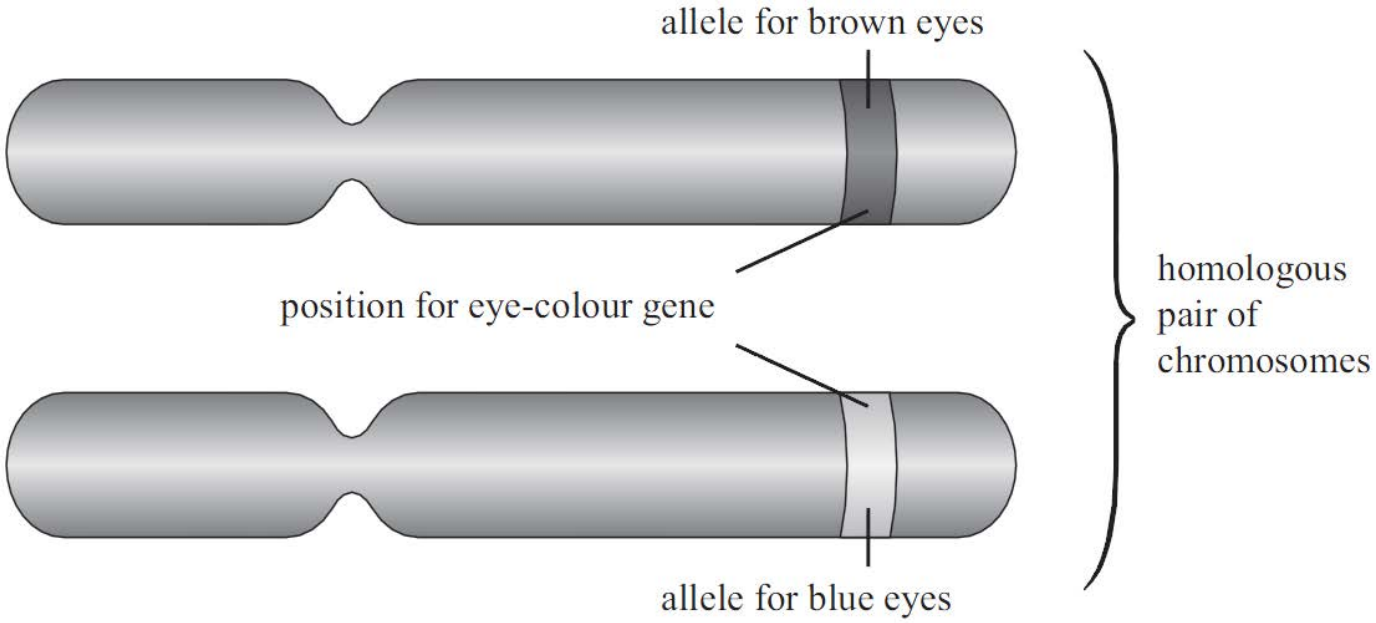
Collated questions

Demonstrate understanding of biological ideas relating to genetic variation

DNA STRUCTURE

THE ROLE OF DNA IN INHERITANCE (2013:2)

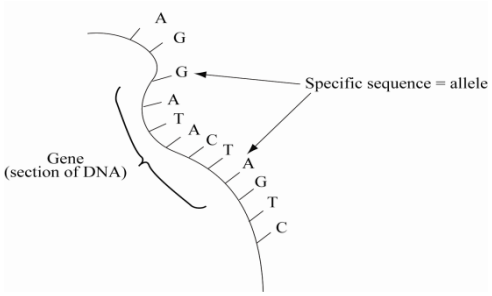


(a) Use the diagram above to help you explain the relationship between chromosomes, genes, alleles, phenotype, genotype, and the molecule DNA.

A labelled diagram may assist you.

Chromosomes are made up of DNA. DNA is a large molecule that is coiled into a double helix (twisted ladder structure). It is responsible for determining the phenotype of an organism. Along this molecule are bases. These bases pair up; A always pairs with T, and G with C.

A sequence of bases which codes for a particular trait (eg, eye colour) is called a gene.



The different versions of each gene are called alleles, and these show the different variations of each characteristic, eg brown / blue eyes. Because chromosomes come in pairs for each trait, there will be two possible alleles. These different versions of genes (alleles) occur as the DNA base sequence is different.

This combination of alleles for each trait is called the genotype; this can be any combination of two of the available alleles. The genotype determines the phenotype (the physical appearance) of the organism. Whichever alleles are present may be expressed. Dominant alleles (B) will be expressed over recessive alleles (b).

(b) The allele for brown eyes (B) is dominant over the allele for blue eyes (b) in humans.
Discuss how it would be possible for a child to have blue eyes, even though both their parents have brown eyes.

In your answer you should:

- use labelled Punnett squares
- link the genotypes and phenotypes of the child, parents, AND grandparents.

For the child to have blue eyes they must have a genotype of bb (ie have both recessive alleles). If a dominant allele, B is present then brown eyes would be seen.

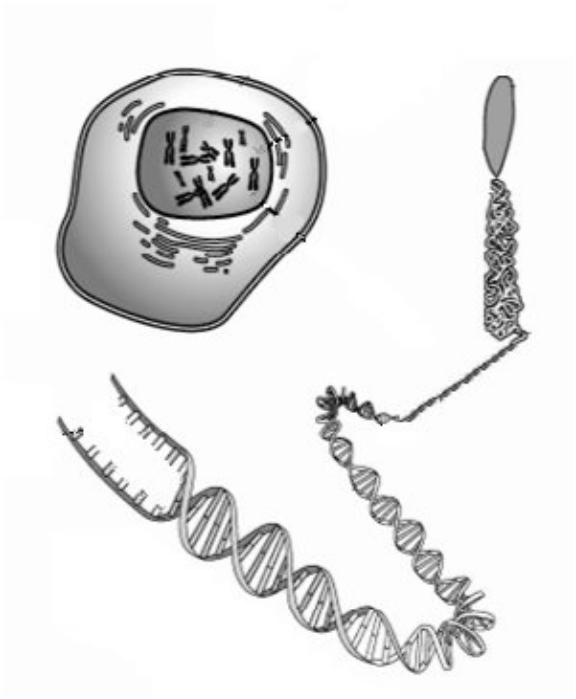
	B	b
B	BB	Bb
b	Bb	bb

In order to have a genotype of bb, each parent must have given a b (recessive allele). Both parents have brown eyes so therefore they both must have a dominant allele (B) and because each parent passes on a recessive allele the genotype of each parent must be Bb. The grandparents could have a genotype of bb, Bb, or BB. It is not possible to say for sure, but at least one of the grandparents on each side must pass on a recessive allele (b) in order for each parent to have a recessive allele to pass on to the child. Punnett square(s) may be used to show this but must be explained.

GENETIC STRUCTURE (2012:1)

The diagram below shows the relationship between chromosomes, genes, and DNA (deoxyribonucleic acid).

(a) Explain the relationships between DNA, chromosomes and genes.
You may add notes and labels to the diagram above to support your answer.

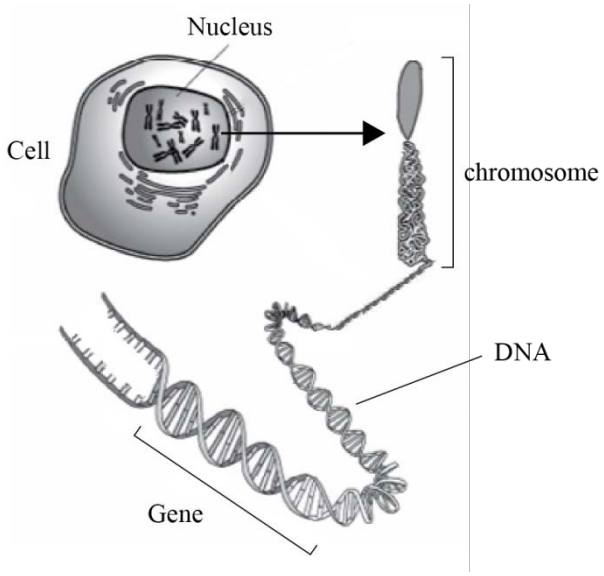


DNA – contains the genetic instructions (used in the development and functioning of proteins).

Gene – a section of DNA that codes for a particular protein / feature/characteristic.

Chromosome – an (organised) structure of DNA (found in the nucleus of a cell).

DNA is the heredity material of the cell which is found in the chromosomes in the nucleus. These are found as strands each one of these strands of DNA is called a chromosome.



(b) Explain how the relationships in your answer to (a) lead to different characteristics and how this contributes to genetic variation.

A gene is a segment of DNA, found in a small section of the chromosome. Along the DNA, base sequences provide the code for building different proteins, which then determine particular features. Slight differences in the sequence of the bases making up a gene are called alleles and they cause the variations in the phenotypes. These differences lead to genetic variation between individuals.

DNA AND VARIATION (SAMPLE 2011:1)

Genes determine many of the features of organisms, such as the colour of the flowers on a plant.
A gene is a part of a DNA molecule.
The type of plant shown in the photograph below can have red or white flowers.

Red flowers are due to a dominant allele and white flowers are due to a recessive allele.

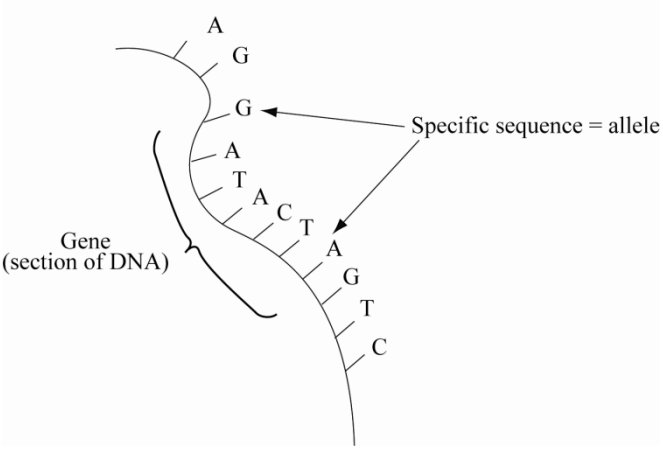
Discuss how information in DNA gives organisms (like the plant above) their individual features, such as red and white flowers. In your answer, you should:

- explain the difference between a gene and an allele
- draw a labelled diagram to show the relationship between a gene and an allele and the structure of a DNA molecule
- explain how the base sequence on DNA determines a particular feature (eg, flower colour) and different forms (variations) of that feature (eg, red and white flowers).

Explanation of difference:

A gene is a section of DNA that codes for a particular feature / protein.

An allele is an alternative form of the gene.

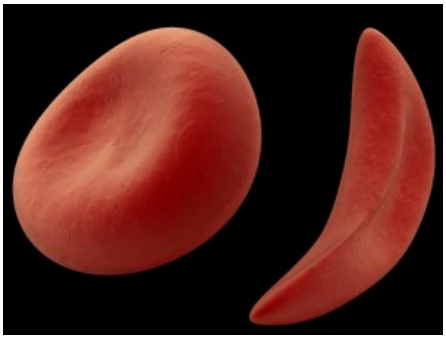


Along the DNA, base sequences provide the code for building different proteins, which then determine particular features. Specific sections of the DNA determine individual features such as flower colour. Variations in the feature (eg red or white flowers) are due to (slight) differences in the sequence of the bases making up a gene. The different variations of a particular gene are called alleles and they cause the variations in the phenotypes.

MONOHYBRID CROSSES

PATTERNS OF INHERITANCE (2012:2)

A blood disorder caused by red blood cells with an unusual curved (sickle) shape is inherited through a single gene with two possible alleles, normal and sickle.



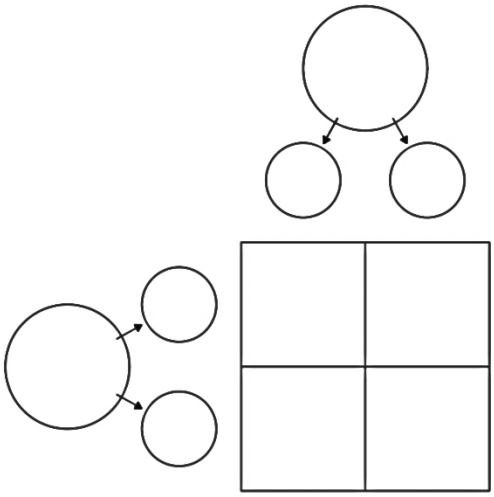
normal red blood cell sickle-shaped blood cell

Use '**H**' to represent the dominant 'normal' allele, and '**h**' to represent the recessive 'sickle' allele.

(a) Explain how two parents with normal blood cells can have a child with sickle-shaped blood cells.

In your answer, you should:

- state the genotype of a child with the sickle-shaped blood cells
- state the genotypes of **both** normal parents
- draw a Punnett square to show how two normal parents can produce a child with sickle-shaped blood cells.



	H	h
H	HH	Hh
h	Hh	hh

Genotype of child with sickle cell anaemia – hh

Genotype of both parents – Hh

(b) The parents in part (a) have four children all with sickle-shaped blood cells. They are expecting a fifth child.

(i) Explain how normal parents could have produced FOUR children with sickle-shaped blood cells.

You should refer to your Punnett square in (a).

Two normal parents Hh x Hh have 1 in 4 (25%) chance of producing a child with sickle cell anaemia by each donating the recessive h allele.

Each child / fertilisation has an equal one in four chance of producing a child with sickle cells. This is because in the process of gamete formation / during meiosis alleles are randomly assorted.

(ii) Explain what the chances are of the fifth child having sickle-shaped blood cells.

Previous conceptions have no effect on future offspring; each is separate event.

Chances of fifth child having sickle cells is still one in four, as previous conceptions have no effect on this child; it is a new random event. Each fertilisation is the result of gamete formation where each of the heterozygous parents may give either the recessive (h) or dominant (H) allele.

SQUASH PLANTS (2011:1)

One trait in squash plants is the colour of the fruit.

White fruit are due to a dominant allele (F) and yellow fruit are due to a recessive allele (f).

(a) Explain the difference between a gene and an allele.

A section of DNA within a chromosome that codes for a trait / phenotype is called a gene.

The gene in this example is squash fruit colour.

An allele is an alternative form of a gene. In this case white or yellow fruit.

Genes can differ slightly in their sequence of bases; these are called alleles.

(b) The alleles for the colour of squash fruit combine to produce THREE different genotypes, but only TWO phenotypes.

Explain how the alleles combine to produce only two different squash colours – white and yellow.

In your answer you should:

- define genotype and phenotype
- state the three different genotypes produced and the phenotype of each.

Two different alleles for squash colour are possible; white (F), which is dominant and yellow (f), which is recessive.

Alleles come in pairs; for each gene there will be two alleles in each plant, one from each parent. The genotype is the combination of these alleles. There are three possible genotypes:

FF homozygous dominant. Ff heterozygous. ff homozygous recessive.

A phenotype is the physical appearance of the genotype. These three genotypes give two possible phenotypes. Because both FF and Ff contain the dominant allele, this will mask any recessive allele and appear as white.

For ff there are two recessive alleles (no dominant to mask), so these plants will have yellow fruit.

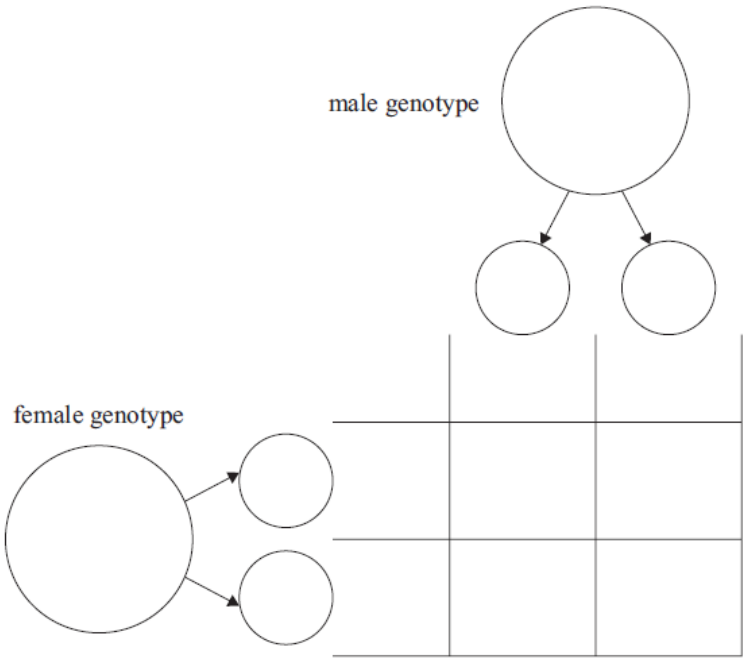
A GIRL OR A BOY? (2011:3)

A couple are expecting their third child. They already have one boy and one girl.

(a) Discuss the likelihood of their third child being a girl.

In your answer you should:

- explain how sex is determined in humans
- complete a Punnett square showing sex inheritance
- explain the relevance of the couple already having children.



Women are XX, so when they create eggs with half the number of chromosomes, both eggs will have an X chromosome. A male is XY so when they create sperm, half will have the X chromosome and half will have the Y chromosome

When the gametes come together (egg is fertilised), there is a 50% probability they will have a baby girl.

		female	
		X	X
male	X	XX	XX
	Y	XY	XY

The sex of the baby is determined by whether it is an X or a Y (sperm) that fertilises the egg.

If it is X it will be female; if it is Y it will be male.

The fact that they already have one girl and one boy has no effect on what the next baby will be. Fertilisation is random at each event, and previous fertilisations have no effect.

(b) One of the parents is a teacher who developed deafness last year as a result of having noisy classes.

Discuss the likelihood of this type of deafness being inherited by the new baby.

In your answer you should consider:

- the reason for the parent's deafness
- what types of characteristics are inherited
- the effects of genetics and the environment on deafness in offspring.

The parent / teacher became deaf because of loud noises related to teaching. Deafness was caused by 'environment', not genetics. (The question makes no reference to him having inherited deafness, nor was he born with it).

Only genetic characteristics can be inherited, not those acquired as a result of environment.

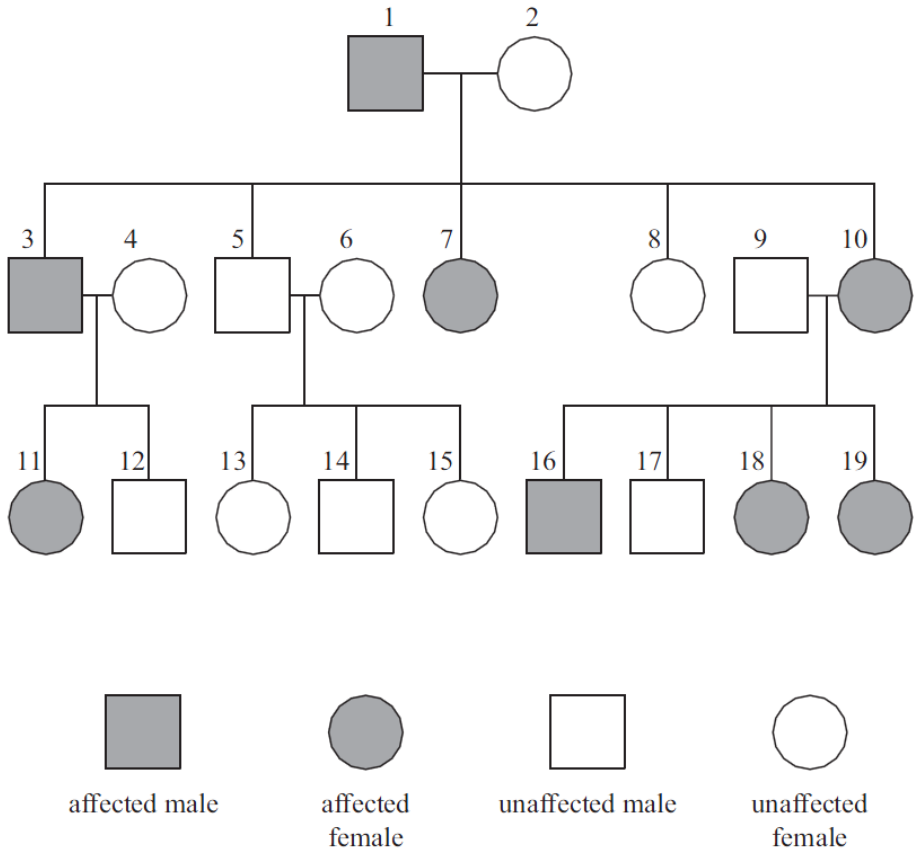
It is unlikely any of his children will be born deaf, as it appears the deafness was caused by environment, not genetics. However, we cannot determine whether they will be deaf at any stage in their life, as deafness can be work-related and it depends on the job they have later in life.

Genetics determines the characteristics you will be born with, but environment then affects these characteristics once you are born.

PEDIGREE CHARTS

PEDIGREES AND PUNNETT SQUARES (2013:1)

Huntington's disease is a genetic disorder in humans. It is caused by a dominant allele (H). The normal allele is recessive (h).



(a) Using H and h, give the two possible genotypes for an individual who has Huntington's disease:

Possible genotypes for an individual who has Huntington's are HH or Hh.

(b) State the genotype of individual 9 in the pedigree chart above. State the genotype of individual 10 in the pedigree chart above. Explain how you worked out the genotype for individual 10. You should support your answer using evidence from BOTH the parents AND children of individual 10.

Genotype of parent 9 (male) is: hh/homozygous

Genotype of parent 10 (female) is: Hh/heterozygous

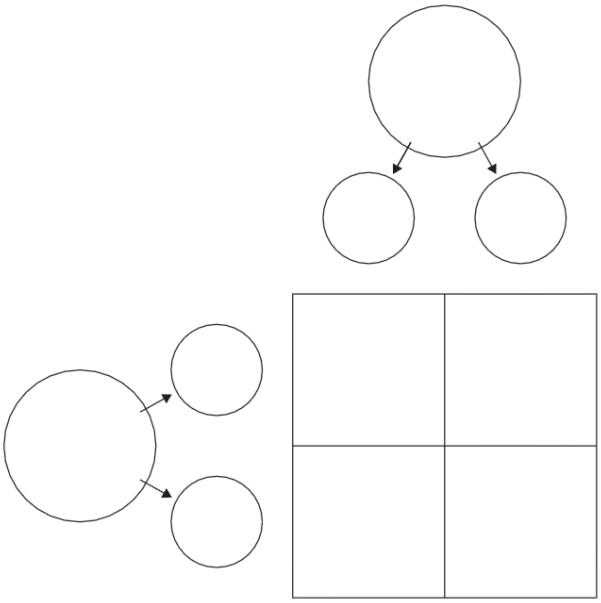
Explains how parent 10 is Hh; eg her parents are Numbers 1 and 2. Her mother (number 2) is unaffected but her father (number 1) has Huntington's disease. Number 2 is hh and number 1 must have at least one dominant allele. Because one allele is inherited from each, parent No. 2 must have given her daughter a recessive allele. She has inherited the dominant allele from her father, No. 1. She cannot be homozygous dominant HH because one of her offspring, No. 17, is not affected by the disease and is therefore hh.

(c) Draw a Punnett square to show the **possible** genotypes of the children from parents 9 and 10.

(i) From **your** Punnett square, predict what fraction of the children would have Huntington's disease and what fraction would not have Huntington's disease.

- Fraction of children with Huntington's disease:
- Fraction of children without Huntington's disease:

	H	h
h	Hh	hh
h	Hh	hh



(ii) Using your Punnett square, show the expected phenotype ratio for the children.

Fraction of children with Huntington's disease is ½ .

Fraction of children without Huntington's disease is ½ . Phenotype ratio is 1:1.

(d) In the pedigree chart the phenotype ratio of Huntington's disease in the children of parents 9 and 10 is not the same as the predicted ratio you have given on the previous page.

Give reasons why the predicted ratio in the Punnett square and the observed ratio in the children may NOT be the same.

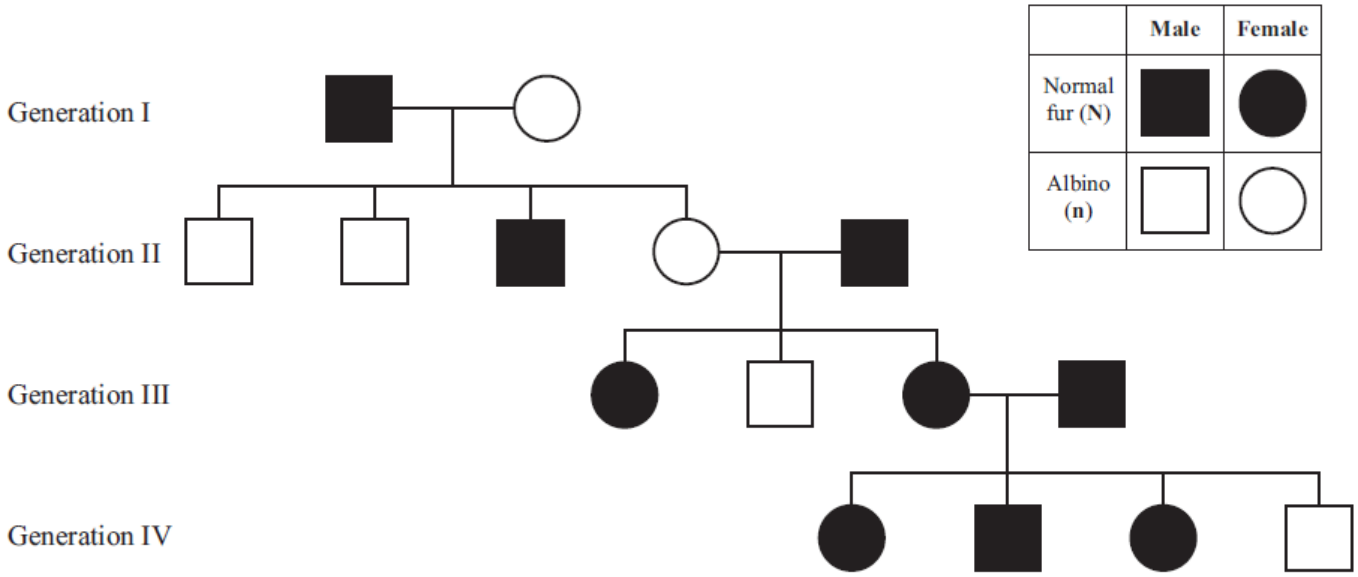
Punnett squares **predict** probable offspring genotypes (thence the expected phenotypes) based on the gametes of the parents. Pedigree charts give the observed (actual) phenotypes. Since each fertilisation is a random event, it is by chance whether the offspring of number 10 inherits the

dominant H allele and therefore has Huntington's or the recessive h allele and does not have the disease. In the pedigree chart 3 of the 4 offspring have the disease but only 2 out of 4 would have been predicted from the Punnett square.

APEING AROUND (2011:2)

Gorillas show an inherited recessive condition called albinism. This results in white fur.

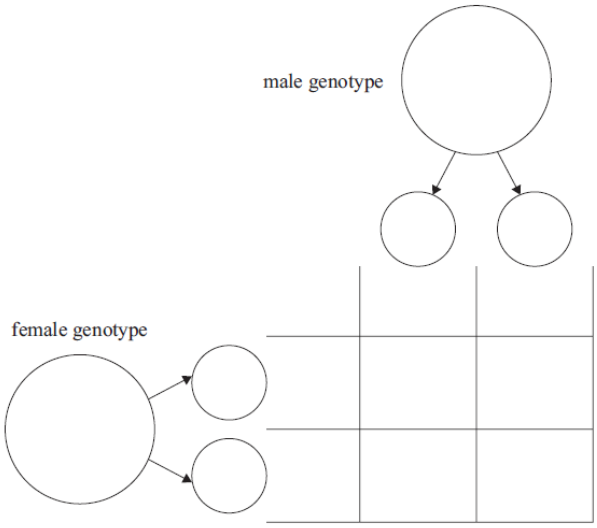
The pedigree chart below shows the inheritance of albinism in a family of gorillas. Normal fur is the dominant allele (N), while albino fur is recessive (n).



(a) Explain how the pedigree chart can be used to show that albinism is a recessive trait.

In your answer you should:

- define the terms dominant and recessive
- state the genotypes of albino and normal gorillas
- complete a labelled Punnett square to support your answer
- explain how your Punnett square shows that albinism is a recessive trait.



Dominant means the trait will be expressed, even if only one allele is present in a pair (heterozygous).

Recessive means the trait will be expressed only if two alleles are present (homozygous). It will be masked in the presence of one dominant allele (heterozygous).

Albinism is a recessive trait. This can be established using Generation III and Generation IV. In Generation III, two normal individuals have three normal offspring and one albino offspring.

The only way this is possible is for Generation III to both be Nn. When two n alleles come together, a homozygous recessive nn (albino) offspring forms.

If normal was recessive, Gen III individuals would be nn. There is no way of forming an individual with N in its genotype.

	N	n
N	NN	Nn
n	Nn	nn

(b) Explain why the genotype of the 3 normal fur offspring in Generation IV cannot be determined, based on the evidence in the pedigree chart and your Punnett square from part (a).

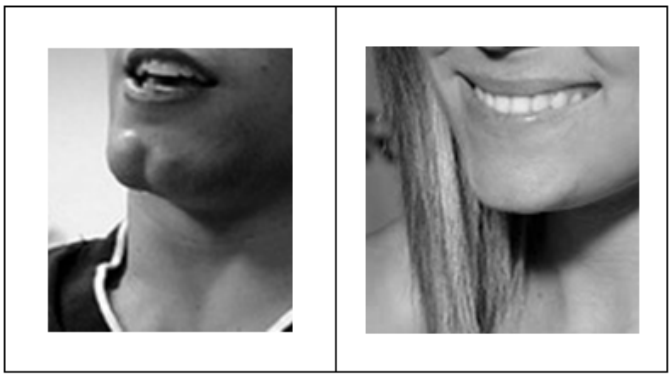
All three Normal fur offspring are either Nn or NN.

Because their parents must have both been Nn to produce albino offspring, there is a 25% chance they are NN and a 50% chance they are Nn.

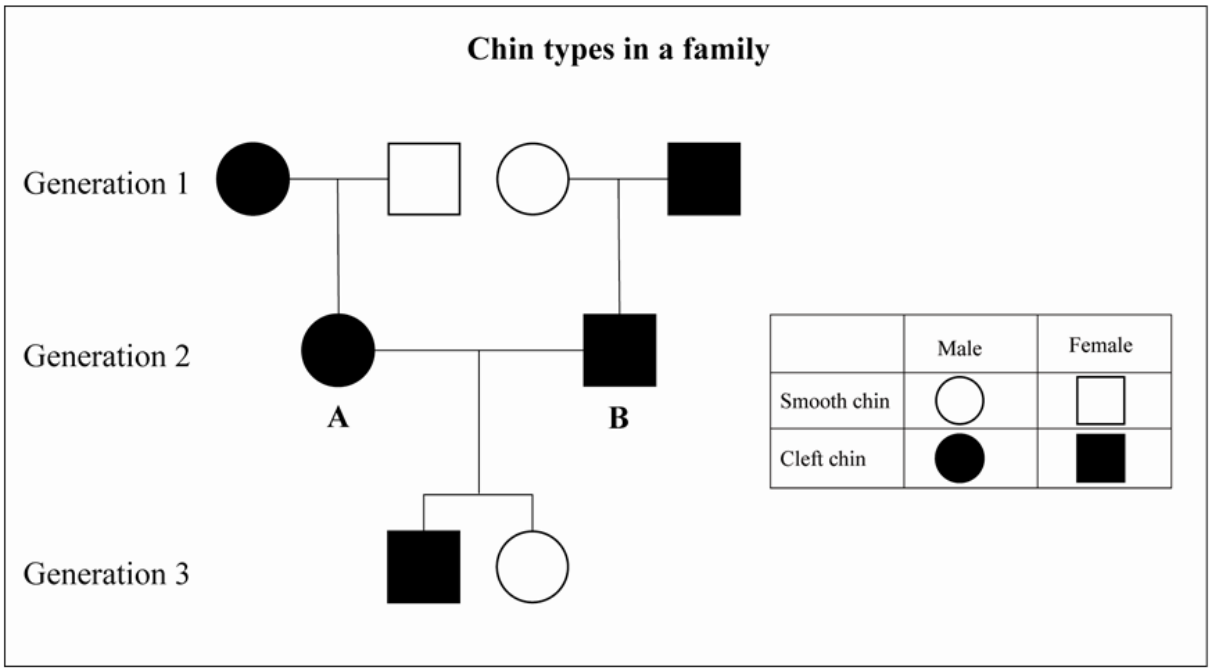
Their genotype cannot be established until they produce offspring.

MONOHYBRID CROSSES (SAMPLE 2011:3)

The allele for a cleft chin (D) is dominant over the allele for a smooth chin (d).



The pedigree diagram shows the chin types in a family.



(a) Explain how evidence in the pedigree diagram on page 6 shows that the cleft chin allele (D) is dominant over the smooth chin allele (d).

In your answer, you should:

- explain what the term dominant allele means
- draw a Punnett square(s) to show your reasoning.

A dominant allele is the form of a gene that is always expressed whether the individual has one or two copies of that allele.

A and B who both have a cleft chin produce a smooth chin child. If the cleft chin allele was recessive they would not be able to produce a child with a smooth allele, so the cleft chin allele must be dominant OR other correct evidence from chart.

(b) Explain why the genotype of male A in generation 2 of the pedigree diagram shown must have the genotype Dd.

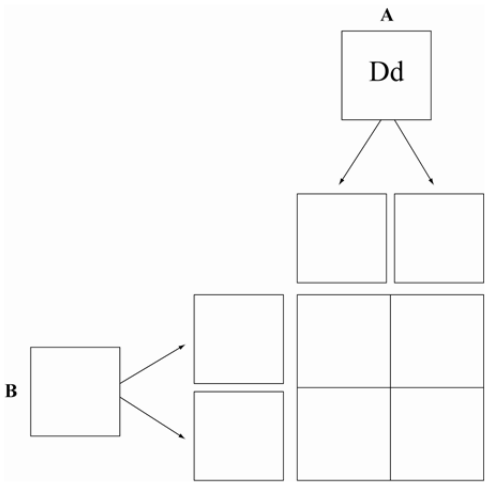
As male A has a cleft chin this must mean that he carries at least one cleft chin allele (D). Male A and female B have a smooth chin boy, which means that A must have passed on a smooth chin allele (d) to the boy as to be smooth chinned the boy must have two recessive alleles, one from each parent.

(c) Individuals A and B, as shown on the pedigree diagram on page 6, decide to have another child. They draw a Punnett square to find what type of chin their child might have.

Explain why the result predicted by the Punnett square may not accurately tell them what type of chin their child will have.

In your answer, you should:

- draw a Punnett square to show the cross between individual A and individual B
- determine the probabilities of the child having a cleft chin and having a smooth chin
- explain why the ratio of children born into the family with cleft and smooth chins may not match the probabilities.



Probability of cleft chin = ¾ or 75% or 3 out of 4

Probability of smooth chin = ¼ or 25% or 1 out of four

Explanation of discrepancy: Random fertilisation of eggs by sperm means that number of offspring showing a particular variation will not always match the probability predicted by a Punnett square, unless the number of offspring is quite large.

	D	d
D	DD	Dd
d	Dd	dd

CELL DIVISION

SEXUAL REPRODUCTION AND SURVIVAL (2012:3)

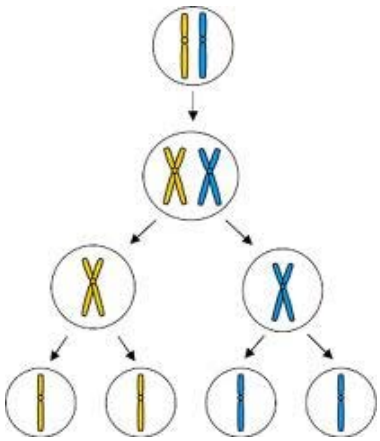
Explain how sexual reproduction causes genetic variation AND how this leads to increased survival of the species.

In your answer you should consider:

- the processes of gamete formation (meiosis) and fertilisation
- how sexual reproduction leads to variation in the population
- the link between genetic variation and survival of a species.

You may use labelled diagrams with notes to support your answer.

Gamete formation by meiosis



Description of meiosis: produces gametes/sex cells which have half the normal number of chromosomes as body cells.

Process of fertilisation: Random male and female gametes fuse (join), producing a unique zygote.

Role of sexual reproduction: Random assortment of chromosomes in meiosis and random fertilisation produce variation between individuals. Sexual reproduction leads to genetic diversity – because of randomness in formation of gamete and fertilisation.

Genetic variation: Genetic variation refers to a variety of different genotypes for a particular trait within a population. Genetic variation leads to increased survival of the species/population.

The advantage of variation to a species is that it may enable some individuals to survive if some threatening event occurs, eg disease or drought, as they will reproduce and pass on favourable phenotypes to strengthen the species.

SEXUAL REPRODUCTION (SAMPLE 2011: 2)

Meiosis is a particular form of cell division that produces male and female gametes.

(a) Describe what gametes are and explain why they are needed for sexual reproduction.

A gamete is a sex cell (eg, an egg or sperm), which has half the normal number of chromosomes as body cells.

It is required in sexual reproduction to ensure that when a sperm fuses with an egg, the resulting first cell of the new organism has the correct number of chromosomes.

Meiosis contributes to genetic variation.

(b) Discuss how meiosis contributes to genetic variation, and why genetic variation is important in a population. In your answer, you should:

- describe what is meant by genetic variation
- explain how the process of meiosis leads to genetic variation
- explain why genetic variation is of benefit to a population.

You may draw labelled diagrams to support your answer.

Genetic variation refers to a variety of different genotypes for a particular trait within a population.

Meiosis produces gametes with half the number of chromosomes. This means that pairs of alleles are separated at meiosis.

At fertilisation, which sperm fertilise which egg is due to chance and this results in new combinations of alleles.

The advantage of variation to a species is that it may enable some individuals to survive if some threatening event occurs. For example, if a new disease arrives, not all individuals will be wiped out.

GENETIC VARIATION

SEXUAL REPRODUCTION (2013:3)

For both plants and animals, there are advantages and disadvantages to sexual reproduction.

(a) Identify TWO **disadvantages** of sexual reproduction in **animals** and explain why they are disadvantages.
Possible disadvantages: need two parents that are able to reproduce, if conditions are stable could introduce variation, which may be counterproductive.

(b) Explain how sexual reproduction contributes to variation in a population of **animals**.

In your answer you should refer to gametes, meiosis and fertilisation.

Gametes are sex cells (sperm and egg) which are formed in the testes and ovaries. During gamete formation (meiosis), the homologous chromosomes are halved and the gamete will inherit one of each pair of chromosomes. Which chromosome is passed on is random due to the process of independent assortment.

During fertilisation, the gametes combine and the resulting offspring will have two alleles – they may inherit two alleles the same, homozygous, and show that characteristic or they may inherit one of each allele, heterozygous in which case they will show the dominant allele in their phenotype.

(c) Discuss why variation caused by sexual reproduction in a population of plants or animals is an **advantage** in a changing environment, such as a period of drought (a period of time of very dry weather, when there is no or very little rain).

Support your answers with examples.

Genetic variation: variety within a population, eg different alleles possible for each gene. The advantage of variation to a population is that it may see some individuals survive if environment changes, in this case if drought occurs. Because of variation, not all individuals will be wiped out. Those with favourable alleles / traits / phenotypes will survive and be able to pass on genetic material to offspring and therefore survival of the species occurs.

THE TASMANIAN DEVIL (2013:4)

Read the information to help you answer the questions.

The Tasmanian devil is known for its aggressive behaviour. Aggressive behaviour is **inherited** in Tasmanian devils.

The aggressive behaviour means that they fight and bite each other leading to injury and possible death. This behaviour has caused Tasmanian devils to become endangered (in danger of dying out).

(a) Variation in phenotype can assist survival. Explain how variation in phenotype may assist the Tasmanian devil to survive in the wild and therefore avoid the species completely dying out (becoming extinct).

In your answer you should:

- define phenotype
- explain how difference in phenotype can aid survival of an individual
- explain why the LESS aggressive phenotype (Tasmanian devils that do not fight and bite) may have a survival advantage for the species.

Definition: Phenotype – an organism's observable characteristics or traits based on the genotype.

The less aggressive Tasmanian devils are biting others less, therefore have less chance of being injured. The less aggressive Devils may expend less energy fighting and spend more time foraging.

Phenotypic ratio is the pattern of offspring based on observable characteristics.

(b) Explain how the survival of certain individuals in the wild within the Tasmanian devil population can change the ratio of aggressive to less aggressive types of Tasmanian devil within the species over time AND relate this to the species avoiding extinction.

The aggressive devils have decreased life expectancy due to increased disease and injury, therefore have fewer breeding cycles and consequently have fewer offspring during their shortened life.

The unaffected devils have a normal life expectancy and therefore more breeding cycles, resulting in more offspring during the lifetime of the individual.

The less aggressive trait has a greater chance of increasing in the population as there will be more of them to reproduce.

GENETIC VARIATION (2012:4)

Light coloured tree

Dark coloured tree

A species of moth has two phenotypes, light and dark. Both light and dark moths are eaten by birds.

Explain how the two phenotypes of the species of moth help the population to survive if the environment changes and all the trees on which the moths live become darker.

In your answer you should:

- define phenotype
- explain how colour helps individual moths to survive
- explain why the environmental change to darker trees, affects the ratio of the phenotypes in the moth population over time.

Definition of Phenotype: The physical expression of genotype / alleles, eg light and dark body / wings.

Explanation – colour: White bodied moths are more visible on a dark background and easily preyed upon. Dark coloured moths are more visible against a light / lichen background. (Camouflage)

Explanation – environment: **Individuals that are best suited to an environment will survive** to reproduce and pass on their genes to future generations. This will lead to increase in numbers of the moth with an advantageous phenotype.

If the environment changes, eg trees become darker, those individuals with dark bodies will have the beneficial characteristic and pass this onto their offspring, while the light coloured moths will stand out and be preyed upon, therefore reducing in number. As a result the **phenotypic ratio will change** to more dark than light **over time**.

VARIATION (2011:4)

Genetic variation is important in a population.

(a) Describe what is meant by the term genetic variation, and explain its importance to a population.

Genetic variation: variety within a population, eg different alleles possible for each gene.

The advantage of variation to a population is that it may see some individuals survive if environment changes, eg drought, insecticides, disease.

Because of variation, not all individuals will be wiped out. Those with favourable alleles / traits / phenotypes will survive and be able to pass on genetic material to offspring.

(b) One process that produces genetic variation is mutation.

Explain what mutations are and how they contribute to genetic variation.

In your answer you should include:

- what a mutation is

- the effect of mutations on genes, alleles and DNA
- whether all mutations are passed on to the next generation.

A mutation is a change in genetic material / DNA / genes of an organism. When a mutation occurs, the base sequence of the gene changes; this results in completely new alleles. If mutations occur in the gametes, these new alleles have the possibility of being passed on to offspring.

If mutation occurs in body cells, only the one individual will show variation – will not be passed on.

Mutations do not always result in variation, but when they do, the variation is often in the form of entirely new alleles. Mutations contribute to variation within a population by referring to formation of new traits / proteins / phenotypes.Mutations are more likely to be passed on if beneficial (natural selection).

SURVIVAL (SAMPLE 2011:4)

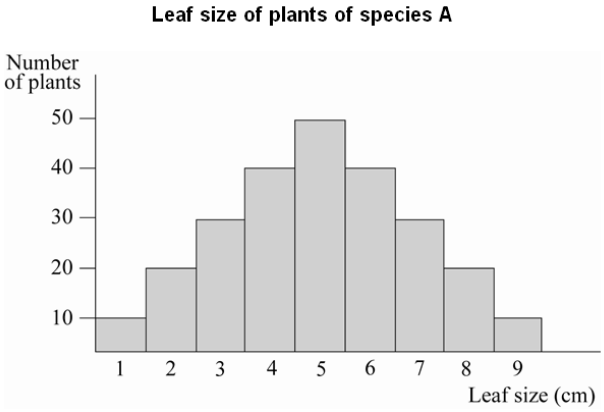
Mutations can occur in DNA during cell division.

(a) Explain whether a mutation could be inherited if it occurred in a skin cell of an individual. In your answer, you should:

- describe what a mutation is
- explain what determines whether a mutation is able to be inherited or not.

A mutation is a change to the base sequence of a gene along the DNA of an organism or a change in the genetic code. If the mutation occurs in a gamete, then it would be inherited. As a skin cell is not a gamete, a mutation in a skin cell cannot be inherited.

(b) A population of plants, species A, living in a certain area shows a lot of variation in its leaf size, from very small to very large as shown in the graph.



Leaf size affects the ability of a plant to absorb sunlight and make food. Plants with larger leaves can live in areas with lower light levels.

A new plant, species B, starts growing in the same area as species A. Species B plants grow taller than species A plants, which reduces the light available to plants growing below species B.

Discuss how variation in leaf size occurs in the starting population of species A and explain how this might help species A to survive when species B starts growing in the same area.

In your answer, you should consider:

- what causes variation within a population
- the effect of reduced light on different individuals of plant species A.

The variation in plants of species A may be due to differences (mutations) in the sequence of bases in a particular gene.

Effect of reduced light: Those individuals of plant species A whose leaves are large will be better adapted to live under lower light levels. They will survive better and produce more offspring, increasing the number of larger leaves plants in the population.

Fate of poorly adapted plants: Plants with average or small leaves will not be able to absorb enough sunlight and make sufficient food. In time these plants might die off.