9. *Weakened strains (attenuated) of bacteria or viruses, neutralised toxins, (toxoids) or a genetically engineered vaccine.*

10.

(a) *No, immunisation is not compulsory although availability of welfare benefits may be linked to child immunisation.*

(b) *Measles, mumps, chicken pox, tetanus, pertussis (whooping cough), diphtheria, rubella, polio, hepatitis A or B, cholera, typhoid, human papilloma virus, influenza, Hib, tuberculosis, meningococcal infection.*

(c) *HIV (AIDS), dengue fever, common cold, malaria.*

(d) *Streptococcus pneumoniae is a bacterium which may cause ear infections, pneumonia and meningitis.*
Hib (Haemophilus influenzae type B) is a bacterium which may cause infection of the meninges (membranes which enclose the brain and spinal cord), the upper respiratory tract and other parts of the body.
Meningococcal C is a strain of bacterium (Neisseria meningitidis) which causes a disease which develops rapidly and may cause brain damage and death. The bacterium can infect the meninges or can be carried to major organs, joints and connective tissue. It causes serious damage to these body parts. Symptoms include high fever, headaches, sleepiness, joint and muscle pain, stiff neck, light sensitivity and rash.

6: MUTATIONS

Terminology

(i) *allele – the alternative form of a gene.*

(ii) *autosomal chromosome (autosome) – a chromosome which is not a sex chromosome. A normal human somatic cell contains 22 pairs (44) of autosomes.*

(iii) *cancer – the uncontrolled growth of cells, often able to spread (metastase) via the circulatory or lymphatic system to invade other tissue.*

(iv) *centromere – the point of attachment of two chromatids. The spindle fibres attach to the centromere during cell division.*

(v) *chromosome – a strand of DNA that carries the genetic information of an organism.*

(vi) *deleterious – having a harmful effect, e.g. smoking tobacco.*

(vii) *DNA replication – the process in which DNA copies itself prior to cell division. This ensures that the cells produced all contain the same genetic information.*

(viii) *gene – a segment of DNA that codes, or determines a particular trait or characteristic.*

(ix) *genotype – the genetic make-up of an organism for a particular trait, e.g. rr or Rr*

(x) *germ line cell – a cell that gives rise to gametes (sperm or ova). If a mutation occurs in a germ cell then it may be inherited by a person in the next generation.*

(xi) *karyotype – is a picture showing the total number and general appearance of chromosomes within the nucleus of a cell. The chromosomes have been stained, matched up for size and shape and organised in pairs in order from the biggest to the smallest (1 to 22 in humans).*

(xii) *meiosis – cell division process that produces gametes (sperm or ova). The normal diploid number of chromosomes is reduced to the haploid number in this process, e.g. in human the diploid number of 46 chromosomes per somatic cell is reduced to 23 chromosomes in each gamete.*

(xiii) *metaphase – the stage in cell division (mitosis or meiosis) in which chromosomes are lined up along the 'equator' of a cell.*

(xiv) *missense mutation – produced by a change in the base sequence of a DNA triplet. This results in an altered protein whose biological function may be affected.*

(xv) *mitosis – the cell division process that produces new cells for growth and repair.*

(xvi) *mutagen – a mutation causing agent, e.g chemicals, radiation, viruses, high temperature.*

(xvii) *mutant – an organism that has a mutation.*

(xviii) *mutation – a sudden and permanent change to the genetic code which makes an offspring different to its parents. When the change occurs in a germ line cell, it may be inherited by future generations.*

(xix) *nitrogenous base – organic compounds containing nitrogen which make up the genetic code in DNA and RNA molecules, e.g. cytosine, guanine, adenine, thymine and uracil.*

(xx) *nonsense mutation – caused by a change in a nucleotide in a codon. It produces a stop codon which affects protein synthesis and related metabolic pathways.*

(xxi) *nucleotide – the structural unit of DNA made up of a sugar molecule (deoxyribose), nitrogenous base and phosphate.*

(xxii) *phenotype – the expression of a particular genotype, e.g. blue eyes, curly hair.*

(xxiii) *silent mutation – occurs when a nucleotide is changed in a base sequence but the change has no effect on the amino acid that is coded for, due to the degeneracy in the genetic code.*

(xxiv) *somatic cell – a non-sex cell (not sperm or ova). Muscle, nerve, connective and epidermal cells are somatic.*

(xxv) *variation – generally a small difference between two members of the same species. Variation in the offspring of sexually reproducing organisms is greater than*

that found in the offspring of asexually reproducing organisms.

Review Questions

1.

(a) *A mutation is a change in the genes or chromosomes of an organism which makes it different to its parents. If it occurs in a gamete, it may be inherited by the offspring.*

(b) *Mutations are relatively rare, spontaneous and random. The rate of mutation can be increased by environmental factors such as certain chemicals (formaldehyde, benzene, aflatoxin, dioxins, asbestos, mustard gas, etc.), UV radiation or nuclear radiation and some viruses. These can cause a change to the sequence of nitrogenous bases in a DNA molecule and affect the way the base sequence is translated resulting in a different amino acid sequence producing a different protein and ultimately, a new allele. Alternatively, they can affect whole genes or segments of a chromosome in which whole or part of a chromosome may be missing or duplicated.*

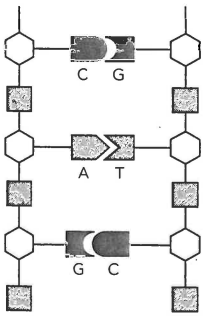
2.

(a) *A germ line mutation is an error that occurs during meiosis. It may result in a change to a gamete. A somatic mutation is an error which occurs during mitosis. It therefore affects normal body cells other than the gametes.*

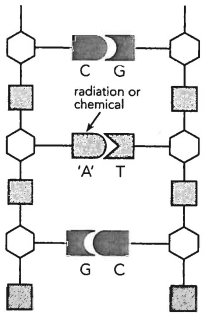
(b) *Because the germ line mutation affects a gamete there is a possibility that it could be passed on to an offspring and become an inherited change affecting future generations. A somatic mutation will not be inherited as it occurs in only body cells. If it does occur early in an embryo's development, it may result in many subsequent cells growing that are different. This may cause a significant change to the developing individual. Sometimes somatic mutations may develop as a cancer. However, most somatic mutations are believed to have no serious effect as they are hidden by normal genes, or just cause the death of an individual cell.*

3.

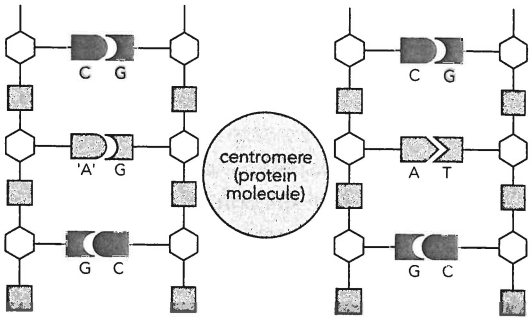
(a) *Before the chromosome replicates, part of it may appear as shown below:*



If radiation or a chemical changes A as shown:



So that A resembles C then when the chromosome 'unzips' and produces two new copies, they will appear as:



Instead of two copied chromosomes being identical, one has a gene that is different to the other. As well as substitution (pictured), bases may also be inserted into or deleted from a strand of DNA.

(b) *If the error is made in the replication of chromosomes (which occurs during interphase) before mitosis, some cells which are produced for growth or repair may be different to the normal cells. If they have a tendency to reproduce rapidly, then they may form a cancerous growth. If the error is made in the replication of chromosomes before a meiotic division, the error may be present in the chromosomes of a gamete. In this case, if the gamete is directly involved in the creation of a zygote, the altered gene will be inherited by the offspring. It will be part of every somatic and germ line cell in that offspring.*

4. *Gene mutations refer to changes in bases, chromosome mutations affect whole chromosomes. A gene or point mutation is one in which bases are affected by either: adding extra bases (insertion), removing (deletion) or replacing (substitution) bases. In chromosome mutations, parts of chromosomes may be broken and inverted, or repeated or reattached to other chromosomes. As well, multiple copies of chromosomes can also be produced, e.g. trisomy 21 (Down's Syndrome).*

5.

(a) *A blood sample is taken. White blood cells are separated and these cells are cultured. The cells begin to divide.*

- *A chemical which inhibits cell division is added and this holds the chromosomes in metaphase.*
- *A hypotonic solution is added which spreads the chromosomes apart.*
- *The chromosomes are then stained and photographed.*
- *The individual chromosomes are cut out of the photograph and glued onto a karyotype analysis sheet in pairs, starting with the longest chromosome and running through to the smallest.*

(b) *Each chromosome consists of two copies (called chromatids) connected by a structure called a centromere. The chromosomes are arranged in pairs. The autosomes (autosomal chromosomes) all look similar (i.e. 22 pairs that look much the same) and the sex chromosomes are put together separately at the bottom of the karyotype.*

(c) *The one on top has two X chromosomes and the one on the bottom has an X and a Y chromosome.*

(d) *The one at the top is from a female; the one at the bottom is from a male.*

(e) *A child with Trisomy 21 (Down's Syndrome) would have three copies of chromosome number 21.*

(f) *One can acquire an extra chromosome from non-disjunction in meiosis, in which the chromosomes do not separate and one gamete ends up containing an extra chromosome.*

6.

(a) *Female.*

(b) *45 chromosomes.*

(c) *Male.*

(d) *47 chromosomes.*

7.

(a) *By mutations.*

(b) *During chromosome replication.*

(c) *The GAG codes for a particular amino acid in the protein haemoglobin, GTG codes for a different amino acid. Therefore, the change results in haemoglobin which has one amino acid in its chain which is different to the normal. This gives the haemoglobin produced different properties. It changes its shape and oxygen carrying capacity.*

(d) *Being heterozygous, Hb^NHb^S, is usually described as having the sickle cell trait. This genotype confers a protection from the pathogen that causes malaria because the parasite cannot invade a sickle-shaped red blood cell (like it does a normal shaped RBC). Therefore individuals with the sickle cell trait are more likely to survive and reproduce than individuals with the normal homozygous genotype, Hb^NHb^N, in those areas of the world where malaria is endemic. Individuals who inherit the homozygous sickle cell genotype, Hb^SHb^S, are described as having sickle cell disease or sickle cell anaemia, a condition that may lead to death in childhood due to the abnormal rate of red blood cell destruction. The disease reduces life expectancy significantly and therefore does not confer an advantage in any part of the world.*

8.

(a) *If the mutation provides the offspring with some feature which gives it an advantage in competition with its rivals, it is an advantageous mutation. The offspring with the mutated gene is more likely to survive, especially in adverse conditions and pass on its mutated gene to its offspring which then has the same advantage.*

(b) *An adaptation.*

(c) *A number of mutations have probably occurred in the human genome which resulted in our ability to walk bipedally. These include changes to our feet, legs, pelvis and vertebrae.*

9.

(a) *Only if the offspring have inherited two recessive deleterious genes, one from each parent.*

(b) *In marriages between close relatives.*

10.

(a) *Radiation – X-rays, nuclear radioactivity; Chemicals – thalidomide; Infections – Rubella virus.*

(b) ***Nuclear radioactivity:** If a foetus is exposed to high levels of nuclear (ionising) radiation, the child may be born suffering from damage to the brain. The risk rises with increasing levels of radiation.*

***Thalidomide:** This is a drug which is currently used to treat abnormalities of the immune system. However, many years ago it was used as a drug to treat nausea (morning sickness) in pregnant women. It was found to cause abnormal growth in foetuses resulting in, children being born with malformed limbs, without limbs and also many miscarriages and still births.*

***Rubella virus:** If an expectant mother contracts this virus during her pregnancy, her child could be born with deafness, blindness, heart and brain damage. The particular damage caused by the virus appears to be determined by which parts of the foetus are developing at the time of the infection.*

11.

(a) *Carriers are individuals that have a particular allele in their genotype but it does not show in their phenotype.*

(b) *Cystic fibrosis is caused by a recessive,*

autosomal gene mutation.

(c) Three letters are missing (AAG).

(d) mRNA 'reads' nucleotides three at a time.

(e) Each group of three nucleotides codes for an amino acid. A chain of amino acids form a protein. So the sequence of bases determines a chain of amino acids which form a particular protein.

(f) Deletion.

7: GENE POOLS

Terminology

(i) allele – an alternative form of a gene that could occupy the same place (locus) on a chromosome.

(ii) allele frequency – see gene frequency.

(iii) autosomal – refers to a chromosome other than a sex (X or Y) chromosome.

(iv) dominant – describes the allele that is always expressed if it is present in a genotype. It is represented by a capital letter, e.g. R.

(v) evolution – (according to Darwin) referred to gradual changes in organisms over a long period of time. This implies that organisms have become increasingly complex over time and that all organisms have shared common ancestry at some time in the past. More recently, evolution is defined as a change in gene frequency.

(vi) Founder effect – refers to the reduction in genetic diversity that occurs when a small group of individuals becomes isolated and establish a new population. It contributes to genetic drift.

(vii) gene flow – the movement of genes from one population to another; comes about by immigration and emigration.

(viii) gene frequency (allele frequency) – the proportion of members of a population with a particular gene, or allele given as a percentage.

(ix) gene pool – the total variety of genes and alleles present in a sexually reproducing population that are available to be passed on to the next generation.

(x) genetic drift – a change in allele frequencies in a population that is due to random events or chance.

(xi) genetic equilibrium – occurs when the allele frequencies of a population do not change from one generation to the next.

(xii) independent assortment – refers to the random lining up of the chromatids in meiosis so that different pairs of genes are passed onto the zygote without being affected by the inheritance of another pair of genes. This results in new genetic combinations in the offspring.

(xiii) migration – movement of organisms from one place to another. Includes immigration (into) and emigration (away from) a place.

(xiv) mutation – a random, permanent change in the DNA of an organism.

(xv) population – a group of organisms that are of the same species, living together in the same place at the same time.

(xvi) recessive – describes the allele that is masked, or hidden by the presence of a dominant allele. The recessive is only expressed in the homozygous condition. It is represented by a lower case letter, e.g. r.

(xvii) selection pressure – an aspect of the environment that determines the chance of survival of a particular characteristic.

(xviii) species – a group of similar individuals which can interbreed to produce fertile offspring under natural conditions or, a collection of populations sharing a common gene pool (within which regular gene flow occurs) which are reproductively isolated from other gene pools.

Review Questions

1. Natural selection, random genetic drift, mutation, Founder effect, immigration and emigration. Cultural isolation of a group within a breeding population.

2.

(a) Variation refers to the differences between members of the same species.

(b) The amount of sunlight that skin is exposed to can affect the degree of melanin development. Wind strength and direction can affect the size and shape that plants grow. Variation (IQ, weight, height) in identical twins separated at birth and raised in different family circumstances.

(c) (i) Mutation. (ii) Crossing over. (iii) Random (independent) assortment of chromosomes across the equator in metaphase in meiosis. (iv) Chance combination of gametes in sexual reproduction (random fertilisation).

3.

(a) Gene flow refers to the movement of genes from one population to another as a result of interbreeding between members of the two different populations. This introduces new variations into the population increasing the range of variations within that group.

(b) Migration (and interbreeding) of European, African and Asian people to Australia.

4.

Event	Increase or decrease gene frequency
Mutation	Increase
Natural selection	Decrease
Migration or gene flow	Increase
Random genetic drift	Decrease or increase
Isolation	Decrease

5.

(a) *A mutation is a sudden, spontaneous change in the base sequence of DNA. Mutations are not always inherited. Somatic mutations which affect body cells are not inherited but may affect a person in their lifetime. Mutations that occur in the ovum or sperm can be passed on to future offspring.*

(b) *Viruses, ionising radiation, ultra-violet light, chemicals such as asbestos, chloroform, PCB's, aflatoxin (from a fungal infection of peanuts), dioxins (TCDD found as a contaminant in Agent Orange), acrylamide, benzene, mustard gas, captan, radon gas, vinyl chloride.*

6. *Random genetic drift describes the random change in genetic frequencies that occurs in the gene pool of small populations from generation to generation. It suggests that variation in gene frequencies of populations can occur by chance rather than by natural selection. In other words, evolutionary change occurs by chance only. It is important in small or isolated populations. In a small population not all the alleles of a species may be present. Chance events like the accidental death of an individual that carries the only example of a particular allele can result in the disappearance of that allele.*
   Similarly, it is also possible for an allele to increase in frequency simply by chance, i.e. random genetic drift means that what happens is unpredictable. As a result a small population could become more, or less, suited to a particular environment.

7. *When members of a population become separated from others of the same population, they may possess only a small selection of genes from the gene pool of their species. Genetic drift that results from this is called the Founder effect, e.g. the Dunkers who migrated from Germany to America. Interbreeding within this group has produced allele frequencies quite different to the German population as well as to the American population amongst whom they live. The same applies to the various Pacific sub-populations – Melanesians, Polynesians, and NZ Maori.*

8.

(a) *Reproductive isolation is a process in which members of a population get separated from other members of the same population so that there is little or no interbreeding. In people it may be due to geographical features or cultural reasons.*

(b) *Gene flow is reduced.*

(c) *Cultural features: economic status, religion, education, occupation. Geographical features: oceans, mountain ranges, deserts.*

9.

(a) *This allele is maintained in this population because of cultural isolation. Members of this population are encouraged to intermarry with people in the same population.*

(b) *They could be told that it is due to a recessive autosomal gene and as parents that obviously do not have the disease; they could be carriers for the condition. If both parents are carriers, i.e. heterozygous for the gene, there is a 25% chance of having a baby with Tay-Sachs disease. It is then their choice whether or not they have a child.*

7.2 Natural Selection

Terminology

(i) *adaptation – a characteristic that an organism has that helps it to survive in its environment. Adaptations may be structural, physiological or behavioural.*

(ii) *reproduction – making more of the same species.*

(iii) *selection – is a process in which genetic or environmental influences determine which types of organism thrive better than others. It is regarded as a factor in evolution.*

(iv) *variation – differences between individuals of the same species.*

(v) *viable – able to live.*

Review Questions

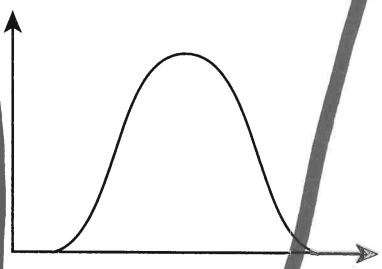
1.

(a) *Differences or variations occur between all members of a population. The members of a population also compete for food, shelter, mates, etc. Some individuals in the population have variations which make them better suited to a particular environment than others. These individuals have a better chance of survival because they compete for the available resources more successfully. Consequently they will be able to reproduce in greater numbers and pass on their favourable features to the next generation, members of which, in turn, are also better suited to the environment. Individuals that are not so well suited to the environment either do not survive, or produce fewer offspring. The genes responsible for the less favourable features (that make them unsuccessful) decrease in frequency.*
   This process, by which the environment favours or selects organisms with suitable features that survive and pass on their genetic information to future generations, is known as natural selection.

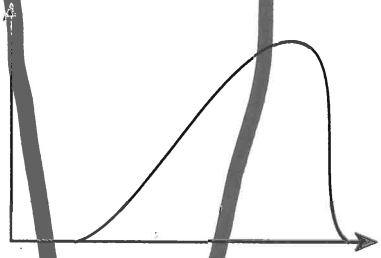
(b) *Over time the suitable features become more common in the population. In other words, the frequency of the genes responsible for*

these features increases and the population becomes recognisably different from its ancestors. Variation within the population decreases. However, mutations produce new variations which are continually tested by natural selection. If the environment does not change, the successful genes will predominate.

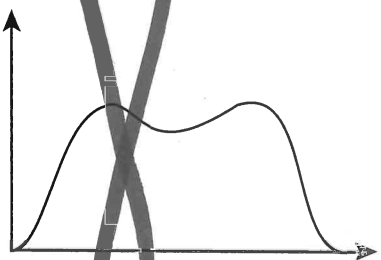
However, if the environment changes, these genes and the features they determine may not be suitable and other genes may be selected.

2. 'Survival of the fittest' means or refers to those organisms that are best suited to the current environmental conditions.
3. Speciation is driven by natural selection and may arise in a population due to (i) geographic barriers such as mountain ranges, changes in river courses or sea levels (allopatric speciation) and (ii) reproductive isolation either as pre-zygotic (before fertilisation) or post-zygotic (after fertilisation). Pre-zygotic isolation between members of a population may occur because they breed at different times, or there may be morphological differences which prevent them breeding, or their courting behaviour is not recognised, or their gametes may become incompatible. Post-zygotic isolation happens because even if fertilisation occurs there may be some problem with the zygote which prevents it from becoming a fertile adult. For example, the offspring may be sterile due to an uneven number of chromosomes stopping meiosis and the formation of sex cells. Example – horse and donkey producing sterile mules. (iii) random genetic drift within a reduced gene pool resulting from (i) or (ii) above. It also may be driven by a change in environmental conditions (sympatric speciation). People who live high in the Andes Mountains have a greater concentration of haemoglobin in their blood compared to people who live at sea level. This means that even though the air they breathe contains less oxygen, their red blood cells can carry more oxygen enabling them to live normally.
4. A genotype is the set of alleles carried by an individual whereas the phenotype is the physical trait produced by the interaction of the genotype and the environment. The human blood group A, in the ABO system is a phenotype. There are two genotypes which can produce this phenotype: they are I^AI^A or I^Ai. The environment is unlikely to affect the phenotype in this example.
5. Some phenotypes in a population are better adapted than others in the population. These phenotypes are more likely to survive and reproduce. A human example of this would be darker skin in equatorial regions of the world. Having darker skin provides protection from UV radiation. It results in less skin cancer among people who have darker skin living in tropical and sub-tropical regions of the world. The phenotype is therefore better suited or adapted for living in these areas.
6. 

Stabilising natural selection favours the average phenotype in the population, e.g. average human birth weights of 3-4 kg.



Directional natural selection favours one extreme phenotype over what is average or the other extreme, e.g. the small stature of Homo floresiensis.



Disruptive natural selection favours the two extremes rather than the average.

7. (a) An adaptation is a feature an organism has which helps it to survive and reproduce in a particular environment.
   (b) Skin colour, facial features, blood groups, hair colour and texture, somatotypes.
   (c) The dark pigment (melanin) in a dark skin will help protect the underlying tissues from damage from ultraviolet light.
   (d) Short stocky builds lose heat more slowly than tall, thin people because they have a smaller surface area to volume ratio.
8. Genetic markers are features determined by a single pair of genes that are not affected by age or environmental conditions, i.e. either you have it or you do not, there is no