of chromosomes. It is the haploid number of chromosomes.

Aneuploidy is the addition of all or part of a chromosome, e.g. in humans there could be 45 or 47 chromosomes instead of the normal 46 chromosomes in the nucleus.

Monosomy is the lack of one chromosome from the normal number of chromosomes.

Trisomy means there are three copies instead of the normal two copies of a particular chromosome.

Nondisjunction

The change in the number of chromosomes can be due to nondisjunction during cell division. Nondisjunction is an error during mitosis or meiosis when both members of a pair of homologous chromosomes or both sister chromatids fail to separate properly.

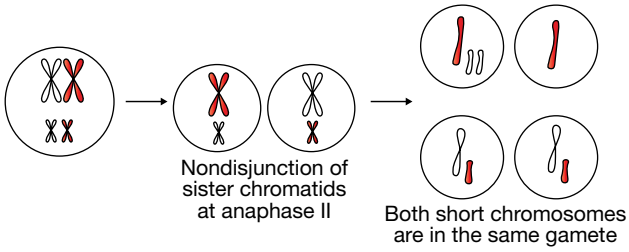


Figure 6.1 Nondisjunction in anaphase II in meiosis.

Nondisjunction leads to an abnormal number of chromosomes in a gamete. If it occurs in anaphase II there will be two affected daughter cells – one with two copies of the chromosome and the other with no copies of that chromosome.

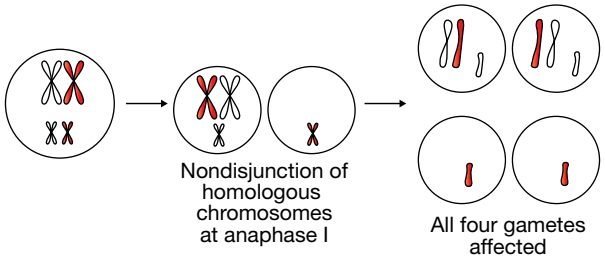


Figure 6.2 Nondisjunction in anaphase I in meiosis.

If the homologous chromosomes do not separate during anaphase I then all four daughter cells are affected – two daughter cells will have an additional chromosome and two cells will be missing one chromosome.

During fertilisation the fusion of an abnormal gamete with a normal gamete will create a zygote with an incorrect number of chromosomes, e.g. missing a chromosome or having an additional chromosome. Many cases of trisomy 21 (Down syndrome) are due to nondisjunction.

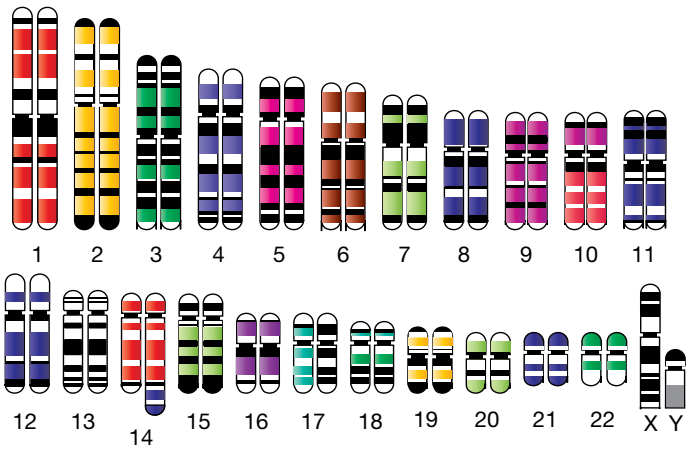


Figure 6.3 Trisomy 21 with translocation to chromosome 14.

Nondisjunction can also occur during mitosis. If the error occurs during early embryonic development then a large number of cells will have the aneuploidy condition. In mosaic Down syndrome the person has some cells with trisomy 21 and some cells with the normal 46 chromosomes in a cell.

Unequal crossing over in meiosis

Chromosome abnormalities can occur when crossing over between adjacent non-sister chromatids in the tetrad in prophase I if meiosis is unequal. In the resulting gametes one cell may have a chromosome with a gene deletion and another cell will have a chromosome with a gene duplication. Transposable elements in the chromosome provides the sites for crossing over in non-sister chromatids.

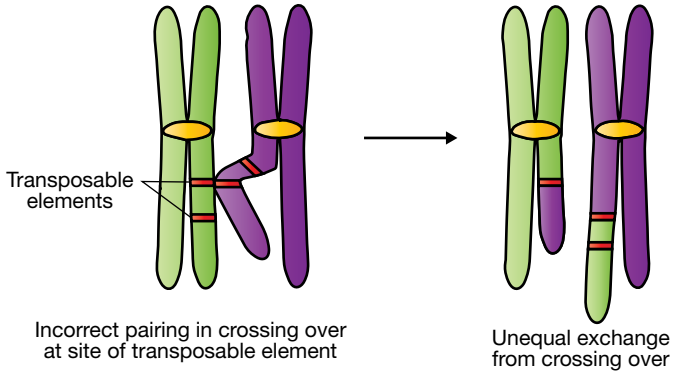


Figure 6.4 Unequal crossing over.

Sex chromosomes and chromosome abnormalities

There are several genetic disorders that occur due to an incorrect number of sex chromosomes. A changed number of sex chromosomes does not seem to upset the genetic balance as much as an aneuploidy autosome condition. The Y chromosome has few genes and in somatic cells the extra copies of the X chromosome are inactivated as Barr bodies.

- **Klinefelter syndrome** is a chromosomal disorder that occurs when males have two or more X chromosomes, e.g. a male has XXY. It is a congenital disorder detected by karyotyping. Males have abnormally small testes and the man is sterile. It is not an inherited disorder and occurs in about 1 in 500 men to 1 in 1000 men in Australia with many not being diagnosed.
- **Turner syndrome** (TS) is a chromosomal disorder that occurs when females are missing part or a whole X chromosome, e.g. a female has XO. It is the only known viable monosomy in humans. The female will have 45 chromosomes instead of 46 chromosomes. The person may show mosaicism with some cells normal and some cells lacking the second X chromosome. Mosaic Turner syndrome occurs in about 20% of cases. Turner syndrome affects about 1 in every 2000 girls born in Australia with about 50% having XO in all their cells. They are sterile as the sex organs do not mature. Oestrogen therapy can aid the development of secondary sex characteristics.
- **XYY syndrome** has an extra Y chromosome in each cell in a human male. Individuals seem to have an increased risk for learning difficulties, often have heavy acne and have increased height. It is estimated to occur in about 1 in 1000 live births. The males have normal fertility.
- **Triple X syndrome** (XXX) has an X chromosome in each cell in a human female with a mosaic form being present in only some cells. Most cases are not diagnosed and since only one X chromosome is only activated at a time due to the formation of Barr bodies, there are few obvious signs of the condition and most are usually fertile.

Cancer and chromosome abnormalities

Some translocations are associated with certain cancers. In chronic myelogenous leukaemia (CML) there is a reciprocal translocation between chromosome 22 and chromosome 9 with a large amount of chromosome 22 exchanging for a small tip of chromosome 9 – this forms the Philadelphia (Ph) chromosome. This rare cancer causes the overproduction of granulocytes.

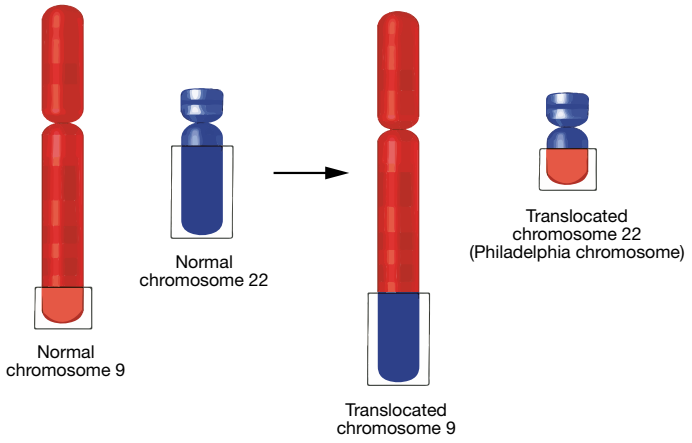


Figure 6.5 Translocation forming the shortened chromosome 22 known as the Philadelphia chromosome.

QUESTIONS

1. Distinguish between polyploidy, monoploidy and aneuploidy.
2. Distinguish between monosomy and trisomy.
3. Define nondisjunction.
4. Explain how nondisjunction leads to abnormal cells.
5. Explain how mosaic trisomy conditions arise.
6. Suggest why a number of anomalies involving the sex chromosomes are in the human population and do not have as great a genetic effect as autosome abnormalities.
7. Construct a table to summarise some genetic disorders caused by abnormalities involving the sex chromosomes.
8. What is the only known viable monosomy in humans?
   (A) Klinefelter syndrome.
   (B) Turner syndrome.
   (C) Down syndrome.
   (D) Edwards syndrome.
9. What will cause cell division to form four daughter cells that are all affected with chromosome abnormalities?
   (A) Nondisjunction in anaphase I of homologues in the tetrad.
   (B) Nondisjunction in anaphase II of sister chromatids.
   (C) Nondisjunction in anaphase I of sister chromatids.
   (D) Nondisjunction in anaphase II of homologues in the tetrad.
10. What is the best term to describe chromosomal abnormalities where there are either extra or missing copies of certain chromosomes?
    (A) Polyploidy.
    (B) Monoploidy.
    (C) Euploidy.
    (D) Aneuploidy.

7 Polyploidy

Ploidy refers to the number of basic sets of chromosomes, e.g. diploid (2*n*) has two sets and hexaploid (6*n*) has six sets.

Polyploidy is the possession of more than two sets of chromosomes per nucleus, e.g. triploid (3*n*), tetraploid (4*n*). In most cases plants are more tolerant of extra chromosome sets than animals. Polyploidy is lethal in humans and fairly rare in the animal kingdom with some insects, fish, amphibians and reptiles being polyploid. In 1999 a tetraploid rat was reported in Argentina.

Autopolyploidy is the spontaneous doubling of the chromosomes, e.g. during meiosis the spindle fails to form properly and does not involve another species.

Allopolyploidy results from two different species interbreeding and combining their chromosomes. The hybrid has two or more sets of chromosomes from each of two different species. Some allopolyploids are fertile producing viable gametes as the homologous pairs can pair during meiosis. Some allopolyploids can self-fertilise but they cannot interbreed with either parent line due to the different number of chromosomes to both parents.

When allopolyploidy occurs the new hybrids can be less competitive than the parent lines and die out, they can compete with the parent lines leading to extinction of one or both parent lines or they can coexist with both parent lines.

Palaeopolyploidy is a genome duplication that occurred at least several million years ago. Rice is a palaeopolyploid as it is believed the main duplication events occurred around 40 to 50 million years ago.

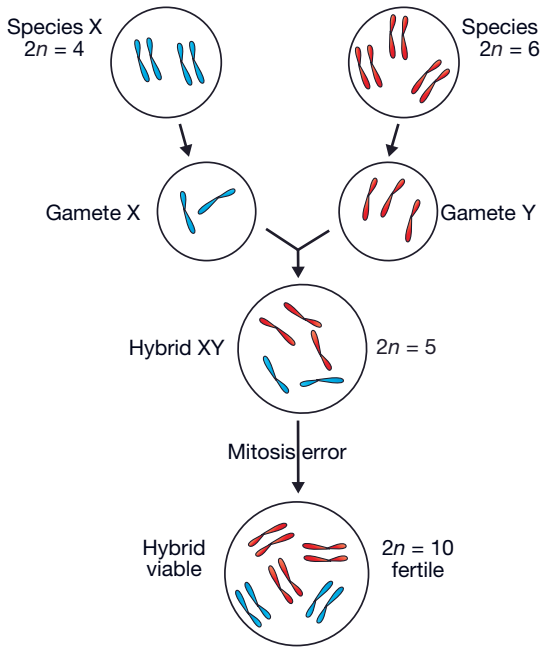


Figure 7.1 Allopolyploidy.

Cause of polyploidy

Multiple sets of chromosomes can occur in several ways.

- Failure of chromosome separation during meiosis.
- Chromosomes divide in mitosis but the cell does not.
- Fertilisation of an egg by more than one sperm.
- The genome of a single species duplicates during the cell cycle.
- The genomes of two species can combine in hybridisation.
- If polyploidy occurs in a sterile hybrid then fertility can be restored as the duplicate chromosomes can pair in meiosis.
- Researchers have found that the use of the chemical colchicine readily induces polyploidy. Colchicine inhibits microtubule polymerisation which is needed in mitosis and colchicine thus acts as a 'spindle poison'. This was first used in the 1920s to artificially cross a radish and a cabbage though it unfortunately had the root of the cabbage and the shoot of the radish.

Occurrence of polyploidy

Polyploidy is common in plants, especially flowering plants with 30% to 70% of today's flowering plants believed to be polyploid.

Allopolyploidy forms a new species in one generation and can be used to account for the rapid evolution of some flowering plants. Once polyploidy has occurred the new species can continue to thrive due to natural selection and it is also used in artificial selection when humans wish to grow plants with a combination of specific features, e.g. apple varieties and potato varieties.

Polyploid plants have several features that increase their chances of survival, e.g. they have larger cells, have larger nuclei, are larger in size, are more vigorous, have larger reproductive organs and fruit and are able to self-fertilise. Larger reproductive organs and fruit are favoured in selective breeding but are not necessarily an advantage in natural ecosystems. Thus selective breeding has artificially created many crop plants and garden flowers.

Due to the high chromosome number polyploid plants also have more possible combinations and therefore show greater variability and range.

Bread wheat

Humans have been selecting wheat varieties to grow since the beginning of the agricultural revolution and modern researchers are trying to breed 'super' varieties with higher yields and specific features such as disease, drought and/or frost resistance.

Modern bread wheat (*Triticum aestivum*) has been created in two separate hybridisations from three different diploid species that each had 14 chromosomes. Bread wheat is hexaploid (6×) with 42 chromosomes ($2n = 42$) that forms 21 pairs of chromosomes ($n = 21$) during meiosis.

One of the original forms of wheat was Einkorn wheat (*Triticum monococcum* $2n = 14$) which was crossed with a wild wheat ($2n = 14$) to form a tetrapod wheat ($2n = 28$), e.g. *Triticum turgidum*. The second hybridisation crossed the tetrapod wheat with a wild wheat species ($2n = 14$) forming modern wheat (*Triticum aestivum* $2n = 42$).

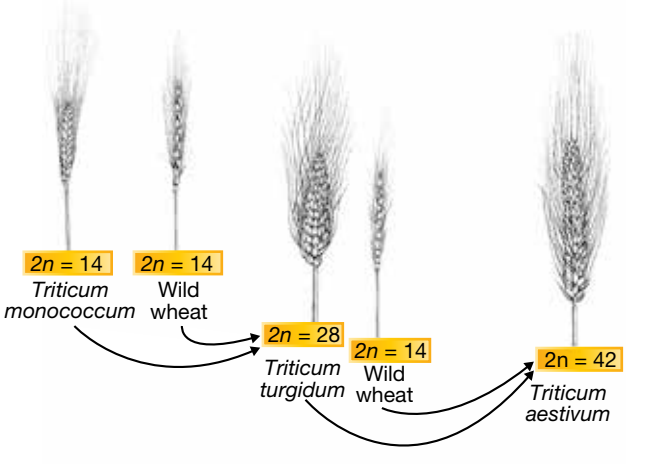


Figure 7.2 Bread wheat is hexaploid.

In Australia wheat breeding is an important aspect of the agricultural industry with techniques such as backcrossing of varieties in the 1940s being introduced to improve yields and improve disease resistance, e.g. against rust. In the 1960s dwarf breeding materials were introduced.

Breeding programs have led to a large range of wheat varieties that are used for specific purposes, e.g. *T. durum* is used to make semolina flour for pasta and Australian prime hard wheat, a high protein milling wheat, is used to make Chinese style yellow alkaline noodles and Japanese ramen noodles. The Victorian State Government and the Australian Government Grains Research and Development Corporation (GRDC) publish Crop Variety Sowing Guides, Crop Summaries and results of National Variety Trials (NVT) that can be used for advice about the best variety to plant and which current crops are grown in Australia.

QUESTIONS

1. Define polyploidy.
2. Define palaeopolyploid.
3. Distinguish between allopolyploidy and autoploidy.
4. Outline how polyploidy can occur.
5. Explain why genetic researchers use the chemical colchicine.
6. Rice is a palaeopolyploid with the main duplication events occurring around 40 to 50 mya and there are currently about 23 species of the rice genus *Oryza* worldwide. Explain why genome sequencing will help our understanding of the genetic history of rice.
7. In 1928 Georgii Karpechenko published in English his findings of a new species he named *Raphanobrassica* from a cross with a cabbage (genus *Brassica*) and a radish (genus *Raphanus*). He wanted to create a 'vegetable of the Proletariat'.
   (a) What type of mutation had Karpechenko created?
   (b) Why was Karpechenko's work both a success and a failure?
8. The diagram shows a type of chromosome abnormality.

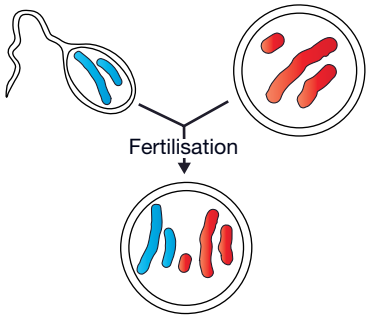


Figure 7.3 Chromosome abnormality.

   (a) Explain what is happening in this situation.
   (b) Would you expect this offspring to be fertile?
9. Australian hard wheat is used to make dough for a range of baked products, e.g. Middle Eastern flat bread and European style pan and hearth breads. This wheat is grown in north-western Victoria.
   (a) How did people create the modern bread wheat *Triticum aestivum*?
   (b) Explain why wheat is polyploid.

Use the diagram for the next TWO questions.

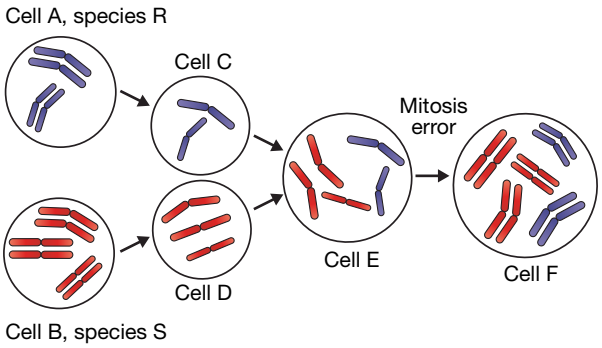


Figure 7.4 Chromosome abnormality.

10. What type of mutation is shown in this diagram?
    (A) Autopolyploidy. (B) Monoploidy.
    (C) Allopolyploidy. (D) Aneuploidy.
11. What is the diploid number of cell A?
    (A) 2 (B) 3 (C) 4 (D) 5

8 Gene Flow

Gene flow occurs when alleles are added to a gene pool or subtracted from the gene pool due to the movement of fertile individuals or gametes, e.g. due to migration of individuals from one population to another. Gene flow may involve natural selection or can occur due to chance.

Gene flow will change allele frequencies and in many instances will reduce the number of differences between populations. The higher the gene flow between two populations the less likely the two populations will evolve into two species. For example, pollen released from plants can be blown by the wind over distances to other populations where they fertilise plants introducing new alleles into that population. If a particular allele did not previously exist in a population then its introduction can have a dramatic effect on natural selection.

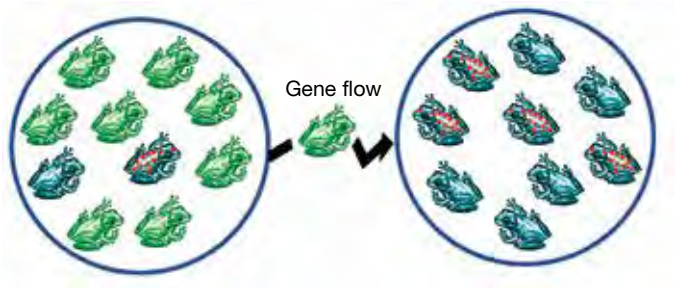


Figure 8.1 Gene flow.

Gene flow does not always require individuals to permanently migrate. For example, during the Vietnam war some US soldiers had children with Vietnamese women which altered the allele frequency of the Vietnamese gene pool.

Factors affecting gene flow

There are several factors that can affect the rate of gene flow between different populations:

- Mobility of fertile individuals or gametes – high mobility increases the chances of gene flow from one population to another.
- Presence of barriers, e.g. mountain ranges, oceans, deserts.
- Behavioural barriers, e.g. different courtship rituals in adjacent populations or the time of reproduction.

Gene flow and natural selection

Gene flow often reduces the number of differences between populations with the higher the gene flow between two populations the fewer the differences and the less likelihood that the two populations will evolve into two species, e.g. pollen released from plants can be blown by the wind over distances to other populations where they fertilise plants introducing new alleles into that population and differences between the populations decrease.

A change in the allele frequencies in a population's gene pool over a number of generations means evolution is occurring. Evolution is a change in the genetic make-up of a population. Scientists are interested in changes in allele frequencies as it provides information about how populations with different size gene pools cope with changing conditions or how populations can change due to genetic drift or gene flow over time.

The size of the gene pool is most important when considering the survival chances of a population in a changing environment. A small gene pool means there is little genetic diversity and if conditions alter and there are no variants with features suited to the new conditions the population becomes endangered and may face extinction. A large gene pool makes a population more robust and able to withstand changing conditions and natural selection.

Barriers to gene flow

Geographical isolation – If two populations occupy different geographical areas or different habitats within the same area, they may not meet and cannot mate to produce offspring. Primary geographical barriers occur with water or mountain ranges separating two sections of a population. Frequently populations extend over a large distance and over time the groups at either end are too far apart for direct interbreeding. A barrier for one species may not be a barrier for another species, e.g. a desert is a barrier to a species that requires a moist environment but not a barrier to a desert species. For example, there are seven species of Australasian treecreepers of family Climacteridae. The white-throated treecreeper *Cormobates leucophaea* is the most common and found on the east coast from Queensland to Victoria. Treecreepers live in forests and woodlands with some species separated by deserts. The distribution of the brown treecreeper, *Climacteris picumnus* overlaps with *C. leucophaea*, yet the brown treecreeper is most closely related to the rufous treecreeper *Climacteris rufus* of Western Australia.

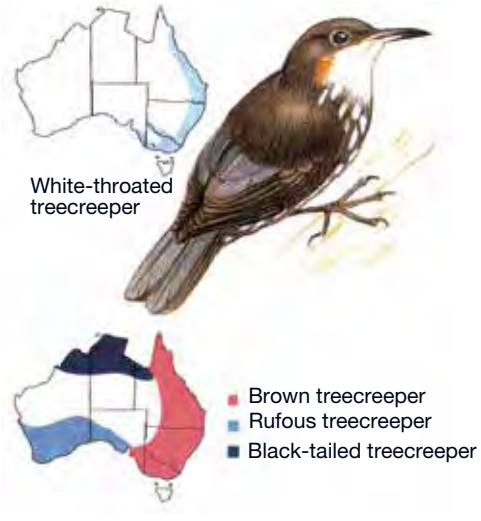


Figure 8.2 White-throated treecreeper.

Hybrid infertility – Different parent species may be able to breed but the hybrid offspring can be infertile. If the parent genotypes have different diploid numbers or the chromosomes vary in structure, the hybrid can be vigorous but unable to produce normal gametes. For example, a female horse ($2n = 64$) is crossed with a male donkey ($2n = 62$) to make a mule ($2n = 63$). The mule is stronger, has hybrid vigour, is longer lived with a much longer working life and is able to survive harsher conditions. However, the mule is sterile as chromosomes cannot correctly pair during meiosis.

Temporal isolation – Temporal isolation refers to species that breed during different times of the day, different seasons or different years. This means that their gametes cannot mix. For example, *Pinus radiata* and *P. muricata* are two species of pine found in California. *P. radiata* releases pollen in early February while *P. muricata* sheds its pollen in April. This means that interbreeding is unlikely to occur between the two pines even though they are found in the same geographical range.

Behavioural isolation – Behavioural isolation means that courtship rituals that attract mates and other behaviours specific to each population produce an effective barrier that prevents mating even between closely related species. Signals can be visual and/or auditory and some species, e.g. insects release pheromones for species identification. For example, several different dabbling ducks live in the same region of eastern USA. The ducks have an elaborate courtship display and the male has distinctive plumage. Both the colourful patterns and behaviour of the males helps the female choose a mate from the correct species, as a mistake results in a hybrid which is less viable.

Artificial insemination and gene flow

Artificial insemination is the injection of male semen into the vagina or cervix of a female without sexual intercourse. Sperm is collected from a stud male, e.g. a champion sheep, bull or horse and then usually the sperm are frozen. Frozen sperm can be stored indefinitely or can be sent anywhere in the world. This means that a male with especially desirable features may provide sperm for many females, and father many offspring in many countries without leaving his home paddock. Artificial insemination has increased gene flow for certain alleles in some species and has not involved migration.

When using artificial insemination it is important to maintain detailed pedigrees of the animals as less favourable genes can also be passed on at the same time and it is important to ensure closely related animals are not crossbred. Overuse of the sperm from one particular breeding line can reduce the genetic diversity, causing problems if recessive characteristics and genetic diseases show up more frequently in the phenotype. Most sheep, cattle and pigs are bred by artificial insemination. Desired features include cattle with more beef or higher milk production, or milk with higher butterfat, or sheep with finer wool or pigs with more lean meat.

Artificial pollination and gene flow

Artificial pollination involves humans taking the pollen from one plant that has certain desired features and placing it on the stigma of another flower. Mendel used artificial pollination in his pea plant experiments and plant geneticists have used the technique to produce a wide range of fruits, vegetables and cereal crops. Humans using artificial pollination have altered allele frequencies in many plant populations.

Desired features can include higher yield, larger fruit, or disease resistance. However, if there is overuse of one strain in a large monoculture, entire areas can become susceptible to a specific pest. For example, the Irish potato famine of 1845 to 1851 was caused by a fungal disease and one million people died due to the potato crop failure.

QUESTIONS

1. Define gene flow.
2. Outline how migration can cause gene flow.
3. Explain why gene flow can reduce the number of differences between populations.
4. Outline some factors that can affect gene flow.
5. Construct a table to summarise how different types of barriers restrict gene flow.
6. The Kaibab squirrel is found on the north side of the Grand Canyon and Abert's squirrel is found on the south side of the Grand Canyon, USA. They are closely related and have a common ancestor. Explain why there is restricted gene flow between these two populations of squirrel.
7. Discuss the advantages and disadvantages of artificial insemination.
8. Describe some examples to show how artificial insemination has been used to change breeding stock.
9. Define artificial pollination.
10. Discuss the advantages and disadvantages of artificial pollination.
11. If all farmers used the same variety of seed produced by artificial pollination, what would happen to the genetic composition of the population over time?
    (A) Diversity increases. (B) Diversity decreases.
    (C) Numbers increase. (D) No change.
12. Artificial insemination has been useful in agriculture. Which of the following shows an example of a use of artificial insemination in cattle?
    (A) Produce identical offspring the same as the parent.
    (B) Overcome infertility in females.
    (C) Create new alleles which give predetermined characteristics.
    (D) Breed offspring when the male is in one country and the female in another.

Answers

1 Assumed Knowledge

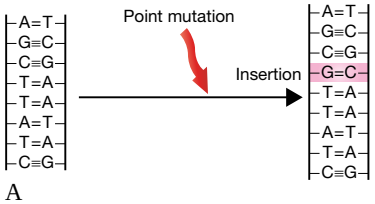
1. (a) Chromosomes are long strands of hereditary information containing genes. Chromosomes are made up of DNA and histone proteins.
   (b) Each gene is found at a particular locus on a chromosome.
2. A gene is a section of DNA coding for proteins that expresses itself as the phenotype for that trait, e.g. gene for plant height whereas an allele is an alternative for a particular trait, e.g. there are two alleles for height in pea plants – tall (T) or short (t). Alleles are the alternative forms of a gene and occupy a particular locus on a chromosome.
3. Autosomes are chromosomes that are not directly involved in determining sex whereas sex chromosomes are responsible for determining the sex of an individual. In humans there are two sex chromosomes, X and Y. Males are XY and females are XX.
4. (a) Person A is female as they have XX.
   (b) Trisomy is the presence of three copies of a homologous chromosome rather than the normal two copies.
   (c) Person A has trisomy 21 with three copies of chromosome 21 – this is Down syndrome.
5. A genome sequence is the order of the As, Ts, Cs and Gs in the DNA code.
6. A gene pool is the total aggregate of genes in a population at any one time.
7. The frequency of an allele in a population refers to the proportion of the population that have that allele.
8. Polyploidy occurs when cells contain more than two haploid (*n*) sets of chromosomes, e.g. triploid (3*n*), tetraploid (4*n*).
9. A mutation is a permanent change in the genetic information. This causes genetic diversity.
10. The genotype shows the genetic make-up or set of alleles of an organism that control a characteristic whereas phenotype is the observable physical and physiological traits of an organism and the outward appearance of an organism. A dominant phenotype can have two dominant alleles, e.g. TT or one dominant allele and one recessive allele, e.g. Tt. The recessive phenotype has two recessive alleles, e.g. tt.
11. Humans are classified as a primate as they have – 1. An opposable thumb. 2. Shoulder joint with high rotating ability. 3. Forward facing eyes with stereoscopic vision. 4. Reduced snout and olfactory centre of the brain. 5. Enlarged skull with large cerebrum. 6. Five digits on limbs. 7. Nails (not claws). 8. Bicuspid teeth.
12. Comparative genomics studies compares genomic features of different organisms, e.g. looking for differences and similarities in DNA sequence, genes, gene order and regulatory sequences.
13. Biodiversity refers to the amount of variation within the group.
14. A fossil is a remain or trace of a pre-existing organism.
15. A species is a group of organisms that can interbreed to produce fertile offspring. They share a common gene pool.
16. Most DNA in eukaryote cells is in the nucleus. DNA is also found in mitochondria and chloroplasts.
17. Diagram shows DNA replication.
18. An index fossil is a distinctive organism that lived for a short time and is found over a wide area.
19. The law of superposition states that the oldest layers are on the bottom and the youngest layers are on top, unless there has been folding or faulting or another form of dynamic Earth movement.
20. Relative dating uses the law of superposition to determine if something is 'older than' or 'younger than' whereas absolute dating gives a date in years with the experimental error of the method used in determining the date.
21. Trilobite D (Redlichids) are suitable as an index fossil as they only existed for a short time and are indicative of the Cambrian period. However, trilobite H was present in the Cambrian through to the Permian which is a very long time frame and is thus not particularly useful as an index fossil – it simply indicates the Palaeozoic era.
22. Natural selection is a process that leads to a change in a population over time due to some phenotypes having more success surviving and reproducing in particular environmental conditions.
23. An example of fossil evidence that shows present-day organisms have developed from different organisms in the past is the fossil history of the horse. The modern day horse *Equus* has evolved from *Hyracotherium* which was much smaller, had more toes, and smaller teeth and lived around 60 million years ago.
24. Evolution occurs when natural selection causes changes in relative frequencies of alleles in the gene pool.
25. The order of evolution of living things was organic molecules, membranes, prokaryotic heterotrophic cells, prokaryotic autotrophic cells, eukaryotic cells, colonial organisms and multicellular organisms.

2 Allele Frequency

1. An allele is an alternative for a particular trait, e.g. there are two alleles for height in pea plants – tall (T) or short (t). Alleles are the alternative forms of a gene and occupy a particular locus on a chromosome.
2. The frequency of an allele in a population refers to the proportion of the population that have that allele.
3. Population genetics studies how populations change genetically over time and the Hardy-Weinberg equilibrium proposes that frequencies of alleles and genotypes remain constant from generation to generation provided that only Mendelian segregation and recombination of alleles occur. Thus the Hardy-Weinberg equilibrium is only useful in population genetics if there is no evolution occurring in the population.
4. A gene pool is the total aggregate of genes in a population at any one time.
5. A population is a group of organisms which can freely interbreed to produce fertile offspring living in a particular area at a particular time.
6. A genome is the complete complement of an organism's genes and genomics studies whole sets of genes and their interactions.
7. Genes are coded DNA which is transcribed into RNA that is used in protein formation. Controlling proteins means that when differentiation occurs the final cell structure and functioning is controlled by which genes are switched on or off in gene expression, e.g. the switching on of genes that control specific proteins such as keratin and collagen and the switching on of particular enzymes that regulate specific reactions will determine the shape, composition and functioning ability of the cell.
8. Phenotype is the observable physical and physiological traits of an organism and the outward appearance of an organism. Allele frequency of particular traits will then show the frequency of particular phenotypes in a population.
9. Phenotype is determine by the alleles present (genotype) and environmental influences.
10. C

3 Point Mutations

1. A mutation is a permanent change in the genetic information and is a cause in genetic diversity.
2. A gene mutation is a permanent change in the genetic information in a gene. The mutation can involve one or more base pairs and can be anywhere in the gene.
3. A point mutation is a change in one base in a single nucleotide in a gene.
4. There are four basic types of point mutation – base pair substitution, insertion, deletion and inversion.
5. A silent mutation is a change that has no effect on the protein being produced.
6. (a) GUU (valine) to GUG (valine) is a silent mutation.
   (b) ACG (threonine – polar amino acid) to AAG (lysine – electrically charged amino acid) is not likely to be a silent mutation.
   (c) GGU (glycine a non-polar amino acid) to GAU (aspartic acid – electrically charged amino acid) is not likely to be a silent mutation.
   (d) CGG (arginine is an electrically charged amino acid) to UGG (tryptophan a non-polar amino acid) is not likely to be a silent mutation.
   (e) GCU (alanine) to GCC (alanine) is a silent mutation.
7. A missense mutation is a changed codon that codes for an amino acid but does not necessarily make the correct sense whereas a nonsense mutation is a point mutation that changes a codon for an amino acid into a stop codon. The stop codon causes translation to stop shortening the polypeptide chain that is being synthesised.

8. A frameshift mutation occurs when a deletion or insertion causes an incorrect reading of the nucleotide sequence. The sequence is read in multiples of three (the code for each amino acid) so the deletion or insertion of one nucleotide will change the reading sequence but it is not affected if the change was an insertion or deletion of three nucleotides.
9. If the point mutation affects the shape of the protein or is at the active site of an enzyme, the change can seriously affect the functioning of the protein. For example, disulfide bridges are formed when two cysteine amino acids are brought close together with their sulfhydryl groups (–SH) on their side chains in close proximity due to the folding of the protein and a change in the code from UGC (cysteine a polar amino) to UUC (phenylalanine a non-polar amino acid) can affect the structure and functioning of the protein. Disulfide bridges are important in maintaining the structure of immunoglobulins (antibodies) and the antigen receptor sites on lymphocytes. Inability to form functional antibodies will reduce the effectiveness of the immune system.
10. The mRNA stop codes are UAA, UAG and UGA.
11. 

12. A
13. D
14. B
15. C

4 Block Mutations

1. A block mutation is a permanent change to a segment of a chromosome that rearranges, deletes or disrupts many loci.
2. Transposons (transposable genetic elements) are DNA segments that can move from one position to another in the chromosome.
3. The term 'jumping genes' is not accurate as it does not explain how transposons move as they do not 'jump'. When the folding of the DNA molecule brings segments near each other transposons follow a 'cut and paste' mechanism to move to a new location or follow a 'copy and paste' mechanism replicating a section of DNA and adding it to another area.
4. Repetitive DNA is multiple copies of DNA sections which are found in the genome.
5. Barbara McClintock discovered transposons when studying corn in 1940s and she was awarded the Nobel Prize in Physiology or Medicine in 1983 for her discovery of 'mobile genetic elements'.
6. Block mutations can be duplications, inversions, deletions, insertions or translocations.
7. Hotspots are places that are more likely to undergo mutation than other places with an observable higher mutation frequency. Hotspots can be single nucleotides or short stretches of repeated nucleotides that have some basic instability or chemical tendency for nucleotide substitution.
8. The diagram shows a block deletion. In this type of mutation a section of DNA is removed from the sequence in a chromosome and in this case several genes have been removed near the centromere.
9.

Change	What happens	Example
Deletion	A section of DNA is removed altering chromosome structure.	Cri du chat syndrome involves a deletion of the end of the short arm of chromosome 5.
Duplication	A section of DNA is repeated and added to the chromosome.	Charcot-Marie-Tooth disease type 1 with duplication of 17p12 – a large section on the short arm of chromosome 17.
Inversion	A section of DNA breaks and is reattached in the reverse orientation and order.	In factor VIII gene on X chromosome causing haemophilia A.
Translocation	A section of one chromosome moves to a non-homologous chromosome. In a reciprocal translocation the non-homologous chromosomes exchange segments.	Myeloproliferative syndrome caused by translocation of genetic material from chromosome 8 to other chromosomes, e.g. t(8;13)(p11;q12).

10. Tandem repeats are copies of a repeat sequence that lie adjacent to each other
11. Satellite DNA or simple sequence DNA is a section of tandem, non-coding DNA that can be thousands of base pairs long and was discovered when DNA was centrifuged and the repetitive units appeared as a distinct band in the tube.
12. Microsatellite DNA is a short region of repeats that are used as genetic markers in DNA fingerprinting. In humans some microsatellites have 20 or more alleles which provides the variation to assist in identifying particular individuals by their DNA.
13. Trinucleotide disorders occur when there are too many trinucleotide repeats in a gene, e.g. Huntington's disease occurs when there are more than 35 CAG repeats on the gene coding for the protein HTT.
14. The diagram shows reciprocal translocation as the exchange is between non-homologous chromosomes (they are different lengths and have different genes) with blocks of DNA being swapped and two new different chromosomes are formed with different sets of genes.
15. (a) Multigene families arise after duplication of one or more genes to make a gene pair and both copies stay in the genome and are inherited by future generations. This means that the genes in the multigene family started with genes that coded for particular proteins which started the multigene family.
    (b) Sine the original genes that were duplicated and started the multigene family were involved in a particular body function, the future forms of the genes in the multigene family are most likely to be involved in the same process.
    (c) Comparative genomics studies compares genomic features of different organisms, e.g. looking for differences and similarities in DNA sequence, genes, gene order and regulatory sequences. Researchers are interested in multigene families such as the globin genes for haemoglobin as differences and similarities in the DNA sequences found in the genomes of different species can provide information about evolution and the interrelatedness of the species.
16. C
17. B
18. B
19. A

5 Transposable Genetic Elements

1. Transposable genetic elements are DNA segments that can move from one position to another in the chromosome.
2. Transposition is the movement of a transposon.
3. A prokaryote does not have membrane bound organelles, e.g. nucleus, while a eukaryote does have membrane bound organelles, e.g. has a nucleus.
4. Transposons have been found in prokaryotes and eukaryotes, being first found in plants.
5. The work of Barbara McClintock was not recognised until 1983 as the discovery of 'jumping genes' in the 1940s was quite ahead of its time. Watson and Crick did not propose the double helix structure of DNA until 1953 and thus the idea of genes moving location and changing the Mendelian laws of inheritance would have been unexplainable at that stage of genetic understanding. With the advent of genetic engineering her work could be fully recognised.
6. In humans, jumping genes have been linked with antibody production by the immune system and some of the rearrangements caused by jumping genes are believed to be the cause of some forms of cancer.
7. Retrotransposons use a 'cut and paste' method to move genetic information from one location on a chromosome to another but they copy RNA rather than DNA.
8. Transposons have been called 'junk' DNA as there is no clear indication how they benefit the host; and they have been called 'selfish' DNA as they often make multiple copies of themselves.
9. Since transposons have been found in all life forms, transposons may have been present in the last universal common ancestor. Or transposons may have evolved independently several times.