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**MENDEL'S 1<sup>ST</sup> LAW, SEX AND BLOOD GROUP DETERMINATION**

**Learner Note:** Mendel's Laws are very important and you must understand the basic concepts of Genetics. Check the Additional Notes in Session 12. You must understand the concepts of dominance and how this plays a role in monohybrid crosses (mono = one = one characteristic or trait). Be aware of confusing the word 'cross/ crossing' with 'crossing over' in Meiosis. You cross individuals and calculate the chances of a characteristic or trait being in the offspring. "Crossing over" takes place in Meiosis during prophase where pieces of chromosomes cross over from the male chromosomes to the female chromosomes to ensure a mix of the characteristics in the offspring. You must be clear of the difference between these two terms. Questions on blood group inheritance and sex determination are often asked during examinations. The more examples of genetic crosses you do, the better you will be.

**SECTION A: TYPICAL EXAM QUESTIONS – PLEASE adhere to the time allocations.**

**Question 1:** (Taken and adapted from Study & Master Biology Grade 11) – 6 minutes

1. Blood typing can be used to identify a parent in that the blood type can prove that a person is not the parent of a child rather than determine without question who the parent is. A, B, AB and O blood groups are the result of three alleles. Allele A and B are incompletely dominant and O is recessive to both A and B. Should the discrepancy continue, tissue typing and DNA fingerprinting will be used. Read through the following information and answer the questions below:

In a maternity ward of a hospital, two newly born babies were mixed up. One baby is blood type O and the other is type A. Both mothers believe the baby with blood type O is their baby. Can you sort it out? On testing the parents' blood it was found that:

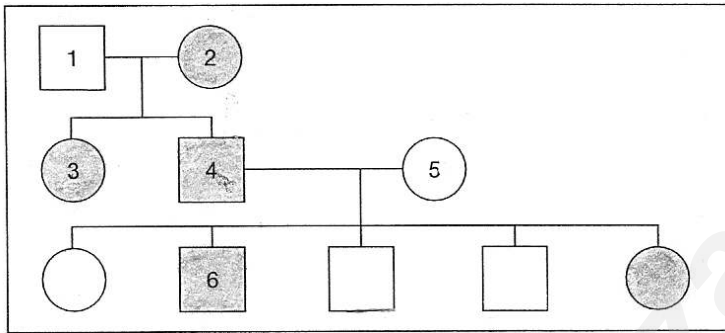
1. Mr. Xhosa is blood group AB and his wife is blood group B
2. Mr. Mbundwini is type A.

Who owns baby 'O' and who owns baby 'A'? Explain / show your reasoning. (6)

**(Remember that in blood groups there are three alleles A, B and O. A and B are co-dominant over O which is recessive. There must be two of the same alleles if a recessive trait is present in the individual)**

**Question 2:** (Taken and adapted from Study&Master Biology Grade 11) - 11 minutes

2. The diagram below shows the inheritance of eye colour in humans. The squares represent men and the circles, women. The individuals represented in shaded symbols have brown eyes and the unshaded symbols have blue eyes. Brown eye colour (B) is dominant over blue eye colour (b).

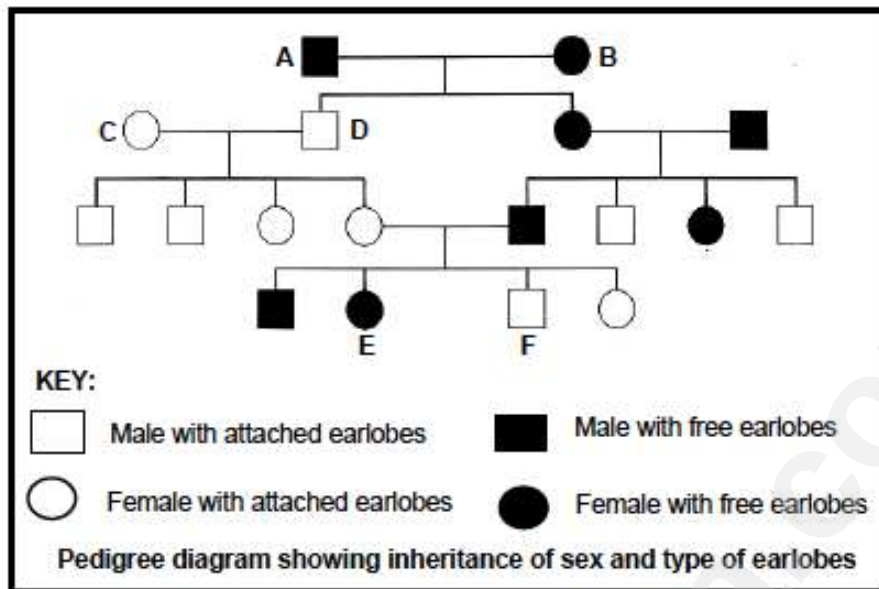


- 2.1. Use the letter B and b as indicated and write down the genotypes of the individuals numbered 1 to 5. (5)  
**(Remember that genotype will be what is in the genes and not what you can see)**
- 2.2. Draw a diagrammatic representation of all the genetic combinations with regard to eye colour, of the descendants when 6 marries a woman with the same genetic composition as 3. Use the letters B and b to show the genotype and phenotype of this F<sup>1</sup> generation. (6)

**(Remember to use a Punnett square. Refer to your notes to check that you write all the information required or you will lose unnecessary marks)**

**Question 3:** (Taken from DoE Additional Exemplar 2008 Paper 1) – 13 minutes

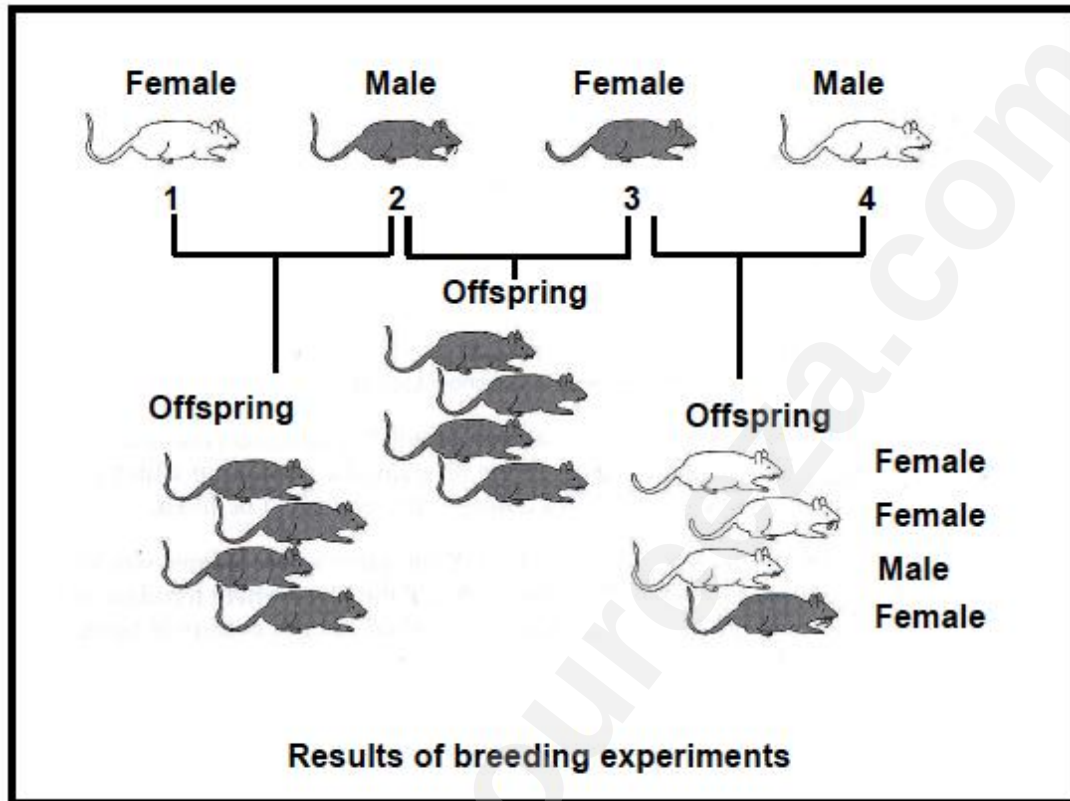
3. Study the family tree below which shows the inheritance of sex and type of earlobes over four generations of a family. In humans, free earlobes (F) is dominant over attached earlobes (f).



- 3.1. How many members of the family have free earlobes? (1)
- 3.2. What proportion of offspring in the fourth generation are females with attached earlobes? (2)
- 3.3. If the genotype of person A is FF, what will be the genotype of person B? (2)
- 3.4. Give a reason for your answer to QUESTION 3.3. (2)
- 3.5. Persons E and F are twins. Were they produced from a single fertilised egg cell or from two separately fertilised egg cells? (1)
- 3.6. Explain your answer to QUESTION 3.5. (2)
- 3.7. Is it possible for individuals C and D to have a child with free earlobes? (1)
- 3.8. Explain your answer to QUESTION 3.7. (2)

**Question 4:** (Taken from DoE Feb/March 2009 Paper 1) – 6 minutes

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



- 4.1. State which sex chromosomes would be present in the gametes of parent mouse 2 and mouse 3, respectively. (2)
- 4.2. 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)
- 4.3. Which of the parent mice (1, 2, 3 or 4) is likely to be homozygous dominant for coat colour? (1)
- 4.4. State why mouse 3 can only be heterozygous for coat colour. (2)

## SECTION B: SOLUTIONS AND HINTS

1.1. Mr. Xhosa -  $I^A I^B$  ✓Wife -  $I^B I^B$  or  $I^B I^O$  ✓ $F^1$  is  $I^A I^B$  or  $I^B I^B$  or  $I^B I^O$  or  $I^A I^O$ . ✓✓✓✓

Baby 'A' is the only possible blood group ✓. Baby 'O' is not a possibility

**(Remember that in blood groups there are three alleles A, B and O. A and B are co-dominant over O which is recessive. There must be two of the same alleles if a recessive trait is present in the individual)**

1.2. Mr. Mbundwini -  $I^A I^A$  or  $I^A I^O$  ✓✓

Wife – Not given, but assume she is recessive ✓. Therefore baby 'O' is the possible blood group ✓ as 'O' cannot be the result of Mr. Xhosa and his wife ✓.

**(Each tick = ½ mark)** (6)

2.1. BB = brown eyes bb = blue eyes - since both 3 and 4 have brown eyes, 2 will be BB.

1 = bb ✓

2 = BB ✓

3 = Bb ✓

4 = Bb ✓

5 = bb ✓

(5)

2.2. Brown = BB Blue = bb

 $P^1$  ✓ Bb x Bb - Meiosis ✓

| Gametes | B  | b  | ✓ |
|---------|----|----|---|
| B       | BB | Bb | ✓ |
| b       | Bb | bb | ✓ |
| ✓       |    |    |   |

Fertilization ✓

 $F^1$  ✓

**Genotype:** 1:4 Homozygous brown BB, 2:4 Heterozygous brown – Bb ✓ ,  
1:4 Homozygous blue – bb ✓

**Phenotype:** 75% brown ✓ {1 homozygous brown + 2 heterozygous brown}  
25% blue ✓ {1 homozygous blue} **(Each tick = ½ mark)** (6)

- 3.1 8 (1)
- 3.2  $25\% \checkmark$  (2)
- 3.3 Ff (2)
- 3.4 Individual B would have **one dominant gene** since he/she has free earlobes  $\checkmark$  and the other gene **must be recessive** since they were able to produce offspring with attached earlobes/the recessive characteristic  $\checkmark$  (2)
- 3.5 Two separate (1)
- 3.6 One is male and the other is female  $\checkmark$   
Identical twins are identical in every respect  $\checkmark$  /from the same sex (2)
- 3.7 No (1)
- 3.8 Since C and D have attached earlobes  $\checkmark$  they have only recessive genes  $\checkmark$  and can therefore have no dominant gene/gene for free earlobes to pass to their offspring  $\checkmark$  .Any (2)
- 4.1 Mouse 2 – XY  $\checkmark$   
Mouse 3 – XX  $\checkmark$  (2)
- 4.2 50%  $\checkmark$  (1)
- 4.3 Mouse 2  $\checkmark$  (1)
- 4.4 A cross between mouse 3 and mouse 4  $\checkmark$  produced offspring with white  $\checkmark$  /recessive coat colour and white colour will only show up if both parents have at least one recessive gene  $\checkmark$  any (2)
- (6)**

### SECTION C: ADDITIONAL CONTENT NOTES

#### 1. Mendel's First Law: The Law of Dominance and Segregation

**Background:** Gregor Mendel (1822–1884) was an Augustinian monk who enjoyed experimenting with plants and investigating the outcome. He is known as the first **biogeneticist**. He studied the characteristics of garden peas grown in the monastery garden and recorded his findings. The laws he wrote are based on these findings and are used by geneticists today.

The Law of Dominance and Segregation: when two individuals with **contrasting homozygous** (pure bred) characteristics are crossed, the individuals of the **F1 hybrid generation** will all resemble the parent possessing the dominant characteristic. This law

shows the principles of dominance and recessiveness using the characteristic of height. Pea plants either grow tall (TT or Tt) or are short plants (tt). Mendel crossed the pure bred homozygous tall and homozygous short varieties to prove his theories.

**P<sub>1</sub> TT x tt - Meiosis**

| Gametes | T  | T  |
|---------|----|----|
| t       | Tt | Tt |
| t       | Tt | Tt |

**Fertilization**

**F<sub>1</sub>**

**Genotype:** 4:4 Tt heterozygous offspring **Phenotype:** 100% Tall

**(Note that the F<sub>1</sub> offspring show characteristics from both parents.)**

The plants of the F<sub>1</sub> grow and mature. When they are ready to reproduce, they produce gametes for tallness (T) and shortness (t) because the **gametes segregate** (T + T + t + t) during meiosis. One half of the gametes will contain the characteristic of one of the parents - for tallness and the other half will contain the characteristic of the other parent plant - for shortness. The characteristic for shortness is the **recessive** characteristic and it will appear in the second cross offspring called the **F<sub>2</sub> generation**.

**P<sub>2</sub> Tt x Tt - Meiosis**

| Gametes | T  | t  |
|---------|----|----|
| T       | TT | Tt |
| t       | Tt | tt |

**Fertilization**

**F<sub>2</sub> Genotype:** 1:4 Homozygous Tall, 2:4 Heterozygous Tall, 1:4 Homozygous short  
(1 TT : 2 Tt : 1 tt)

**Phenotype:** 75% tall {1 homozygous tall + 2 heterozygous tall}  
25% short {1 homozygous short}



**Learner Note:** Mendel's First Law is a MONOHYBRID cross because only ONE characteristic is focussed on per generation. The more examples you do, the better you will be. Check the mark so that you don't forget to write information and loose marks for this. You need to understand that the punnett square shows the **possible** combination of the gametes. Check the notes given in Session 12 about the terms each time the term is mentioned in an monohybrid cross and check that you remember, e.g: homozygous – Homo = same and zygous = zygote. When two alleles of a pair of genes are the same for one trait e.g.: both alleles are for red flowers, the cross will result in a **pure breed** for red.

## 2. Sex Determination

In humans, the **somatic cells** are diploid and contain 23 pairs of chromosomes in each nucleus of which:

- 22 pairs of autosomes
- 1 pair of sex chromosomes: females - **XX** sex chromosomes and males - **XY** sex chromosomes

Gametes are formed by **gametogenesis** in the **ovaries** and **testes**. The egg cell (female gamete) can only ever contain one X chromosome, but half the sperm cells will have X and half will have Y chromosomes. When fertilization occurs, there is a 50 % chance that the zygote is male and a 50 % chance that the zygote is female:

$$\begin{array}{rclcl} \text{X} & + & \text{X} & = & \text{XX} \text{ or} \\ \text{X} & + & \text{Y} & = & \text{XY} \end{array}$$

P<sub>1</sub>

Meiosis

| gametes | X  | X  |
|---------|----|----|
| X       | XX | XX |
| Y       | XY | XY |

Fertilization

F<sub>1</sub>

**Genotype:** X X 2:4 X Y 2:4

**Phenotype:** 50% males and 50% females

### Sex-linked genetic diseases:

**Haemophilia** (the inability of the blood to clot) and colour blindness are disorders that are sex-linked characteristics. The alleles of the genes for these disorders are **recessive** and located on the **X chromosome** of the female. Females are generally 'carriers' of the gene, with the gene masked by the normal allele gene.



Males have only ONE X chromosome, so if the gene is present, there is NO masking allele and they will inherit and display the trait.

Sex-linked genetic crosses:

**H = normal (dominant)**

**h = haemophilia (recessive)**

Carrier female:  $X^H X^h$  where H = normal (dominant) and h = haemophilia (recessive)

Normal male:  $X^H Y$  there is no 'arm' on the chromosome to carry the allele

**P<sub>1</sub>  $X^H X^h \times X^H Y$  - Meiosis**

| Gametes | $X^H$     | $X^h$     |
|---------|-----------|-----------|
| $X^H$   | $X^H X^H$ | $X^H X^h$ |
| Y       | $X^H Y$   | $X^h Y$   |

**Fertilization**

**F<sub>1</sub>**

**Genotype:** 1:4  $X^H X^H$ , 1:4  $X^H Y$ , 1:4  $X^H X^h$ , 1:4  $X^h Y$

**Phenotype:** 25% normal female ( $X^H X^H$ )  
 25% normal male ( $X^H Y$ )  
 25% carrier female ( $X^H X^h$ )  
 25% male with haemophilia ( $X^h Y$ )

### 3. Blood Group Inheritance

Each human has blood flowing through their blood vessels. Blood is determined by **THREE alleles** and not two (one on each chromosome) as for all other characteristics and traits. The fact that there are three possible genes is termed **multiple alleles**. Specific proteins that are present on the surface of the red blood cells determine the blood type. Protein A and protein B are coded by **alleles A and B**. If no protein A or B is present, then these cells will be coded by the **allele O**. Any two of these alleles (genes A, B or O) will occur in combination in an individual. The alleles A and B are **co-dominant** (both dominate equally) over O, which is **recessive**.

The **rhesus factor** plays a further role in determining blood type. **Rh-positive** - the presence of the rhesus **antigen** on the surface of the red blood cell. **Rh-negative** – the **absence of the antigen**. Blood groups are classified by the gene and also the rhesus factor, e.g.: A+ or A–. **Rh-negative** individuals have the ability to produce an antibody called **anti-Rh** as part of the immune response when their blood comes into contact with Rh-positive blood. The rhesus system can cause complications during blood transfusions, pregnancy and birth

**Pregnancy:** if a Rh-negative mother carries an Rh-positive baby, the mother will produce **antibodies**. The antibodies react with the antigens present in the baby's blood and a condition called **haemolysis** will occur (breaking down of red blood cells) so baby has **fewer red blood** cells and less **oxygen carrying capacity**. The baby will look blue at birth = called a 'blue baby'. The first pregnancy is not really a problem, as the mother does not produce enough antibodies to cause real harm to the foetus. With following pregnancies, the mother will produce more antibodies. Medical Science has developed a substance that is injected into the mother to remove Rh-positive foetal cells before they stimulate the production of more antibodies to protect the next Rh-positive foetus.

#### Types of blood groups:

| Phenotype | Genotype |
|-----------|----------|
| A         | AA       |
| A         | AO       |
| B         | BB       |
| B         | BO       |
| AB        | AB       |
| O         | OO       |

#### Did you know?

An organism's full set of genes on all its chromosomes is known as its genome. There are certain methods that scientists have used since the late 1980s to find the sequence of base pairs of the DNA, and to identify the position of genes on human chromosomes. This is called the **Human Genome Project**.

**SECTION D: HOMEWORK EXERCISES** *(questions taken and adapted from Study&Master)*

***(Refer to the Additional Content notes from session 12 and this if you get stuck. If you still can't answer the homework questions, then refer it to your class teacher)***

1. In guinea-pigs, the gene for black coat is dominant to the gene for white. Two heterozygous black guinea-pigs are crossed.
  - 1.1. By means of a diagram, show the genotypic results that would be expected in the  $F^1$  generation. (7)
  - 1.2. One of the white  $F^1$  offspring was crossed with its black parent. By means of a diagram show the expected  $F^1$  genotypic results of this new cross. (6)
2. In horses black coat colour (B) is dominant over white (b). A white mare mates twice with the same black stallion. She produces a white foal on the first occasion and a black foal on the second occasion. Use the letters B and b as indicated above and write down the genotypes of:
  - 2.1. the mare and stallion (2)
  - 2.2. the first and second foal (4)

**SECTION E: SOLUTIONS TO SESSION 14 HOMEWORK**

No homework given in session 14.