Terminology

These are some of the terms from this section which you should know. Write the meaning of each term in the space provided.

(i) allele

(ii) autosomal chromosome (autosome)

(iii) cancer

(iv) centromere

(v) chromosome

(vi) deleterious

(vii) DNA replication

(viii) gene

(ix) genotype

(x) germ-line cell

(xi) karyotype

(xii) meiosis

(xiii) metaphase

(xiv) missense mutation

(xv) mitosis

(xvi) mutagen

(xvii) mutant

(xviii) mutation

(xix) nitrogenous base

(xx) nonsense mutation

(xxi) nucleotide

(xxii) phenotype

(xxiii) silent mutation

(xxiv) somatic cell

(xxv) variation

Review Questions

1. (a) What is a mutation?

(b) What may cause a mutation?

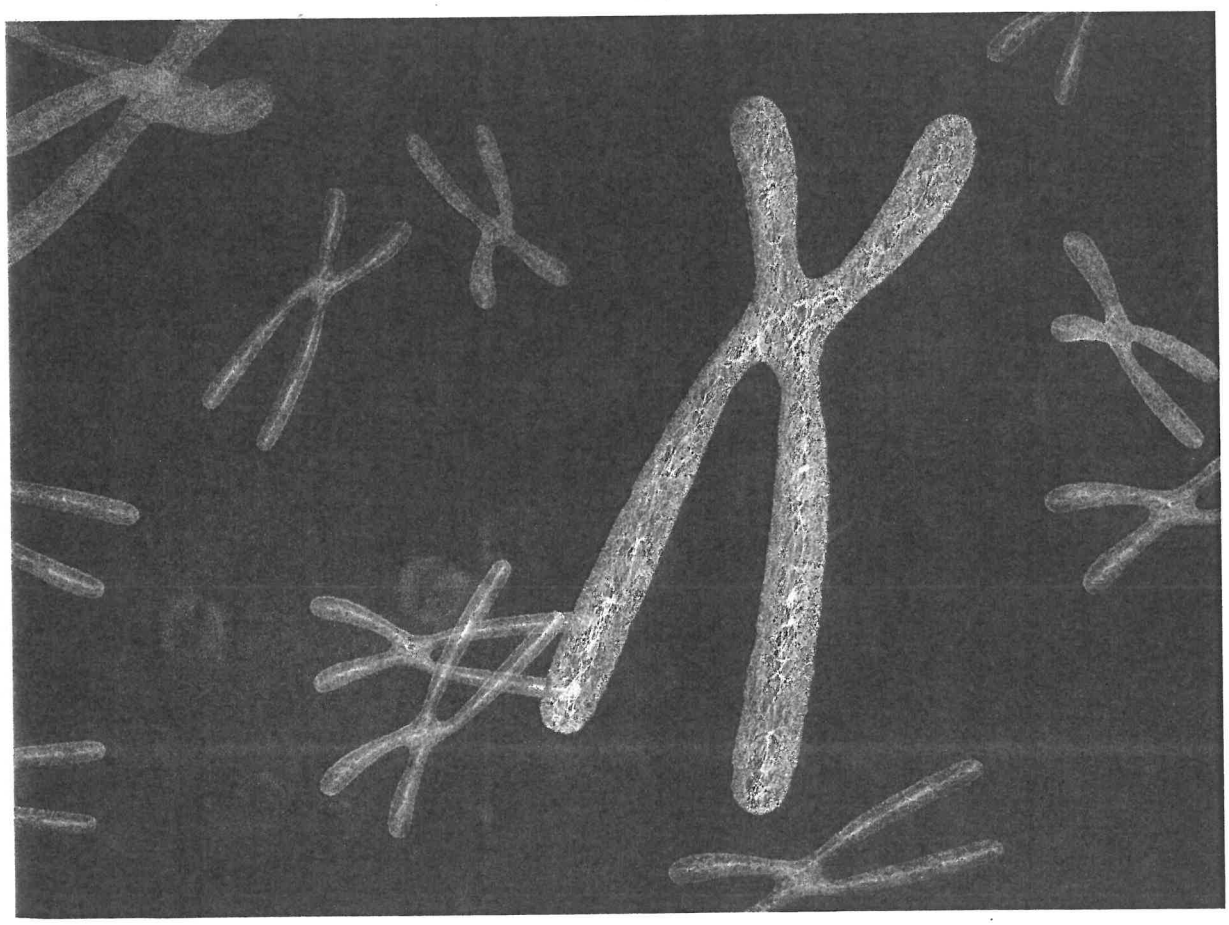
2. (a) What is the difference between a germ line mutation and a somatic mutation?

(b) Discuss the most significant difference in the possible outcome of each of these mutation types.

3. (a) How might an environmental agent change the sequence of nitrogenous bases of a chromosome?

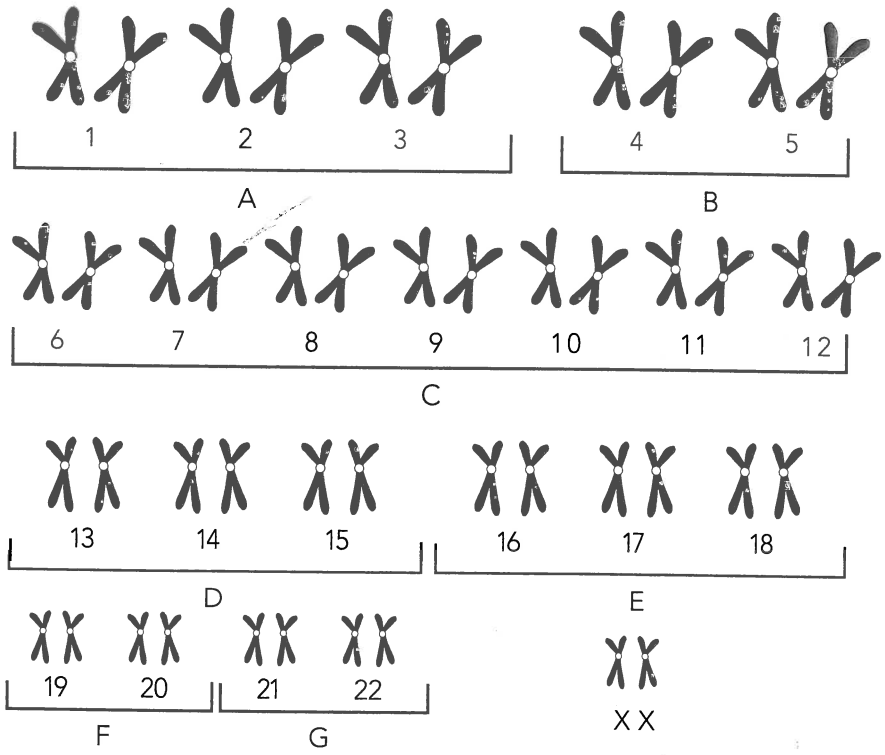
(b) How can a 'mistake' in the sequence of nitrogenous bases affect a person?

4. Describe some differences between gene mutations and chromosome mutations.

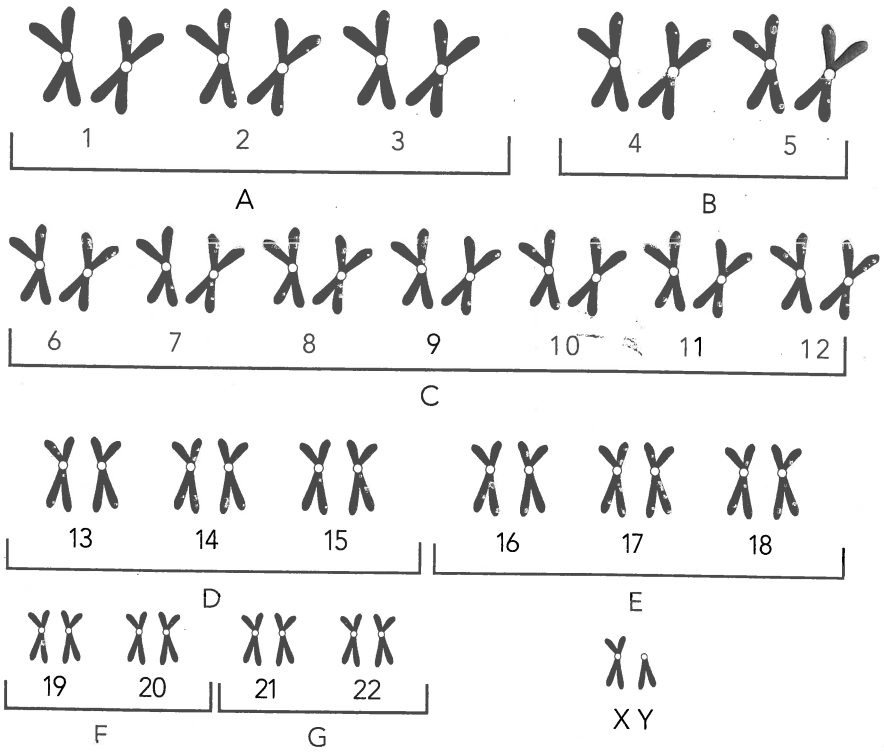


5. The following diagrams are karyotypes from two different individuals.

Individual 1



Individual 2



(a) How is a karyotype prepared?

(b) In what ways are the karyotypes shown similar to each other?

(c) In what ways are these karyotypes different to each other?

(d) To which sex does each karyotype belong?

(e) How would these karyotypes differ if it was from a child with Down's Syndrome?

(f) How could a gamete acquire an extra chromosome?

6. Sometimes the extra chromosomes gained (or lost) in zygotes are sex chromosomes rather than autosomes. Two human conditions caused by gaining or losing sex chromosomes are Turner and Klinefelter's Syndromes.

A person with Turner Syndrome has only one X chromosome. Instead of XX they can be represented as XO.

(a) What gender would such a person be? _____

(b) How many chromosomes would they have in one somatic cell?

_____

A person with Klinefelter's Syndrome has two X's and one Y chromosome, shown as XXY.

(c) What gender would such a person be?

_____

(d) How many chromosomes would they have in one somatic cell?

_____

7. (a) How are new alleles created?

_____

(b) When do most mutations occur?

_____

Sickle-cell anaemia results from the replacement of an adenine, A, by a thymine, T, in the codon which is part of the gene that codes for haemoglobin. Instead of the codon being GAG, it becomes GTG, the T replacing the A.

(c) Why does this mutation cause a problem?

_____

_____

_____

_____

_____

_____

_____

_____

_____

_____

(d) In some parts of the world, being heterozygous for the sickle-cell condition is an advantage. Explain why this is the case.

8. Most mutations are neutral, i.e. they do not affect the survival and viability of the offspring. Some mutations are harmful and some are helpful.

(a) How could a mutation be useful to an offspring?

(b) If a mutation results in an improved phenotype, what is this improvement called?

(c) Give an example of such changes which may have occurred in human evolution.

9. (a) There are many deleterious genes in the human genome. However, most of these are recessive and since we have two genes for each trait, the deleterious genes are not expressed. When would such deleterious genes be expressed in an offspring?

(b) When is this combination of deleterious genes most likely to occur?

10. (a) List three mutagens and give an example of each.

(b) Describe some of the effects that these examples may have on a developing child.

11. One mutation that occurs on chromosome 7 in humans results in cystic fibrosis. This affects the production of a protein that is involved in the movement of salt in and out of cells and as a consequence, thick, sticky mucus is produced in the lungs and pancreas. A child with cystic fibrosis suffers shortness of breath, repeated chest infections and coughs. As well, enzymes produced from the pancreas and used in digesting food may be blocked from being released. So far, there is no cure. Affected children use nebulisers, daily physiotherapy, antibiotics and take enzyme tablets with meals. They are encouraged to have a good diet and exercise regularly. For a child to be born with cystic fibrosis, both parents must be carriers.

(a) What are carriers?

(b) How could you describe the inheritance of cystic fibrosis?

Consider the following sequence of bases on DNA.

(c) If TACGTAGTTAAGCCGTCATGA becomes TACGTAGTTCCGTCATGA, what has happened?

Deleting nucleotides in this way would affect the way that ribosomes read messenger-RNA in translation as part of protein synthesis.

(d) How do ribosomes 'read' transfer RNA?

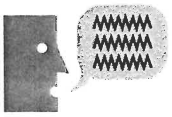
(e) How does that normally relate to protein formation?

(f) If a sequence of nucleotides is different to the original sequence, as in (c), what is this situation called?

(g) What effect does it have?

(h) Cystic fibrosis is a very common mutation in Caucasians. Despite its effects, it has persisted in the population for a long time. How could you explain that?

7.1 GENE POOLS



Terminology

These are some of the terms from this section which you should know. Write the meaning of each term in the space provided.

(i) allele

(ii) allele frequency

(iii) autosomal

(iv) dominant

(v) evolution

(vi) Founder effect

(vii) gene flow

(viii) gene frequency

(ix) gene pool

(x) genetic drift

(xi) genetic equilibrium

(xii) independent assortment

(xiii) migration

(xiv) mutation

(xv) population

(xvi) recessive

(xvii) selection pressure

(xviii) species

Review Questions

1. List four ways in which changes in gene frequencies can occur.

2. (a) What is meant by variation?

(b) Describe an example of variation caused by environmental factors.

(c) List four ways in which variation occurs independently of environmental factors.

(i)

(ii)

(iii)

(iv)

3. (a) Explain how gene flow affects variation in a population.

(b) Give an example of migration into Australia which could change the gene frequency in the Australian population.

4. Indicate on the table below how each event alters gene frequencies.

Event	Increase or decrease gene frequency
Mutation	
Natural Selection	
Migration	
Random Genetic Drift	
Isolation	

5. (a) What is a **mutation?**

(b) List three factors that can speed up the rate of mutation.

6. Describe random genetic drift. What effect does it have on gene frequency?

7. What is the Founder effect? Describe an example of it.

8. (a) What is meant by reproductive isolation?

(b) What effect does this have on gene flow?

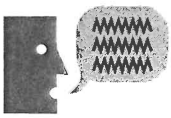
(c) List four possible causes of isolation in human populations.

9. In some isolated populations there is a greater incidence of some genetic diseases than in the rest of the general population, e.g. Tay-Sachs disease in the Ashkenazi Jewish population. This is due to the lack of an enzyme in the brain resulting in damage to the neurons in the brain. Children born with this condition gradually develop symptoms from the age of 3 to 6 months, becoming paralysed, blind and deaf and die by five years of age.

(a) Why might the frequency of this allele be higher amongst these people than in the general population?

(b) There is no cure for a child born with Tay-Sachs disease which is caused by a recessive autosomal gene. If you were a genetic counsellor with this community, what could you tell a couple who were planning to have children?

7.2 NATURAL SELECTION



Terminology

These are some of the terms from this section which you should know. Write the meaning of each term in the space provided.

(i) adaptation

(ii) reproduction

(iii) selection

(iv) variation

(v) viable

Review Questions

1. (a) Describe the process of natural selection.

(b) What effect does it have on gene frequency?

2. What do you think 'survival of the fittest' means?

3. Outline some possible mechanisms that could lead to speciation.

4. Explain the difference between the terms 'genotype' and 'phenotype' using a human example.