Chapter 6

Mutations and gene pools

Mutations

- mutations in genes and chromosomes can result from errors in DNA replication, cell division or from damage caused by mutagens
- different genotypes produce a variety of phenotypes, which are acted on differently by factors in the environment, producing different rates of survival
- mutations are the ultimate source of variation introducing new alleles into a population: new alleles may be favourable or unfavourable to survival

Gene pools

- populations can be represented as gene pools that reflect the frequency of alleles of a particular gene; gene pools can be used to compare populations at different times or locations
- gene pools are dynamic, with changes in allele frequency caused by:
  - mutations
  - differing selection pressures
  - random genetic drift, including the founder effect
  - changes in gene flow between adjoining groups
- the incidence of genetic diseases in particular populations illustrates the effects of different factors on the dynamics of gene pools, including the incidence of Tay-Sachs disease, thalassemia (α and β) and sickle-cell anaemia
- natural selection occurs when factors in the environment confer a selective advantage on specific phenotypes to enhance survival and reproduction
- the mechanisms underpinning the theory of evolution by natural selection include inherited variation, struggle for existence, isolation and differential selection, producing changes to gene pools to such an extent that speciation occurs

MULTIPLE-CHOICE QUESTIONS

1. *(2016:1.04)*

Evolution occurs through a process that involves the following events:

i. changes to gene pool

ii. struggle for existence

iii. survival of the fittest

iv. variation

v. sexual reproduction.

The correct order of these events is

(a) V, IV, II, III and I.

(b) V, II, III, IV and I.

(c) I, V, III, II and IV.

(d) I, V, II, IV and III.

2. *(2016:1.08)*

In the relatively isolated region of Lake Maracaibo in northwest Venezuela, there is an unusually high incidence of the genetically-inherited nerve disorder known as Huntington's disease. Around the world Huntington's disease is estimated to affect 5–7 people in 100000. In Lake Maracaibo, it occurs at a rate of 700 people in 100000. All of the individuals in Lake Maracaibo who carry the allele for Huntington's disease can trace their ancestry to one of the original 19th century inhabitants of the region.

This example is an illustration of

(a) natural selection.

(b) the founder effect.

(c) a high rate of new mutations occurring.

(d) a natural disaster causing genetic drift.

3. *(2016:1.09)*

Which of the following statements describes speciation?

(a) A mutation occurs in an individual then produces a new species.

(b) Two different species interbreed to produce a different species with combined features.

(c) A population of one species diverges genetically and produces one or more new species.

(d) A small population can no longer interbreed with others in the larger population, producing a new species.

4. *(2016:1.13)*

The genetic disease known as Tay-Sachs has been the subject of much scientific debate over the evolutionary mechanisms that have produced the patterns of inheritance of the disease. Different theories, all of which have sound scientific reasoning, have linked Tay-Sachs to the founder effect, genetic drift and natural selection.

Which of the following would be the **best** reasoning to link the inheritance of Tay-Sachs to genetic drift?

(a) The original populations carried a high incidence of the allele.

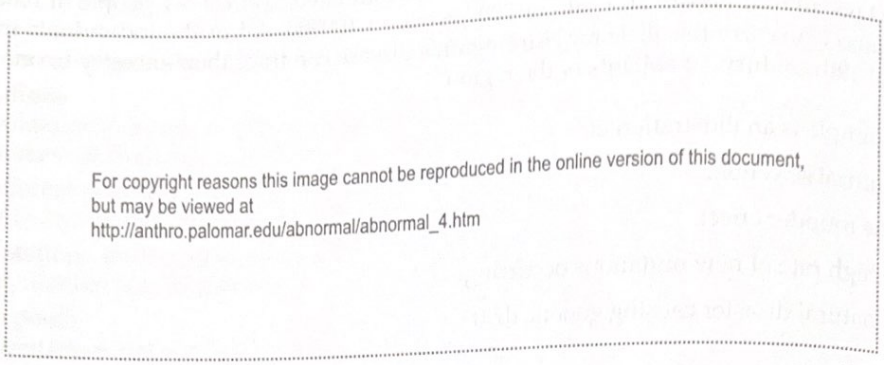
(b) Affected populations tend to be small and reproductively isolated.

(c) Sufferers of the disease reproduce at greater rates than non-sufferers.

(d) Carriers of the allele have a survival advantage over non-carriers.

The next two questions refer to the information and diagram shown below.

Down syndrome is an example of a disorder caused by a mutation. This is known as trisomy-21 and is shown below in the karyotype of an affected female.



5. *(2016:1.24)*

This type of mutation is a

(a) point mutation.

(b) somatic mutation.

(c) chromosomal mutation.

(d) gene mutation.

6. *(2016:1.25)*

The cause of this mutation is

(a) non-disjunction.

(b) deletion.

(c) duplication.

(d) translocation.

7. *(2017:1.11)*

A ship is wrecked and the survivors, a group of ten men and ten women, manage to reach an island. They are never found and so have had no contact with the outside world. Of the survivors, 12 have blue eyes, 4 have green eyes and 4 have brown eyes. After a while, the population on the island grows and after more time the only eye colour of the population is blue.

The process by which a population's gene pool changes significantly from the original population is known as

(a) founder effect.

(b) random genetic drift.

(c) natural selection.

(d) mutation.

8. *(2017:1.17)*

The following statements relate to natural selection.

(i) Some genotypes provide a better chance of survival in specific environments.

(ii) Members of different species show some similarities in phenotype.

(iii) Some characteristics are heritable and are passed on to offspring.

(iv) Members of the same species show variation in phenotype.

Which of the statements support the theory of natural selection?

(a) (i), (ii), (iv)

(b) (ii), (iv)

(c) (i), (iv)

(d) (i), (iii), (iv)

9. *(2017:1.26)*

Which of the following statements **best** describes evolution?

(a) Evolution can occur in a population without causing any changes in gene frequency.

(b) The development of new features occurs when a species settles into a new, uninhabited territory.

(c) Evolution alters the genetic variation in a population.

(d) The process occurs more readily in larger populations.

10. *(2017:1.28)*

A gene pool is

(a) all the different genes found in an ecosystem.

(b) all the different alleles in an interbreeding population.

(c) the alleles in a group of individuals that cause a genetic disease.

(d) the total number of genes found in a species.

11. *(2018:1.03)*

Thalassemia and sickle-cell anaemia are both genetic diseases that

(a) result in sickle-shaped haemoglobin.

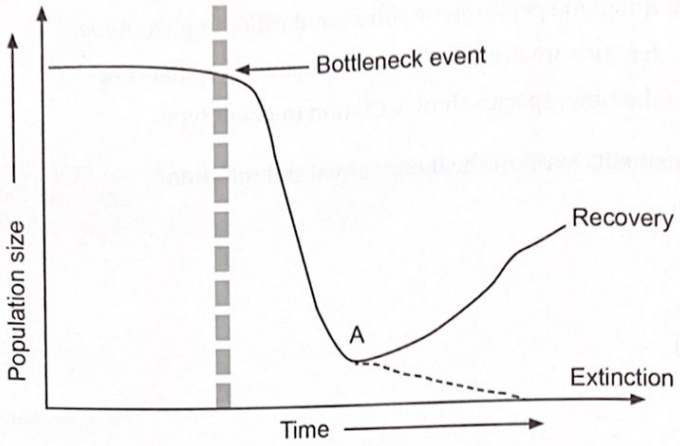
(b) are caused by a genetic mutation associated with haemoglobin formation.

(c) occur at high incidences in some populations.

(d) are shown to have a heterozygote advantage in some circumstances.

The next question refers to the diagram below.

Population change with bottleneck effect



12. *(2018:1.10)*

The bottleneck effect shown in the diagram is a catastrophic event that occurs to a population, resulting in a sharp reduction to a gene pool. At point A on the graph, the population has a chance of either recovery or extinction. What is the most likely characteristic of the population, after the bottleneck event, that creates this situation?

(a) The population must still be recovering from the catastrophic event and not have strong alleles present in the gene pool.

(b) A small population size means the gene pool contains highly-varied alleles producing offspring not well suited to the environment.

(c) Members of the population now vary too much and cannot produce fertile offspring.

(d) A small population size means it is more susceptible to random genetic drift.

13. *(2018:1.16)*

Around 14 000 years ago, about 70 humans crossed a land bridge from Siberia to North America. DNA studies have shown that these 70 humans are the ancestors of all the Native American tribes found in North America. On the basis of this information, characteristics shared by all Native Americans can be best described as being a result of

(a) random genetic drift.

(b) natural selection.

(c) speciation.

(d) founder effect.

14. *(2019:1.10)*

Which of the following pair of responses identifies correctly the difference between sickle cell anaemia and sickle cell trait?

	Sickle cell anaemia	Sickle cell trait
(a)	provides resistance to malaria	provides no resistance to malaria
(b)	people are carriers of the genetic disease	people are not carriers of the genetic disease
(c)	involves two affected alleles for the condition	involves only one affected allele for the condition
(d)	people have malformed haemoglobin	people have crescent-shaped haemoglobin

15. *(2019:1.14)*

Gene flow occurs when

(a) species migrate to new areas.

(b) genes are exchanged during fertilisation.

(c) individuals migrate to new areas.

(d) mating occurs between related individuals.

16. *(2019:1.20)*

The 'Toba catastrophe theory' suggests that the human population was almost completely wiped out around 75000 years ago. It is believed that a super-volcano near Lake Toba in Indonesia erupted, creating a global volcanic winter that lasted 10 years. This event killed most humans, leaving as few as 40 breeding pairs of adults. According to the theory, this small population went on to repopulate the entire human species.

If true, this repopulation would be best described as an example of

(a) natural selection.

(b) random genetic drift.

(c) gene flow.

(d) selective breeding.

17. *(2020:1.03)*

The term 'selectively-advantageous mutation' means the mutation

(a) provides a survival advantage in a particular environment.

(b) will always be passed to the offspring.

(c) is always changing to adapt to new environments.

(d) provides a benefit to heterozygote individuals in populations.

(2021:1.13)

18.

Which of the following are all selection pressures which could reduce genetic variation in a population?

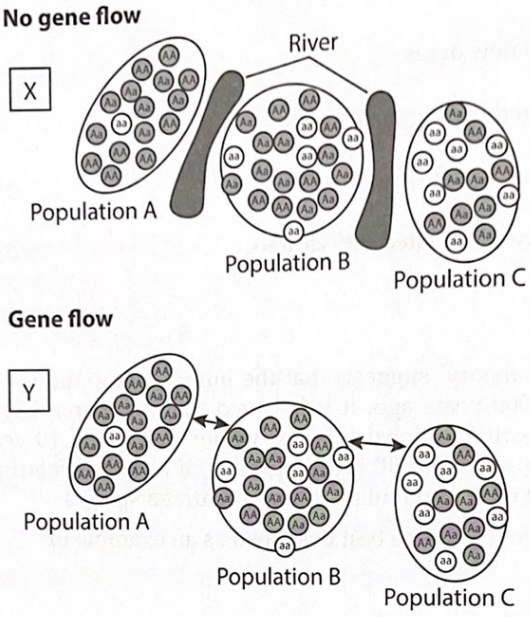
(a) disease, increased competition and climate change

(b) increased food availability, disease and climate change

(c) increased competition, reduced environmental pollutants and disease

(d) reduced land availability, increased food availability and climate change.

The following question refers to the diagram shown below.



19. *(2021:1.16)*

On the basis of the theory of natural selection, the **best** prediction for the future of populations 'A', 'B' and 'C' is that

(a) Y indicates gene flow is maintained between populations A, B and C so the populations would expect to evolve separately into unique species.

(b) X indicates gene flow is not maintained between populations A, B and C due to large water bodies, but this should have little impact on the evolution of the different species.

(c) X indicates that, over time, populations A, B and C could become different species due to geographical barriers and reproductive isolation between the populations.

(d) Y indicates that populations A, B and C should become one species in the future as gene flow is maintained and reproductive isolation is not evident between populations.

20. *(2021:1.22)*

Which of the following statements does **not** support the theory of natural selection?

(a) Organisms reproduce at a rate greater than can be supported by the environment.

(b) Genotypic variation is exhibited in the phenotypes of individuals.

(c) Selective agents act on the alleles in a gene pool.

(d) Similar individuals mate and produce offspring with favourable traits.

21. *(2021:1.28)*

Alpha and Beta Thalassemia have similarities and differences in their inheritance patterns. Which of the following is correct?

(a) Both are autosomal dominant conditions controlled on multiple gene loci.

(b) Alpha Thalassemia is fatal for homozygote individuals while Beta Thalassemia is not.

(c) Both are autosomal recessive conditions that result in affected individuals having four defective globin genes.

(d) Beta Thalassemia is fatal for heterozygote individuals while Alpha Thalassemia is not.

SHORT ANSWER QUESTIONS

22. [11 marks] *(2016:2.38)*

Anthony Clifford Allison, a South African geneticist, conducted studies on genetic resistance to malaria. This led him to discover the link between malaria and the frequency of the sickle-cell allele in the African population. He found that the majority of Africans with the sickle-cell trait did not develop malaria. The majority without the sickle-cell trait did develop malaria. Malaria can be life threatening so those individuals who had the sickle-cell trait were able to live longer lives than those without the trait.

(a) People who have both alleles for the sickle-cell have the disease sickle-cell anaemia. What is sickle-cell anaemia? [2]

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(b) People who carry only one sickle-cell allele are said to have the sickle-cell trait. State **one** disadvantage sickle-cell trait has for people with this condition. [1]

(c) The sickle-cell trait has an advantage for people living in malaria-prone areas due to a selectively advantageous mutation. Explain what is meant by a selectively advantageous mutation. [3]

(d) (i) Describe how the link between the sickle-cell allele and malaria can lead to changes in the allele frequencies in a population. [4]

(ii) What term is used to describe the process given in part (d)(i)? [1]

23. [10 marks] *(2017:2.39)*

Since World War I, mustard gas has been used as a weapon in chemical warfare. Within 24 hours of exposure to mustard gas, victims develop chemical burns, which appear as large blisters to the exposed skin and respiratory surfaces.

In 1940, Auerbach and Robson completed the first research into the other potential effects of mustard gas. They exposed Drosophila flies to mustard gas and then examined the chromosomal damage in the flies' offspring over several generations. Their results showed a dramatic increase in the number of chromosomal mutations compared to the control group.

Najafi and others (2014) studied human victims 25 years after their exposure to mustard gas during the Iraq-Iran War. It found there were 122 different mutated genes in the respiratory pathways of the victims.

(a) The studies described above outline how mustard gas has been shown to cause mutations. Mustard gas can therefore be classed as a [1]

(b) Auerbach and Robson were studying chromosomal mutations. Describe **two** types of these mutations that can occur in organisms. [4]

(c) The 2014 study examined mutated genes. Describe how this type of mutation differs from a chromosomal mutation. [2]

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(d) Explain why Auerbach and Robson studied the offspring flies, not the parent flies, and what information this gave about the type of mutations that occurred. [3]

24. [12 marks] *(2018:2.36)*

Evolution of a species can occur through either genetic drift or natural selection.

The evolution of hominids probably occurred through the process of natural selection as the environment around them changed.

(a) Describe mechanisms underpinning the theory of natural selection that produce changes in a gene pool, leading to speciation. [5]

Genetic drift is another evolutionary mechanism that can cause a change in gene pools.

(b) What is random genetic drift? [2]

Nigeria has the highest incidence of children born with sickle-cell anaemia, which can cause premature death.

(c) Explain, using the most likely scenario (random genetic drift or natural selection), how sickle-cell anaemia became common in Nigeria. [5]

25. [9 marks] *(2019:2.33)*

'The Radium Girls' of the 1920s were women who worked in United States factories applying glow paint to the dark dials of watch faces. The women used paint containing radium to create the numerals on the watch faces. The women were told to use their lips and tongue to pinch the paint brush tip in order to create a fine painting point. As a result, the women were accidently consuming radium. The women suffered many devastating negative effects, including bone disease and various cancers.

(a) Radium is known to cause mutations in the DNA. Therefore, radium can be classified as what type of substance? [1]

(b) Studies of the types of mutations that radium causes have shown that it can result in a change to DNA, known as aneuploidy. Examples of aneuploidy included trisomy-21 and Turner's syndrome. In these cases, there is either an extra number or a reduced number of chromosomes found in daughter cells when compared to normal cells.

(i) When would the error occur to cause aneuploidy, including genetic disorders such as trisomy-21 and Turner's syndrome? [1]

(ii) Is aneuploidy an example of a gene or chromosomal mutation? Justify your answer. [2]

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(iii) Is aneuploidy an example of a somatic or germline mutation? Justify your answer. [2]

(c) Explain how mutations can lead to changes in the allele frequencies of gene pools. [3]

26. **[7 marks]** *(2019:2.39)*

A genetic condition which is often associated with the Founder Effect is Tay Sach's disease. The disease occurs in much higher incidence among Ashkenazi Jews and members of the Cajun community of Louisiana.

(a) Explain how the Founder Effect could account for the occurrence of Tay Sach's disease in these populations. [4]

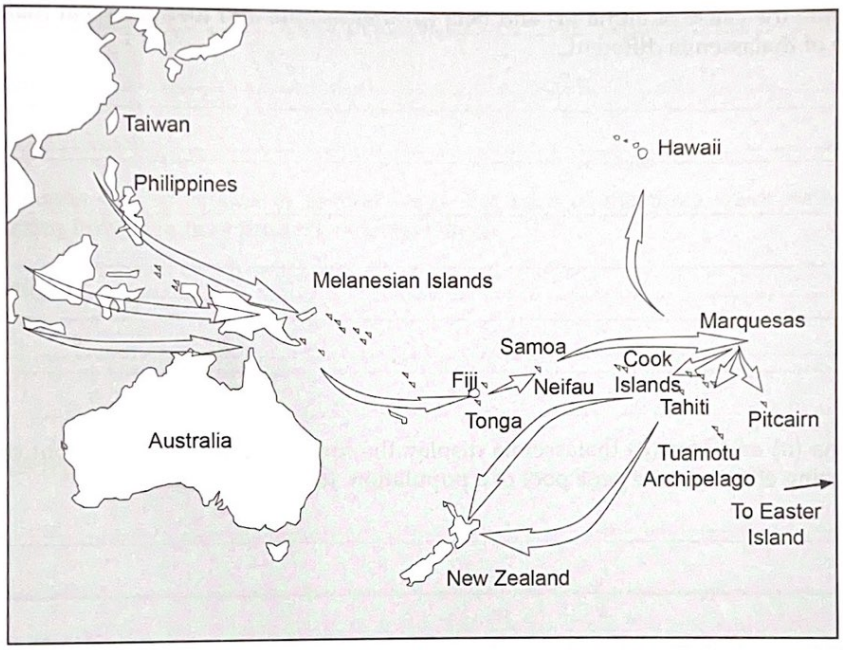
Although Tay Sach's disease is fatal, the allele has been maintained in gene pools. This is believed to be because it provides a heterozygote advantage.

(b) (i) What is meant by the term 'heterozygote advantage'? [2]

(ii) Identify the heterozygote advantage that Tay Sach's is believed to provide. [1]

27. [5 marks] *(2020:2.35)*

The image below depicts a possible settlement pattern for the previously uninhabited Polynesian islands.



(a) In relation to the following, describe the effect on the gene pools each time a small group of people settle onto a new Polynesian island.

(i) Gene flow: [2]

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(ii) Impact of selection pressures on alleles: [3]

(2020:2.36)

28. [8 marks]

Thalassemia is a genetically inherited disorder that affects the haemoglobin of blood. Two types of thalassemia are alpha (α) and beta (β). One of the treatments of both types of thalassemia involves drugs that are administered to help remove excess iron from the blood. A research team wanted to investigate the effectiveness of the different modes of delivery of these drugs.

(e) Outline the cause of alpha (α) and beta (β) thalassemia and identify what makes each type of thalassemia different. [3]

(f) Alpha (α) and beta (β) thalassemia display the same inheritance pattern but can have differing effects on the gene pool of a population. Justify this statement. [5]

29. [14 marks] *(2021:2.32)*

In 2009, the University of California discovered a mutated gene that has been shown to produce naturally short sleepers. The gene was found in a family who all normally went to bed around 11 pm and woke up naturally around 5 am. The mutated gene, known as DEC2, is believed to affect the circadian rhythm and results in people who don't need as much sleep as the average person.

(a) DEC2 is a gene mutation. How do gene and chromosomal mutations differ? [2]

(b) In the study, the mutation was found in several family members. What does this tell you about where the original mutation occurred? Justify your response. [2]

(c) Mutations can be caused in several ways. For each of the three ways stated below, describe how they may produce new mutations. [6]

Mutagens:

DNA replication:

Cell division:

CONTINUED NEXT PAGE

(e) A mutation, such as the DEC2 gene, could be favourable to the human population. Explain how a favourable gene like this could lead to changes in allele frequencies of a gene pool. [4]

30. [9 marks] *(2021:2.34)*

Approximately 1 in 25 people from the Ashkenazi Jewish community in Australia will be a genetic carrier for Tay-Sachs as well as other genetic conditions, such as cystic fibrosis. There are several theories as to why the frequency of these genetic conditions is high in the Ashkenazi Jewish population.

(a) State what is Tay-Sachs. [1]

(b) Explain how the high incidence of Tay-Sachs within the Ashkenazi Jewish community in Australia is an example of the founder effect. [4]

During the Second World War, tuberculosis (TB) ran unchecked in Eastern European Jewish settlements. Often, healthy relatives of children with Tay-Sachs disease did not contract TB, even when exposed repeatedly.

(d) Using the theory of natural selection, explain why Tay-Sachs disease still exists in the Ashkenazi Jewish populations today. [4]

EXTENDED ANSWER QUESTIONS

31. [20 marks] *(2017:3.41)*

For each inherited genetic disease, identify a population in which the disease is prevalent, outline the cause and main symptoms experienced by affected individuals and describe how the disease is inherited and the effect it has on the gene pool of populations.

(a) sickle-cell anaemia [7]

(b) Tay-Sachs disease [6]

(c) thalassemia [7]

32. [20 marks] *(2018:3.41)*

Mutations in the human genome can result from a variety of different causes. For each of the scenarios below explain how mutations are caused and the types of mutations produced. Provide an example of each to help clarify your answer.

(a) Errors in DNA replication [7]

(b) During cell division [7]

(c) From damage caused by mutagens [6]