JENN
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Province of the
EASTERN CAPE
EDUCATION

SUBJECT: LIFE SCIENCES

GRADE 12

AUTUMN CLASSES

TEACHER AND LEARNER CONTENT MANUAL

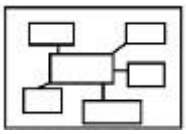
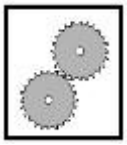
Topics

GENETICS

1. Monohybrid
2. Sex linked disorder pedigrees
3. Dihybrid cross mutation
4. Genetic engineering

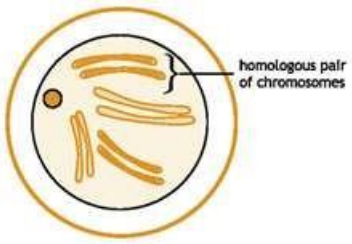
CONTENTS **PAGE**

ICON DESCRIPTION

 MIND MAP	 EXAMINATION GUIDELINE	 CONTENTS	 ACTIVITIES
 BIBLIOGRAPHY	 TERMINOLOGY	 WORKED EXAMPLES	 STEPS/HINTS

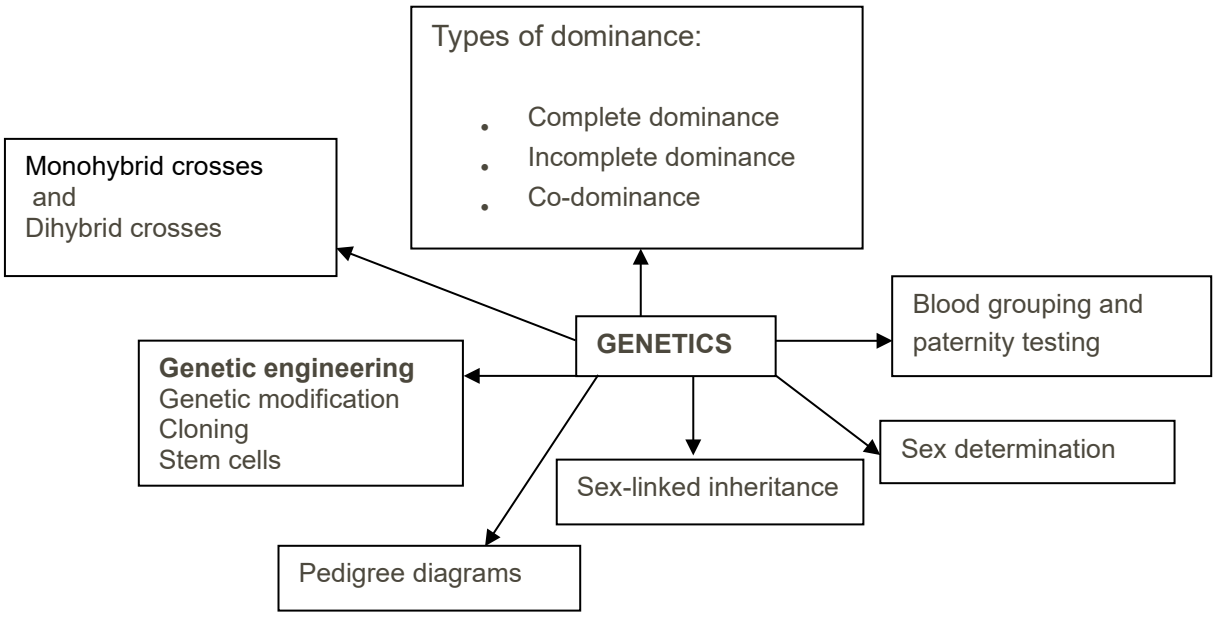
Introduction

Heredity refers to the transmission of characteristics from parents to their offspring.
Genetics refers to the study of heredity and the variations that occur in the transmission of hereditary characteristics.



The somatic cells of angiosperm and gymnosperm plants, and mammals contain two sets of chromosomes (not two chromosomes!) i.e. they are diploid.
One of these sets is of maternal origin and the other is of paternal origin. In humans we have two sets of 23 different types of chromosomes (i.e. the diploid or 2n number is 46) where one set (i.e. 23 chromosomes) comes from our male parent and the other set from our female parent A pair of identical chromosomes where one is of maternal origin and the other of paternal origin is referred to as a homologous pair of chromosomes.

Genetics mind map



TOPIC 1: TERMINOLOGY AND COMPLETE DOMINANCE

Outcomes

By the end of this topic you should be able to:

- Give a definition for each term
- Know how to solve a Monohybrid cross on Complete dominance
- Know the ratios of crosses between homozygous and heterozygous parents
- Use the template for solving monohybrid crosses
- Understand the difference between Incomplete and Co-dominance
- Solve sex-determination crosses
- Understand multiple alleles and blood groups
- Explain how blood groups are used in paternity cases
- Solve blood groups crossings (multiple alleles)

Examination guideline

GENETICS AND INHERITANCE Paper 2: 48 marks	Term 1 & 2	3½ weeks

CONTENT	ELABORATION
Introduction	❑ Mention of Mendel as the 'father' of genetics
Concepts in inheritance	❑ Chromatin and chromosomes ❑ Genes and alleles ❑ Dominant and recessive alleles ❑ Phenotype and genotype ❑ Homozygous and heterozygous ❑ The Law of Dominance- • When two homozygous organisms with contrasting characteristics are crossed, all the individuals of the F_1 generation will display the dominant trait • An individual that is heterozygous for a particular characteristic will have the dominant trait as the phenotype.
Monohybrid crosses	❑ Format for representing a genetics cross ❑ Mendel's Principle of Segregation –An organism possesses two 'factors' which separate or segregate so that each gamete contains only one of these 'factors' ❑ Types of dominance: • Complete dominance – one allele is dominant and the other is recessive, such that the effect of the recessive allele is masked by the dominant allele in the heterozygous condition • Incomplete dominance – neither one of the two alleles of a gene is dominant over the other, resulting in an intermediate phenotype in the heterozygous condition • Co-dominance – both alleles of a gene are equally dominant whereby both alleles express themselves in the phenotype in the heterozygous condition ❑ Genetics problems involving each of the three types of dominance ❑ Proportion and ratio of genotypes and phenotypes
Sex determination	❑ 22 pairs of chromosomes in humans are autosomes and one pair of chromosomes are sex chromosomes/gonosomes ❑ Males have XY chromosomes and females have XX chromosomes ❑ Differentiate between sex chromosomes (gonosomes) and autosomes in the karyotypes of human males and females ❑ Representation of a genetic cross to show the inheritance of sex

Sex-linked inheritance	❑ Sex-linked alleles and sex-linked disorders ❑ Genetics problems involving the following sex-linked disorders: • Haemophilia • Colour-blindness
Blood grouping	❑ Different blood groups are a result of multiple alleles ❑ The alleles I^A, I^B and **i** in different combinations result in four blood groups ❑ Genetics problems involving the inheritance of blood type
Dihybrid crosses	❑ Mendel's Principle of Independent Assortment – The various 'factors' controlling the different characteristics are separate entities, not influencing each other in any way, and sorting themselves out independently during gamete formation. ❑ Dihybrid genetics problems ❑ Determination of the proportion/ratio of genotypes and phenotypes
Genetic lineages/pedigrees	❑ A genetic lineage/pedigree traces the inheritance of characteristics over many generations ❑ Interpretation of pedigree diagrams
Mutations	❑ Definition of a mutation ❑ Effects of mutations: harmful mutations, harmless mutations and useful mutations ❑ Mutations contribute to genetic variation ❑ Definition of gene mutation and chromosomal mutation ❑ Two types of mutations that can alter characteristics leading to genetic disorders: Gene Mutations • Haemophilia – absence of blood-clotting factors • Colour-blindness – due to absence of the proteins that comprise either the red or green cones/photoreceptors in the eye Chromosomal mutation • Down syndrome – due to an extra copy of chromosome 21 as a result of non-disjunction during meiosis

CONTENT	ELABORATION
Genetic engineering	❑ Biotechnology is the manipulation of biological processes to satisfy human needs. ❑ Genetic engineering is an aspect of biotechnology and includes: • Stem cell research – sources and uses of stem cells • Genetically modified organisms – brief outline of process (names of enzymes involved are not required) and the benefits of genetic modification • Cloning – brief outline of process and benefits of cloning
Paternity testing	❑ The use of each of the following in paternity testing: • Blood grouping • DNA profiles
Genetic links	❑ Mutations in mitochondrial DNA used in tracing female ancestry

1. MONOHYBRID CROSSES

1.1: Terminology

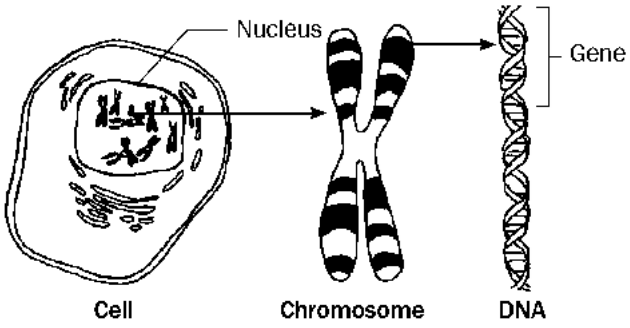
Differentiate between each of the following:

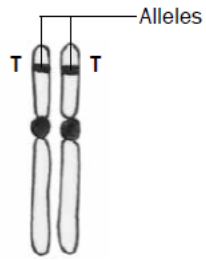
Chromatin and chromosomes

Genes and alleles

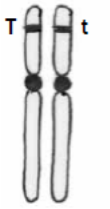
- Phenotype and genotype
- Dominant and recessive alleles
- Homozygous (pure breeding) and heterozygous (hybrid)
- Monohybrid cross and dihybrid cross

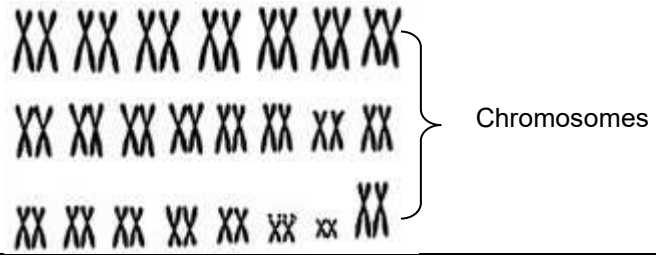
Make use of the following table to explain the concepts to your learners.

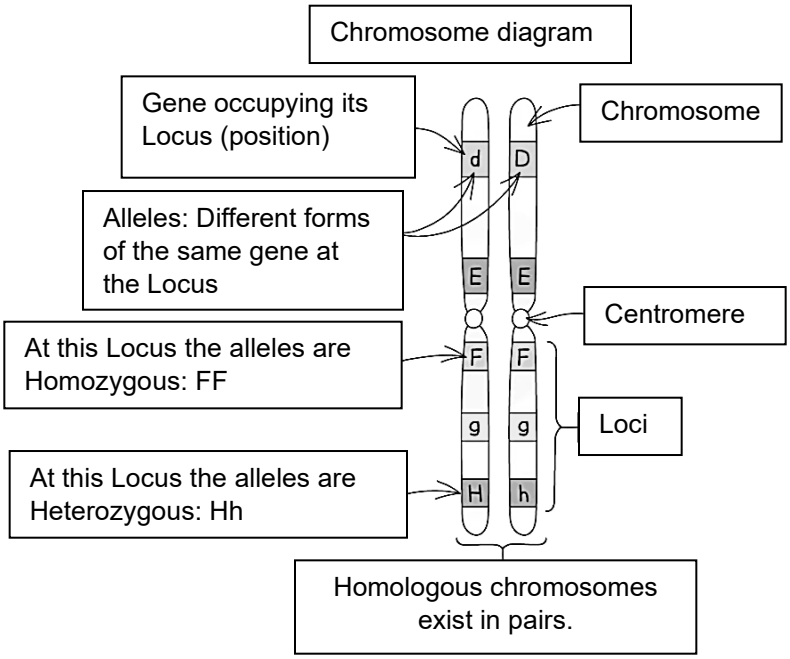
<table>
<tr><th>Term</th><th>Explanation</th><th>Diagram/Additional notes</th></tr>
<tr><td>Gene</td><td>A small portion of DNA coding for a particular characteristic.</td><td rowspan="4"> 

Dominant allele (t) – tall plant
Recessive allele (t) – short plant
• Homozygous dominant (both alleles are dominant)
• Genotype TT
• Phenotype tall
 

• Homozygous dominant (both alleles are dominant)
• Genotype TT
• Phenotype – tall</td></tr>
<tr><td>Alleles</td><td>Different forms of a gene which occur at the same locus (position) on homologous chromosomes.</td></tr>
<tr><td>Genotype</td><td>Genetic composition (make- up) of an organism.</td></tr>
<tr><td>Phenotype</td><td>The physical appearance of an organism determined by the genotype, e.g. tall, short.</td></tr>
<tr><td>Dominant allele</td><td>An allele that is expressed (shown) in the phenotype when found in the heterozygous (Tt) and homozygous (TT) condition.</td></tr>
</table>

Recessive allele	An allele that is masked (not shown) in the phenotype when found in the heterozygous (Tt) condition. It is only expressed in the homozygous (tt) condition.	 • Homozygous recessive (both alleles are recessive) • Genotype tt • Phenotype – short	
Heterozygous	Two different alleles for a particular characteristic, e.g. Tt.	 • Heterozygous (one dominant and one recessive allele) • Genotype Tt • Phenotype – tall	
Homozygous	Two identical alleles for a particular characteristic, e.g. TT or tt.		
Monohybrid cross	Only one characteristic or trait is being shown in the genetic cross.	*Example*: Flower colour only, e.g. yellow flower or white flower OR shape of seeds only, e.g. round seeds or wrinkled seeds.	
Complete dominance	A genetic cross where the dominant allele masks (blocks) the expression of a recessive allele in the heterozygous condition.	In this type of cross the allele for tall (T) is dominant over the allele for short (t). The offspring will therefore be tall because the dominant allele (T) masks the expression of the recessive allele (t).	Tall (TT) x short (tt) Tall (Tt)
Incomplete dominance	A genetic cross between two phenotypically different parents produces offspring different from both parents but with an intermediate phenotype.	*Example:* If a red-flowered plant is crossed with a white-flowered plant and there is incomplete dominance – the offspring will have pink flowers (intermediate colour).	Red flower x White flower RR X WW RW Pink flowers (intermediate phenotype)
Co-dominance	A genetic cross in which both alleles are expressed equally in the phenotype.	*Example:* If a red-flowered plant is crossed with a white-flowered plant and there is co-dominance the offspring has flowers with red and white patches.	Red flower × White flower RR WW RW Flowers with red and white Patches (both alleles expressed in phenotype)
Multiple alleles	More than two alternative forms of a gene at the same locus.	*Example:* Blood groups are controlled by three alleles, namely I^A, I^B and i. *This is the ONLY acceptable notation for the blood group alleles*	

Sex-linked characteristics	Characteristics or traits that are carried on the sex chromosomes.	*Examples:* Haemophilia and colour-blindness The alleles for haemophilia (or colour-blindness) are indicated as superscripts on the sex chromosomes, e.g. X^HX^H (normal female), X^HX^h (normal female), X^hX^h (female with haemophilia), X^H Y (normal male), X^hY (male with haemophilia).
Karyotype	The number, shape and arrangement of all the chromosomes in the nucleus of a somatic cell.	
Cloning	Process by which genetically identical organisms are formed using biotechnology.	*Example:* Dolly the sheep was cloned using a diploid cell from one parent; therefore, it had the identical genetic material of that parent.
Genetic modification	The manipulation of the genetic material of an organism to get desired changes.	*Example:* The insertion of human insulin gene in plasmid of bacteria so that the bacteria produce human insulin.
Human genome	The mapping of the exact position of all the genes in all the chromosomes of a human.	*Example:* Gene number 3 on chromosome number 4 is responsible for a particular characteristic.
Autosomes	All the chromosomes except the sex chromosome	*Example:* In the human there are 22 pairs (44) autosomes
Gonosomes	The sex chromosomes X and Y	XX female XY male



Activity 1

1.1 Give the correct **biological term** for each of the following descriptions. Write only the term next to the question number (1.2.1 – 1.2.2) in the ANSWER BOOK.

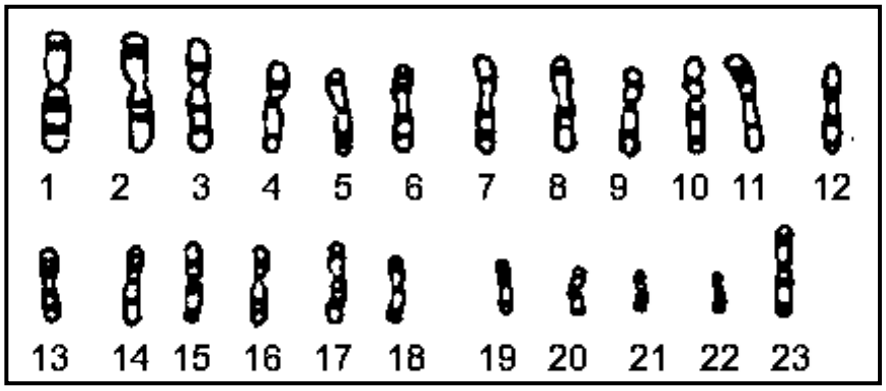
1.1.1 A section of a DNA molecule that codes for a specific characteristic
1.1.2 Type of dominance in which both alleles are expressed in the phenotype
1.1.3 The position of a gene on a chromosome
1.1.4 The physical or functional expression of a gene (4 x 1) **(4)**

1.2 Indicate whether each of the statements in COLUMN I applies to **A only, B only, both A and B** or **none** of the items in COLUMN II. Write **A only, B only, both A and B, or none** next to the question number (1.2.1 – 1.2.3) in the ANSWER BOOK.

	COLUMN I		COLUMN II
1.2.1	The type of dominance where both alleles of a gene are expressed in the phenotype in the heterozygous condition	A B	Co-dominance Complete Dominance
1.2.2	The pair of chromosomes in a diploid organism that have the same size and shape	A B	Homologous Homozygous
1.2.3	The study of heredity and variation in organisms		Inheritance Genetics

(3 X 2 **(6)**

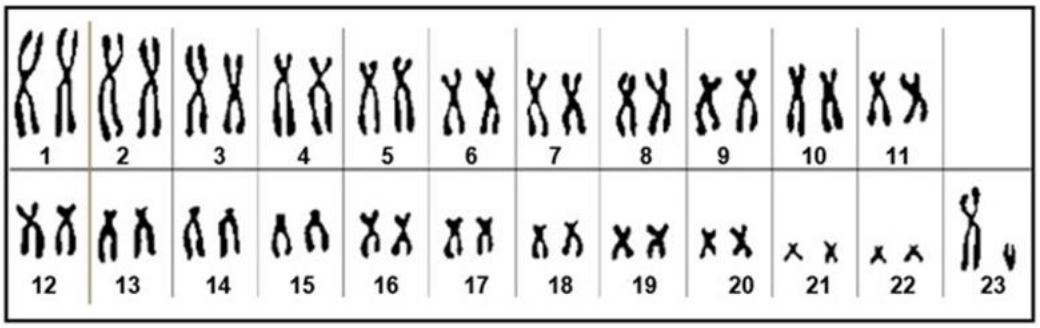
1.3 The diagram below represents the chromosomes from a sperm cell.



1.3.1 According to the karyotype how many autosomes are present in the sperm cell? (1)

From;: DBE question paper June 2016

1.4 The diagram below shows a karyotype.



1.4.1 How many of the following are present in the karyotype?

(a) Chromosomes (1)
(b) Autosomes (1)
(c) Gonosomes (1)

1.4.2 How many chromosomes would be present in the gametes produced by this individual? (1)

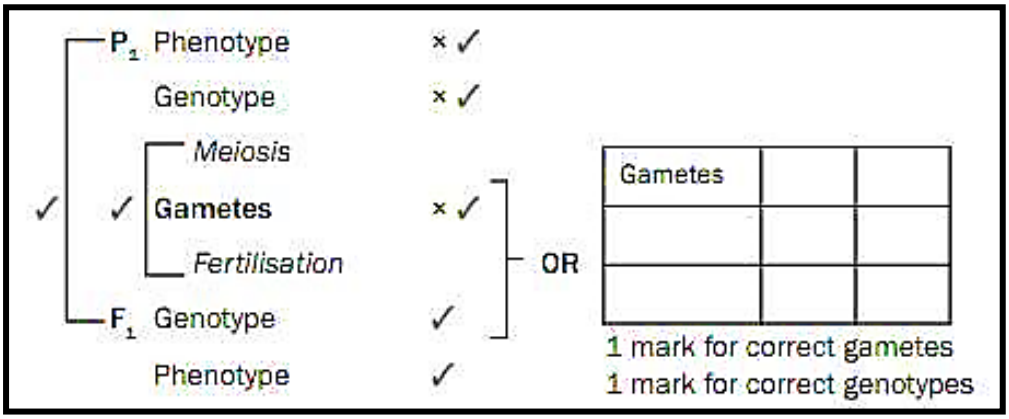
1.4.3 Is the karyotype in the diagram that of a male or a female? (1)

1.2: Complete dominance crosses

Complete dominance – one allele is dominant, and the other is recessive, such that the effect of the recessive allele is masked by the dominant allele in the heterozygous condition.
A monohybrid cross is a cross between two individuals that looks at only ONE characteristic
e.g. The height of a plant

A specific template is used to solve monohybrid crosses.

- Use the following genetic problem format or template to solve all monohybrid genetic problems:



NOTE: The template MUST always be written in order as shown above to earn the marks for P1 & F1 and Meiosis and Fertilization. Do NOT use FUSION for FERTILISATION.

The following problem represents a genetic cross which shows complete dominance:

Two homozygous brown eyed parents are crossed. Brown eyes (B) is dominant and blue eyes (b) is recessive

Parents (P1) **Phenotype** Brown Brown
Genotype BB BB
Meiosis
Gametes B B

Fertilisation

	B	B
B	BB	BB
B	BB	BB

Offspring (F1) **Genotype** BB
Phenotype Brown

1. Homozygous x Homozygous

P1 Genotype Brown Blue
Phenotype BB bb
Meiosis Gametes B B

Fertilization

	B	B
b	Bb	Bb
b	Bb	Bb

F1 Genotype Bb
Phenotype All brown 100%

2.Homozygous X Heterozygous (dominant)

P1 Genotype Brown Brown
Phenotype BB Bb
Meiosis
Gametes B b

Fertilization

	B	B
B	BB	BB
b	Bb	Bb

1:1
F1 Genotype Bb:BB
Phenotype All 100% brown

Heterozygous X Homozygous (recessive)

P1 Genotype Brown Blue
Phenotype Bb bb
Meiosis Gametes B b

Fertilization

	B	b
b	Bb	bb
b	Bb	bb

F1 Genotype Bb bb
Phenotype 50%brown 50% blue

3. Heterozygous X Heterozygous

P1		Genotype	Brown	Blue
		Phenotype	Bb	Bb
	Meiosis	Gametes	B;b	B;b
	Fertilization		B	b
		B	BB	Bb
		b	Bb	bb
F1			1:2:1	
		Genotype	BB: Bb: bb	
		Phenotype	3 brown : 1 blue	
			75%: 25%	

Teacher Activity: Work through the following example with the learners on the board

In humans the ability to roll the tongue is due to a dominant allele. A man who is heterozygous for tongue-rolling and a woman who cannot roll her tongue have children. Use the symbols **T** and **t** for the alleles of the tongue-rolling characteristic and represent a genetic cross to determine the possible genotypes and phenotypes of the children. (6)

HINT: Identify the phenotypes of the man and the woman (parents/P), i.e. the man is a tongue-roller and the woman is not,

Activity 2

2.1 Various options are provided as possible answers to the following questions. Choose the correct answer and write only the letter (A – D) next to the question number (1.1.1 – 1.1.2) in your ANSWER BOOK, for example 1.1.3 D.

2.1.1 Which ONE of the following monohybrid crosses where complete dominance applies will result in a phenotypic ratio of 3:1?

A Both parents are heterozygous
B Both parents are homozygous for the dominant characteristic
C One parent is heterozygous and the other is homozygous recessive
D One parent is heterozygous and the other is homozygous dominant

2.1.2 Which ONE of the following is TRUE about genes?

Genes …
A code for a particular characteristic.
B are made up of amino acids.
C are made up of RNA.
D are found in a ribosome.

2.1.3 The table below shows the results of the F_1 generation after an investigation into the inheritance of eye colour in fruit flies was carried out.

EYE COLOUR	NUMBER OF FLIES
Red	182

The genotypes of the parents were …

A RR x Rr.
B Rr x rr.
C Rr x Rr.
D RR x rr.

2.1.4 Two red-eyed fruit flies were mated, and they produced 150 flies with red eyes and 48 flies with white eyes. From this information we can reasonably conclude that the ...

A white-eyed condition is recessive, and both parents are heterozygous.
B red-eyed condition is dominant, and both parents are homozygous for red eyes.
C white-eyed condition is recessive, and both parents are homozygous for red eyes.
D red-eyed condition is recessive, and both parents are heterozygous.

2.2 Give the correct **biological term** for each of the following descriptions. Write only the term next to the question number (2.2.1 – 2.2.4) in the ANSWER BOOK.

2.2.1 Characteristics controlled by genes which are located on the sex chromosomes
2.2.2 A genetic cross involving one characteristic only
2.2.3 An allele that is expressed phenotypically only in the homozygous condition
2.2.4 Chromosomes that are similar in structure and code for the same characteristics

(4 x 1) **(4)**

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2.3 In rabbits, black fur is produced by the allele (**B**) and white fur by the allele (**b**).

The table below shows the genotypes of some rabbits.

RABBIT	GENOTYPE
1	BB
2	Bb
3	bb

2.3.1 What is the phenotype:

(a) Produced by the recessive allele (1)

(b) Of rabbit **2** (1)

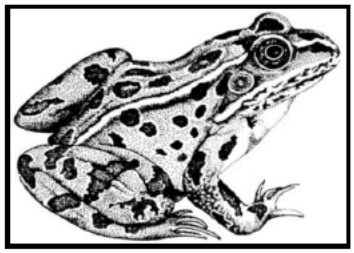
2.3.2 Give the NUMBER only (**1**, **2** or **3**) of the rabbit(s) that is/are:

(a) Pure-bred

(b) Homozygous dominant (3)

2.3.3 Use a genetic cross to show the percentage chance of rabbits **1** and **3** having offspring with white fur. (6)
(11)

2.4 The back of the leopard frog *(Rana pipiens)* can be spotted, as shown below, or be without spots.



Spotted frogs were allowed to interbreed and they produced 150 spotted offspring and 50 offspring without spots.

2.4.1 Which phenotype is dominant? (1)

2.4.2 Explain your answer to QUESTION 2.4.1. (2)

2.4.3 A frog that is heterozygous for spotted back was crossed with a frog without spots.

Using the letters **D** and **d**, represent a genetic cross to show the expected genotypes and phenotypes of the F_1 generation. (6)
(9)

1.3 Incomplete dominance

Incomplete dominance – neither one of the two alleles of a gene is dominant over the other, resulting in an *intermediate phenotype* in the heterozygous condition. (red x white gives pink)

NB Use 2 sets of symbols (e.g R -red, W- white)

This refers to a genetic cross between two phenotypically different parents producing an offspring different from both parents but with an intermediate phenotype. The following problem represents a genetic cross that shows incomplete dominance.

Teacher Activity: Work through the following example with the learners on the board

A homozygous snapdragon plant with red flowers (R) was cross-pollinated with a homozygous snapdragon plant with white (W) flowers. All the plants that grew from the cross had red and white flowers (6)

Activity 3

3.1 (DBE Life Sciences May-June 2021)
In rabbits, fur colour may be black, white or grey. The inheritance of fur colour is controlled by two alleles namely:

Black fur (**B**) and White fur (**W**)

3.1.1 Explain why fur colour in rabbits is an example of inheritance with incomplete dominance. (2)

3.1.2 Use a genetic cross to show the expected genotypes and phenotypes of the offspring when a grey male mates with a black female. (6)
(8)

3.2 In certain marine invertebrates the colour of the shell is under the control of one gene with three alleles. In different combinations, the three alleles produce four phenotypes: orange, yellow, orange-yellow and black.

The table below shows the results of the offspring produced from crosses involving parents of different phenotypes.

CROSS	PHENOTYPES OF SHELLS	
	PARENTS	OFFSPRING
1	Yellow x yellow	27 yellow: 9 black
2	Black x black	All black
3	Orange x orange	30 orange: 10 black
4	Orange x yellow	All orange-yellow

3.2.1 Name and describe the type of dominance shown by cross **4**. (3)

3.2.2 Which shell colour is controlled by the recessive allele? (1)

3.2.3 Use information in the table to support your answer to QUESTION 3.2.2 (2)
(6)

3.3 Various options are provided as possible answers to the following questions. Choose the correct answer and write only the letter (A – D) next to the question number (3.3.1 – 3.3.2) in your ANSWER BOOK, for example 3.3.3 D.

3.3.1 A plant with red flowers is crossed with a plant with white flowers. The offspring include plants with red flowers (1/4), pink flowers (1/2), and white flowers (1/4). Which allele is dominant?

A Complete dominance
B Multiple alleles
C Codominance
D Incomplete dominance (2)

3.3.2 George observed a plant in his garden. He hypothesized that the stem height showed incomplete dominance. To check for this he created true-breeding (homozygous) lines of tall and short plants. He then crossed these and all the offspring had intermediate height.He crossed the plants with intermediate height and sample 1000 of the offspring.

Which of the following results matches his hypothesis?

A 500 tall plants, 250 intermediate plants and 250 short plants
B 250 tall plants, 500 intermediate plants and 250 short plants
C 250 tall plants,250 intermediate plants and 500 short plants
D 125 tall plants, 750 intermediate plants, 125 short plants (2)

1.4 Co-dominance

This refers to a genetic cross in which both alleles of a gene are equally dominant whereby both alleles express themselves in the phenotype in the heterozygous condition. Both alleles of the characteristic are expressed (red x white -give red flowers with white spots)

Teacher Activity: Work through the following example with the learners on the board

A homozygous plant with red flowers (R) is crossed with a homozygous plant with white (W) flowers. All the plants that grew from the cross had red apink flowers (6)

Activity 4

4.1.1 When a red horse (RR) is crossed with a white horse (WW), the offspring are all roan (RRWW). Roan colour pattern is characterised by a mixture of red and white hairs on the body.
This type of inheritance is known as:

A Complete dominance
B Multiple alleles
C Codominance
D Incomplete dominance (2)

4.2. In rabbits, black fur is produced by the allele (**B**) and white fur by the allele (**W**). The table below shows the genotypes of some rabbits.

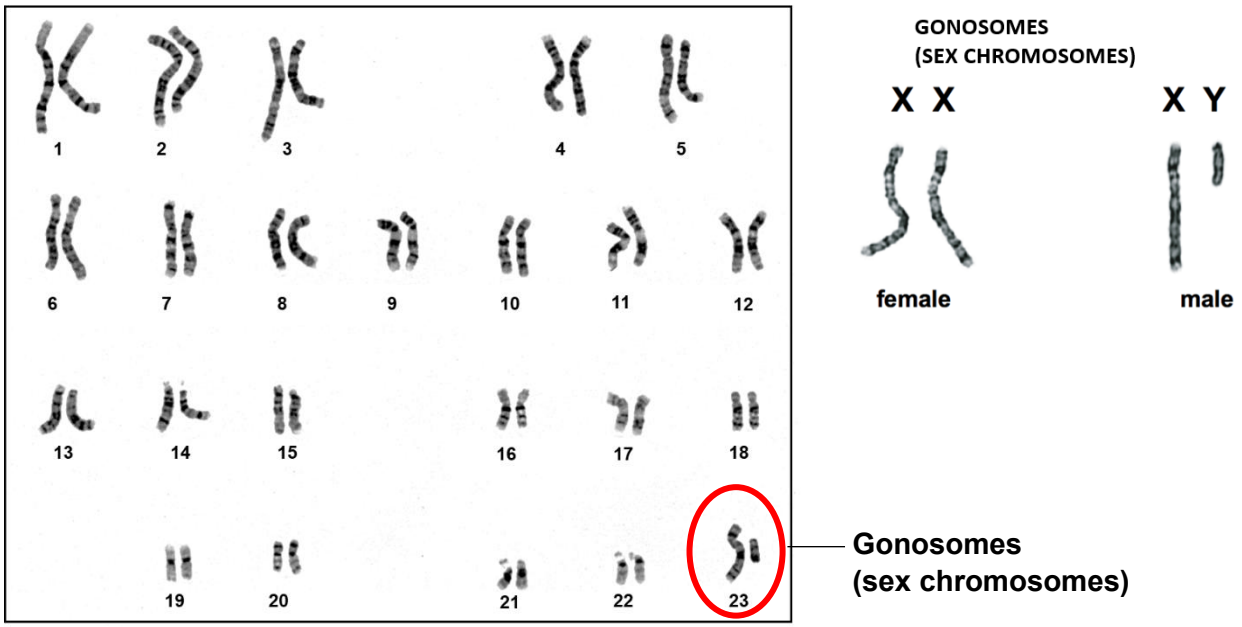
RABBIT	GENOTYPE
1	BB
2	BW
3	WW

Use a genetic cross to show the percentage chance of rabbits **1** and **3** having offspring with white fur.

4.3. A farmer has an orchard of apple trees. Each apple produced expressed red and yellow colour equally (red-yellow apples). To extend his apple orchard, the farmer collected seeds from the red-yellow apples and grew them. When the new trees matured, he found that some of the trees produced red apples (R), others produced yellow apples (Y) and the rest produced apples that were red-yellow. Use a genetic cross to explain his results in the F_1 generation

1.5 Inheritance of sex

S*ex chromosomes (gonosomes)* are pair number 23 in the human karyotype
It is the X chromosome and the Y. Chromosome XX indicate a woman XY indicates a male.



Worked example

The following represents a cross that shows inheritance of sex,

P1	Phenotype	Male	x	Female	Step 1
	Genotype	XY	x	XX	Step 2
	Gametes	X and Y	x	X	Step3

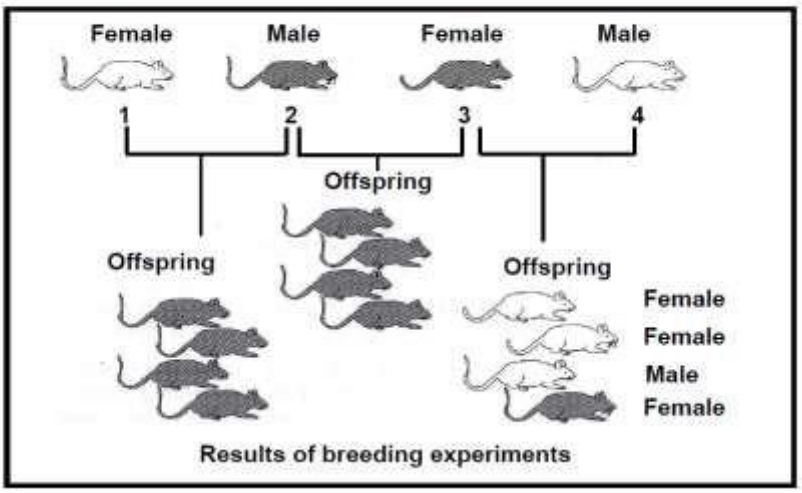
Fertilisation

	X	Y
X	XX	XY
X	XX	XY

	Genotype	XX		XY	Step 4
F1	Phenotype	FEMALE		MALE	Step 5
	Ratio	50% probability	:		

Activity 5 (DBE March 2015 Life Sciences P1)

5.1 Study the diagrams below that show some breeding experiments on mice. A single pair of alleles showing complete dominance controls coat colour (white or grey) in these mice.



5.1.1 State which sex chromosomes would be present in the gametes of parent mouse **2** and mouse **3**, respectively. (2)

5.1.2 If mice **3** and **4** had a second set of offspring, what is the percentage chance that the first mouse born would be female? (1)

5.1.3 Which of the parent mice (**1, 2, 3** or **4**) is likely to be homozygous dominant for coat colour? (1)

5.1.4 State why mouse **3** can only be heterozygous for coat colour. (2)

1.6 Blood grouping

- Different blood groups are a result of multiple alleles
- Human blood type is determined by three different alleles, known as I^A, I^B, and i.
- The I^A and I^B alleles are co-dominant.
- The i allele is recessive.
- The possible human phenotypes for blood group are type A, type B, type AB, and type O.
- Type A and B individuals can be either homozygous (I^AI^A or I^BI^B, respectively), or heterozygous (I^Ai or I^Bi), respectively.

Phenotype	Genotype	
Blood group	Homozygous	Heterozygous
A	I^AI^A	I^Ai
B	I^BI^B	I^Bi
AB		I^AI^B
O	ii	

Let's look at the following question (DBE Life Sciences Dec 2019 P2) –

NOTE: LIFE SCIENCES PAPERS NO LONGER HAVE ESSAYSSINCE 2021 BUT YOU MAY BE ASKED PARAGRAPH QUESTIONS

Sometimes the paternity of a son or a daughter is disputed.

Describe sex determination in humans and explain how blood grouping and DNA profiling are used in paternity testing.

Content: (17)
Synthesis: (3)
(20)

Sex determination (S)

- Females have XX chromosomes✓
- thus produce an ovum which will always carry the X chromosome✓
- Males have XY chromosomes✓
- thus a sperm will either carry X✓
- or Y✓ chromosome
- If a sperm carrying the X chromosome fertilises the ovum carrying the X chromosome✓
- then a female child results✓
- If a sperm carrying the Y chromosome fertilises the ovum carrying the X chromosome✓
- then a male child results✓
- Therefore it is the father's gamete carrying X or Y chromosome that determines the sex of the child✓
- There is a 50% chance that the child can be a boy or a girl✓ Any 7 (7)

Blood grouping (B)

- The blood group of a child is determined by the alleles received from both parents✓
- The blood group of the mother, the child and the possible father is determined✓
- If the blood group of the mother and possible father cannot lead to the blood group of the child✓
- the man is not the father✓
- If the blood group of the mother and the possible father can lead to the blood group of the child✓
- the man might be the father✓
- This is not conclusive✓
- because many men have the same blood group✓ Any 5 (5)

DNA profiling (P)

- A child received DNA from both parents✓
- The DNA profiles of the mother, child and the possible father are determined✓
- A comparison of the DNA bands of the mother and the child is made✓
- The remaining DNA bands are compared to the possible father's DNA bands✓
- If all the remaining DNA bands in the child's profile match the possible father's DNA bands✓
- then the possible father is the biological father✓
- If all the remaining DNA bands in the child's profile does not match the possible father's DNA bands✓
- then the possible father is not the biological father✓ Any 5 (5)

Content: (17)
Synthesis: (3)
(20)

Worked example (DBE Life Sciences exemplar 2011 P1)
Human blood type is determined by three different alleles, known as I^A, I^B, and i. The I^A and I^B alleles are co-dominant, and the i allele is recessive.
A woman with type A blood and a man with type B blood could have offspring with blood type:

A A and B only
B B and AB only
C O only
D A, B, AB or O

Activity 6

DBE Life Sciences March 2017

6.1 Human blood groups are controlled by multiple alleles.

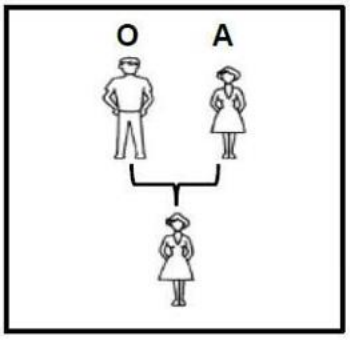
6.1.1 How many alleles control blood groups? (1)

6.1.2 Which TWO alleles are codominant in the inheritance of blood groups? (2)

6.1.3 A man is heterozygous for blood group A and marries a woman who has blood group O. Use a genetic cross to show the phenotypic ratio of their offspring. (7)
(10)

6.2 (DBE Life Sciences March 2016)

The diagram below shows the blood types of two parents.



The only possible blood type(s) of the offspring of the first generation (F_1) is/are …

A AB and O.
B A and O.
C A only.
D A and B.

6.3 A man with blood group **A** and a woman with blood group **B** have children. Their first child has blood group **AB** and the second child has blood group **O**.

Which prediction about the blood groups of future children is CORRECT?

A Future children have a 50% chance of having blood group **AB** and a 50% chance of having blood group **O**.
B All will have blood group **A** or **B**.
C Each future child will have an equal chance of having blood group **A**, **B**, **AB** or **O**.
D None of the future children will have blood group **A**.

(DBE Life Science P2 Nov 2016)

6.4 Human blood groups are controlled by multiple alleles.

6.4.1 Name ALL the alleles that control human blood groups. (3)

6.4.2 How many of the alleles named in QUESTION 6.4.1 can any individual inherit? (1)

6.4.3 Give a reason for your answer to QUESTION 6.4.2 (2)

6.4.4 A man has blood group **A** and his wife has blood group **B**. Their first child has blood group **AB** and the second child has blood group **O**.

6.4.5 What can one conclude about the blood groups of their future children? (3)
(9)

(DBE Life Sciences June 2016 P2)

6.5 THREE babies (**X**, **Y** and **Z**) from three different sets of parents were born in a hospital. TWO of the babies were accidentally swopped. Blood groups of the parents were used to establish which baby belonged to which set of parents.

The blood groups of the parents and the babies are shown in the table below.

PARENTS	BABIES	BLOOD GROUPS OF PARENTS AND BABIES		
		Mother	Father	Baby
Mr and Mrs Pule	X	B	A	A
Mr and Mrs Chaka	Y	AB	B	O
Mr and Mrs Tau	Z	O	B	AB

6.5.1 Which TWO babies (from **X**, **Y** and **Z**) were swopped? (2)

6.5.2 Give the surnames of the biological parents of the two babies that were swopped. Write the correct surnames of the parents next to the letter (**X**, **Y** or **Z**). (2)

6.5.3 Give the possible genotype(s) of Mr Pule that could have produced baby **X**. (2)
(6)

TOPIC 2: SEX-LINKED INHERITANCE AND PEDIGREE DIAGRAMS

Outcomes

By the end of this topic you should be able to:

Know what a sex-linked allele is
Know how sex-linked alleles can cause sex-linked genetic disorders
Solve genetic problems with sex-linked alleles
Understand the sex-linked disorder Hemophilia and solve genetic problems
Understand the sex-linked disorder on colour blindness
Interpret and understand pedigree diagrams
Understand that a pedigree diagram traces the inheritance of characteristics over many generations.

Examination guideline

CONTENT	ELABORATION
Sex-linked inheritance	❑ Sex-linked alleles and sex-linked disorders ❑ Genetics problems involving the following sex-linked disorders: • Haemophilia • Colour-blindness
Genetic lineages/pedigrees	❑ A genetic lineage/pedigree traces the inheritance of characteristics over many generations ❑ Interpretation of pedigree diagrams

2.1 Inheritance of sex-linked characteristics

Sex-linked characteristics are characteristics (traits) that are carried on the **X** sex chromosome. Sex linked alleles can cause some sex-linked genetic disorders like

- Haemophilia – absence of blood clotting factors
- Colour- blindness – absence of the proteins that comprise either the red or green cones/photoreceptors in the eye

The following problem represents a genetic cross which shows the inheritance of sex-linked characteristics.

Worked example

Haemophilia is a sex-linked hereditary disease that occurs as a result of a recessive allele on the X-chromosome (X^h). A father without haemophilia and a mother who is heterozygous for haemophilia have children. Represent a genetic cross to determine the possible genotypes and phenotypes of their children.

The alleles for haemophilia are indicated as superscripts on the sex chromosomes, e.g. X^HX^H (female without haemophilia), X^HX^h (female without haemophilia), X^hX^h (female with haemophilia), X^HY (male without haemophilia), X^hY (male with haemophilia).

P1	**Phenotype**	**Male without haemophilia**	**x**	**Female without haemophilia**	**Step 1**
	Genotype	X^HY	x	X^HX^h	Step 2
Meiosis					
	Gametes	X^H , Y	x	X^H , X^h	Step3

Fertilisation

	X^H	Y
X^H	$X^H X^H$	$X^H Y$
X^h	$X^H X^h$	$X^h Y$

	Genotype	$X^H X^H$, $X^H X^h$	$X^H Y$	$X^h Y$	Step 4
F1	Phenotype	2 daughters without haemophilia	1 son without haemophilia	1 son with haemophilia	Step 5

Activity 1 (DBE Life Science March 2017 P 2)

1.1 Haemophilia is a genetic disorder caused by a recessive allele on the X-chromosome.

A haemophiliac female marries a male without haemophilia. Explain why all their sons will be haemophiliacs (4)

(DBE Life Sciences March 2013 P 1)

1.2 A rare form of rickets in humans is caused by a sex-linked dominant allele (**R**) which is carried on the X-chromosome. An affected female, whose father was unaffected, married an unaffected male.

1.2.1 Determine the possible genotypes and phenotypes of their offspring by representing a genetic cross. (6)

1.2.2 What is the percentage chance that they will have a child who is an unaffected male? (2)

1.2.3 Explain why this disorder, although it is sex-linked, does NOT affect males only. (2)

2.2 Pedigree diagrams

A pedigree diagram/ genetic lineage traces the inheritance of characteristics over many generations in a family. A pedigree diagram is also called a family tree.

Pedigree diagrams can show autosomal dominant and recessive alleles but also sex-linked alleles (Alleles on the X chromosome/gonosome)

- In a pedigree diagram symbol are used to show the status of an individual. (you must know this symbols) Squares represent males and circles represent females.

Female without the disorder

Female with the disorder

Male without the disorder

Male with the disorder

To solve/ interpret a pedigree diagram we follow a sequence of steps:

Step 1: Study any key and opening statement/s and look for dominant and recessive characteristics and phenotypes.

Step 2: Write in the phenotypes of all the individuals as given in the problem.

Step 3: Fill in the genotype of all the individuals with the recessive condition – it must have two recessive alleles (two lower case letters, e.g. ff).

Step 4: For every individual in the diagram that has the recessive condition, it means that each allele was obtained from each of the parents. Work backwards and fill in one recessive allele for each parent.

Step 5: If the parents showed the dominant characteristic, fill in the second letter which represents the dominant allele (a capital letter, e.g. F).

Step 6: Any other individual showing the dominant characteristic will most likely be homozygous dominant (FF) or heterozygous dominant (Ff).

Worked example

The pedigree diagram below shows inheritance of eye colour in humans over three generations of a family. Brown eye colour (B) is dominant over blue eye colour (b). Study the diagram and then answer the questions that follow.

Note the following in the pedigree diagram

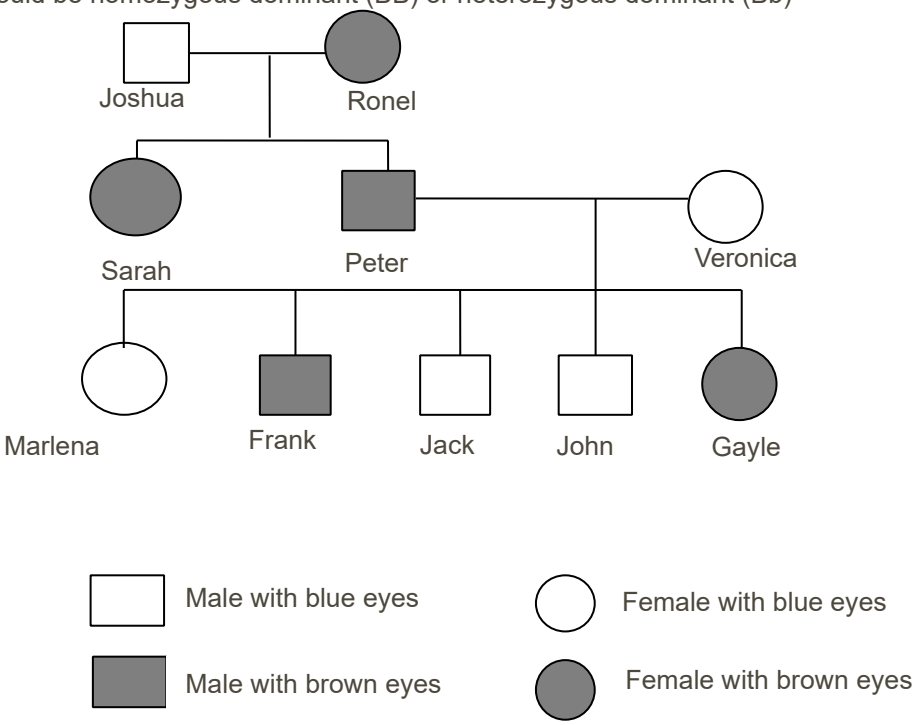
- Squares represent males and circles represent females.
- The horizontal line between a square (Joshua) and a circle (Ronel) shows that they have mated.
- The vertical line flowing from the horizontal line represents the offspring (Sarah and Peter) of the two parents (Joshua and Ronel).

- **Brown eye colour (B) is dominant over blue eye colour (b)** – stated in problem. ..... **Step 1**
- Joshua, Jack and John are males with blue eyes.
- Veronica and Marlena are females with blue eyes.

Step 2

- Peter and Frank are males with brown eyes.
- Ronel, Sarah and Gayle are females with brown eyes.
- Joshua, Veronica, Marlena, Jack and John will have the genotype 'bb'. The recessive characteristic only shows up in the homozygous condition. . . **Step 3**
- *Example:* The genotype of Peter is 'Bb' – working backwards from the offspring Marlena or Jack or John who are homozygous recessive. This means that one of the recessive alleles of Marlena, Jack and John, i.e. 'b', must have come from parent Peter and the other one from parent Veronica **Steps 4 and 5**

Ronel could be homozygous dominant (BB) or heterozygous dominant (Bb)



Pedigree diagram showing inheritance of eye colour

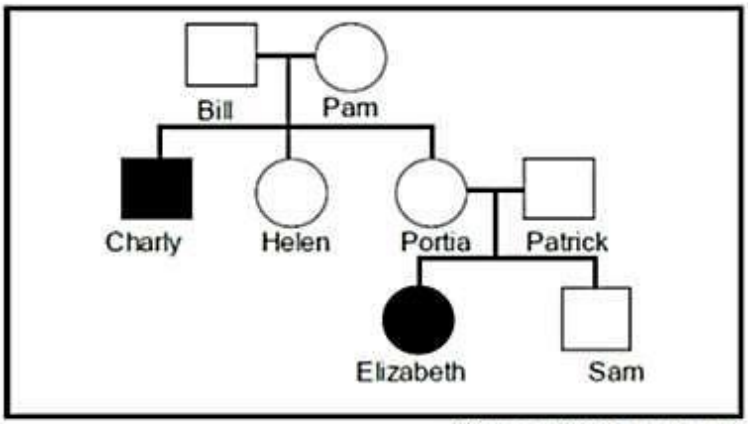
1. How many members of the family have blue eyes? (1)
2. Is Veronica homozygous or heterozygous for eye colour? (1)
3. Write down the genotype of
   - (a) Joshua (2)
   - (b) Ronel (2)
   - (c) Frank (2)
4. If Frank marries a woman with the same genetic composition as Sarah, what is the percentage probability of them having a child with brown eyes? (1)

ACTIVITY 2

DBE Life Science March 2017 P2

2.1 Tay-Sachs disease is caused by an autosomal recessive allele (**n**). Children with Tay-Sachs disease lose motor skills and mental functions. Over time, the children become blind, deaf, mentally retarded and paralysed. Tay-Sachs children die by the age of five.

The pedigree diagram below shows the inheritance of Tay-Sachs disease in a family.



[Source: www.tay-sachs.org]

2.1.1 Give:

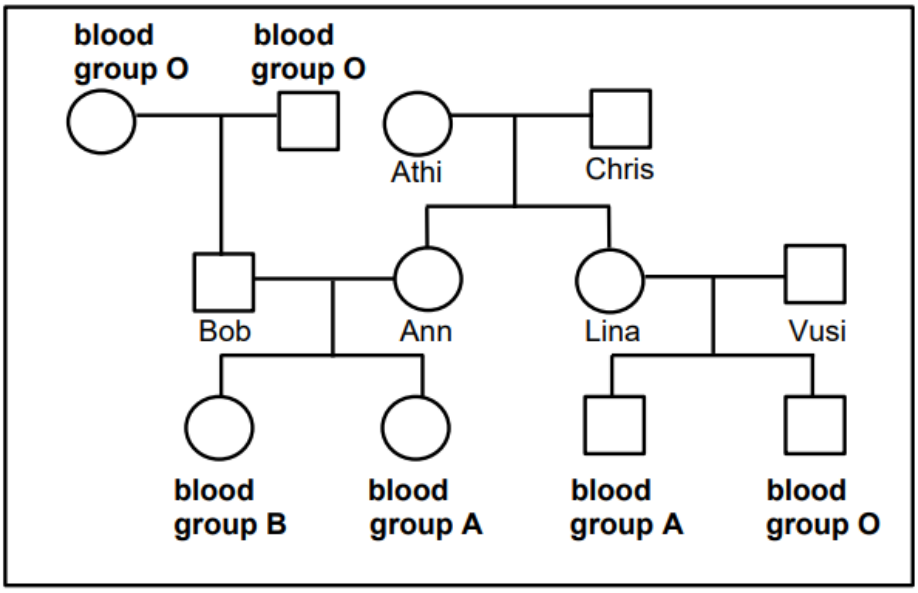
(a) Charly's phenotype (2)

(b) Portia's genotype (2)

(c) Bill's genotype (2)

2.1.2 Explain why Patrick is normal, but a carrier of Tay-Sachs disease. (2)
(8)

2.2 The diagram below shows the inheritance of blood groups in a family.



2.2.1 Name the type of diagram shown. (1)

2.2.2 Give the number of alleles that control blood groups. (1)

2.2.3 How many generations are represented in the diagram? (1)

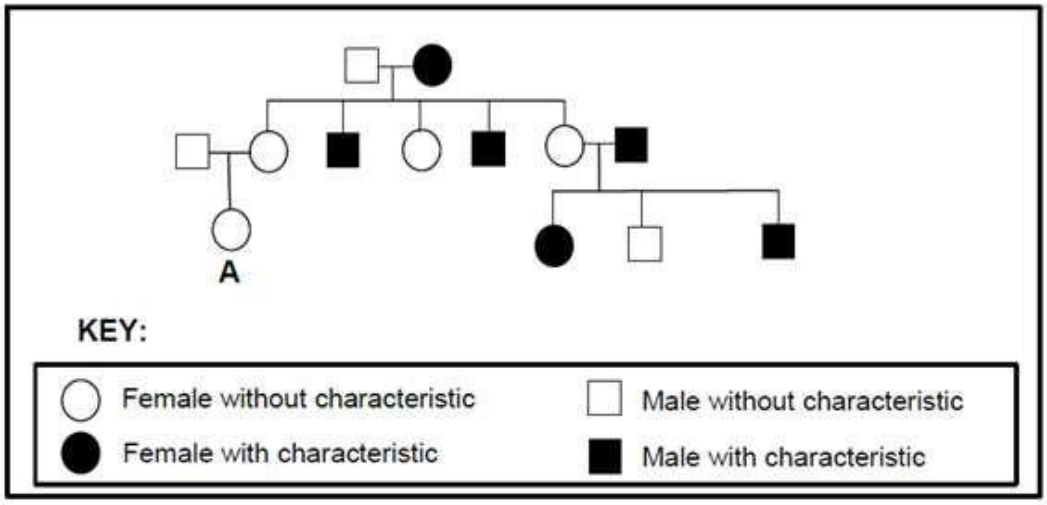
2.2.4 Lina's genotype is $I^A i$.

State ALL the possible genotypes of Vusi. (2)

2.2.5 Give the genotype of Bob. (1)

2.2.6 Give the name of the individual which displays co-dominance. (2)
(8)

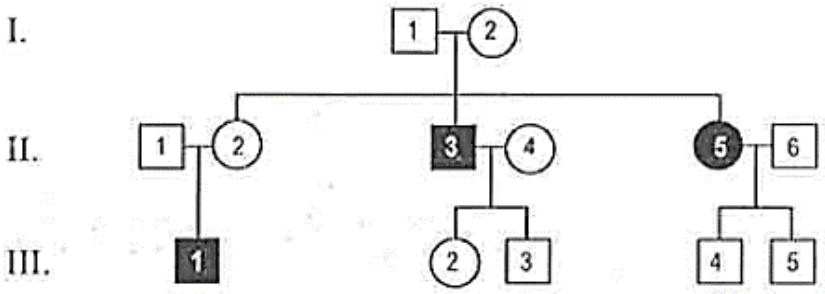
2.3 The pedigree diagram shows inheritance of a certain characteristic.



2.3.1 Use X^N and X^n to represent the relevant alleles of the characteristic. The possible genotype(s) of individual **A** will be …

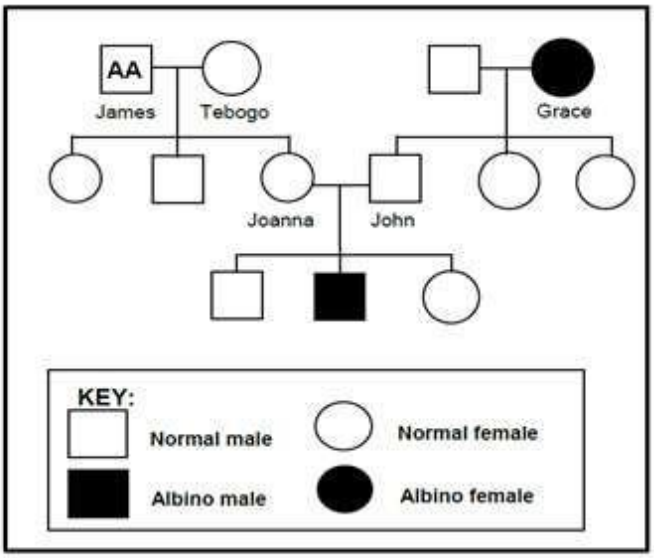
A X^NX^n only.
B X^NX^N only.
C X^NX^N and X^NX^n.
D X^NX^N and X^nX^n.

2.3.2



The shaded circles or squares in a pedigree indicate …
A individuals who have produced offspring.
B individuals who do not have the trait bing shown.
C individuals who siblings.
D Individuals who have the trait being shown.

2.4 Albinism is an inherited condition caused by a recessive gene mutation. This mutation results in the absence of the protein melanin in the skin. The pedigree diagram below shows the inheritance of albinism in a family. The genotype of James is shown in the diagram.



2.4.1 How many grandsons do James and Tebogo have? (1)

What is:

(a) Grace's phenotype (1)

(b) John's genotype (2)

2.4.2 John and Joanna wish to have another child. What is the percentage chance that the child will:

(a) Be a girl (1)

(b) Have albinism (1)
(6)

END OF AUTUMN SCHOOL 2024

TOPIC 3: DIHYBRID CROSSES AND MUTATIONS

Outcomes

By the end of this topic you should be able to:

Know what is meant by a dihybrid cross
Understand Mendel's principle of Independent Assortment
Determine the proportions/ratios of the genotypes and phenotypes in a dihybrid cross
Know how to determine the gametes of the parents in a dihybrid cross
Define a mutation
Know the effects of mutations
Explain how mutations contribute to genetic variation
Explain how mutations lead to altered characteristics resulting in genetic disorders like Haemophilia, Colour-blindness and Down syndrome.

GENETICS AND INHERITANCE Paper 2: 45 marks	**Term 2**	**4 weeks**
Dihybrid crosses	❑ Mendel's Principle of Independent Assortment ❑ Dihybrid genetics problems ❑ Determination of the proportion/ratio of genotypes and phenotypes	
Mutations	❑ Definition of a mutation ❑ Effects of mutations: harmful mutations, harmless mutations and useful mutations ❑ Mutations contribute to genetic variation ❑ Definition of gene mutation and chromosomal mutation ❑ Mutations lead to altered characteristics in each of the following genetic disorders: • Haemophilia – absence of blood-clotting factors • Colour-blindness – due to absence of the proteins that comprise either the red or green cones/photoreceptors in the eye • Down syndrome – due to an extra copy of chromosome 21 as a result of non-disjunction during meiosis	

3.1 Dihybrid cross

A dihybrid cross involves the inheritance of two characteristics. Mendel explained the results obtained from dihybrid crosses according to his Principle of Independent Assortment.

According to **the Principle of Independent Assortment**, the various 'factors' controlling the different characteristics are separate entities, not influencing each other in any way, and sorting themselves out independently during gamete formation.

This means that the two characteristics are transmitted to the offspring independently of one another.
•The above law only applies if the genes for the two characteristics are not on the same chromosome.

Worked Example

In pea plants, the allele for tallness (T) is dominant and the allele for shortness (t) is recessive. The allele for purple flowers is dominant (P) and the allele for white flowers is recessive (p). Two plants, heterozygous for both tallness and purple flowers, were crossed.

<table>
<tr><th>Step</th><th>What to do generally</th><th>What to do in this problem</th></tr>
<tr><td>Step 1</td><td>Identify the phenotypes of the two plants for each of the two characteristics.</td><td>According to the statement of the problem, both parents are tall and have purple flowers.</td></tr>
<tr><td>Step 2</td><td>Choose letters to represent the alleles for the gene responsible for each characteristic.</td><td>Use the letters, e.g. t for tall, t for short, P for purple, and p for white as provided in the question.</td></tr>
<tr><td>Step 3</td><td>Write the genotypes of each parent.</td><td>According to the statement of the problem, both parents are heterozygous for each characteristic. Their genotype will therefore be ttPp.</td></tr>
<tr><td>Step 4</td><td>• Determine the possible gametes that each parent can produce.
• Remember that each parent will have two alleles for each gene.
• The gametes of each parent will have only one allele for each gene because of segregation during meiosis.
• Remember that because of the principle of independent assortment an allele for one gene could appear in the same gamete with any of the alleles for the other gene.</td><td>Each parent has the genotype TtPp.
If we represent the alleles for each gene in the following format, then we can see how these alleles could come together randomly (principle of independent assortment) to form the four types of gametes: TP; Tp; tP and tp as shown below.
<table>
<tr><td>Alleles</td><td>T</td><td>t</td></tr>
<tr><td>P</td><td>tP</td><td>tP</td></tr>
<tr><td>p</td><td>tp</td><td>tp</td></tr>
</table></td></tr>
<tr><td>Step 5</td><td>Enter the possible gametes at the top and side of a Punnett square.</td><td>Please refer to the solution that follows.</td></tr>
<tr><td>Step 6</td><td>Because of random fertilisation, gametes from both parents could fuse in different combinations to form the offspring.

In the punnet square, write down the genotypes of the offspring that will result</td><td>Please refer to the solution that follows.</td></tr>
<tr><td>Step 7</td><td>Determine the phenotypes of the offspring from the genotypes obtained in the punnet square.</td><td>Please refer to the solution that follows.</td></tr>
</table>

NOTE: 1. You may not mix the letters of the different characteristics when writing a genotype of an individual in a dihybrid cross. E.g. TPtp instead of TtPp

2. Do not leave a space between the letters constituting a genotype of an individual in dihybrid cross.

E.g. Tt Pp instead of TtP

Solution to the problem

P 1 Phenotype Tall, Purple × Tall,,Purple ... Step 1

Genotype TtPp × TtPp .... Step 2,3

Meiosis

Fertilisation

gametes	tP	tp	tP	tp
tP	TTPP	TTPp	TtPP	TtPp
tp	TTPp	TTpp	TtPp	Ttpp
tP	TtPP	TtPp	ttPP	ttPp
tp	TtPp	Ttpp	ttPp	ttpp

Steps 4-6

F 1 Genotype 9 different genotypes, as in the table above

Phenotype 9 tall, purple flowered plants (T–P–);
3 short, purple flowered plants (ttP–);
3 tall, white flowered plants (T–pp), and
1 short, white flowered plant (ttpp). ..... . Step 7

Worked example 2 In hamsters, the allele for blackcoat colour (B) and is dominant over the allele for white coat colour (b).

The allele for rough coat (R) is dominant over the allele for coat (r).

If you cross a hamster that is heterozygous black and homozygous rough, with one that is Heterozygous black and heterozygous rough, what will be the phenotypes and genotypes of the offspring? (Use the steps 1–7 to arrive at an answer)

Answer

P 1 Phenotype Black, Rough coat × Black, Rough coat ...

Genotype BbRR × BbRr .....

Meiosis

Fertilisation

gametes	BR	BR	bR	bR
BR	BBRR	BBRR	BbRR	BbRR
Br	BBRr	BBRr	BbRr	BbRr
bR	BbRR	BbRR	bbRR	bbRR
br	BbRr	BbRr	bbRr	bbRr

F 1 Genotype 6 different genotypes, as in the table above

Phenotype 12 with a black, rough coat and 4 with a white, rough coat .....

Activity 1

DBE Life Sciences NOV 2014 P2

1.1 About 70% of people get a bitter taste when a substance called PTC is placed on their tongue. They are referred to as 'tasters'. All other people are unable to taste PTC and are referred to as 'taste-blind'. The 'taster' allele is dominant and the 'taste-blind' allele is recessive.

Also in humans, normal skin pigmentation is dominant to the albino condition (no pigmentation).

The letters in the key below must be used to represent the alleles for the different characteristics above.

Key:

T – taster **t**
– taste-blind
N – normal skin pigmentation
n – no skin pigmentation
(albino)

A man who is heterozygous for both tasting PTC and skin pigmentation marries a woman who is taste-blind for PTC and is an albino.

1.1.1 State why the example above represents a dihybrid cross. (1)

1.1.2 Write down:

(a) The genotype of the woman (1)

(b) ALL the possible gametes of the man (2)

1.1.3 The man and woman have a child whose genotype is **ttNn**. What is the child's phenotype? (2)

1.1.4 A man and a woman are only able to produce children with the genotype **TtNn**. The woman's genotype is **ttnn**. State the only possible genotype of the man. (2)

(8)

1.2 DBE Life Science June 2016

1.2 In rice plants the allele for high yield (**H**) is dominant over the allele for low yield (**h**). The allele for a tall stem (**T**) is dominant over the allele for a short stem (**t**).

There are two varieties of rice plants, **A** and **B**.
The genotype of variety **A** is **HHtt**.
The genotype of variety **B** is **hhTT**.

A plant breeder wants to produce a rice plant variety with a high yield and a short stem.

1.2.1 Give the phenotype of variety **A**. (2)

1.2.2 Give ALL the possible genotypes of the gametes of variety **B**. (1)

1.2.3 Give the genotype(s) of the variety the plant breeder wants to produce. (2)

1.2.4 Explain why the plant breeder would want to produce a rice plant with a short stem. (1)

1.2.5 Describe how the plant breeder would be able to produce rice plants with a high yield and short stems only. (2)
(8)

DBE Life Science Nov 2016 P2

1.3 The leaf colour in a plant is controlled by two alleles, green (**G**) and yellow (**g**). Thorns on plant stems are controlled by two alleles, presence of thorns (**T**) and no thorns (**t**).

Two plants with the genotypes **GGTT** and **ggtt** were crossed. Their offspring were then left to pollinate each other.

The table below shows the possible genotypes of the offspring of the second generation. Genotypes **(i)** and **(ii)** have been left out.

Gametes	GT	Gt	gT	gt
GT	GGTT	GGTt	GgTT	GgTt
Gt	GGTt	GGtt	**(i)...**	Ggtt
gT	GgTT	GgTt	ggTT	ggTt
gt	GgTt	Ggtt	ggTt	**(ii)...**

1.3.1 Give the:

(a) Genotype of **(i)** (1)

(b) Phenotype of **(ii)** (2)

1.3.2 List the FOUR genotypes of the offspring of the second generation that would be phenotypically different from the original pair of parents. (4)
(7)

3.2 Mutations

- A mutation *is a sudden change in the genetic composition of an organism*. Mutations occur suddenly and randomly.
- A gene mutation *is a change in the sequence of nitrogenous bases or nucleotides* in DNA
- A Chromosomal mutation *is a change in the normal structure or number of chromosomes*. May be due to no separation of a chromosome during Anaphase I and II in Meiosis

NOTE: DESCRIPTIONS OF POINT AND FRAMESHIFT MUTATIONS ARE NOT REQUIRED

Effect of Mutations

Harmful mutations
Cause changes in DNA that can cause errors in protein sequencing, that can result in partially or completely non- functional proteins.

Harmless mutations have no effect on the structure or functioning of the organism.

Useful mutations can be advantageous to the organism, and they are passed on from parent to offspring.

Mutations result in new genotypes as we move from one generation to the next. This leads to variation within a species.

Gene mutations can cause genetic disorders:

- Haemophilia: Absence of the protein needed for the formation of blood clots due to a mutant gene.
- Colour blindness: Absence of the proteins that make up either the red or green cones/photoreceptors in the eye.

Activity 3

3.1 Describe how a gene mutation may result in the formation of a protein that is different from the one that is required. (6)

3.2 Read the information below.

> A gene, VKORC1, codes for a blood-clotting factor in humans. This gene codes for a protein that is made up of 163 amino acids.
>
> A mutation occurred that affected amino acid 128 and 139, the sequence CTG changed to CAG and the TAT became TCT. This mutation has been transmitted as an autosomal dominant characteristic through the generations.
>
> The mutation has resulted in resistance to Warfarin drugs in humans. Warfarin is used in the treatment of thrombosis. Thrombosis results in the formation of a blood clot in the artery. Warfarin causes the thinning of blood to break down the blood clot.

3.2.1 Give ONE piece of evidence from the information that shows that the mutation for this gene occurred in the DNA molecule. (1)

3.2.2 How many nitrogenous bases code for the VKORC1 gene? (2)

3.2.3 Describe what is meant by an autosomal dominant allele (3)

3.2.4 The table below shows the amino acids and their corresponding codons.

CODONS	AMINO ACID
GAC	Leu
UCU	Ser
AUA	Try
GUC	Gln
AGA	Arg
ACA	Trp
CAG	Gln
UAU	Phe

Explain:

(a) How the mutation on the VKORC1 gene resulted in resistance to Warfarin in humans (5)

(b) The effect of this mutation on humans with thrombosis (3)

(14)

TOPIC 4: GENETIC ENGINEERING

Outcomes

By the end of this topic you should be able to:

Know how genetic engineering uses biotechnology to satisfy human needs

Know what stem cell research is it sources and uses

Outline the process of genetic modification

Know the benefits of genetic modification and genetic modified organisms

Outline the process and benefits of cloning

Examination guideline

GENETICS AND INHERITANCE Paper 2: 45 marks	Term 2	4 weeks

CONTENT	ELABORATION
Genetic engineering	❑ Genetic engineering uses biotechnology to satisfy human needs: • Stem cell research – sources and uses of stem cells • Genetically modified organisms – brief outline of process (names of enzymes involved are not required) and benefits of genetic modification • Cloning – brief outline of process and benefits of cloning

Genetic engineering is the process whereby the genes on the DNA are changed, transferred or manipulated to produce a different organism.

Four disadvantages of genetic engineering:

- Expensive / research money could be used for other needs
- Interfering with nature / immoral
- Potential health impacts
- Unsure of long-term effects

Four advantages of genetic engineering:

- Production of medication / resources cheaply
- Control pests with specific genes inserted into a crop
- Using specific genes to increase crop yields / food security
- Selecting genes to increase shelf-life of plant products
-

4.1 Stem cell Research

What is a stem cell?
S**tems cells** are **undifferentiated cells** that have the **ability to develop into any tissue or organ in the body**.
These **stem cells** are called **embryonic stem cells**.

Uses of Stem Cells:

- They can be used to **replace diseased or damaged cells**.
- At **present** it is used for the **treatment of some diseases** such as **diabetes, leukemia, Alzheimer's disease, osteoporosis and sickle cell anemia**. But it is only being **used to a limited extent**.

Harvesting of Embryonic Stem Cells:

As the name suggests **embryonic stem cells come from the embryo**.

- Usually when couples undergo **in-vitro fertilization** some **embryos are not implanted**
- The embryos not used are frozen and stored if. needed in the future
- Embryonic stem cells are harvested from these frozen embryos • But once the stems cells are removed the embryos are destroyed.
- But in a very positive development it was found that…
- If one cell is removed when the embryo was in the 8 cells stage the embryo survives and continues to develop as normal.
- In this case a stem cell is harvested without causing harm to the embryo
- **The cord blood is a rich supply of stem cells.**

A second source *is from the umbilical cord* which is cut after birth and stem cells are collected. These stem cells are stored in certain facilities. The stem cells stored may be used to develop tissues or organs that may need to be replaced from that child in the future.

Worked example

Describe what stem cells are, and give TWO sources from which human stem cells may be harvested. Also explain TWO arguments, with reasons, for the use of stem cells and TWO arguments, with reasons, against the use of stem cells in humans.

Answer

Stem cells are (actively) dividing cells that are not yet differentiated/ not yet mature could give rise to different types of cells

Sources:

Embryo
Blood in umbilical cord/cord blood
Placenta
Bone marrow

Arguments for use of stem cells

- Provide replacements for tissues /organs damaged by age/trauma/disease/improve quality of life
- Used for research to see whether it can cure different diseases e.g. cancer/more reliable results when human stem cells are used
- Stem cells from e.g. the blood from the umbilical cord can be stored when needed in future because it would not be rejected by the body's immune system

Arguments against use of stem cells

- Expensive, research money could be used for other needs
- Only rich people /expensive can afford to store stem cells for later use
- Interfere with religion /culture/creation because it is immoral/ unethical/we cannot play God
- Moral /ethical objection because we are destroying a human life
- The dangers of using stem cells are unknown and may be a risk
- Can lead to illegal trade in embryos /the placentas of new-born babies/ to make money
- Embryos conceived and then aborted abandoned/ to use the stem cells from the placenta

Activity 1
DBE Life Sciences Nov 2022 P2

1.1 Read the extract below.

> When a child is born, the umbilical cord is cut and stem cells can be obtained from it. Many people think that the stem cells for treating human conditions should be obtained from umbilical cords, rather than from human embryos.
>
> Recently, stem cells have also been obtained from bone marrow. These stem cells are used to treat conditions such as heart disease and spinal injuries.

1.1.1 Name THREE sources of stem cells mentioned in the extract. (3)

1.1.2 Explain why the characteristics of stem cells make them useful for treating some disorders. (2)

1.1.3 Name ONE condition in the extract that can be treated with stem cells. (1)
(6)

4.2 Genetic modification

- **Genetic Engineering:** the process where scientists alter, swap or manipulate the genes on the DNA, to produce a different organism. Genetic engineering involves the **transfer of genes** from one organism to an unrelated species.

Diabetics are people who cannot produce their own **insulin**. Insulin is a **hormone** needed to regulate blood sugar. Biochemists have devised a way in which to produce artificial insulin. A similar process is used to genetically modify many different organisms. Make sure you know the basic process:

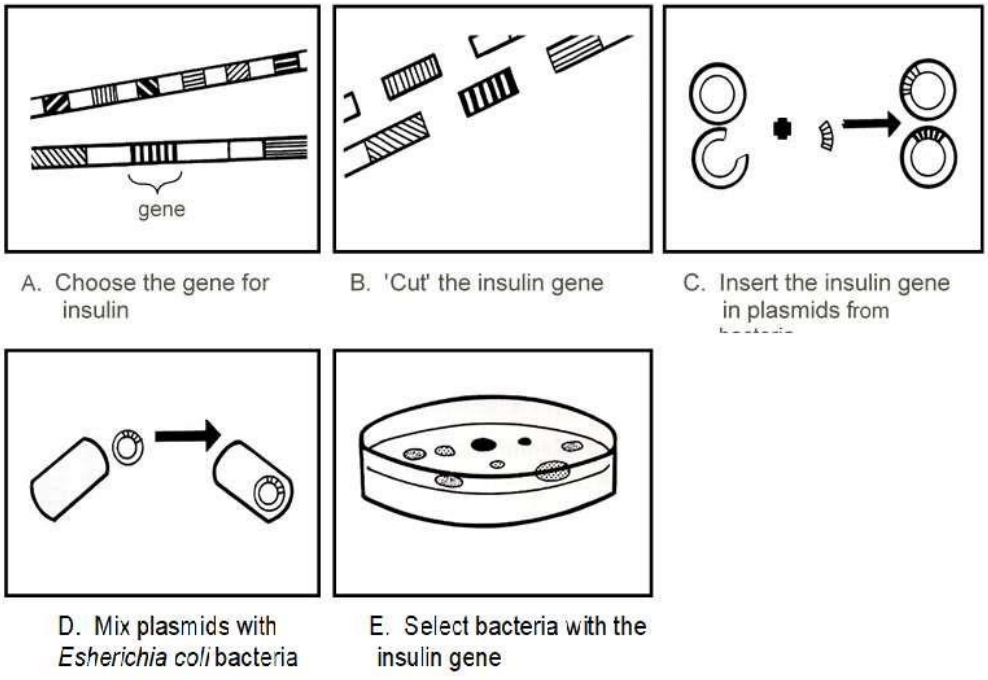
An example is were bacteria is modified to produce insulin

- **Genetic Engineering:** the process where scientists alter, swap or manipulate the genes on the DNA, to produce a different organism. Genetic engineering involves the **transfer of genes** from one organism to an unrelated species.

Diabetics are people who cannot produce their own **insulin**. Insulin is a **hormone** needed to regulate blood sugar. Biochemists have devised a way in which to produce artificial insulin. A similar process is used to genetically modify many different organisms. Make sure you know the basic process:

- Bacteria produce **restriction enzymes** that 'cut' DNA molecules. These restriction enzymes are extracted from the bacteria.
- DNA is removed from a healthy person's pancreas cells.
- Restriction enzymes are used to 'cut' out a piece of DNA, which contains the **genes** that produce insulin.
- When the genes are transferred from one organism's cells to another, the DNA in the recipient cell is called **recombinant DNA**. Insulin is produced by using recombinant DNA in the bacterium called *Escherischia coli* (*E. coli*), which lives in the human gut.
- **Plasmids** are taken out of a bacterium and cut open with the restriction enzyme.
- The human genes are inserted into the plasmid. The healthy bacterium absorbs the plasmids.
- The piece of human DNA continues to produce proteins, which make insulin, inside the bacterium.
- The insulin is then extracted from the bacterium cell.
- Diabetics inject themselves with this insulin everyday, so that they can regulate their blood sugar.

The diagram shows the production of Insulin



Advantages of GM crops	Disadvantages of GM crops
• Better nutritional value • Greater crop yield resulting in higher food production and long term reduction in costs • Crops are better adapted to less favourable environments and climates • Disease and pest resistant means less insecticides and pesticides are released into the environment • Increased biodiversity as new varieties are developed • Efficient use of scarce agricultural land as well as land that may previously not have been fit	• Possible increase in allergen and carcinogen levels • Nutritional changes and possible toxicity • Traits can be transferred to other species like weeds • Cause un-natural selection pressure • Expensive start-up costs • DNA alteration of the new varieties are owned and controlled by biotechnology company which may lead to issues with ethics and ownership • Lack of transparency regarding what is in our food

Activity 2

DBE Life Sciences Nov 2016 P2

2.1 Read the passage below.

GENETIC MODIFICATION OF CROPS

Genetic modification (GM) of crops began with the discovery that the soil bacterium *Agrobacterium* could be used to transfer useful genes from unrelated species into plants.

The gene called **Bt**, which produces a pesticide toxin that is harmless to humans, but is capable of killing insect pests, is one of the genes most commonly inserted into crop plants. Many new GM crops, such as maize, potatoes and tomatoes, are modified to be pest, disease or weed-killer resistant.

GM foods could have unforeseen effects. Toxic proteins may be produced or antibiotic-resistance genes may be transferred to human gut bacteria. Modified crops could become weed-killer resistant 'super weeds'. Modified crops could also 'accidentally' breed with wild plants or other crops.

[Adapted from GM Organisms www.newscientist.com]

2.1.1 According to the passage, how did genetic modification of crops begin? (1)

2.1.2 Explain why a plant, which is modified to be weed-killer resistant, could be a problem for farmers. (2)

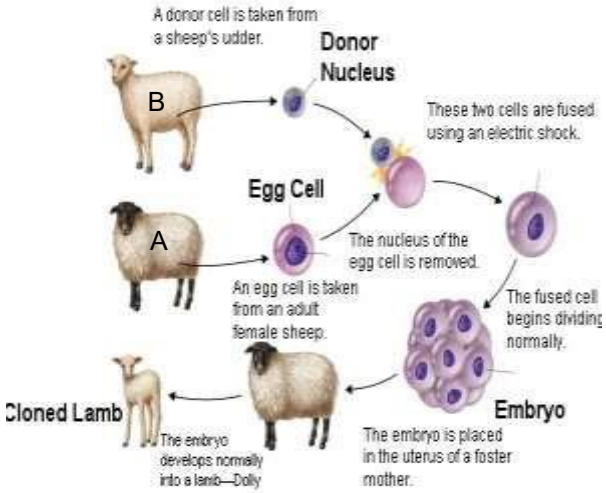
2.1.3 Give TWO examples in the passage of the use of GM crops that may be a potential threat to human health. (2)
(5)

4.3 Cloning

Cloning is the production of an individual which is genetically identical to the one from which it was produced, using biotechnology. Cloning of mammals such as rabbits, guinea pigs, cows, sheep and even humans following the process described below:

The Process of cloning

- They removed an ovum from a sheep by surgery.
  Let us call this Sheep A.
- They then removed the nucleus of this ovum.
- They then removed an actively dividing somatic cell from another sheep of the same species. Let us call this sheep B.
- They carefully took out the nucleus from a cell of sheep B.
- The nucleus of sheep B was inserted into the ovum of sheep A.
- The ovum, now with a diploid number of chromosomes, was put back into sheep A.
- This ovum behaved just like a fertilised egg. It became attached to the 'womb' and developed into a new individual. In other words,

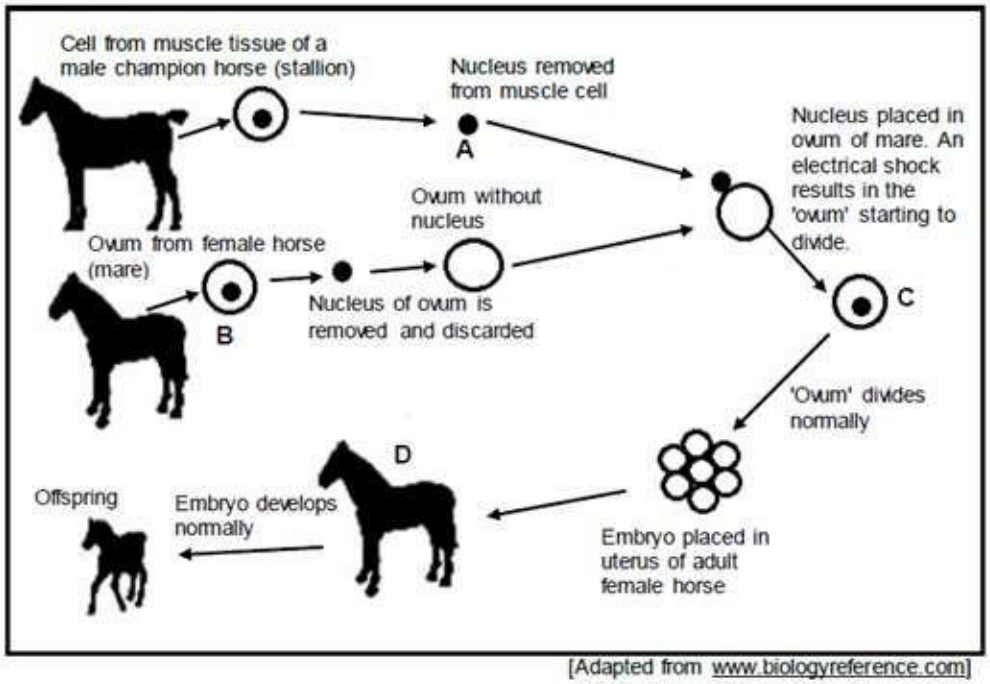


- Remember that since the nucleus came from rabbit B, the new individual is therefore a **clone** of rabbit B.

Advantages of Cloning	Disadvantages of Cloning
Produce individuals with desired traits	Objections to interfering with God's creation
Better yields	Reducing the gene pool by reducing variation
Resistant to diseases	Cloned organisms may have developmental problems
Organisms produced in a shorted time	Costly process
Saving endangered species	Generate experimental waste
Produce body parts/organs for transplant	May lead to killing clones for organs/body parts
Produce offspring when organisms are infertile	Cruelty to animals and inhumane behaviour

Activity 3
DBE Life Sciences March 2016 P2

3.1 The diagram below shows a genetic engineering process. A donor cell was taken from the muscle cell of a male champion horse (stallion) to create a new offspring.



[Adapted from www.biologyreference.com]

3.1.1 Name the:

(a) Genetic engineering process shown in the diagram above (1)

(b) Process that produced ovum **B** (1)

3.1.2 Why is the donor cell extracted from a champion horse? (2)

3.1.3 Explain why only the nucleus of the donor cell is used. (2)

3.1.4 A somatic cell in a horse contains 64 chromosomes.

3.1.5 How many chromosomes would there be in:

(a) Structure **A** (1)

(b) Ovum **B** (1)

(c) A muscle cell in organism **D** (1)

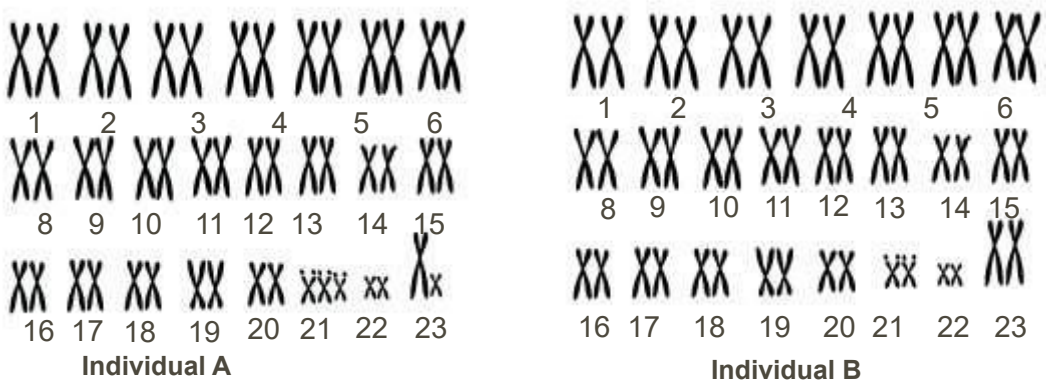
3.1.6 Explain why the 'ovum' labelled **C** cannot be considered a gamete. (2)
(11)

Summative Task

Pedigree diagrams , Karyotypes and Genetic disorders

ANSWER ALL THE FOLLOWING QUESTIONS

1. The diagrams below show the sets of chromosomes (karyotypes) in two human individuals, A and B. Study the diagrams and answer the questions that follow.



Karyotypes of two individuals

1.1 Describe what is a karyotype of an individual. (2)

1.2 Which individual (A or B) is female? (1)

1.3 Give a reason for your answer to QUESTION 1.2. (1)

1.4 How many chromosomes are present in each cell of Individual A (1)

1.5 How many chromosomes is present in each cell of a normal person. (1)

1.6 Identify which individual (A or B) has an abnormal number of chromosomes. (1)

1.7 Name the genetic disorder that the individual in QUESTION 1.6 has. (1)

1.8 Explain the abnormal chromosome number of the disorder named in QUESTION 1.7 (2)

1.9 Give THREE symptoms of a person that suffers from the disorder named in QUESTION 1.7. (3)

(13)

2. A group of Grade 12 learners did a survey about the frequency of genetic disorders in a small human population. The results are shown in the table below: The number of people in the population is 1 200.

Results of the survey on the frequency of genetic disorders in a population

GENETIC DISORDERS	FREQUENCY IN POPULATION (%)
Albinism	7
Down's syndrome	**x**
Sickle cell anaemia	10
Without any disorder	78

2.1 According to the table, which disorder is most common in this community? (1)

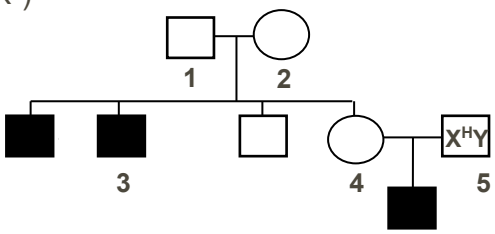
2.2 Calculate the number of people with Down's syndrome. Show ALL working. (2)

2.3 Give ONE reason why genetic counseling would be important for parents who are expecting children that may be sufferers of a disorder such as Down's syndrome. (2)

(5)

3. In humans a gene responsible for clotting of blood is carried on the X-chromosome. Males who carry the recessive gene, bleed easily and are called hemophiliacs. Females will be only have hemophilia if they have two recessive genes. The diagram below shows the occurrence of hemophiliacs in a certain family after surveys were carried out.

Note: H = normal clotting
(X^H) h = hemophilia (X^h)



Non-hemophiliac male

Hemophiliac male

Non-hemophiliac female

3.1 Write down the genotypes of individuals numbered 1 to 4. **(8)**

4.1 A grade 12 learner planned an investigation to establish the attitudes and beliefs of high school learners about albinism. She asked THREE questions in the survey:

1. Should mothers alone take the blame for children born with albinism?
2. Are people with albinism cursed and need to be hidden from public view?
3. Should people with albinism be employed?

The results of the survey are shown in the table below. Study the table and answer the questions that follow.

Results of survey on people's attitudes and beliefs about albinism

	Number of people	
	Yes	**No**
Question 1	40	10
Question 2	30	20
Question 3	5	45

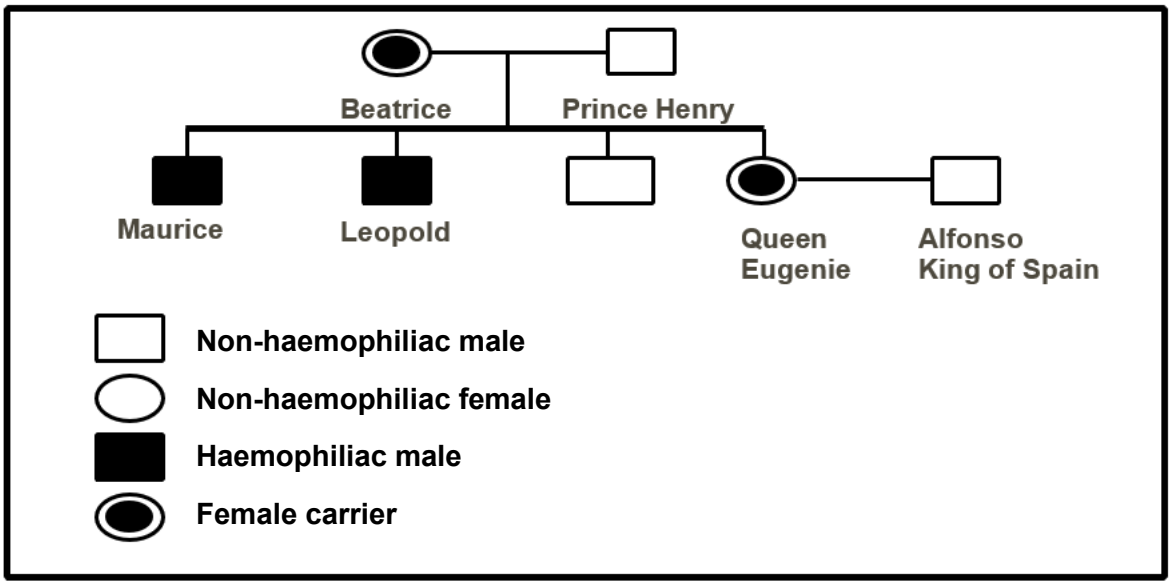
4.1 How many people participated in this survey? (1)

4.2 Draw a bar graph of the results. (10)

4.3 State conclusions from the results of each the THREE questions asked (6)

(17)

5 Study the pedigree diagram, which shows the inheritance of haemophilia and answer the questions that follow.



5.1 What is haemophilia? (2)

5.2 What causes this disease? (3)

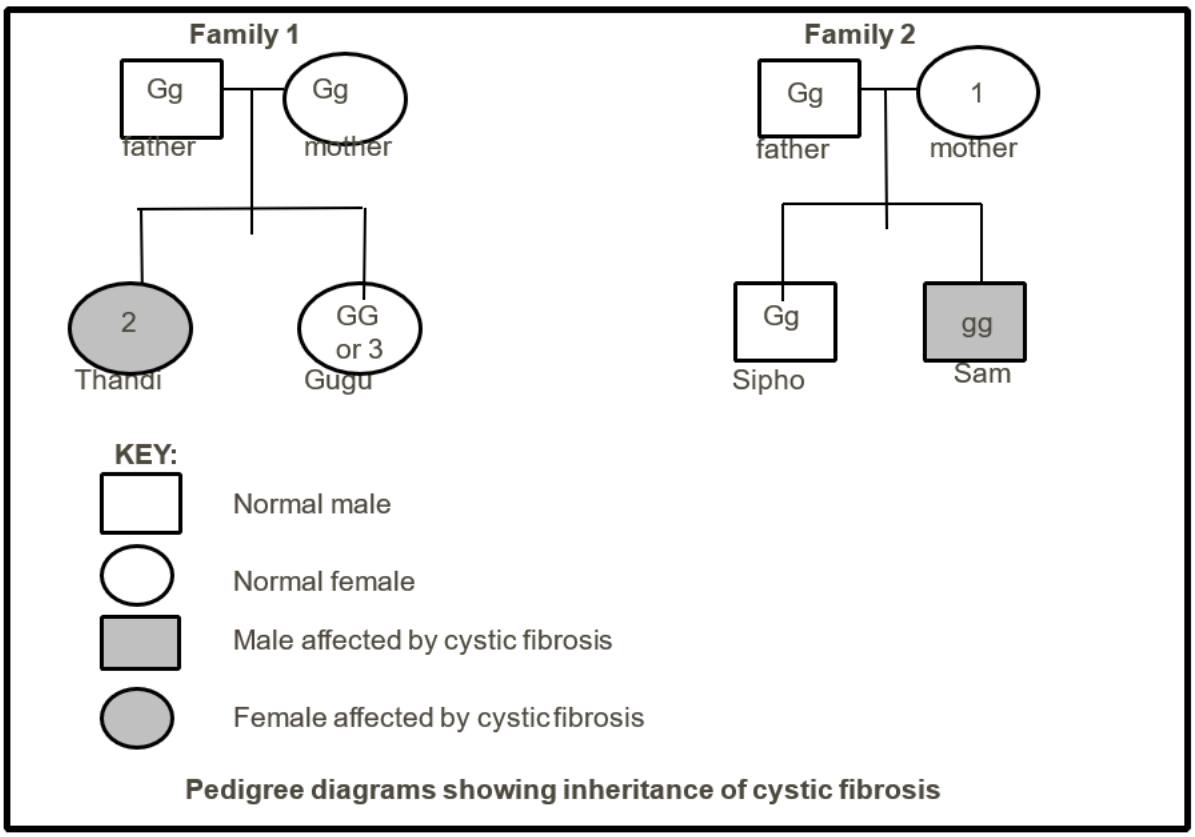
5.3 Why is Queen Eugenie only a carrier but does not suffer from haemophilia? (2)

5.4 Why is this disease said to be sex-linked? (2)

5.5 Explain why one male in 10 000 gets this disease whereas only one female in 100 000 000 gets it? (2)

5.6 Explain why the pedigree above confirms your answer to QUESTION 5.5. (2)
(13)

6 Cystic fibrosis is an inherited disorder of the human body caused by a recessive gene. This disorder affects mucus production causing blockage of tiny air passages in the lungs. Study the pedigree diagrams below and answer the questions that follow.



Pedigree diagrams showing inheritance of cystic fibrosis

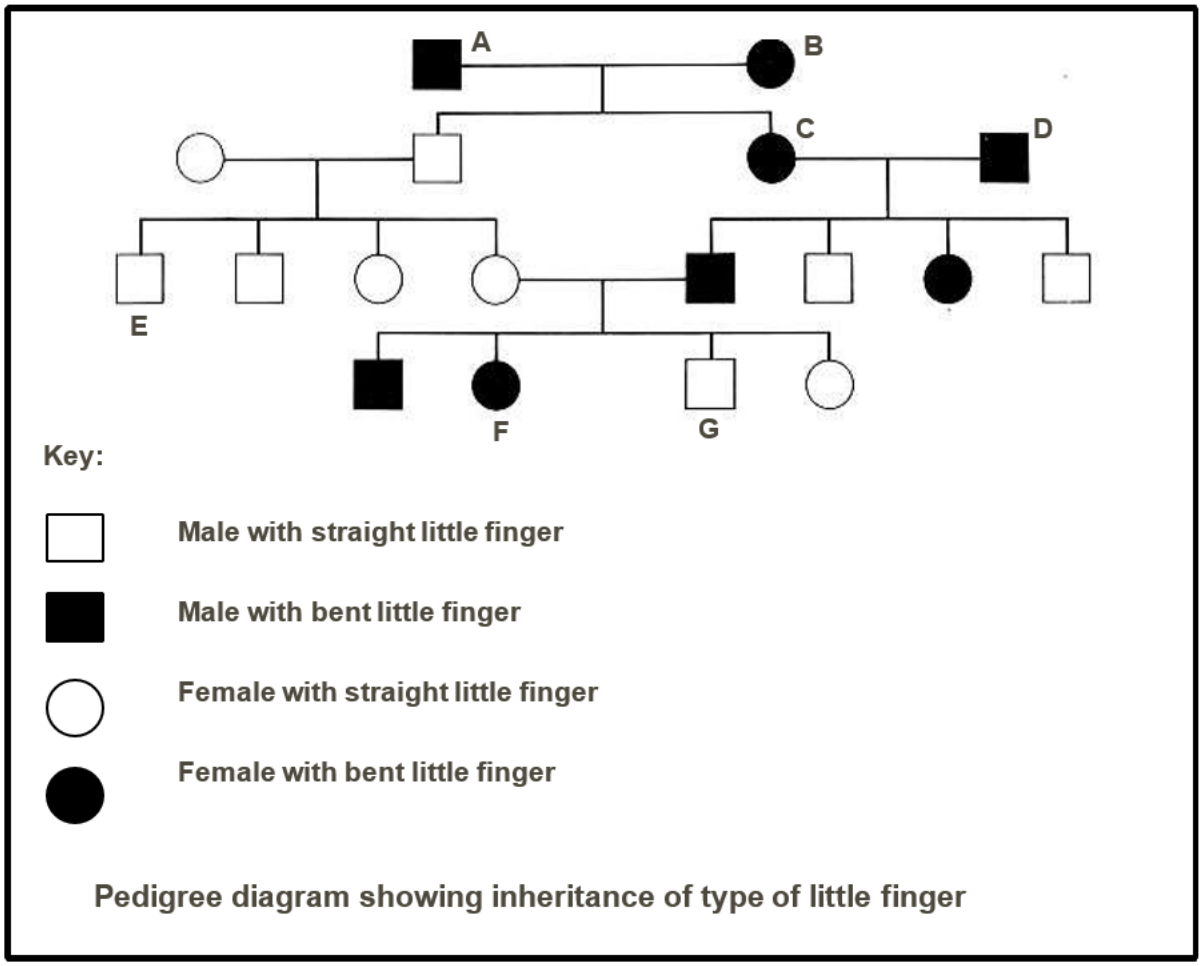
6.1 Name the genotypes represented by 1 and 3 in the diagrams respectively (2)

6.2 What is Thandi's genotype? (1)

6.3 Does Thandi suffer from cystic fibrosis (1)

6.4 Thandi and Sipho intend getting married. Show, using a pedigree diagram and the key above, ALL the possible genotypes of any sons they might have. (2)
(6)

7 Study the pedigree diagram below which shows inheritance of type of little finger over four generations in a family.



Pedigree diagram showing inheritance of type of little finger

7.1 How many members of the family have straight little fingers? (1)

7.2 Is person **E** homozygous or heterozygous for the type of little finger? (1)

7.3 Which type of little finger is controlled by the dominant gene? (1)

7.4 Explain your answer to QUESTION 7.3. (3)

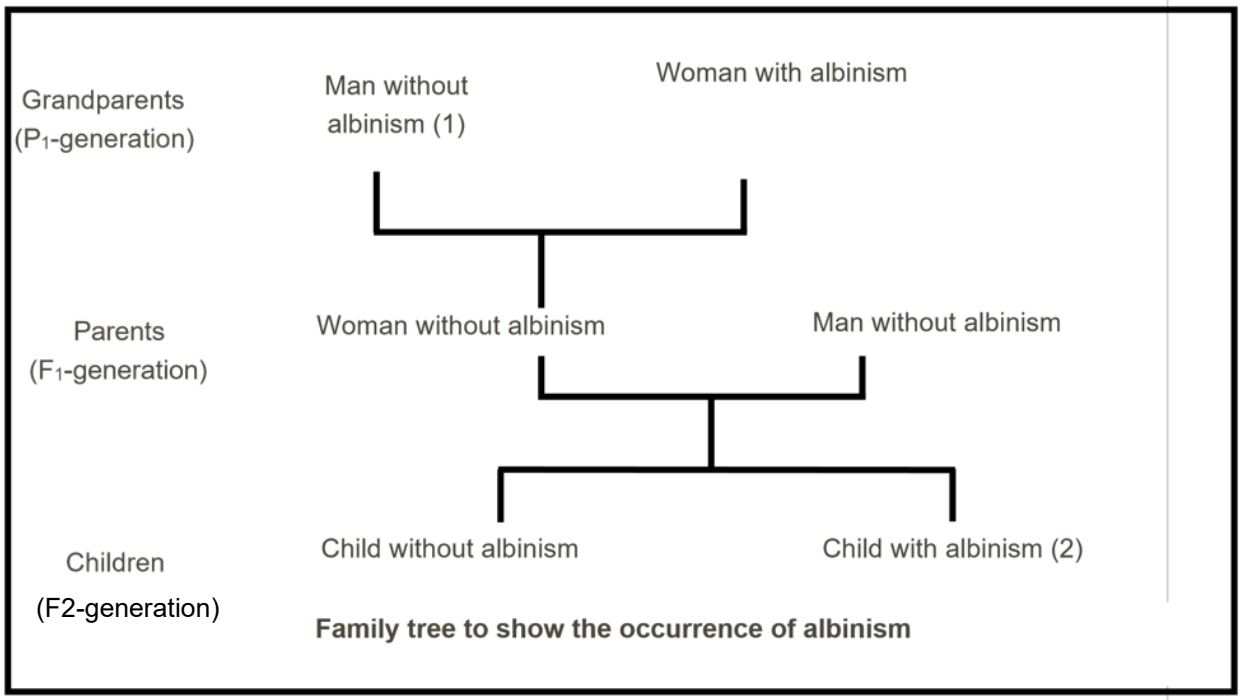
7.5 In the fourth generation of the family what proportion will be female with straight little finger? (2)

7.6 Persons F and G are twins. Were they produced from a single fertilized egg cell? (1)

7.7 State TWO reasons based on the phenotype to support your answer in QUESTION 7.6 (2)
(11)

8 People with albinism are unable to produce the dark pigment, melanin, in their skin. This condition is caused when an individual is homozygous recessive for this characteristic.

The pedigree diagram below shows the occurrence of albinism over three generations.



Family tree to show the occurrence of albinism

8.1 Indicate whether each of the individuals below could be homozygous dominant, homozygous recessive or heterozygous.

(a) 1
(b) 2

(3)

8.2 Explain your answer to QUESTION 8.1 (a) (2)

Bibliography

Bowie, M; Johannes, A; Mhlongo, R Pretorius, E. 2011. *Life Sciences Grade 12 Learner's book*. Via Afrika

Joyce, BR; Weil, M & Calhoun, E, 1986. *Models of teaching* (Vol. 499). Englewood Cliffs, NJ: Prentice-Hall.

Isaac, Chetty S et al.: 2011 Life Sciences grade 12 Pulse