SUGGESTED TEXTBOOK ANSWERS

Chapter 12 Mutations and gene pools

The following are suggested answers only. Other answers to the same questions may also be correct.

Science inquiry

Activity 12.1 The incidence of cancer in Australia

Many cancers arise through mutations in somatic cells, and sometimes these mutations are caused by mutagens to which the patient has been exposed. Use references to find out:

- the incidence of different cancers in Australia
- whether there is any relationship between type of cancer and locality in Australia
- what groups of Australians are exposed to mutagens that can cause cancer
- what is being done to limit exposure of Australians to cancer-causing mutagens
- the age groups at which particular cancers are more common in Australia
- whether there are any upward or downward trends in the incidence of particular cancers in Australia.

Answer: There is a wide range of material that a student may present in response to these dot points. Teachers need to ensure that students present recent statistics and have used reliable sources of data.

Activity 12.2 Venusians

Interpreting your data

1 How has this activity shown that mutations that increase an individual's chance of survival and reproduction can affect the proportions of particular characteristics in a population?

Answer: Advantageous mutations are likely to aid survival and reproduction, so they will be retained in a population and their frequency in the gene pool will probably increase.

2 What has happened to the proportion of the allele b in the population? Has it been entirely eliminated? Do you think it ever will be?

Answer: The proportion of the *b* allele in the population declined, because two of the alleles are lost from the population when homozygotes die in infancy.

The allele *b* will not be entirely eliminated, as it has a survival advantage for those with the heterozygous genotype.

3 Summarise how this chance mutation has helped the survival of the Venusian population.

Answer: The chance mutation has increased survival chance of the heterozygotes. They have the protective dark skin and are also protected from the insect bite. When two heterozygotes mate, there is a 50% chance that their offspring will also be heterozygous, contributing to the survival of the Venusian population.

Review questions

1 a Define a population.

Answer: A population is a group of organisms of the same species living together in a particular place at a particular time.

b What do scientists mean when they speak of a 'gene pool'?

Answer: A gene pool is the sum of all the alleles in a given population.

2 a Define 'mutation'.

Answer: A mutation is a random spontaneous change in the DNA. It results in a new variation with characteristics that differ from either parent.

b Explain the difference between somatic and germline mutations.

Answer: Somatic mutations occur in the DNA of body cells and cannot be passed on to offspring; whereas germline mutations occur in the gametes and are passed on through reproduction.

3 a Distinguish between gene mutations and chromosomal mutations.

Answer: A gene mutation is a mutation in a single gene whereas a chromosomal mutation is when all or part of the chromosome (and therefore many genes) is affected.

b Give an example of a congenital disorder (a disorder present from birth) that can be caused by a gene mutation and one that can be caused by a chromosomal mutation.

Answer: Congenital disorders resulting from gene mutations include albinism; Duchenne muscular dystrophy; cystic fibrosis; haemophilia; Tay-Sachs disease; and sickle-cell anaemia.

Chromosomal mutation examples include Patau syndrome; Down syndrome; Klinefelter's syndrome; Turner's syndrome; and Cri du chat.

4 Describe four different types of chromosomal mutations.

Answer: A student's description should include four of the following:

- deletions – the loss of part of a chromosome
- duplications – part of a chromosome occurs twice
- inversions – breaks occur in a chromosome and the broken piece rejoins the chromosome the wrong way around
- translocations – part of a chromosome breaks and rejoins, but rejoins the wrong chromosome
- non-disjunction – when a homologous pair of chromosomes during the first meiotic division, or a pair of chromatids during the second meiotic division, fail to separate and both move to the same pole of the dividing cell. This results in two or more gametes having either more or less chromosomes than is normal.

5 a What are mutagens (or mutagenic agents)?

Answer: Mutagens are agents that increase the rate at which mutations occur.

b List five examples of mutagenic agents.

Answer: Five examples of mutagenic agents are: mustard gas, sulfur dioxide, some antibiotics, formaldehyde, ionising radiation (UV light, X-rays, cosmic rays, nuclear fallout or radiation from radioactive waste).

c Why does special care need to be taken when pregnant woman require X-rays?

Answer: Special care needs to be taken when pregnant women require X-rays as radiation from the X-rays can cause mutations in the developing foetus, especially in the first three months of pregnancy.

6 What is a lethal recessive?

Answer: A lethal recessive is a recessively inherited allele that results in the offspring dying either during embryonic or foetal development, or later in life. The offspring must inherit two of the lethal recessive alleles – one from the mother and one from the father.

7 Summarise the pattern of inheritance that occurs in genetic disorders such as Duchenne muscular dystrophy. When there is no history of such disorders in a family, how are they thought to arise?

Answer: Duchenne muscular dystrophy is a condition that is X-linked recessive; that is, the recessive allele for the condition is carried on the X chromosome. Males can therefore show the disorder with only one recessive allele, because they only have one X chromosome. Females must inherit two recessive alleles to have the disorder. When there is no history of such a disorder in a family, they are thought to arise from a mutation in the mother that is then passed on to her children.

8 a Distinguish between trisomy and monosomy.

Answer: Trisomy is when there are three copies of a chromosome instead of the normal two (an extra chromosome); whereas monosomy is when there is one copy of a chromosome instead of the normal two (missing a chromosome).

b Give an example of each condition.

Answer: Trisomy examples: Trisomy 21 or Down syndrome; Klinefelter's syndrome, Patau Syndrome (trisomy 13).

Monosomy example: Turner's syndrome

Partial monosomy example: Cri du chat syndrome

9 Explain how Klinefelter's syndrome and Turner's syndrome come about.

Answer: Klinefelter's syndrome and Turner's syndrome arise from non-disjunction during meiosis. If one of the gametes formed from that meiotic division is involved in fertilisation, the resulting individual will have either trisomy or monosomy. Klinefelter's syndrome is trisomy XXY; Turner's syndrome is monosomy X.

10 Explain how mutations could change the proportion of certain alleles in a gene pool.

Answer: If a mutation resulted in the formation of a lethal recessive allele, then the death of an individual with these recessive alleles before they were passed on to the next generation would result in the removal of those alleles from the gene pool. Over time, the proportion of these lethal recessive alleles in the gene pool would gradually reduce.

If a mutation gave the individual with the mutant allele a survival advantage, that individual would be more likely to survive, reproduce and pass on the mutation than an individual with the normal allele. Over time the proportion of the mutant alleles in the gene pool would increase.

Apply your knowledge

1 Discuss why mutations occurring in the reproductive cells are considered more important than those occurring in the body cells. In your discussion, describe the possible long-term effects of the two situations.

Answer: Mutations in the reproductive cells can be passed on to offspring. Mutations in body (somatic) cells cannot be passed on. The mutation originating in the reproductive cells could have long-term consequences if it is continually inherited. The somatic mutation affects only that individual and cannot be passed on to offspring.

Mutations in the reproductive cells are also important, because they contribute to variation and they could give rise to a favourable variation that increases the survival chance of the individual with that mutation.

2 A large number of mutagenic agents can be found in the environment. Consider those that could possibly affect you in your lifetime and discuss steps that you can take to minimise any risks from exposure to those agents.

Answer: During a lifetime you could be affected by the following mutagenic agents.

- X-rays: these should only be used if absolutely necessary.
- Sulfur dioxide: consider the environment where you live – avoid living near a gold mine or smelter.
- Mutagenic chemicals such as formaldehyde: use protection such as breathing apparatus when exposed to such chemicals.
- Antibiotics: discuss the possible mutagenic effects with a medical practitioner.

3 The more often cells divide, the greater the risk of errors and mutations. For this reason, scientists have hypothesised that when a baby is born with a congenital disorder caused by an error in cell division, the father is the parent more likely to have contributed the gene with the mutation. Compare the number of eggs produced by a female with the number of sperm produced by a male and explain why scientists have proposed this hypothesis.

Answer: The average number of sperm in just one ejaculation is more than 100 million. A female foetus has several hundred thousand primary oocytes at birth. Once sexual maturity is reached, usually only one egg completes meiosis in each monthly cycle. Scientists have therefore proposed this hypothesis because male reproductive cells are undergoing many more meiotic divisions than reproductive cells in a female.

4 Some naturally occurring viruses are considered mutagenic, because they can insert themselves into host DNA. Explain why this ability would make them mutagenic.

Answer: Naturally occurring viruses that are able to insert themselves into host DNA result in the host having altered DNA. This may result in a change in the organism's characteristics. Thus, the virus has caused a mutation and is therefore termed mutagenic.

5 What is the sex of the individual whose karyotype is shown in Figure 12.5?

Answer: The individual is a male because the karyotype shows one X and one Y chromosome.

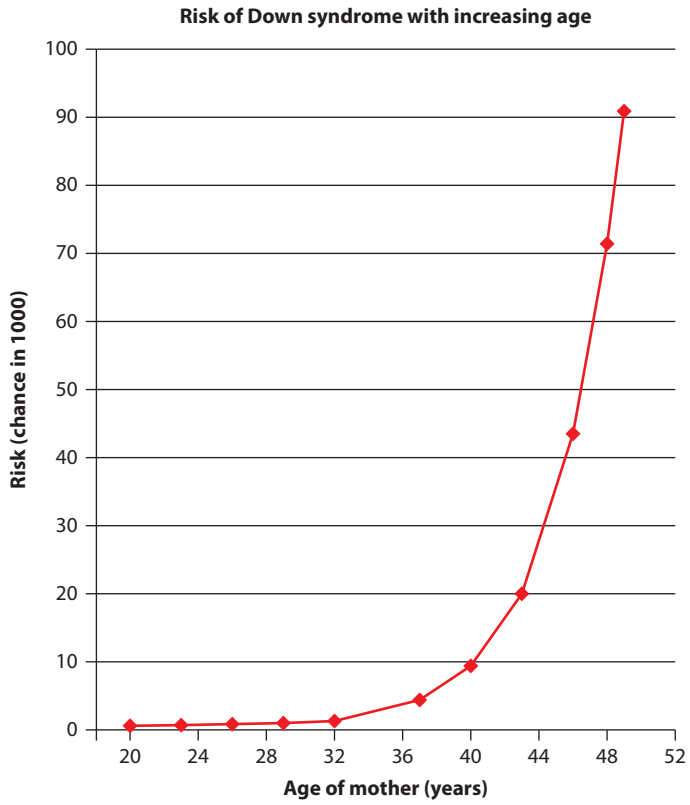
6 Lethal recessive alleles result in the death of an individual. How would this affect the allelic composition of the gene pool?

Answer: The allelic composition of the gene pool is affected by the death of an individual with lethal recessive alleles as most individuals with such alleles die before they are able to reproduce and pass their alleles on to the next generation. Therefore, such a death results in the removal of these alleles from the gene pool. Over time, the proportion of these lethal recessive alleles in the gene pool would gradually reduce, thus changing the composition of the gene pool.

7 The risk of a having a baby with Down syndrome increases as the mother gets older. Table12.1 shows the relationship between Down syndrome and maternal age.

a Draw an appropriate graph to display the data in Table 12.1.

Answer:



Students may draw a column graph, which is perfectly acceptable.

b The risk of a baby having any chromosome abnormality increases dramatically with increasing maternal age. Suggest reasons for this.

Answer: The ova begin to form before a female child is born. Thus in a 40-year-old woman the eggs are 40 years old. The DNA becomes older and less stable so there is an increased chance of mutations and errors during meiosis.

8 Figure 12.8 shows the sequence of the genes A to M on a chromosome. What type of chromosomal mutation is represented by each of **a**, **b** and **c**?

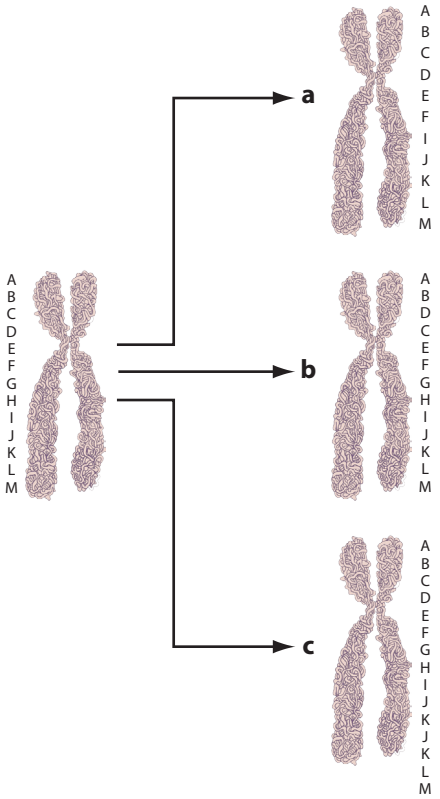


Figure 12.8 Some types of chromosomal mutations

Answer:

a Deletion

b Inversion

c Duplication