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Answers: Chapter 9 Evolution produces changes across generations

Questions 9.1

RECALL KNOWLEDGE

**1** Define ‘mutagen’ and list three examples.

*Answer:* An agent that is known to increase the rate of occurrence of mutations. Some examples include: mustard gas, formaldehyde, sulfur dioxide and some antibiotics, all forms of ionising radiation are also mutagenic.

**2** Describe the effects of these different types of mutation:

**a** missense mutation

*Answer:* Missense mutations cause a change in the amino acid, and therefore the protein produced.

**b** nonsense mutation

*Answer:* Nonsense mutations change the base sequence to the code to STOP. This means the synthesis of the protein will stop, and a shorter protein is produced that is unlikely to be able to fulfil its function.

**c** neutral mutation

*Answer:* A neutral mutation causes a change in an amino acid, however the amino acid is of the same type and does not change the structure of the protein enough to change its function.

**d** silent mutation.

*Answer:* A silent mutation does not cause any change in the amino acid, and therefore in the protein produced.

**3** Draw and label diagrams to demonstrate how an inversion changes the base sequence on DNA.

*Answer:* Students may also refer to Figure 9.10 on page 239 for a chromosome inversion mutation.

Original strand of DNA: A T G C T A G C T A G G C A A T C G A T G C A T

Inversion mutation: A T G C T A G C T A C T A A C G G G A T G C A T

**4** Explain why only germline mutations are passed on to the next generation.

*Answer:* Germline mutations are mutations that have occurred in the gametes. In these instances, the individual cells are rarely affected, but all their gametes will contain the mutated DNA.

**5** List two conditions due to:

**a** gene mutations

*Answer:* Duchenne muscular dystrophy, cystic fibrosis, sickle-cell anaemia, Tay-Sachs disease

**b** chromosomal mutations.

*Answer:* Trisome-21 (Down syndrome), Klinefelter’s syndrome, Turner’s syndrome, Cri-du-chat syndrome

**6** Is Down syndrome an example of a gene mutation or a chromosomal mutation? Explain your answer.

*Answer:* Down syndrome is a chromosomal mutation. It is a result of non-disjunction of the 21st chromosome, resulting in a person having three of the 21st chromosomes.

APPLY KNOWLEDGE

**7** Use a diagram to explain why the insertion of a single nucleotide can cause a frameshift mutation, resulting in a change in many amino acids.

*Answer:* Refer to Figure 9.9a on page 238 to show an insertion and frameshift mutation.

**8** The original base sequence of a small section of DNA is shown below.



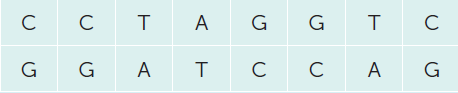
Name the point mutation that has occurred in each of the following.

**a**

****

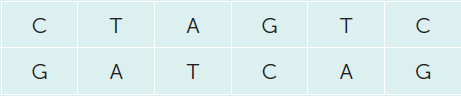
*Answer:* Substitution

**b**

****

*Answer:* Insertion

**c**

****

*Answer:* Deletion

**9** Use Tay-Sachs disease as an example to explain how a gene mutation can be lethal.

*Answer:* Tay-Sachs disease is a disorder of lipid metabolism, inherited in an autosomal recessive pattern. The mutation occurs in the HEXA gene that codes for a protein to break down toxic substances (a fatty acid called GM2 ganglioside) in the brain and spinal cord. A baby born with two recessive alleles will develop normally for the first few months and then will die in early childhood.

**10** Some mutations result in a STOP codon, preventing any more amino acids being added to the chain. These are known as nonsense mutations. Figure 9.7 shows the base sequence on the mRNA, which is a copy of the coding strand of the DNA but containing the base uracil instead of thymine. Describe the mutation that could have occurred to each of the following original base sequences on the coding strand to result in a nonsense mutation.

**a** A A A

*Answer:* Two substitutions would need to occur to result in the DNA sequence reading A T T or A C T. This will produce U A A or U G A respectively which are both STOP codons.

**b** T C G A

*Answer:* Two substitutions would need to occur to result in the DNA sequence reading A C T A. This would give the mRNA code of UGA which is a STOP codon.

**c** T A T

*Answer:* An inversion of the DNA sequence to read A T T will result in the mRNA of A U A which is a STOP codon.

Questions 9.2

RECALL KNOWLEDGE

**1** Define ‘gene flow’.

*Answer:* Gene flow is the movement of genetic material from one population to another.

**2** Describe how migration facilitates gene flow.

*Answer:* Migration is when individuals move between populations and enabling gene flow. The people entering or exiting the population move their genes with them.

**3** List four barriers to gene flow.

*Answer:* Geographical barriers include oceans, mountain ranges, large lake systems, deserts and expansive ice sheets. Sociocultural barriers include economic status, educational background and social position.

**4** Describe how religion may be a barrier to gene flow.

*Answer:* Some religions do not allow marriages outside the religion. This will stop gene flow.

APPLY KNOWLEDGE

**5** Explain why geographical barriers have less influence on gene flow in today’s populations than in previous times.

*Answer:* In previous times the only way to migrate was to walk or use simple machines. Smaller distances would be covered in a longer period of time. In today’s world we can fly vast distances in a comparatively short period of time, or use machines (boats, cars, trains) to travel vast distances.

**6** Describe an example where barriers to gene flow would be:

**a** an advantage

*Answer:* If a population had a high incidence of a lethal recessive gene, it would be a benefit to limit gene flow so as to not influence the gene pools of other populations.

**b** a disadvantage.

*Answer:* A major disadvantage to a barrier to gene flow is the in-breeding that can occur in small isolated populations. As such the frequencies of unusual characteristics can be much higher than in larger populations that exhibit gene flow.

Questions 9.3

RECALL KNOWLEDGE

**1** Define ‘natural selection’.

*Answer:* The process by which a species becomes better adapted to its environment; those individuals with favourable characteristics have a survival advantage and so pass on those characteristics to subsequent generations.

**2** Describe the phenotype of individuals who have each of the following genotypes for sickle-cell anaemia:

**a** homozygous

*Answer:* The individuals will have red-blood cells that, when oxygen is removed or lowered, will change shape to a sickle-shape.

**b** heterozygous.

*Answer:* The individual will show no ill-effects, unless oxygen is in limited supply, in which their red-blood cells will show mild sickling.

**3** List the steps involved in evolution through natural selection.

*Answer:*

* Variation exists in the species
* More offspring are produced than can possibly survive to maturity
* There is a struggle for existence
* Individuals with characteristics best suited to the environment have a better chance of surviving and reproducing
* Favourable characteristics are passed on to the next generation

**4** Describe what a selective agent is, and give an example.

*Answer:* A selective agent is the environmental factor acting on the population. Examples include malaria or tuberculosis.

APPLY KNOWLEDGE

**5** Explain why variation is crucial for natural selection.

*Answer:* Variation is crucial because it provides different physical characteristics for the selective agent to act on. If there was no variation in a population, the selective agent could act to remove all members of the species.

**6** Use a flow chart to summarise the history of our understanding of natural selection.

*Answer:*

**7** Describe how natural selection may have led to an increased occurrence of Tay-Sachs disease in areas where tuberculosis occurs.

*Answer:* Individuals who were heterozygote for Tay-Sachs had a resistance to tuberculosis. They were able to survive and reproduce, passing the Tay-Sachs allele on to subsequent generations. An individual who is homozygote for Tay-Sachs dies in childhood, and an individual who does not have the Tay-Sachs allele will likely die from exposure to tuberculosis. As such, over time, the allele for Tay-Sachs is maintained in populations where tuberculosis is prevalent.

**8** Suggest the selective agent for each of the following characteristics.

**a** Bacteria becomes resistant to antibiotics.

*Answer:* Antibiotics

**b** Inuit people tend to be short limbed and long bodied.

*Answer:* Temperature

**c** Prickly pear cacti have thorns on their flesh.

*Answer:* Water availability

**9** Explain how malaria can lead to an increased frequency of the allele for sickle-cell anaemia.

*Answer:* Individuals who are heterozygote for sickle-cell anaemia (have sickle-cell trait) have a resistance to malaria. They do not suffer any ill-effects from sickle-cell trait and as such are able to survive and reproduce, passing the sickle-cell allele on to their offspring. The heterozygote offspring will also be resistant to malaria. The presence of the selective agent of malaria acts to increase the prevalence of the sickle-cell allele.

**10** Use a Venn diagram to compare and contrast alpha thalassemia and sickle-cell anaemia.

*Answer:*

Questions 9.4

RECALL KNOWLEDGE

**1** Define:

**a** genetic drift

*Answer:* The occurrence of characteristics in a small population as a result of chance rather than natural selection.

**b** founder effect

*Answer:* A type of genetic drift where a new population is formed by a small number of individuals. The founding population shows marked allele differences from the original population.

**c** bottleneck effect.

*Answer:* An extreme form of genetic drift that occurs when the size of a population is severely reduced due to a sudden event such as a natural disaster. The allele frequencies of the survivors may not reflect that of the original population.

**2** Explain how genetic drift is different from natural selection.

*Answer:* Genetic drift is random and non-directional change in allele frequency in a population. It occurs by chance and does not require a selective agent to drive the change in allele frequency. Random genetic drift is observed in small populations.

**3** Use an example to explain how the founder effect may result in a population having different characteristics from another population of the same species.

*Answer:* The Dunkers in Pennsylvania originally came from Hesse, Germany. They do not marry outside of their group due to their religion, and as such are an isolated population within the total population of the United States. Traits including mid-digital hair, left and right handedness, ABO, Rh and MN blood groups were studied, showing the Dunkers displaying a greater variation in allele frequency form the present-day population of Hesse, and also from the surrounding Pennsylvanian population. This is not attributed to natural selection, as there is no difference in environmental factors where the Dunkers and the surrounding American population live.

APPLY KNOWLEDGE

**4** Explain why genetic drift is unlikely to have a significant effect on the allele frequency of a large population.

*Answer:* Genetic drift occurs in large populations but is more pronounced in small populations because they have less variation and a lower ability to respond favourable to changing conditions. Small populations will change quickly in fewer generations. Genetic drift does not take into account allele benefit or harm; therefore, in a small population, beneficial alleles may be lost, or a harmful allele may become fixed purely by chance. This would not occur in a larger population.

**5** Explain how it is possible that bushfires that affected areas of Victoria and New South Wales in early 2020 may reduce the genetic variation in koalas.

*Answer:* This is an example of the bottleneck effect, where a large percentage of a population is lost due to a natural disaster. The remaining koalas now make up the founding population of the areas. Due to the reduced number, there is reduced genetic variation in the population.

Questions 9.5

RECALL KNOWLEDGE

**1** Define ‘speciation’.

*Answer:* The process of producing two species.

**2** List the steps involved in speciation.

*Answer:*

* Variation
* Isolation
* Selection
* Speciation

**3** List three situations that may lead to isolation of groups.

*Answer:* A geographical barrier including an ocean, large lake system or mountain range may split a species. Reproductive isolation may occur from sexual selection, or sociocultural reasons including religion, language or economic status.

APPLY KNOWLEDGE

**4** Explain why variation is necessary for speciation.

*Answer:* Variation is needed for speciation so that when the populations are split and different selective agents act on the populations, different traits are selected for, changing the gene pools of the split population.

**5** Do you think it is likely that humans will form a new species in the future? Explain your answer.

*Answer:* Student responses will differ.

Affirmative students may say:

Yes, as the sociocultural divide increases between developed and developing countries, those with greater access to medical care may live longer, reproduce and pass genes on to their offspring. Those without the same access to medical care will die, potentially not passing on their genes to subsequent generations. This forms a sociocultural barrier to limit gene flow.

Students opposing this may say:

No, due to geographical barriers no longer posing the same limitation to gene flow. People are able to migrate and interbreed more than ever before, increasing the variation in these populations and limiting the isolation.

**6** Is mutation, natural selection or genetic drift the most important process in speciation? Justify your answer.

*Answer:* Mutation is the most important. Mutation provides a source of variation, without variation natural selection and genetic drift will have no differing effects leading to speciation.

**7** Would two groups of a species that are isolated in environments that are similar form new species? Explain why or why not.

*Answer:* It is unlikely they would form new species, as the selective agent/environmental factor is similar. The selective agent would select the same traits to survive so no change would occur in the isolated gene pools.

Chapter 9 Activities

ACTIVITY 9.1 Investigating the effect of ultraviolet radiation on *Saccharomyces cerevisiae*

Discussion

**1** What is your independent variable?

*Answer:* The independent variable is the time the plates are exposed to light.

**2** What is the range of your independent variable?

*Answer:* The range of the independent variable is 0–20 minutes.

**3** What is your dependent variable?

*Answer:* The dependent variable is the amount of damage is done/ amount of growth observed.

**4** What are your control variables and how did you control them?

*Answer:* Temperature and time of incubation are control variables. These are controlled by incubating all plates together.

**5** What type of mutation does the UV-sensitive yeast portray?

*Answer:* UV-sensitive yeast portrays a physical mutagen that occurs as a result of ionising radiation (UVB) and naturally occurring mutagens. UVB can break the chemical bonds in DNA, making it mutagenic and carcinogenic. The most common result of UV radiation is the production of pyrimidine dimers (cross-linked nucleotides) which prevent normal replication and transcription; affecting both gene products and the cell cycle.

**6** Compare your results with others in your class. Were the results consistent?

*Answer:* Student answers will vary. However, inconsistencies in the results are likely to occur due to human error. There may be variation in the inoculation procedure and exposure time. Incidences of cross contamination will also affect the results.

**7** Did your experiment support or refute your hypothesis, or were your results inconclusive?

*Answer:* Student answers will vary.

**8** Based on your findings, how does UV light impact the two different yeast strains? Do they differ? If they do, explain why.

*Answer:* Based on the results of the experiment, students should surmise that the UV-sensitive yeast sustains greater damage as a result of UV light exposure in comparison to wild-type Saccharomyces cerevisiae. The UV-sensitive yeast strain does not incorporate all the genes necessary for effective repair of photochemically damaged DNA. As a result, the exposure to sunlight kills this strain of yeast more quickly than wild type *Saccharomyces cerevisiae*.

**9** Suggest how your findings might relate to evolution.

*Answer:* The results of this experiment show how different strains of yeast have evolved to survive in different environmental conditions.

ACTIVITY 9.2 Venusians: Investigating natural selection

**Interpreting your data**

**1** How has this activity shown that mutations that increase an individual’s chances of survival and reproduction 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 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.

ACTIVITY 9.3 Modelling natural selection

**Studying your results**

**1** Which colour frog became the most frequent in the population? Why do you think this was the case?

*Answer:* Responses may vary, however one would expect the green frogs to be the most frequent, because they experienced less predation.

**2** Which colour frog was eliminated first? Explain why this occurred.

*Answer:* The yellow frogs would be expected to be eliminated first, because they were exposed to the greatest amount of predation.

**3** Compare your results with other groups in the class. Have all groups obtained similar results? How much variation was there in the results between the different groups?

*Answer:* One would expect the green frogs to be the most frequent in all groups. There would be variation between the groups because the colour of offspring, and the colours taken by predators, depend on the number displayed when the die is thrown – that number is a matter of chance.

**Interpreting your results**

**1** How has this activity modelled the process of natural selection? In your answer, describe what was creating the selection pressure on the population of frogs.

*Answer:* This activity has modelled natural selection as there was variability in the colour of the frogs and there was different selection pressure dependent on their colour. That is, the chance of survival (and reproduction) depended on the colour of the frog. The selection pressure for the frogs was the predation by birds.

**2** Explain why there was variability, if any, between the groups in your class.

*Answer:* There was variability due to chance (that is, rolling of the die determined the colour of offspring and the colour taken by predators).

**3** What changes would you have to make to predation by the water birds to achieve a completely orange population of frogs? Repeat the activity with your changed parameters. Was your prediction correct?

*Answer:* You would need to change the rate of predation. In this case, make the orange frogs the least preyed on, and increase the predation on the green frogs.

**4** Over several generations, what would happen to the composition of the frog population if water birds preyed equally on the three frog colours?

*Answer:* If predation were equal across the three different colours of frog, then the proportions of different colours in the frog population would be expected to remain the same. In this situation there would be no natural selection – all colours of frog would be selected equally.

**5** Write a summarising paragraph, using the principles of natural selection, to link the breeding patterns of the frogs and predation by water birds.

*Answer:* The paragraph would need to include such things as:

• the variation in frog colour

• the differing rates of predation by birds

• the selective advantage of a particular colour

• the difference in survival rate

• that only frogs that survive long enough to reproduce are able to pass on their favourable alleles to the next generation.

Activity 9.4 Investigating sickle-cell haemoglobin

**Interpreting the results**

**1** What was Allison’s dependent variable? What was his independent variable?

*Answer:* Allison’s dependent variable was whether the participant did or did not develop malaria, while the independent variable was the presence or absence of the sickle-cell allele.

**2** What factors did Allison appear to control in his experiment?

*Answer:* Age; African (where from); no previous malarial infection; sex (male); 40-day observation time; none of the volunteers had been in an area where malaria occurred for at least 18 months; all inoculated with malaria; all inoculated at around the same time.

**3** Which group of subjects was the control group, and which the experimental?

*Answer:* Control group: lacking a sickle-cell allele

Experimental group: with a sickle-cell allele

**4** Did Allison’s results support his hypothesis? Explain why you think so.

*Answer:* Yes. The majority of those with the sickle-cell allele did not develop malaria, whereas a majority of those without the sickle-cell allele did develop malaria.

**5** Do the results Allison obtained suggest a reason why the sickle-cell allele has survived in Africa?

*Answer:* Yes, because this is a region where malaria is prevalent, and the presence of the sickle-cell allele, although a disadvantage in most parts of the world, appears to have a selective advantage where malaria is prevalent.

**6** Refer to Figures 9.21 and 9.24. Does the information provided in these figures support your answer to Question 5? Give reasons for your answer.

*Answer:* Yes. These figures indicate that in areas where malaria is still common, sickle-cell anaemia is still prevalent, or, there is a close correlation between the parts of the world where malaria is endemic and the areas where sickle cell anaemia occurs.

**7** Explain how the high incidence of the sickle-cell allele in parts of Africa could be considered an example of natural selection.

*Answer:* In parts of Africa where there is still a high frequency of the sickle-cell allele, the people who survive malaria are usually those in possession of a sickle-cell allele. Therefore, when they reproduce they pass on this advantageous allele. Those who do not have this allele may contract malaria and die, thus removing alleles for normal red cells from the population. This is a good example of natural selection in human populations. Because individuals with two alleles for sickle-cell anaemia may die from the condition, and individuals without a sickle cell allele may die of malaria, it is the heterozygotes that have the best chance of survival.

**8** Would a university ethics committee today be likely to approve an experiment such as the one that Allison performed? Give reasons for your answer.

*Answer:* It is highly unlikely that a university ethics committee today would approve Allison’s experiment, because there was little supporting evidence at the time for his hypothesis. To do such an experiment today, Allison would have to explain the risks and benefits of the experiment to all the participants, any possible long-term effects and then obtain their informed consent.

Chapter 9 Review questions

Recall

**1** 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.

**2** 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.

**3 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** List the ways that the DNA may be changed in a mutation.

*Answer:*

* A gene may be altered by a point mutation where a single base is substituted, inserted or deleted.
* More than one gene may be altered by a chromosome mutation where sections of DNA may be deleted, duplicated, translocated, inverted or non-disjunction may occur.

**c** 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.

**d** Give an example of a congenital disorder 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 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).

**5** 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.

**6 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

**7** Briefly describe the significance of the founder effect in human evolution.

*Answer:* The founder effect is significant in studies of human evolution because, where a new isolated population is begun by a small number of people, the frequency of alleles in that small population may be quite different from the original population. The characteristics of the newly founded population will then be different from the original population.

Teachers may wish to point out that as humans spread around the world, there would have been many small founding populations, which would have contributed to differences between peoples in different areas.

**8 a** Define ‘gene flow’.

*Answer:* The movement of genetic material from one population to another.

**b** List the common barriers that may lead to the isolation of one gene pool from another, and give examples of each type.

*Answer:*

• Geographical barriers: oceans, mountains, lakes, deserts

Students may quote many examples such as the Sahara Desert separating populations in northern and southern Africa, or the Pacific Ocean separating peoples in the Americas from Australia and Asia.

• Sociocultural barriers: education, language, religion, social position

Students may quote many examples such as Jewish people in Israel marrying within their own faith or the Basque people of the Pyrenees being united and separated by their language and other aspects of culture.

**c** List five different kinds of sociocultural barriers to gene flow, and describe how each is thought to act.

*Answer:*

• Education: People tend to marry and/or reproduce with someone from similar educational backgrounds.

• Religion: People tend to marry and/or reproduce with someone with the same religious beliefs.

• Language: It is more likely that people will meet and marry and/or reproduce with someone who speaks the same language.

• Economic status: People tend to marry and/or reproduce with someone of the same economic status.

• Social status: People tend to meet and marry and/or reproduce with someone with the same social status.

• Ethnic group: People are more likely to meet and marry and/or reproduce with someone within the same ethnic group.

In all these cases people are more likely to meet a potential partner who has similar education, religion, language and so on, rather than individuals who are quite different. It is human nature to socialise with people who are similar to us.

**9** Outline the main points of Darwin’s theory of natural selection. Include an explanation of the terms ‘struggle for existence’ and ‘survival of the fittest’.

*Answer:* The principles of evolution through natural selection include the following:

• All members of a species vary.

• Variations are passed on from one generation to the next.

• Organisms reproduce at a rate far greater than that which their available food supply and other resources allow, leading to competition for survival. This may be referred to as a ‘struggle for existence’.

• More organisms with favourable characteristics survive and reproduce, passing on their favourable alleles. Many of those with unfavourable characteristics die before they have an opportunity to reproduce and do not pass on their unfavourable alleles. This may be known as ‘the survival of the fittest’.

• The proportion of favourable alleles in the gene pool gradually increases.

Explain

**10** 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.

**11** 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.

**12 a** Explain what random genetic drift is.

*Answer:* Random genetic drift is when characteristics of members of a population occur by chance, rather than by natural selection. It only occurs in small populations that are genetically isolated.

**b** Select a modern population in which genetic drift is thought to have had an effect and describe why this might be the case.

*Answer:* One example of random genetic drift in a modern population is the Dunker population in North America. Individuals are not allowed to marry (or reproduce) outside their own population, with its particular beliefs. This has led to a small genetically isolated population.

Students may also quote isolated populations of Australian Aborigines, Pitcairn islanders, the Finns and the inhabitants of Tristan da Cunha.

**13** Using the example of Tay-Sachs disease, explain how genetic diseases can lead to changes in allele frequencies in a population.

*Answer:* Genetic diseases may result in changes to allele frequencies in a gene pool. An allele causing an inherited, fatal disease would be expected to be gradually eliminated from a population because people with the allele would die and would not pass it on to the next generation. In the case of Tay-Sachs disease, the heterozygous form is thought to have a selective advantage in areas where TB is present. People with one copy of the allele are more likely to survive and reproduce and to pass the allele on to future generations. People who do not have the allele are more likely to die from TB.

**14** People of short stature tend to live in cold climates, and people with long limbs and short torsos tend to live in hot climates. Explain how these adaptations to cold and hot environments could have come about.

*Answer:* Originally, there would have been a range of variants for stature in all human populations. Through natural selection, those with characteristics that best suited them to the climate in which they lived would have survived and reproduced, passing on their favourable alleles to their offspring. People of short stature with a smaller surface area in relation to body volume lose heat at a slower rate giving them a survival advantage in cold climates. Those with long limbs and short torsos have a survival advantage in hot climates because they have a larger surface area for heat loss.

**15 a** What is sickle-cell anaemia?

*Answer:* Sickle-cell anaemia occurs when the red blood cells have an abnormal crescent or sickle shape.

**b** Explain why sickle-cell anaemia is usually lethal.

*Answer:* Sickle-cell anaemia is usually lethal because the sickled red blood cells clump together causing organ damage and decreasing life expectancy.

**c** List the advantages and disadvantages of having the sickle-cell trait in an area where malaria is prevalent.

*Answer:*

|  |  |
| --- | --- |
| **Advantages** | **Disadvantages** |
| Malarial resistance | Red cells sickle when oxygen concentration is low  Complications from sickled cells |

**16** How could isolation lead to selection and speciation?

*Answer:* Isolation over a considerable period of time may result in a population undergoing many changes in allele frequency through natural selection, as well as the possibility of genetic drift and mutation. This may result in that population becoming so different from the original that it is no longer able to reproduce with the original population. It has become a different species.

Apply

**17** Why does special care need to be taken when pregnant women 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.

**18** 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.

**19** 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.

**20** 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.

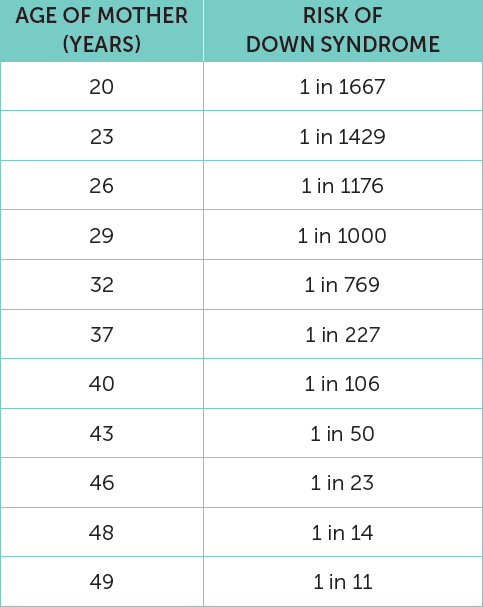
*Answer:* The average number of sperm in just one ejaculation is more than 100 million. A female foetus has several hundred thousand primary oocytes at birth. Once sexual maturity is reached, usually only one egg completes meiosis in each monthly cycle. Scientists have therefore proposed this hypothesis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

**21** 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.

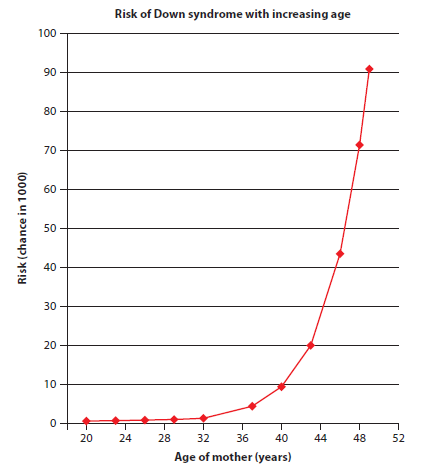
**22** The risk of having a baby with Down syndrome increases as the mother gets older. The following table shows the relationship between Down syndrome and maternal age.

Mother’s age and risk of having a baby with Down syndrome



**a** Draw an appropriate graph to display the data in the table.

*Answer:*

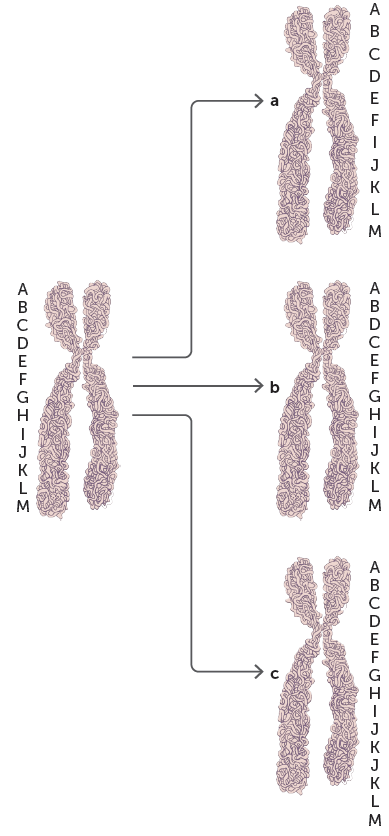


Students may draw a column graph, which is perfectly acceptable.

**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.

**23** The following figure 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?



*Answer:*

**a** Deletion

**b** Inversion

**c** Duplication

**24** During the 14th century, plague epidemics drastically reduced the human population of Europe. Use this as an example to describe the way natural selection operates so that only the fittest tend to survive.

*Answer:* There is variation in all human populations and those of 14th century Europe would have had some individuals who possessed alleles that gave them a survival advantage when exposed to the micro-organism causing the plague epidemics. These people would have survived the epidemics and passed on their favourable alleles to their offspring. Those that died did not possess alleles that gave them a survival advantage and could not pass on their alleles. Thus, in this case, the fittest individuals survived.

**25** According to a recent report, 13% of Scotland’s population are redheads. Two out of every five Scots carry the allele for red hair. However, only 2% of the world’s population are estimated to be natural redheads.

**a** Suggest a reason for the high frequency of the allele for red hair in the gene pool of the Scots.

*Answer:* The original reason for the high frequency of the allele for red hair in the Scottish population may have been the founder effect or random genetic drift. The high frequency of the allele has been retained because Scots would tend to reproduce with other Scots.

**b** In the population of Scotland, what do you think will happen to the frequency of the allele for red hair over time? Give reasons for your answer.

*Answer:* The frequency of the allele for red hair will probably stay about the same because there would be no selective advantage conferred by the red hair allele and people living in Scotland are more likely to meet and marry other people living in Scotland. If there were a significant migration into the Scottish population, then the allele frequency would change.

**26** A team of American scientists has been trying to develop a vaccine to give permanent immunity against malaria. What do you think will happen to the frequency of the sickle-cell gene within a population if this vaccine is effective? In writing your answer, ensure that you explain the adaptive value of the various genotypes and the selection pressures on each.

*Answer:* If an effective vaccine were to be discovered to prevent malaria, one would expect the frequency of the sickle-cell allele to decrease as it would no longer have a selective advantage. Genotypes would reflect the decrease in selective pressure that previously favoured the retention of the sickle-cell allele. Those that possess two of the sickle-cell alleles would still die from sickle-cell disease, and those with two normal alleles would no longer succumb to malaria. Their survival would increase the frequency of the normal allele for haemoglobin in the population. The heterozygotes would no longer have a survival advantage and, over time, the frequency of the sickle-cell allele would decrease and possibly be eliminated from the population altogether.

Extend

**27** Western Australia has been a world leader in the application of carrier detection to reduce the incidence of Duchenne muscular dystrophy.

**a** What does carrier detection involve?

*Answer:* A blood sample, swab from inside the mouth or a saliva sample will be taken and screened for the mutations in the DNA that are known to be associated with Duchenne muscular dystrophy. They are used to determine whether an asymptomatic person may have the irregular gene sequence associated with the disease.

**b** What takes place following the detection of a carrier?

*Answer:* Genetic counselling would occur, informing the individuals of the results of the carrier test and the potential risks in conceiving children naturally.

**c** What is preventing the complete elimination of Duchenne muscular dystrophy?

*Answer:* It is due to the heterozygote nature of the X chromosome where the mutation for DMD can be found. Female carriers are usually asymptomatic so unless there is a family history of the disease, they are unlikely to be tested.

**28** Malthus claimed that species of organisms always produce more offspring than the existing resources can support. Is this true of the human species in the past or at present? Is it likely to be true of the human species in the future?

*Answer:* This claim by Malthus was probably true for the human species in the past, especially in traditional hunter-gatherer societies. The development of agriculture meant that populations were able to increase without starvation occurring. Technological advances such as the green revolution have enabled the human species to keep up with increasing demand for food. In some regions of the world where resources are very limited, or are limited at particular times through drought, famine and floods, the population does exceed existing local food resources.

In the future, ever-increasing population growth could result in more offspring being produced than resources could support. However, technology may keep pace with population growth in ways that are yet to be imagined.

**29** Describe the barriers to gene flow that exist for the following populations:

**a** groups in South Africa

*Answer:* Mountains; deserts; cultural barriers (for example, different tribal groups tend not to intermarry).

**b** groups in the islands of Polynesia, such as New Zealand, Tahiti and Hawaii

*Answer*: Ocean barriers.

**c** Jewish people.

*Answer*: Sociocultural barriers; in this case, religion.

**30** Using analysis of mitochondrial DNA, researchers have determined that all humans are descended from a woman who lived in Africa 200 000 years ago – the so-called mitochondrial Eve. If we are all descended from a common ancestor, how is it that there are so many different types of humans today? Describe the processes that must have taken place to produce the differences between present-day groups of humans.

*Answer*: Even though present-day humans may have all descended from a common ancestor, dispersal into different environments exposed those migrating groups to different environmental selective pressures. Natural selection would have operated on these groups as they settled in differing environments, favouring different allele combinations and leading to different phenotypes. In small populations, especially in the case of small groups of hunter-gatherers, genetic drift may have resulted in different allele combinations. Similarly, the founder effect may have influenced the characteristics of many of the populations in the small islands in the Pacific Ocean. In the past, isolation and a lack of gene flow over considerable periods of time would have resulted in distinctive phenotypes.

**31** Speculate on what might be the long-term effect on allele frequencies if a mutation suddenly produced a favourable allele that gave a natural resistance to all forms of heart disease.

*Answer:* Over time, the allele frequency for natural resistance to all forms of heart disease would be selected for and increase significantly within the population. This would see a gradual decrease in the incidence of the disease, as those dying from the disease would have their alleles removed from the gene pool. However, if death occurred after an individual had reproduced and had passed on their unfavourable alleles, the speed at which the allele frequencies changed would be very much reduced.

**32** One of the best-researched investigations into natural selection is the work of a British geneticist, Henry Bernard Davis Kettlewell, on the peppered moth, *Biston* *betularia.* The peppered moth gets its name from the scattered dark markings on its otherwise pale wings and body. The moth flies at night and rests by day on tree trunks. These trunks are usually encrusted with lichens, and the pale-coloured moth is practically invisible against this background.

However, in 1849, a coal-black mutant form of the moth was found near Manchester in England. Within a century, this black form had increased to 90% of the population in this region. The change in allele frequency that occurred in this example is a good model of how natural selection takes place. Find out:

**a** how the black form of the moth became the more prevalent variant

*Answer:* The black variant became more prevalent because the environmental factor the moths lived with, selected for the black mutant form. The lighter coloured moths were more easily seen against the darkened walls and tree trunks of the city in Manchester and were eaten by predators (birds). The black variants were better camouflaged and were able to survive to reproduce with other black variants passing the black trait onto the subsequent generations.

**b** which form is the most prevalent today.

*Answer:* Due to controls to reduce air pollution and to improve air quality, the tree trunks became lighter and lichen growth increased. The normal Peppered Moths were camouflaged and the black variants were more noticeable. The normal Peppered Moths have become more common than the black forms.