Biology Notes
Topic: Inheritance / Genetics

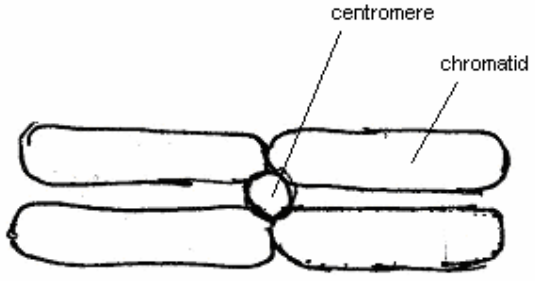
Objectives:
At the end of this topic the students should be able to:
1. Define the terms inheritance and genetics.
2. Describe and use the terms chromosomes, genes, heredity, allele, haploid, diploid.
3. Outline the inheritance of sex in humans in terms of X and Y-chromosomes.
4. Define the terms mitosis and meiosis.
5. Describe the stages of mitosis.
6. Describe the concepts of monohybrid inheritance.
7. Use the following terms as they relate to monohybrid inheritance: genotype, phenotype, homozygous, heterozygous, dominant, recessive.
8. Show the results of simple genetic crosses involving 1:1 and 3:1 ratios.
9. Describe co-dominance and the inheritance of the ABO blood groups.

Inheritance

Inheritance or heredity is the passing on of characteristics from parents to offsprings. The characteristics that are passed on are called genes and these genes are what determine why children resemble their parents, grandparents and other relatives. **The study of inheritance is called Genetics**. Genetics studies the effect of genes on growth and development.

Chromosomes

Chromosomes are the vehicles of inheritance. They are present in the nucleus of the cell as long, fine thread-like structures. The cells of the human body are of two types – **body cells** (also called somatic cells) and **sex cells** (also called gametes). Body cells are the cells that make up the body tissues such as blood cells, muscle cells etc. while sex cells are the reproductive cells such as sperms and eggs. Each **body cell contains 46 chromosomes** in its nucleus (arranged in 23 pairs) while each **sex cell contains 23 chromosomes** in its nucleus (arranged in 11 pairs plus1single sex chromosome). A **chromosome is a chain of thousands of genes**. The chromosomes in the nucleus of each body cell contain the coded instructions for the growth and development of a complete human being. The sex cell on the other hand contains half of the instructions, hence the need for two sex cells – male and female, to form a complete human being.
Chromosomes exist in identical pairs **(homologous chromosomes)** and each chromosome is made up of two identical strands - each strand is called a **chromatid.**



Structure of a typical chromosome

Genes

Genes are the units of inheritance, which are located on chromosomes. A **gene is a segment of a chromosome**, which contains the code for a particular characteristic. There is a gene for every characteristic possessed by an individual e.g. there is a gene for eye colour, for blood group, for skin colour etc. **Genes are made up of a chemical substance called DNA – deoxyribonucleic acid**. The instructions for the growth and development of a human being are encoded in DNA. Genes always occur in pairs with each member of a pair occupying exactly the same position on their respective chromosomes and affecting the same characteristic.

Alleles

Alleles are pairs of genes that are responsible for a pair of contrasting characters e.g. tallness and shortness in height. They are alternate forms of the same gene. They occupy the same position on a pair of homologous chromosomes (i.e. identical pairs of chromosomes) and they control the same characteristic (e.g. height in the example given above). Although both genes of a pair affect the same characteristic, they may vary in their effect e.g. the gene for hair colour may give rise to brown, black, blonde or red hair. **Alleles are sometimes used interchangeably with genes**.

Haploid and diploid

Cells that contain half of the full amount of chromosomes for that species are said to contain the **haploid number** of chromosomes. **In the case of humans the full amount is 46** so the haploid number would be 23. Cells that contain the full amount of chromosomes are said to contain the **diploid number** of chromosomes. Body cells will contain the diploid number of chromosomes while sex cells will contain the haploid number of chromosomes.

Sex inheritance

The sex of an individual is determined by sex chromosomes. There are two sex chromosomes, one is called **X** and the other is called **Y**. All body cells in a **female** contain two X chromosomes (**XX**) while all body cells in a **male** contain one X and one Y chromosome (**XY**). Each female sex cell (gamete) contains only an X chromosome while each male sex cell will contain either an X or a Y chromosome. The sex of an individual is determined at the moment of conception and depends on whether a sperm containing an X or a Y chromosome fertilized the egg, which has an X chromosome. If an X sperm fertilizes the egg, the zygote will contain **two X chromosomes and this will develop into a female**. On the other hand if a Y sperm fertilizes the egg, the zygote will contain **an X and a Y chromosome and will develop into a male.**

Types of cell division

Cell division is the process by which new cells are formed from existing cells.This process takes about two hours to complete and involves the chromosomes. There are two types of cell division – **mitosis and meiosis**.

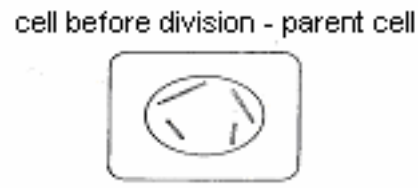
Mitosis is the type of cell division carried out by body cells. This type of cell division produces new cells for growth and for replacement of worn out or damaged body cells. **Mitosis also takes place during asexual reproduction**. Cells produced

by mitosis have the same number of chromosomes as the parent cell i.e. 46, the diploid number.

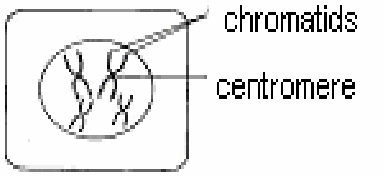
Meiosis (reduction division) is the type of cell division that takes place during the formation of sex cells or gametes. This type of cell division takes place only in the ovaries and the testes and produce cells with half the number of chromosomes that are in the parent cell i.e. the haploid number (23).

Stages of mitosis

Cell division by mitosis takes place in four stages – prophase, metaphase, anaphase and telophase.

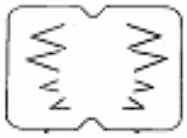


Before the cell begins to divide, each chromosome produces an exact copy or replica of itself. The original chromosome and its replica are called **chromatids**, and they are held together by a structure called the **centromere**. The cell then enters into **prophase** where the chromosomes become visible and is seen to consist of two threads (chromatids) joined at the centromere.

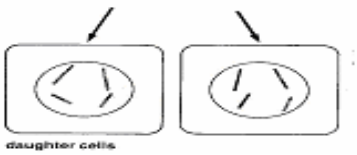


In **metaphase**, the chromosomes line up across the middle of the cell.

In **anaphase**, the chromatids separate and move to opposite ends of the cell and the cell starts to split into two.



In the final stage of division called **telophase**, the chromatids become chromosomes and the cell now completely splits into two. Each new cell has the same number of chromosomes as was present in the original (parent) cell.



Monohybrid inheritance

Monohybrid inheritance is the inheritance of a single characteristic e.g. eye colour. It is important to understand certain terminologies before one can understand monohybrid inheritance.

Genotype – the genes (or alleles) that an organism possesses for a particular characteristic; letters of the alphabet are used to represent genes and two of the same letters of the alphabet (upper case and/or lower case) are used to represent a genotype e.g. AA, Aa or aa. One of each gene of the genotype comes from each parent.

Phenotype – the physical expression of the genotype e.g. if the gene R produces red flowers, then an organism with the genotype RR would have red flowers.

Homozygous – a genotype that contains identical genes or alleles for the same characteristic e.g. BB or bb.

Heterozygous – a genotype that contains two different alleles/genes for the same characteristic e.g. Dd

Dominant (strong) – This term is used to describe a gene whose effect is expressed short; if the gene for tallness is T and the gene for shortness is t, then an individual with the genotype TT or Tt would be tall. As a general rule, upper case symbols are used to represent dominant genes.

Recessive (weak) – This term is used to describe a gene whose effect is not expressed in the presence of the dominant gene e.g. in the example above, the gene for shortness (t) is not expressed, hence it is said to be recessive. As a general rule, lower case symbols are used to represent recessive genes.

In summary

- genes are represented by letters of the alphabet
- capital (upper case) letters represent dominant genes
- common(lower case) letters represent recessive genes
- two identical genes are described as homozygous
- two different genes for the same characteristic are said to be heterozygous
- two dominant genes in one genotype is called homozygous dominant
- two recessive genes in one genotype is called homozygous recessive
- one dominant and one recessive gene in one genotype is called heterozygous dominant
- **if neither of the two genes in a genotype is dominant then the term co-dominant or incomplete dominance is used.**

Genetic crosses and Checkerboard diagrams / Punnett Squares

Monohybrid inheritance is shown by using diagrams called genetic crosses or checkerboard diagrams (also called Punnett Squares). These diagrams show the likely

outcome for the offsprings when mating takes place between two individuals. In constructing these diagrams, certain symbols are used which were explained above.

Method

1. *Write down the genotypes of the parents. A suitable key is to be used if one is not given.*
2. *Write down all the gametes showing the gene each contains.*
3. *Show all the possible outcomes by combining the gametes from each parent. This may be done using the crosses or the checkerboard. The outcome is called the first generation or F_1. If there is mating among the F_1 to produce offsprings, those offsprings form the second generation or F_2.*
4. *Determine the ratio of the genotypes (genotypic ratio) and the ratio of the phenotypes (phenotypic ratio). Fractions and percentages are sometimes used.*

Illustration

The ability to roll the tongue is due to a dominant gene, which we will call **T**. The alternate recessive gene for non-tongue rolling is **t**.

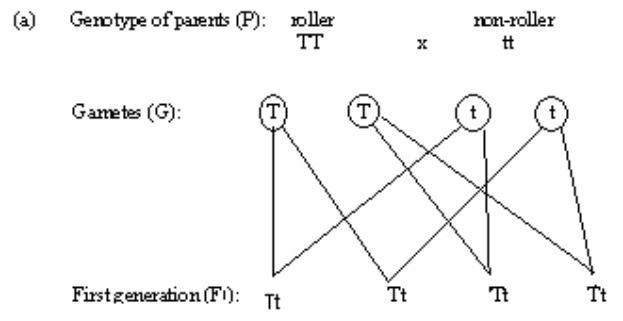
What will be the outcome if:

(a) a homozygous tongue roller mates with a non-tongue roller

(b) a heterozygous tongue roller mates with another heterozygous tongue roller

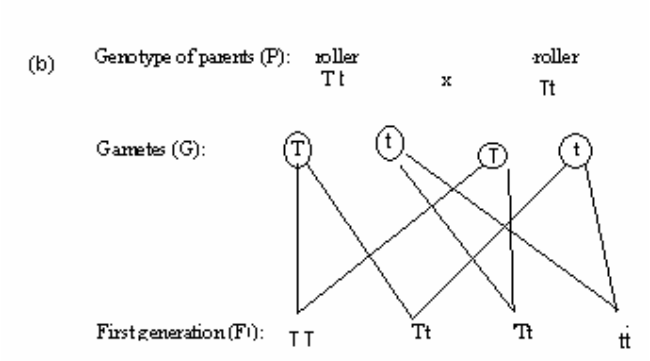
(c) a heterozygous tongue roller mates with a non-tongue roller

Solution



Genotypes: all Tt (100% Tt)
Phenotypes: all rollers (100% rollers)

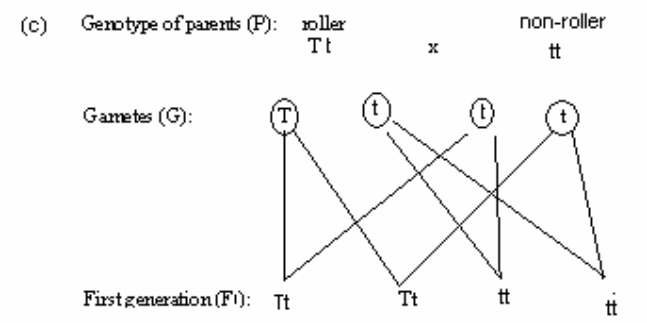
The four possible ways in which the gametes can unite will, in this case, result in all the children of these parents being rollers.



Genotypic ratio 1TT : 2Tt : 1tt or 1:2:1
Phenotypic ratio 3 rollers : 1 non-roller or 3:1

The outcome of this mating will be that 3 out of every 4 children are likely to be rollers. In terms of the four possible genotypes, 1 will be TT, 2 will be Tt and 1 will be tt.
It is important to note that these outcomes are likely outcomes and not definite outcomes. These results only give the probability of each outcome and may vary from the actual outcome. The results can also be expressed using fractions or percentages.
The genotypes could therefore be written as: ¼ TT: ½ Tt : ¼ tt
or 25% TT: 50%Tt :25%tt

The phenotypes could be written as: ¾ rollers : ¼ non-rollers
or 75% rollers : 25% non-rollers



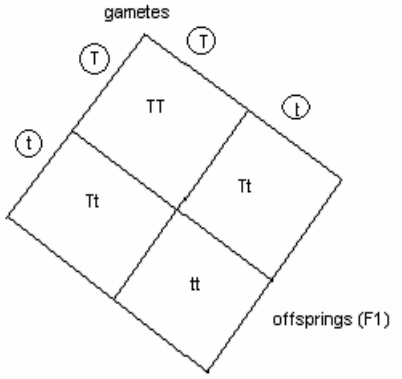
Genotypic ratio 2Tt: 2tt which simplifies to 1:1 or ½ Tt: ½ tt or 50%Tt: 50%tt
Phenotypic ratio 2 rollers: 2 non-rollers or 1:1

The diagrams used above are called **genetic crosses**. The same information can be presented using checkerboard diagrams or Punnett Squares as shown below with example (b).

P: roller x roller
Tt Tt

G: T t T t

The gametes from one parent are put at one side and those from the other parent are put on the other side. The genes are then brought together in the boxes.



The results are treated in the same way as the previous method.

Codominance and the ABO blood groups.
Humans have what is called the **ABO blood grouping system** and there are four types of blood in this system, namely A, B, AB and O. Each blood type contains a different set of antigens. (Refer to Notes: Transport in Humans 2 - Blood)

Type A blood contains antigen A on the red cells and antibody B in the plasma.
Type B blood contains antigen B on the red cells and antibody A in the plasma.
Type AB (least abundant) contains both antigens A and B on the red cells but neither antibodies A nor B in the plasma
Type O (most abundant) blood contains neither antigen A nor B on the red cells but contains both antibodies A and B in the plasma.

There are three genes - A, B and O (hence ABO system) that determine the type of antigen contained on the red blood cells in each blood type. A person must have two of these genes (one inherited from each parent) to exhibit a certain blood type as shown below.

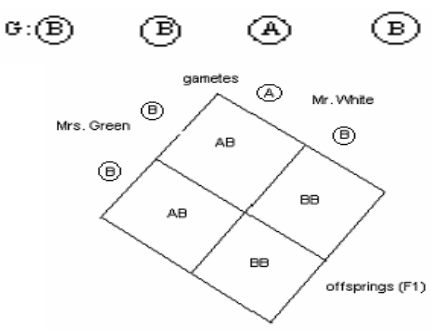
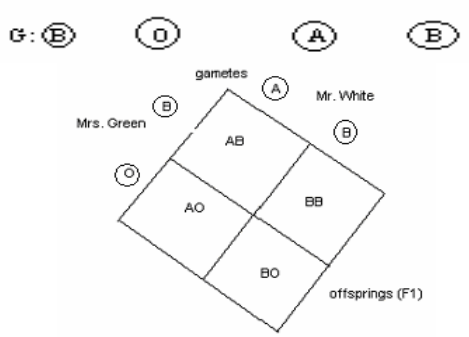
Blood Type	Possible Genotypes
A	AA or AO
B	BB or BO
AB	AB only
O	OO only

Neither the A nor the B genes are dominant over each other. In this case they are said to be **codominant.** In other words, once these two genes occur together, they produce an entirely new phenotype called blood type AB. Genes A and B, however, are both dominant to gene O. To obtain blood type O a person must have inherited both O genes – one from each parent. The concept of blood group inheritance is sometimes used in court cases to determine disputed paternities.

Example
Mrs. Green, blood type B, claims that Mr. White, blood type O, is the father of her child, who is blood type AB. Is it possible for Mr. White to be the father of her child?

Solution

Mrs. Green's genotype could be **BB** or **BO** while Mr. White's genotype can only be **AB**. Let us examine the two possible outcomes.

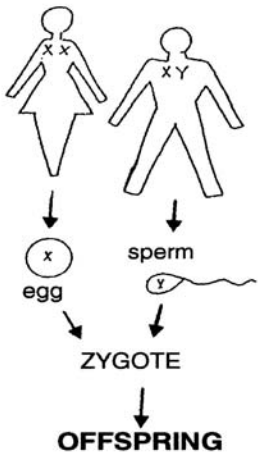
Outcome 1	**Outcome 2**
P: Mrs. Green Mr. White BB x AB 	P: Mrs. Green Mr. White BO x AB 

The results show that nowhere among the possible outcomes is a genotype OO. It is therefore **not possible** for Mr. White to be the father of the child.

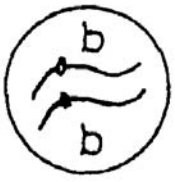
Questions

1. (a) Compare the process of mitosis with that of meiosis. (6)
   (b) Which type of cell division produces the egg and the embryo? (2)
   (c) Do the egg and the zygote have the haploid or the diploid number of chromosomes? Give the reason for your answer in each instance. (4)
   (d) (i) What is codominance? (2)
   (ii) How is the concept of dominance illustrated in the inheritance of the A, B, O blood groups? (6)

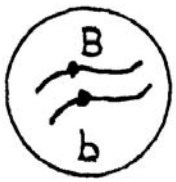
2. The diagram shows the sex chromosomes, which are passed from parents to the offspring. Study the diagram and answer the questions that follow.



   (a) (i) In which part of the cell are chromosomes found? (1)
   (ii) How many chromosomes are found in the egg and the zygote? (2)
   (iii) What is the sex of the offspring? Give the reason for your answer. (2)
   (b) The diagram shows two alleles of a gene for eye colour. B is the dominant allele (black eyes), b is the recessive allele (brown eyes)



P

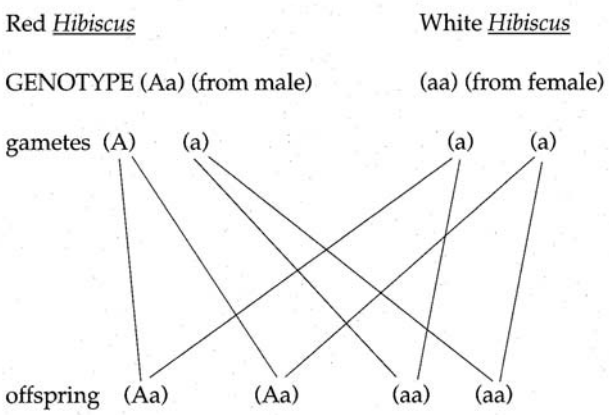


Q

(i) P is a homozygous individual for eye colour. What does the term 'homozygous' mean? (1)

(ii) What colour eyes would the individuals represented by P and Q have if they had the genotypes shown above? Give the reason for your answer in each instance. (4)

3. The diagram shows a cross between a red *Hibiscus* flower and a white *Hibiscus* flower on two different plants.



(a) (i) Name the part of the red flower that produces the gametes. (1)

(ii) What type of division produces the gametes? (1)

(b) (i) Write down the ratio of red to white flowers in the offspring. (1)

(ii) What percentage of the offspring shown has the dominant gene? (1)

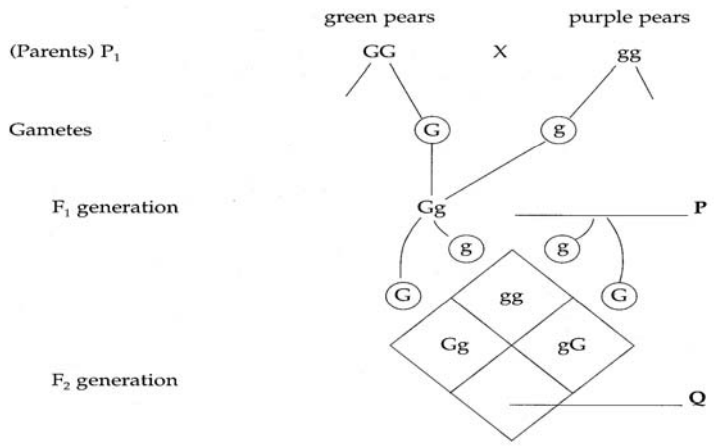
(c) What ratio of flower colour would result from a cross between two heterozygous *Hibiscus* flowers? (no working needs to shown). (2)

(d) Indicate whether the cells present in each of the following has the haploid and/or the diploid number of chromosomes.

(i) leaf (iii) stigma of flower

(ii) female gamete of flower (iv) fruit (4)

4. The diagram shows a cross between two avocado pear trees: one, which produces green pears and the other which produces purple pears. The trait for green pear colour shows complete dominance.



(a) Which symbol represents the dominant allele? (1)
(b) Write the correct symbols for the genotypes indicated by P and by Q. (2)
(c) (i) What is the phenotype of the F_1 pear shown? (1)
(ii) Which term describes the genotype of the F1 pear shown? (1)
(d) If there were 100 pear trees with purple pears produced in the F2 generation, approximately how many pear trees with green pears will be produced in that generation? (1)
(e) A pear tree of genotype Gg is crossed with a pear tree of genotype gg and they produce 400 offsprings. What would be the phenotype of the offspring? State the approximate number of each phenotype. (4)

5. The diagram shows part of the results of across between TWO parents. One parent was blood type A and the other parent blood type B.

Parents:	Blood Type A	Blood Type B
Offsprings		
		OO

(a) (i) Copy and complete the genetic diagram above to show the other possible genotypes of the offspring. (3)
(ii) What are the genotypes of both parents? (2)
(b) Give the term that describes the genotype of the offspring OO. (1)
(c) Sickle cell anaemia is an inherited blood disorder. A person with the genotype HH has normal haemoglobin. A person with the genotype hh has sickle cell anaemia.
If the two parents above have sickle cell trait:
State the percentage of their offspring that you would expect to have:
(i) sickle cell anaemia
(ii) sickle cell trait
(iii) normal blood as far as haemoglobin is concerned.
Show all working. (4)