✓ GENETICS

- ✓ It's a branch of Biology that deals with the study of inheritance. The science of genetics attempts to explain why organisms differ from one another and at the same time show similarities within the same species.
- ✓ Members of the same family may differ in appearance of face, shape of the nose, ears, skin colour etc this is due to a variety of hereditary factors that each of them acquires from their parents.
- ✓ **Variation**

- ✓ It refers to observable differences among living organisms. There are two types of variations among individuals of a given species;
- ✓ **Discontinuous variation**

- ✓ In this type of variation there are definite/distinct groups of individuals with no intermediate forms e.g.
- ✓ In human population an individual is either a male or a female
- ✓ The ABO blood group system in man-an individual can only belong to one of the groups A,B, AB or O
- ✓ Ability to roll the tongue into a U-shape-some people can roll their tongues while others cannot
- ✓ The presence of long hair in the nose and in the ear pinna
- ✓ Some people have a free ear-lobe while in others it's attached.
- ✓ Finger prints-In humans there are 4 main types of finger print patterns i.e. arch, loop, whorl and double whorl. Each individual inherits only one of the 4 main types of finger prints.
- ✓ Ability to taste a chemical substance called phenylthiocarbamide (PTC).Some individuals are able to taste (tasters) while others are unable (non-tasters).

- ✓ In plants, a pawpaw tree is either male or female
- ✓ **NB** Discontinuous variation is basically determined by the genetic factors.
- ✓ ***Practical Activities***
    - ***Activity 1-Investigating tongue-rolling in humans***

- ✓ *Members of the class to try and roll their tongues*
- ✓ *Count and record the number of rollers and non-rollers and tabulate your results*

Tongue-rolling	*Tally*	*Frequency*
Tongue-roller		
Non- Tongue-roller		

- **Activity 11-Investigating finger prints in humans**

- ✓ *Place the tip of your right thumb on the ink-pad and press lightly on it*
- ✓ *Press the inked thumb gently on the plain paper to make an impression of your thumb print. Members of the class to make a print on the same piece of paper.*
- ✓ *Observe the finger prints using a hand lens. Identify each print and name it using the figure below as reference.*

Finger print type	*Tally*	*Frequency(No of individuals)*
Arch		
Loop		
Whorl		
Double-whorl		

- ✓ **Continuous variation**

- ✓ This type of variation exhibits a wide range of differences for the same characteristic from one extreme end to the other e.g.
- ✓ Height- Height within a group of people ranges from the shortest to the tallest with several intermediates.

- ✓ Skin colour (pigmentation) - Some people are very dark-skinned while others have very light-skins with several intermediates
- ✓ Body weights.
- ✓ **NB** Continuous variations arise from the interactions between the genetic and environmental factors e.g. a plant possessing genetic factors for tallness may fail to grow due to unsuitable soil and climate.
- ✓ **Causes of variation**
- ✓ **Gamete formation**

- ✓ Two processes contribute to variations;
- ✓ **Independent Assortment**
- ✓ During Metaphase1 the homologous chromosomes come together in pairs and subsequently segregate into daughter cells independently of each other. This produces a wide variety of gametes. The number of combinations is 2^n where n is the haploid number of chromosomes. In man this is 2^{23} which is 8388608. *That's why even brothers are not exactly alike!*
- ✓ **Crossing-Over**
- ✓ During prophase1 when the homologous chromosomes are in intimate contact with one another, the chromatids of the homologous chromosomes break and rejoin at certain points called chiasmata
- ✓ During Crossing-Over important genetic exchanges take place at the chiasma resulting in more variations.
- ✓ **Fertilization**
- ✓ It permits parental genes to be brought together in different combinations. This way different quality of parents can be combined in the offspring.

- ✓ **Mutations**
- ✓ These are Spontaneous changes in the genetic make up of an organism
- ✓ **The Chromosome**
- ✓ Chromosomes contain the hereditary material or factors that are transmitted from the parents to the offspring. These factors are called genes.

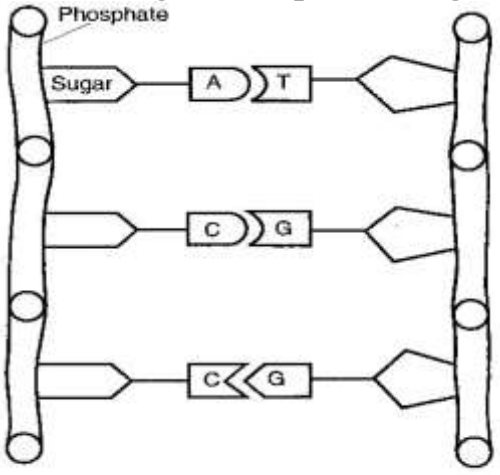
- ✓ There is a definite constant number of chromosomes in each cell for every species of animal or plant e.g.

Species	Common name	No. of Chromosomes Somatic Cells (2n)	Gamete(n)
Ovis auries	Sheep	56 (28 pairs)	28
Bos taurus	Cow	60 (30 pairs)	30
Drosophila melanogaster	Fruit fly	8 (4 pairs)	4
Zea mays	Maize	20 (10 pairs)	10
Pisum sativum	Garden pea	14 (7 pairs)	7
Mus musculus	Mouse	40 (20 pairs)	20

- ✓ **GENES AND DNA**

- ✓ Genes occupy definite position on the Chromosome known as gene loci (gene locus).

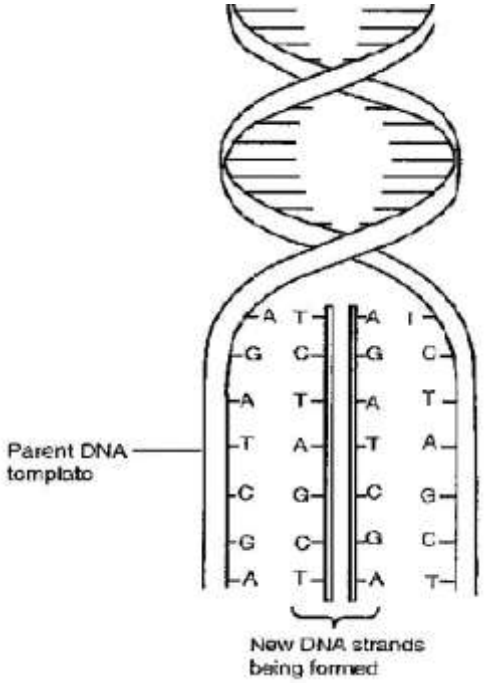
- ✓ The gene is chemical in nature and it is in the form of a nucleic acid molecule called Deoxyribonucleic acid (DNA).
- ✓ **DNA**
- ✓ It's a complex molecule composed of 3 different components i.e.
- ✓ a 5-Carbon sugar
- ✓ a Phosphate Molecule
- ✓ a Nitrogenous base- There are 4 types of bases i.e.
- ✓ Adenine (A)
- ✓ Guanine (G)
- ✓ Thymine (T)
- ✓ Cytosine (C)
- ✓ A combination of the 3 above form a nucleotide. Nucleotides join together to form long chains called DNA strands. Two parallel DNA strands twist on one another forming a double helix.
- ✓ Adenine always combines with Thymine (A-T) while Cytosine combines with Guanine (C-G) when forming the double helix structure of the DNA because they are equal. Therefore DNA is like a twisted ladder with Nitrogen bases forming the steps or rungs e.g.



The untwisted DNA strand

- ✓ **The Role of DNA**
- ✓ Stores genetic information in a coded form.

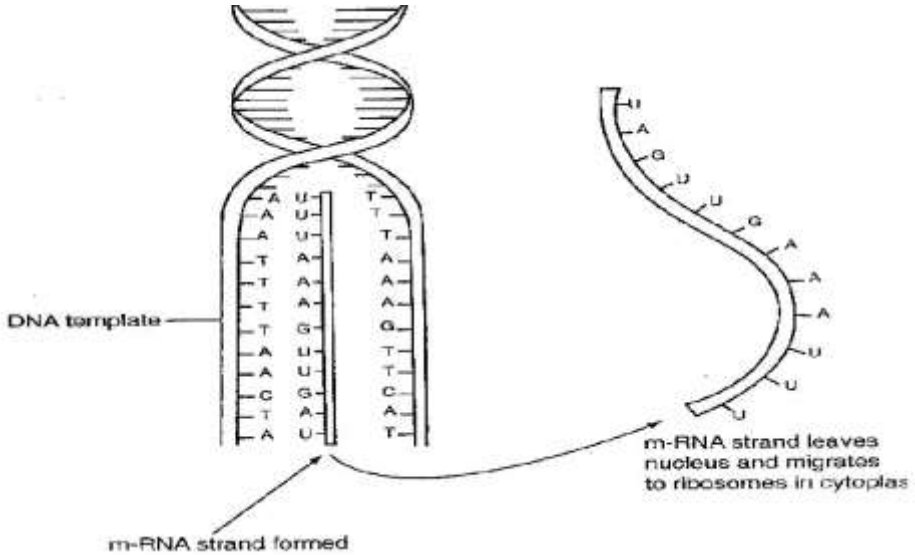
- ✓ Enables the transfer of genetic information unchanged to daughter cells through replication.
- ✓ Translates the genetic information into the characteristics of an organism through protein synthesis.
- ✓ **DNA Replication**
- ✓ This is the process through which a DNA molecule forms an exact replica of itself.
- ✓ **Mechanism of Replication**
- ✓ DNA double helix consists of two long separate strands joined together by the base pairs. When the molecule is due to replicate the double helix unwinds and the two strands unzip themselves. This is made possible by the presence of the weak Hydrogen bonds that link the bases of the two strands.
- ✓ After unzipping the information on the DNA strands (base sequence) is copied out onto a new DNA structure using the parent DNA as template e.g.



- ✓

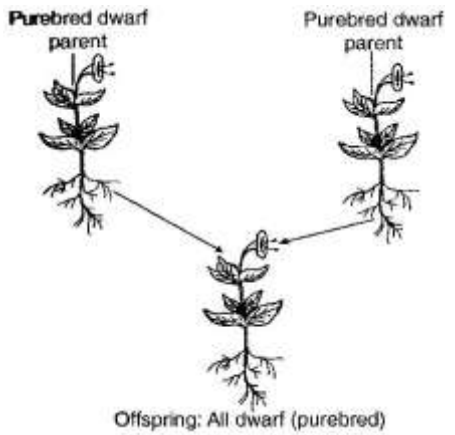
- ✓ The replication makes it possible to pass on the DNA molecule together with its exact genetic information to the daughter cells during cell division in the organism.
- ✓ **ROLE OF DNA IN PROTEIN SYNTHESIS**

- ✓ The sequences of the bases along the DNA strands are important. It acts as the alphabet or code that spells out the sequence of the amino acids when they join up to form protein polypeptide chains.
- ✓ The set of a base triplet is known as a codon and is said to code for a particular amino acid of a protein molecule e.g.
- ✓ AAA-Codes for amino acid Phenylalanine
- ✓ TTT- Codes for amino acid Lysine
- ✓ The cell has got a special molecule that mediates between the DNA and the cytoplasm. This molecule is also a nucleic acid molecule and is known as Ribonucleic acid (RNA). Since its role is to carry genetic information from the DNA to the site of protein synthesis in the cytoplasm, it is referred to as Messenger RNA (M-RNA) and is formed from the DNA strands.
- ✓ In the formation m-RNA an appropriate section of the DNA strand serves as a template. The double helix of the DNA unzips and free nucleotides align themselves opposite the template. The base sequence of the template is copied onto a new strand which then becomes an RNA strand.
- ✓ In the RNA, Thymine is replaced by the base Uracil (U). The transfer of DNA base sequence onto the m-RNA strands is described as ***transcription.***
- ✓ After its formation, m-RNA leaves the nucleus with the full instructions from the DNA about the kind of Protein to be synthesized by the cell. This instruction is in the form of base triplets or codons which are used to assemble the amino acids on the protein polypeptide chains.

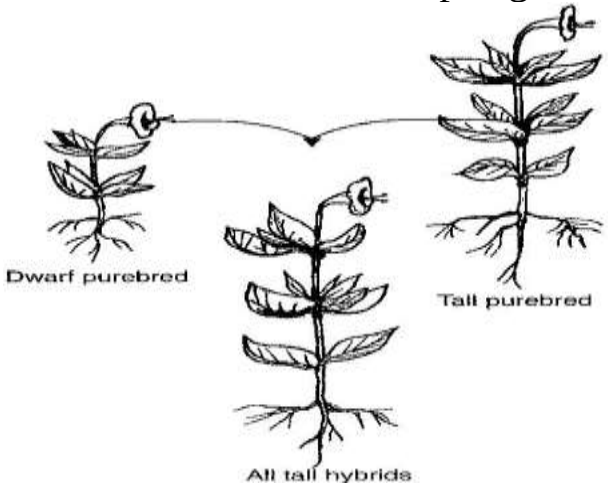


- ✓ Information on the m-RNA is translated by ribosome and is used to assemble the amino acids into specific proteins molecules. Proteins molecules determine the inherited characteristics in organisms.
    - **The First Law of Heredity**
    - **Mendel's Experiments**

- ✓ An Austrian monk known as Gregory Mendel is considered to be the father of genetics. He carried out various breeding experiments and observed variations in different characteristics of the garden pea.
- ✓ Mendel selected a group of dwarf plants & self pollinated them by dusting mature pollen grains onto stigmas of the same plant. He then collected the resulting seeds and planted them. He then noted that these seeds germinated and grew into dwarf plants only.
- ✓ He repeated these experiments for several generations and observed the same results. This showed that dwarf garden pea plants could only produce only their own type i.e. they were pure breeds i.e.

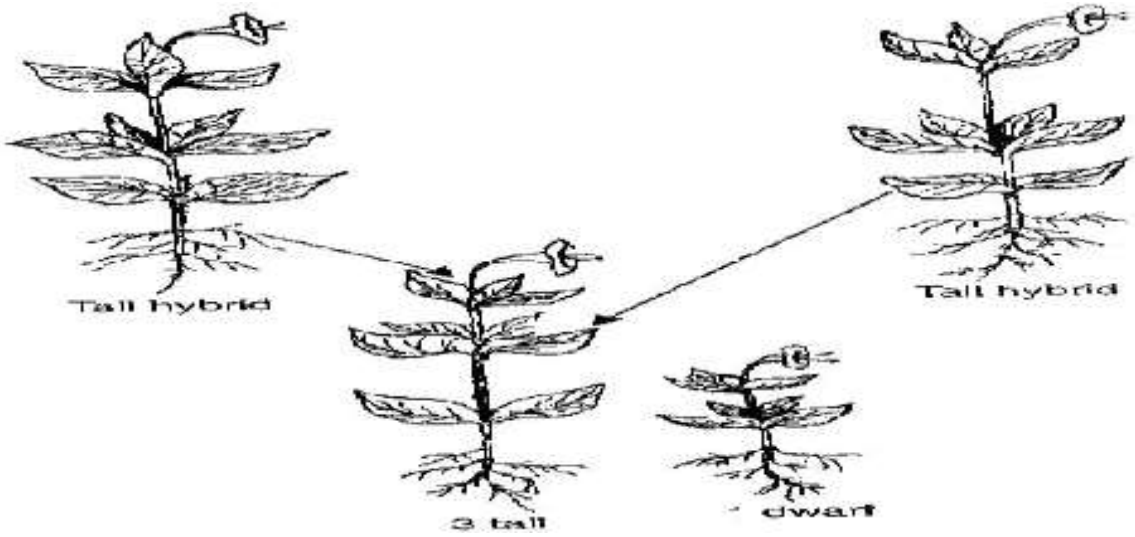


- ✓ In another experiment, Mendel selected tall plants and self pollinated them. He then planted the resulting seeds and observed that they grew into a mixture of tall and dwarf plants.
- ✓ He took seeds from the tall offspring only and repeated the experiment, for many generations until he obtained tall plants only.
- ✓ He then cross-pollinated pure bred tall garden pea plants with the pure bred dwarf variety. He planted the resulting seeds and observed that the offspring were all tall plants e.g.



- ✓ He crossed two of these tall offspring and planted the resulting seeds. He observed that this 2nd generation consisted of a mixture of tall and dwarf plants. He counted these plants

and noted that the ratio of the tall to dwarf plants was approximately 3: 1 respectively.

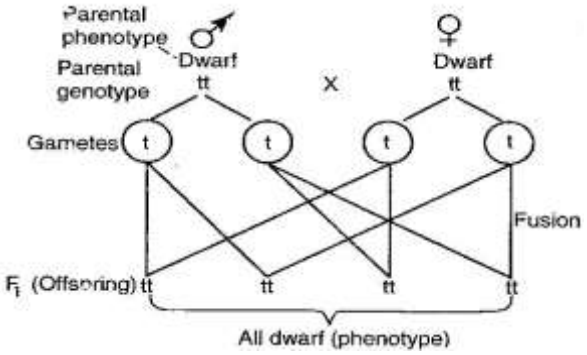


✓

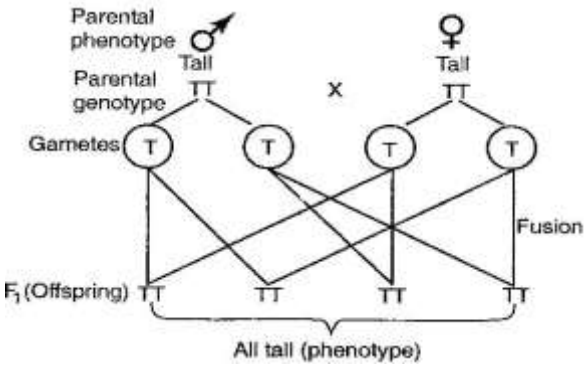
- ✓ Mendel concluded that the characteristics of an organism are determined by hereditary factors which occur in pairs. Only one of a pair of such factors can be represented in a single gamete. This is known as Mendel's 1st law, the law of segregation.
- ✓ This law states "The characteristics of an organism are determined by genes which occur in pairs. Only one member of the gene pair can be carried in a single gamete"
- ✓ Mendel's successes can be attributed to the following;
- ✓ He used favourable materials i.e. garden pea which is self-fertilised.
- ✓ His study focused on particular traits.
- ✓ He kept accurate data on all his experiments.
- ✓ The pea plant he used had several observable contrasting characteristics (traits)
    - **Monohybrid inheritance**
- ✓ In his experiments, Mendel postulated that the inheritance of one characteristic like height in garden pea plant is controlled by a

single pair of hereditary factors contributed by both parents. This type of inheritance is known as Monohybrid inheritance.

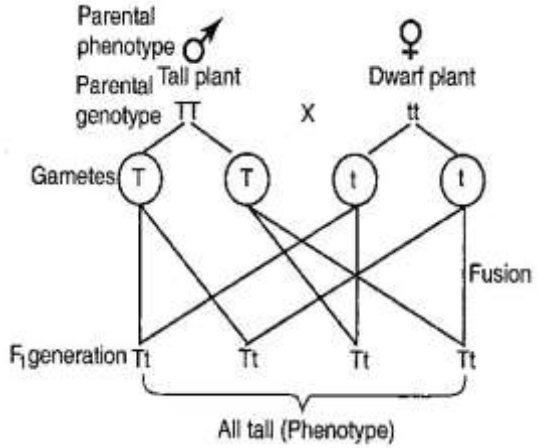
- ✓ It involves the transmission of just one pair of contrasting characteristics (traits) e.g. tallness and dwarfness for height
- ✓ A single pair of hereditary factors known as genes controls such a trait.
- ✓ Genes occur in pairs on the chromosomes and such gene pairs are known as alleles.
- ✓ The genetic constitution of an organism is known as genotype while the outward appearance of the organism is called phenotype.
- ✓ The genotype of an organism is represented using paired letter symbols where the capital letter represents the dominant gene while the small letter represents the recessive gene.
  NB The conventional symbol for male is ♂ while that of a female is ♀
- ✓ The inheritance of dwarfness in garden pea plant can be illustrated diagrammatically by a genetic cross e.g.
- ✓ **Cross between two pure-bred dwarf plants**



- ✓ **Cross between two pure-bred tall plants**

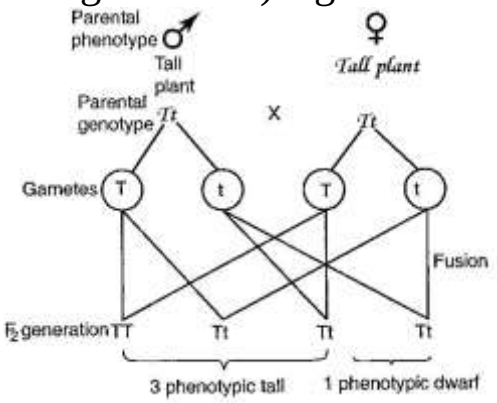


✓ **Cross between a pure-bred tall and pure-bred dwarf plants**



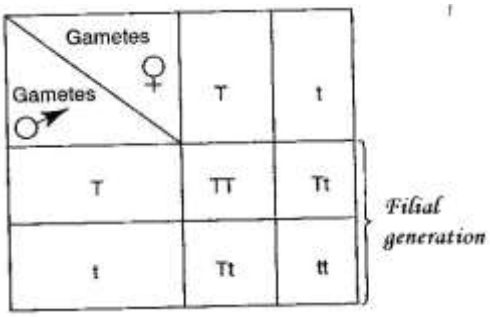
✓ **NB** In the genetic cross above, the male plant is tall and the female plant is dwarf. If the cross is reversed so that the female is tall and the male a dwarf, this is referred to as reciprocal cross. The F1 results will be the same for either cross.

✓ However, when these F1 offspring are self-pollinated, they produce offspring that grow into a mixture of tall and dwarf plants. These offspring are known as 2nd filial generation (F2 generation) e.g.



- ✓ From the genetic crosses above, the following components of a genetic cross are obtained;
- ✓ Parental phenotypes.
- ✓ Parental genotypes- Crossing (X) should be shown between two genotypes.
- ✓ Gametes –Must be circled.
- ✓ The fusion process (fertilization).
- ✓ The filial generation types.

- ✓ An alternative method of making genetic crosses is a punnet square i.e.



- ✓
- ✓ When the allelic genes are identical as in **TT** and **tt**, the condition is known as homozygous. An individual with such condition is referred to as a homozygote.
- ✓ When the allelic genes are not identical as in **Tt** the condition is known as heterozygous. An individual with such condition is referred to as a heterozygote.
- ✓ An individual with genotype **Tt** will be phenotypically tall because the gene **T** is dominant over the gene **t.** The allele **t** is recessive.
- ✓ A dominant gene is that which can express itself in both its homozygous (**TT**) and heterozygous (**Tt)** state, while a recessive gene is that which can only express itself in its homozygous (**tt**) state. Therefore the genotypic condition **TT** is referred to as homozygous dominant while **tt** is homozygous recessive.

- ✓ The ratio 3 tall: 1 dwarf in the F2 generation is characteristic of monohybrid inheritance where one gene is completely dominant over the other. This is complete dominance. The monohybrid crosses are based on Mendel's 1st law, which states that, the characteristics of an organism are determined by internal factors (genes) which occur in pairs. Only one of a pair of such factors can be represented in a single gamete.
    - **Ratios and probability**

- ✓ The 3:1 ratio in monohybrid inheritance can be presented in the form of probability i.e.
- ✓ ¾ or 75% of the offspring are tall while ¼ or 25% are dwarf.
- ✓ The inheritance of characteristics involves probability. The chance that a particular gamete will fuse with another is a random occurrence. In genetic crosses this is done by showing all possible fusions.
- ✓ Similar monohybrid results as those of Mendel have been obtained by using a common insect, the fruit fly (Drosophila melanogaster). The fruit fly is a suitable organism for genetic study due to;
- ✓ (i) The female lays many eggs resulting in a large number of offspring. This increases the sample size.
- ✓ (ii) Have many observable characteristics that are distinct and contrasting.
- ✓ (iii) It is easily bred in the laboratory with minimum requirements.
- ✓ (iv) It has a short generation time i.e. 10-14 days. Therefore many generations can be studied in a short period.
- ✓ (v) Offspring can be crossed with their parents at will (back crossing).
- ✓ (vi) The flies are safe to handle because they do not transmit any known human diseases.
    - **Practical activities**
    - **Homozygote X Heterozygote**

- ✓ 1. Label 4 beakers 1-4
- ✓ 2.Put 50 blue beads into beaker 1
- ✓ 3.Put 25 blue and 25 red beads into beaker 2 and mix them up thoroughly.
- ✓ 4. Close your eyes and take a bead from each beaker. Put the beads together (offspring). Open your eyes.
- ✓ 5. If the offspring is homozygous (either blue or red) then put it into beaker 3 and if heterozygous then put it into beaker 4.
- ✓ 6. Repeat steps 4 and 5 until all beads have been transferred.
- ✓ 7. Count and record the number of;
- ✓ (i) Homozygous offspring……...
- ✓ (ii) Heterozygous offspring……
- ✓ What do the beads represent? genes
- ✓ 8.What conclusion do you make from the results obtained in 7 above?
- ✓ 9. Assume that the characteristics shown by the blue bead is dominant and is represented by B.
- ✓ Complete the cross below;
- ✓ Parents
- ✓ Genotype ……. ……
- ✓ Gametes …… …….
- ✓ Offspring (F1)
- ✓ Genotypes …… ……..
- ✓ Ratio ……. ……..
- ✓ **Heterozygote X Heterozygote**

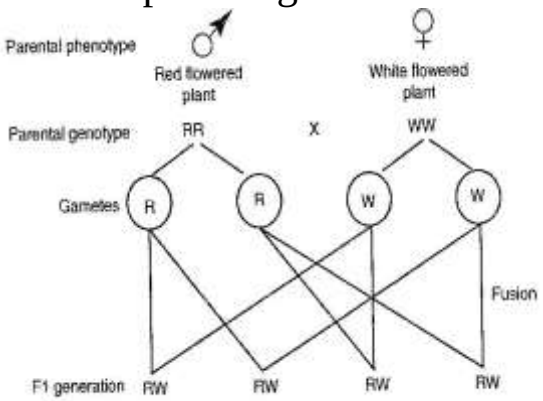
- ✓ Put 50 blue and 50 red beads into each of the two beakers 1 and 2 and mix them thoroughly.
- ✓ Close your eyes and then take a bead from each beaker. Put the two beads together.
- ✓ If the offspring is homozygous dominant (both blue) put them into beaker 3 and if heterozygous put into beaker 4. if homozygous recessive (both red) put into beaker 5.
- ✓ Repeat steps 2 and 3 until all the beads have been transferred, a pair at a time into beakers 3, 4 and 5.

- ✓ Now count the number of;
  - Homozygous dominant offspring……...
  - Heterozygous offspring……
- ✓ Homozygous recessive offspring……

- ✓ What conclusion do you make from the results obtained in 5 above?
- ✓ Assume that the characteristics shown by the blue bead is dominant and is represented by B.
- ✓ Complete the cross below;
- ✓ Parents(heterozygous)
  - Genotype …Bb …Bb……
  - Gametes …Bb Bb…….
  - Offspring (F1)
  - Genotypes 1BB……2Bb 1bb
  - Ratio …

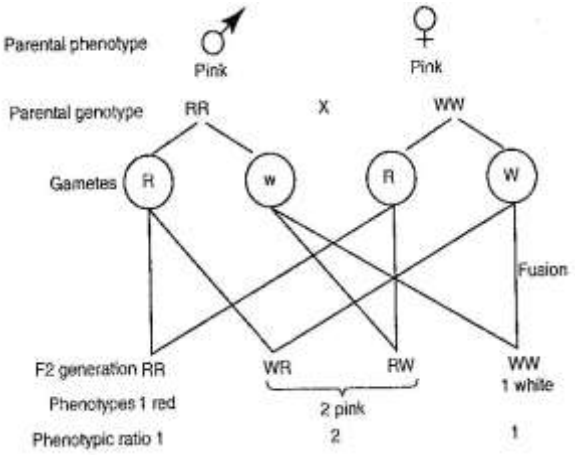
- ✓ Why is it important to to keep your eyes closed when picking the beads?
- ✓ **Incomplete dominance**
- ✓ In the experiments above, the tall plant with the genotype TT (homozygous) could not be distinguished from an individual with the genotype Tt (heterozygous). Thus although the genotypes were different, the phenotypes were identical, indicating that the gene determining tallness is completely dominant over that determining dwarfness.
- ✓ However, in some plants such as Mirabilis jalapa (4 o'clock plant) a cross between red and white flowered plants results in F1 generation with all the offspring bearing pink flowers thus showing the blending of colour.
- ✓ Selfing of F1 individuals gives a phenotypic ratio of 1 red; 2 pink; 1 white. This shows that there is no allele which completely dominates the other. This is an example of incomplete dominance.

- ✓ **NB** in genetic crosses involving incomplete dominance, two different capital letters are used to represent the 2 genes. E.g.
- ✓ Let **R** represent gene for red flower colour
- ✓ Let **W** represent gene for white flower colour



- ✓ Phenotype : all pink

- Selfing F1



- ✓ An example of incomplete dominance in animals ;
- ✓ In short-horned cattle, the gene for the red coat colour is co-dominant to white coat colour. A cross between them produces a mixture of red and white coat colour referred to as roan.
  - **Assignment**

- ✓ 1. In a particular species of tropical beetle, the wings had either red or orange marks. A cross between red marked beetles with orange marked beetles produces offspring with yellow marks

only. When the F1 offspring were selfed, they produced F2 generation in the ratio of 1 red; 2 yellow; 1 orange.

- ✓ (a) Explain the absence of red and orange marks in the F1 offspring.
- ✓ (b) Using a genetic cross show how F2 generation was obtained.
- ✓ In a plant breeding experiment, a tall pea plant was crossed with a dwarf pea plant. All the F_1 generation plants were tall. The F_1 generation plants were selfed that resulted in 325 dwarf plants out of 898 plants.
    - **(a)** (i) **How** many **plants** in **F_2**generation were tall (1 Mark)

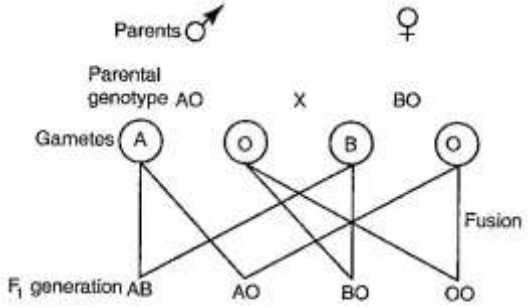
- ✓ **What** was the dominant character in the cross above?
- ✓ (b) Using letter **T** to denote the gene for height, **work out** the genotypes of F_2 generation (3 Marks)
- ✓ **Inheritance of ABO blood groups**

- ✓ In man ABO blood group system is an inheritable characteristic that is transmitted from the parents to the offspring.
- ✓ The ABO blood groups are determined by 3 genes (multiple alleles) and they are responsible for the presence of antigen types on the red blood cells.
- ✓ These are;
- ✓ Gene A responsible for the presence of antigen A.
- ✓ Gene B responsible for the presence of antigen B.
- ✓ Gene O responsible for no (zero) antigen on the red blood cells.
- ✓ Genes A and B have equal degree of dominance i.e. they are co-dominant and both express themselves when present together as in AB blood group. Gene O is recessive and will only express itself in the homozygous condition.
- ✓ The genotypes for the 4 blood groups in the ABO system are thus formed by allelic pairs of genes e.g.

✓ Blood	✓ Genotype	✓ Antigens

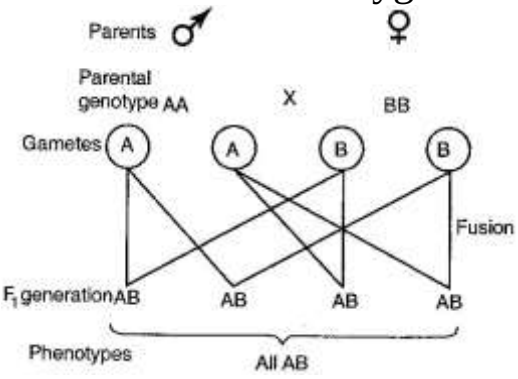
group (Phenotype)		
✓ A	✓ AA, AO	✓ A
✓ B	✓ BB, BO	✓ B
✓ AB	✓ AB	✓ A&B
✓ O	✓ OO	✓ O(zero)

- ✓ A marriage between a man of blood group A and a woman of blood group B. If both parents are heterozygous, they will produce;



- ✓ Phenotypes 1AB 1A 1B 1O

- ✓ A man who is homozygous for blood group A married to a woman who is homozygous for blood group B would produce;



- ✓

- ✓ A Mrs. Abuto claims that her child was wrongly identified at the hospital so that she took away the wrong baby. Both Abuto and

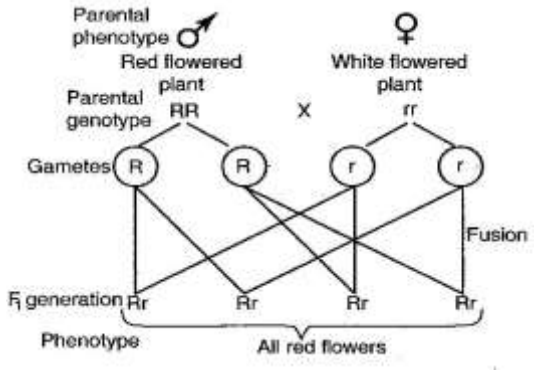
his wife are blood group A. the child's group is O. Is Mrs. Abuto justified in her claim? Make a genetic cross to explain your answer.

- **Inheritance of Rhesus factor**

- ✓ In human beings, possession of rhesus antigen makes one Rhesus positive (Rh+) and this is dominant over absence of Rhesus antigens which is Rhesus negative (Rh-). This is an example of complete dominance in man.
- ✓ If a woman who is Rh- is married to a Rh+ man, when she becomes pregnant the child will be Rh+. The Rhesus antigens cross the placenta into the mother's blood stream prompting the mother's immune system to produce Rhesus antibodies.
- ✓ When the antibodies get into the foetal circulation an antigen-antibody reaction takes place and the red blood cells of the foetus are destroyed (haemolysed). When the baby is born it is very pale.
- ✓ In the 2^{nd} pregnancy the antibodies are more and cause a lot of damage to the foetal red blood cells resulting in death. This is called haemolytic disease of the new born (Erythroblastosis foetalis)

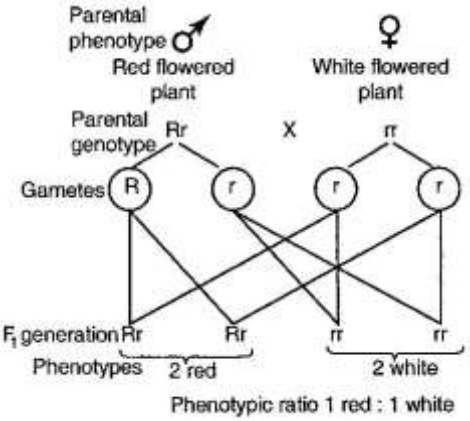
- **Determining unknown genotypes**

- ✓ In the garden pea the gene that determines red colour is dominant over that determines white colour, hence a plant with red flowers may either be homozygous or heterozygous for this characteristic.
- ✓ To establish the genotype it is crossed with a homozygous recessive plant. If the offspring bear red flowers then it indicates that the red flowered plant is homozygous (pure line) e.g.



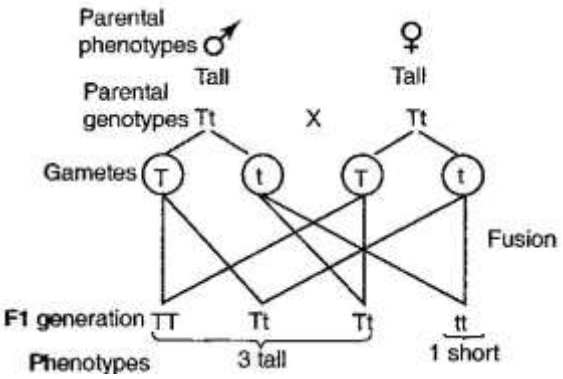
✓

- ✓ If the offspring bear a mixture of red and white flowers in equal proportions giving a ratio of 1:1 this indicates that the red flowered plant was heterozygous e.g.



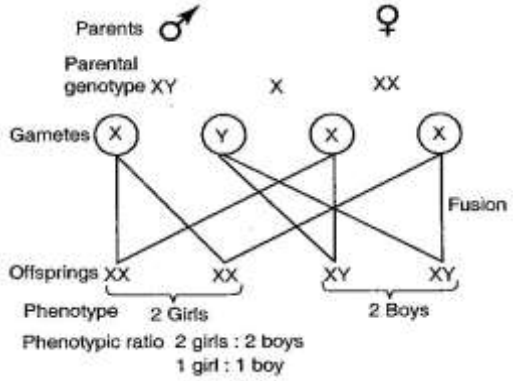
✓

- ✓ A cross between an individual of unknown genotype with an individual of recessive genotype is known as a **test cross**. A test cross where an offspring is crossed with one of its parent is known as **back cross.**
- ✓ Unknown genotypes can also be determined by carrying out selfing experiments e.g. a phenotypically tall plant is either genotypically homozygous or heterozygous for this trait. If selfed and all its offspring are phenotypically tall, then the parental genotype is TT (homozygous dominant).
- ✓ If after selfing both tall and dwarf offspring are produced in the 3:1ratio, then the parental genotype is Tt (heterozygous) e.g.



- **Sex determination**

- ✓ Sex determination in higher animals is controlled by a specific pair of chromosomes e.g. in man, there are 23 pairs of homologous chromosomes in every body cell. The genes that determine whether a child becomes a male or female are located on the specific pair of sex chromosomes called X and Y chromosomes (named after their shape).
- ✓ The remaining 22 pairs of chromosomes are called autosomes and are responsible for other inheritable characteristics.
- ✓ A male human being carries XY (heterogametic) while a female carries XX (homogametic) combination.
- ✓ After meiosis in a male, the spermatozoa can either contain the X or Y chromosome while the female ova will contain only the X chromosome.
- ✓ The sex of a child is a chance occurrence because it depends on whether the ovum is fertilised by an X or Y chromosome therefore there is a 50% chance that fertilization can result in either XY (boy) or XX (girl) e.g.

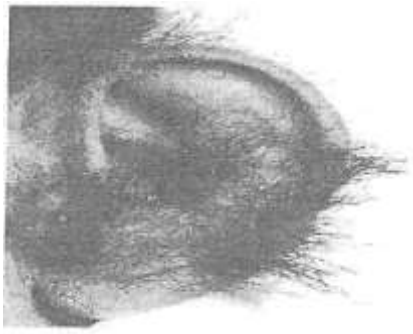


- ✓ In the fruit fly (Drosophila melanogaster) sex determination is exactly as in human beings i.e.
- ✓ Males XY
- ✓ Females XX
- ✓ **Practical activities; To study sex determination in human beings**
- ✓ **Materials**
- ✓ 2 coins of similar denomination
- ✓ 4 labels
- ✓ **Procedure**
- ✓ Obtain 2 coins
- ✓ Label one side of the coin X and the other as Y
- ✓ Label the other coin X on both sides
- ✓ Spin the 2 coins simultaneously and note the letter on top when they stop. Enter the results in the table below. Repeat the procedure 50 times.

Sperm	Ovum	Tally	Totals
X	X		✓
✓ Y	✓ X	✓	✓

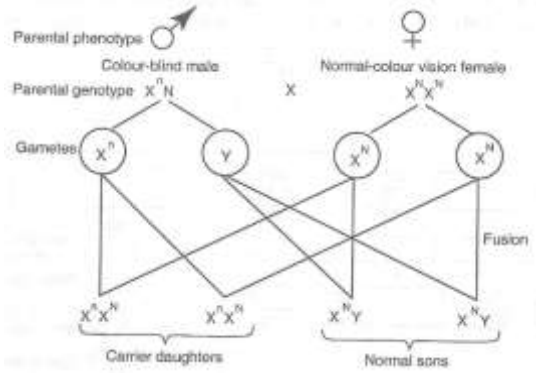
- ✓ Which is the approximate ratio of XX: XY?
- ✓ In birds male is XX and female XY; and in some insects the female is XX and the male is XO; the Y chromosome is missing altogether.
- ✓ **Linkage**
- ✓ Genes located on only 23 pairs of chromosomes control all the thousands of characteristics in human beings. Those genes found on the same chromosome are called linked genes. All the linked genes constitute a linkage group.
- ✓ Linked genes are inherited together and therefore do not segregate during meiosis.
- ✓ **Sex-linked genes**

- ✓ All genes located on the sex chromosome are said to be sex-linked and therefore are transmitted together with those that determine sex.
- ✓ Most sex-linked are carried on the X-chromosome whereas the Y chromosome carries very few genes and is almost empty.
- ✓ In human there are a few genes located on the Y chromosome which control the characteristics that are exclusively male e.g.
- ✓ -Premature baldness
- ✓ -Tuft of hair in the ear pinna and in the nose.

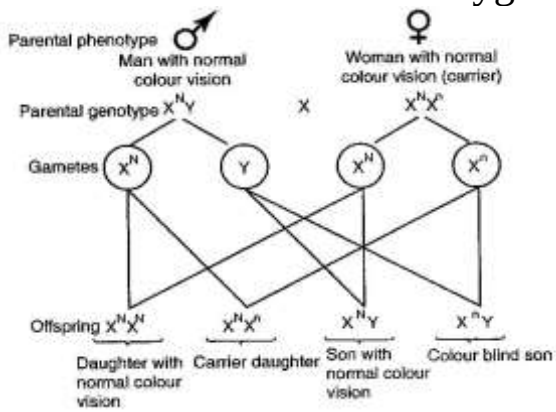


- ✓ Characteristics controlled by genes located on the X-chromosome include;
- ✓ -Colourblindness
- ✓ -Haemophilia
- ✓ These characteristics can arise in either males or females.
- ✓ **Sex-linked genes in man**
- ✓ **Colourblindness**
- ✓ It's the inability to distinguish red and green colours by some people. This trait is linked to the X-chromosome.

- ✓ The genes that determine normal colour vision is dominant over that for colour blindness.
- ✓ A marriage between a colourblind man and a woman homozygous for normal colour vision results in their daughters being carriers, but with normal colour vision. The daughters are described as carriers because they are heterozygous. All the sons of the two parents however are normal e.g.
- ✓ Let **N** represent gene for normal colour vision
- ✓ Let **n** represent gene for Colourblindness
- ✓ Since the gene is linked to X chromosome, its alleles are represented as X^N and X^n.
- ✓

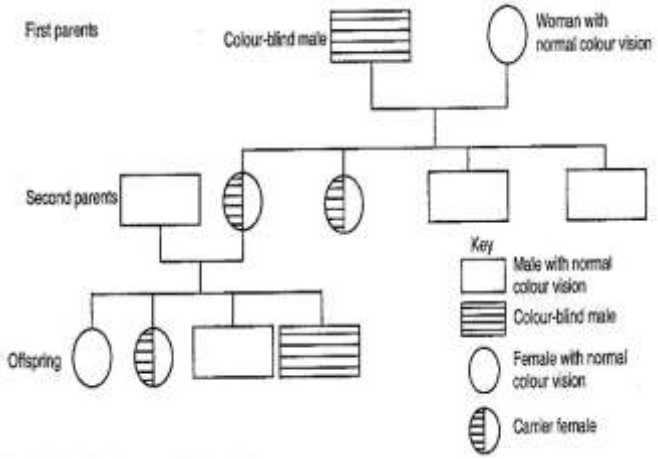


- ✓ If a carrier daughter from the above parents married a normal man, some of their sons will suffer from Colourblindness while the daughters will be either carriers or homozygous for normal colour vision e.g.



- ✓ The above example shows that the gene for Colourblindness is passed from the mother to the sons. This is because the only X chromosome the male offspring inherits is from the mother.

- ✓ There are more male sufferers in a population compared to females. Females only suffer when in homozygous condition of the recessive gene.
- ✓ Inheritance of Colourblindness through several generations can be illustrated using a pedigree. Pedigree is a record in a table form showing the distribution of one or more traits in different generations of related individuals e.g

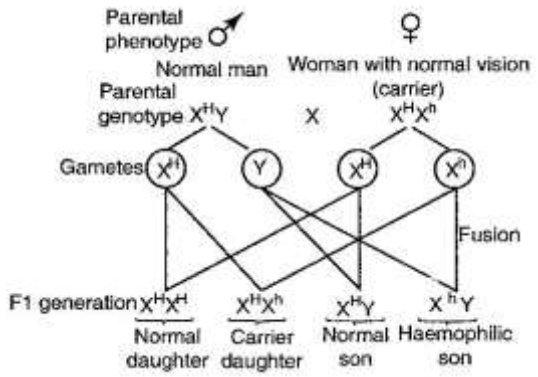


- ✓

Haemophilia

It's also referred to as bleeder's disease. The blood of a person suffering from the disease takes an abnormally long time to clot in the event of a cut, resulting in prolonged bleeding.

- ✓ Haemophilia is caused by a recessive gene on the X chromosome e.g. if a normal man marries a carrier woman for Haemophilia, there is a probability of ¼ that if their child is a boy he will be haemophiliac and if a daughter she will be a carrier e.g.
- ✓ Let **H** represent gene for normal clotting condition
- ✓ Let **h** represent gene for Haemophilia
- ✓ Both genes are located on the X chromosomes eg X^H or X^h



- ✓
- ✓ **Assignment**
- ✓ If a haemophiliac man marries a carrier woman ;
- ✓ (a) Use a genetic cross to work out the product of the above marria
- ✓ (b) What is the probability that their daughter will be haemophiliac
- ✓ (c) The above disorder is more common in males than females. Explain
- ✓ X

- ✓ Also X and Y chromosomes bring about the primary and secondary characteristics e.g. feminine characteristics are controlled by genes on the X chromosome e.g
  - -Enlargement of the breasts
  - -Widening of hips
  - -Growth of pubic hair
  - -Onset of menstrual cycle

- ✓ In males secondary sexual characteristics include;
- ✓ -Breaking of the voice
- ✓ -Growth of pubic hair and beard
- ✓ -Widening of shoulders (masculine characteristics).
- ✓ Other traits influenced by Y-chromosome include;
  -"Porcupine man"- This was the historical man who had patches of hard spiny skin like a porcupine. This trait was passed to sons only.
  -"Hairy ears"- This is where a tuft of hairs sprout out from the pinna of the ear and in the nose. This trait is common in parts of India and is confined to males only.

Premature baldness is limited to males only.

- ✓ **Effects of crossing-over on linked genes**
- ✓ Linked genes are carried on the same chromosome but some of the linked genes separate and are transmitted on different chromosomes. This happens during crossing-over when sections of chromatids of a bivalent intertwine and may break off.
- ✓ Some of these sections get rejoined to different chromatids thus separating genes that were previously linked.
- ✓ Fusion of the few gametes containing chromatids whose genes have changed places in this way will produce new combinations (Re-combinants).
- ✓ However majority of the gametes that fuse contain chromatids whose gene linkage has not been interfered with by crossing-over e.g.
- ✓ X **KLB PG 28 Fig 1.26**
- ✓ **Mutations**
- ✓ These are the sudden changes which occur on the chromosome or genes resulting in change in the individual's genetic make-up. Such an individual is called a mutant e.g.
- ✓ In man, a haemophiliac might suddenly be produced from quite normal parents.
- ✓ Resistance to DDT by some insects e.g. mosquitoes.
- ✓ Mutations are normally due tom recessive genes hence mutations occur naturally but are extremely rare. However they can be induced by certain factors of environment called mutagenic agents e.g.
- ✓ Exposure to gamma rays, ultra-violet (UV) and a variety of chemicals such as colchicines and mustard gas.
- ✓ **Types of mutations**
- ✓ There are two types i.e.
  -Chromosomal mutations/chromosome aberrations
  -Gene mutations
- ✓ **Chromosomal /chromosome aberrations**
- ✓ Involve changes in the structure or number of chromosomes.
- ✓ **Types of Chromosomal mutations**

- ✓ **Deletion**
- ✓ This occurs when some sections of homologous chromatids break off and fail to reconnect to any of the chromatids. In this case, these sections are completely lost and genetic material they contain is said to be deleted out.
- ✓ The consequence includes gross interference in the structure and development of an individual.
- ✓ Most deletions are lethal since the offspring may lose genes responsible for the synthesis of some vital protein molecule e.g.
- ✓ X **KLB PG 29 Fig 1.27(a)**
- ✓ **Duplication**
- ✓ A section of chromatid replicates and adds an extra length to itself, this adds a set of genes e.g.
- ✓ X **KLB PG 29 Fig 1.27(b)**
- ✓ If the gene duplicated were responsible for certain traits, these traits may be over-emphasized in the organism.
- ✓ **Inversion**
- ✓ It occurs when a chromatid breaks at two places. When rejoining, the middle piece rotates and rejoins in an inverted position. This reverses the gene sequence along the chromatid e.g.
- ✓ X **KLB PG 29 Fig 1.27(c)**
- ✓ Inversion might bring closer together genes whose combined effects are advantageous or disadvantageous.
- ✓ **(iv) Translocation**
- ✓ This occurs when a section of one chromatid breaks off and becomes attached to another chromatid but of the non-homologous pair. Hence translocation involves movement of genes from one non-homologous chromosome to another.
- ✓ X **KLB PG 30 Fig 1.27(d)**

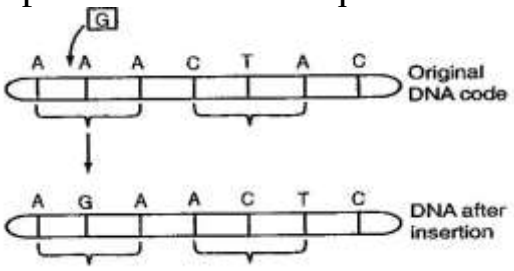
  (v) Non-disjunction
- ✓ This is the kind of Chromosome abnormality that is caused by addition or loss of one or more whole chromosomes. It occurs during anaphase of the 2^{nd} meiotic division when two homologous chromosomes fail to segregate and move on into the same gamete

cell. This results in half of the gametes containing two of the same chromosome whilst the others have none e.g.

- ✓ X **KLB PG 31 Fig 1.28(c)**
- ✓ The fusion of the gametes with two of the same chromosome with a normal gamete of the opposite sex will result in an individual with three such chromosomes i.e. the normal homologous pair plus one extra chromosome (trisomic).
- ✓ A number of human diseases are known to come as a result of non-disjunction e.g.
- ✓ **Down's syndrome/mongolism**
- ✓ This is where there is an extra somatic chromosome number 21.
- ✓ The term mongolism was applied to the disease because the affected individuals have slit-eyed appearance typical of the Mongolian race.
- ✓ Other characteristics are;
- ✓ Reduced resistance to infection.
- ✓ Reduced physical and mental development.
- ✓ Thick tongue.
- ✓ Cardiac malfunctions
- ✓ Short body with stubby fingers
- ✓ Flat nasal bridge
- ✓ Higher incidence is among children of older mothers above 40 years old and may be due to depletion of nutrients in the eggs.
- ✓ Fathers aged 55 years old and above also have increased risk of producing offspring with Down's syndrome.
- ✓ **Klinefelter's syndrome**
- ✓ In this case, individuals have an extra sex chromosome hence they have a total of 47 chromosomes in their cells i.e. XXY (male) and XXX (female).
- ✓ This results when a gamete with an extra sex chromosome fuses with a normal gamete from the opposite sex e.g.
- ✓ X
- ✓ **Symptoms of Klinefelter's syndrome**
- ✓ People with this disorder are externally male (XXY) but they have female features e.g.

- ✓ Female-like breasts, a condition called gnaecomastia.
- ✓ Testes are underdeveloped and produce very few or no sperms (infertility)
- ✓ Reduced facial hair
- ✓ Taller than average with signs of obesity.
- ✓ XXX (females) are females who appear relatively normal in most characteristics.
- ✓ **Turner's syndrome**
- ✓ It's where an individual lacks one sex chromosome hence there are only 45 chromosomes (XO) or (YO) in the cells instead of the normal 46 chromosomes.
- ✓ YO zygotes do not develop due to the absence of many vital genes.
- ✓ Individuals with Turner's syndrome (XO) are females that show underdeveloped female characteristics e.g.
- ✓ -Are infertile due to lack of ovaries and small uterus.
- ✓ -No breast development
- ✓ -Short in stature
- ✓ Other abnormalities arising from non-disjunction of the sex chromosomes lead to the genotype XYY. Individuals with this genotype are males and are known to have a predisposition for violence.
- ✓ **Polyploidy**
- ✓ This refers to the presence of more than two sets of chromosomes in a cell. It can come about due to the failure of a cell to divide after the 1^{st} stage of meiosis.
- ✓ If it divides into two after the 2^{nd} stage of meiosis, diploid gamete results. Fusion of a diploid gamete with a normal haploid gamete of the opposite sex results in an individual whose cell has 3 sets of chromosomes i.e. triploid (3n).
- ✓ If two diploid gametes fuse, the result is tetraploid (4n). This is what is called polyploidy. Polyploidy can also occur if the whole set of chromosomes doubles after fertilization.

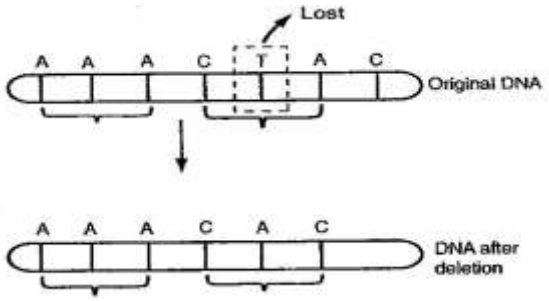
- ✓ Polyploidy is rare in animals but is common in plants e.g. in species of wheat and rice it has various advantages e.g.
- ✓ Increased yields
- ✓ Early maturity
- ✓ Resistance to drought, pests and diseases
- ✓ Polyploidy can artificially be induced by using chemical called colchicines which prevents spindle formation during mitosis thus leading to a cell with double the number of chromosomes (4n).
- ✓ **Gene mutation (point mutation)**
- ✓ A gene mutation arises as a result of a change in the chemical nature of the gene. The change may involve some alteration n the DNA molecule.
- ✓ Types of gene mutation
- ✓ **Insertion**
- ✓ Its the addition of an extra base onto an existing DNA strand e.g. if the base Guanine (G) is inserted between the 1st two Adenines (A) at the beginning of the DNA chain, the resulting M-RNA base triplet and the subsequent amino acid alignment will be altered e.g.



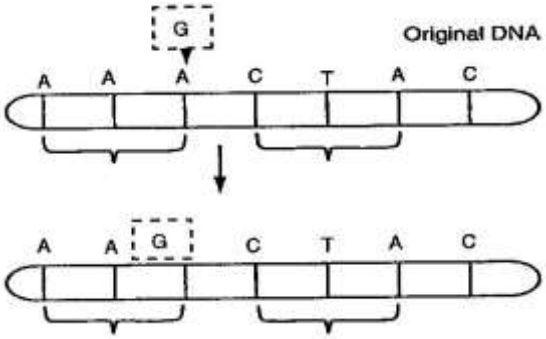
- ✓ By inserting Guanine at the position indicated, no polypeptide chain is formed because none of the intended amino acids have been linked.

(ii) Deletion

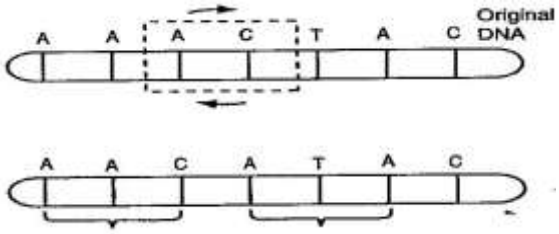
- ✓ It's the removal of a gene portion. If the base thymine (T) is deleted from its position at the indicated section of the DNA strand, the base sequence becomes altered at this point e.g.



- ✓ This results in the alteration of the sequence of the amino acids on the polypeptide chain hence production of a wrong protein.
- ✓ **Substitution**
- ✓ This is the replacement of a portion of the gene with a new portion. If Adenine (A) is substituted with Guanine (G) on a DNA strand the base sequence is altered at this particular portion e.g.



- ✓ **Inversion**
- ✓ This is where the portion of the DNA strand rotates through 180^{o}. The inversion results in the alteration of the base sequence at this point e.g.



- Q. The following are short messages (SMS) on a cell phone communication and can be used as analogies of gene mutation.

Intended	Actual	Mutation

message	message	
Buy me a skirt	Buy me a shirt	Substitution
Mary went shopping	Mary went hopping	Deletion
This is my team	This is my mate	Inversion
Auntie is staying	Auntie is straying	Insertion

- ✓ (a) For each of these messages identify the type of gene mutation illustrated
- ✓ (b) In the messages above show the changes that cause the distortion in the intended message
- ✓ **Disorders due to gene mutation**
- ✓ **Albinism**
- ✓ This is a condition where the synthesis of the skin pigment melanin, fails and is characterized by;
    - -A light skin
    - -White hair
    - -Pink eyes

- ✓ Such an individual is described as an albino.
- ✓ Melanin is a derivative of an amino acid phenylalanine and tyrosine and is synthesized through a series of reactions. Each of these reactions is controlled by a specific gene.
- ✓ In albinism, one of these genes (designated by letter A) is substituted by a recessive gene (designated by letter a). gene **a** in the homozygous condition **aa** blocks in one or two places of the chain reactions involved in the synthesis of melanin, hence melanin is not formed resulting in albinism.
- ✓ The genotype of an albino is homozygous recessive **aa**. A carrier for the trait is heterozygous with the genotype **Aa** and has normal skin pigmentation e.g.
- ✓ X

- ✓ The skin of an albino person is susceptible to sunburn and the eyes are sensitive to bright light.
- ✓ Use of sunglasses and sunscreen lotions help them to lead a normal life.
- ✓ **Sickle-cell anaemia**
- ✓ Normal adult humans contain haemoglobin A in their red blood cells and have the genotype Hb $^{A}Hb^{A}$. In the sickle-cell anaemia, haemoglobin **A** is substituted by a different type called haemoglobin S. such patients have genotype Hb $^{S}Hb^{S}$.
- ✓ Haemoglobin S is defective and has a marked difference from the normal one e.g.

Normal haemoglobin (Hb ^{A})	Defective haemoglobin (Hb ^{S})
A position in each polypeptide chain is occupied by glutamic acid	A similar position in each polypeptide chain is occupied by valine
Does not easily crystallize in low O_2 concentration	Easily crystallizes in low O_2 concentration
The haemoglobin is efficient in O_2 loading and transportation	The haemoglobin is not efficient in O_2 loading and transport
Red blood cells carrying them have the normal biconcave	Red blood cells carrying them have are crescent or sickle shaped.

shape	

- ✓ Most of the individual's red blood cells are therefore sickle shaped and the person frequently experiences oxygen shortage to the body tissues hence cannot carry out strenuous physical exercises.
- ✓ The sickle-shaped cells are not able to squeeze through capillaries; thus they end up clogging blood vessels preventing normal blood flow. This blockage results in to severe pain in the joints, arms, legs and the stomach.
- ✓ With close medical attention, sickle cells persons lead a relatively normal life. However, most deaths are due to infections from other diseases or damage to tissues.
- ✓ A less serious condition is the **sickle cell trai**t. This is a heterozygous condition where less than half the number of red blood cells is sickle shaped. The rest of the cells are normal and are efficient in O_2 loading.
- ✓ Individual with sickle cell trait experiences a mild case of anaemia but leads a normal life. Such individuals have an adaptive advantage in surviving malarial attacks as compared to those who have normal haemoglobin.
- ✓ Inheritance of sickle cell trait
- ✓ X
- ✓ **Haemophilia**
- ✓ It's due to a recessive gene on the X-chromosome produced by gene substitution. The condition is caused by a haemophiliac gene that prevents the production of the necessary clotting factors especially clotting factor viii called antihaemophiliac globulin (AHG).
- ✓ Remedies include the introduction of clotting factors e.g. factor viii and ix from blood donated by normal persons into haemophiliacs.
- ✓ **Colour-blindness**
- ✓ This is where an individual is not able to distinguish between red and green colours.
- ✓ Colour-blindness is brought about by the presence of a gene in its recessive form causing total absence or shortage of the respective cones for colour perception.

- ✓ The gene for colour vision is located on the X chromosome. Due to the occurrence of the gene on the X chromosome, there are more male sufferers than females.
- ✓ Older parents transmit a slightly greater number of mutations to their offspring than younger parents. This increased chance maybe as a result of X-rays or other radiations used in medical diagnosis and treatment.
- ✓ **Achondroplasia**
- ✓ It's a disease that is characterized by a shortened body, legs and hands. Its transmitted by a dominant gene thus both the homozygous dominant (AA) and heterozygous (Aa) individuals show the disease. Homozygous recessive individuals are perfectly normal.
- ✓ Approximately 80% of achondrodystrophic dwarfs die within one year of birth but those that survive show normal mental development and can have children.
- ✓ **Effect of environment on heredity**
- ✓ The development of an organism depends on its genetic make-up or genotype and environment.
- ✓ The interaction between the genotype of an organism and the environment will modify the phenotype of the organism.
- ✓ The degree of modification or influence of gene expression by the environmental factors such as disease and food can be observed and measured e.g. the effects of protein and vitamins in the development of young children has been observed.
- ✓ Mental development and performance depends on both the inherited patterns and the total effect of the environmental influences.
- ✓ **Practical application of genetics**
- ✓ **Plant and animal breeding**
- ✓ Offspring resulting from a cross between two genetically dissimilar lines often posses' beneficial characteristics not shown by either of the parents. This principle is known as hybrid vigour and it has been put to good use in plant and animal breeding e.g.

- ✓ In cattle, Hereford, an English breed contributes high beef production and quick maturation. The boran from Kenya contributes disease resistance and the ability to feed and grow on dry pastures. A cross between Hereford bull and a boran cow yields a suitable hybrid with all these qualities.
- ✓ Early maturity in both plants and animals.
- ✓ Resistance to diseases e.g. Cassava resistant to cassava mosaic
- ✓ Increased length of productive season e.g. chicken breeds with long egg laying durations and kales which can be harvested over a long period.
- ✓ Adaptations to local conditions e.g. maize varieties for various ecological zones.
- ✓ vi) Flowers such as roses, orchids etc are selectively bred for their Colour, shape and aroma.
- ✓ **Blood transfusion**

- ✓ For transfusion, we consider only the effect that the recipient's antibodies in blood plasma will have on the donor's antigens on the red blood cells. The plasma of the donor will be so diluted that it will not affect the red blood cells of the recipient. Blood typing is necessary before transfusion. This includes both the ABO and the Rhesus antigens.
    - **-Settling legal disputes**

- ✓ By matching the blood groups of a baby with those of alleged parents is sometimes possible to settle legal disputes about parentage e.g. an unmarried girl gives birth to a child and accuses a well known politician of being the biological father. The girl has blood group A, genotype AO and the baby is blood group O. The accused man has blood group AB. Could the accused man be the biological father?
- ✓

- ✓ A more advanced method of settling such disputes is to match the DNA of the baby to that of the alleged parents. Such a match will show regions where the base sequence of the baby matches perfectly with those of its parents. This is called DNA fingerprinting.
    - **-Crime detection**

- ✓ The DNA is unique to each individual. Through a series of genetic techniques, the pattern of DNA base sequence is prepared and produced in film. The pattern for each individual is very specific and therefore a "DNA fingerprint".
- ✓ At the scene of crime, a specimen from the suspect e.g. hair, blood, semen in the case of rape is obtained and DNA extracted from the developed hairs or blood cells.
- ✓ By comparing this DNA pattern, it's possible to isolate the culprit from several suspects.
- ✓ **Genetic counseling**
- ✓ This is the use of genetic information to advice couples who have hereditary diseases about the chances of their offspring inheriting the diseases. Hereditary diseases include;
- ✓ -Albinism
- ✓ -Sickle-cell anaemia
- ✓ -Haemophilia
- ✓ -Haemolytic disease of the newborn
- ✓ **Examples**
- ✓ Susan is a carrier of Haemophilia and is worried that her children will turn out to be haemophiliacs. What would you advice her if she marries;
- ✓ **a normal man**
- ✓ X (skip 5 lines)
- ✓ She will have a normal daughter and son, carrier daughter and haemophiliac son.
- ✓ **a haemophiliac man**
- ✓ X (skip 5 lines)

- ✓ She will have a normal son, carrier daughter, and haemophiliac son and daughter.
- ✓ **Advice; It would be better if she marries a normal man**
- ✓ 2. A woman about to be married had a brother who died of sickle-cell anaemia. When samples of her blood were taken and placed in low oxygen concentration, her red blood cells became sickled. This means she is heterozygous. However, her prospective husband's blood remains normal. What would you advise her about her children?
- ✓ X
- ✓ Some children will be normal while others will be affected in low oxygen concentration.
- ✓ It's advisable for people to go for medical tests for blood groups, hereditary diseases and defects to avoid complications in future.
- ✓ **(d) Genetic engineering**
- ✓ This deals with the identification of a desirable gene, altering, isolating and transferring it from one organism to another.
- ✓ Genetic engineering has also made it possible to produce genetically modified organisms (transgenic) which have resulted in increased production in crops and domestic animals.
- ✓ Genetic engineering has been applied in the following fields;
- ✓ **Farming**
- ✓ Genetically modified maize and Soya beans have been produced which have resistance against insect pests.
- ✓ Attempts have been made to transfer the gene for nitrogen fixation from bacteria into cereal crops to increase yields without use of fertilizers.
- ✓ Bovine somatotrophin is a hormone that increases milk production in cows.
- ✓ **Medicine**
- ✓ A strain of *Escherichia coli* has been made which makes insulin. The gene in man that codes for insulin is transferred to the bacterium thereby producing insulin that is purer and in large quantities.

- ✓ Human somatotrophic hormone (human growth hormone) for treatment of dwarfism is now extracted from genetically modified strain of *Escherichia coli*.
- ✓ Sheep have been genetically modified to produce milk which contains medicinal proteins used to relieve Haemophilia and emphysema patients.
- ✓ Effective vaccines from the viruses can be made which contain only outer coats of viruses instead of weakened viruses. Viral coats are mass produced and used in the development of vaccines that do not pose a danger of causing the disease.
- ✓ Some genetically modified plants e.g. bananas are being targeted for the production of orally administered vaccines against rabies and cholera.
- ✓ **Biological warfare-"genetic guns"**
- ✓ *Bacillus anthracis* causes anthrax in cattle, sheep and goats and through them can reach man. It forms spores which are not easily destroyed since they can withstand boiling, freezing and destructive digestive juices.
- ✓ The bacterium affects internal and external parts of the body. If inhaled it can cause pneumonia with bleeding in lungs, ulcers of the stomach and intestines. Internal infections can cause death in 72 hours. Thrown into enemy territory it cause havoc.
- ✓ Other micro-organisms used in warfare include;
- ✓ *Vibrio cholerae*- causes cholera
- ✓ *Clostridium tetani*- causes tetanus
- ✓ *Salmonella typhi*- causes typhoid
- ✓ **Gene therapy**
- ✓ It's the replacement of faulty genes with normal ones aimed at correcting genetic disorders.
- ✓ In somatical gene therapy, genetically modified organisms e.g. viruses are used to carry the normal gene and introduce it into the affected tissue cells. The defective gene in the tissue cell is thereby corrected by the genes in the carrier. This is being used in the treatment of lung cystic fibrosis.
- ✓ **Cloning**

- ✓ This is a type of reproduction where a group of cells arise from a single individual cell without fertilization. The offspring are called clones and are genetically identical.
- ✓ In the recent past, cloning was carried out in sheep. In this process, a nucleus from a fertilized ovum was removed and replaced with a diploid nucleus of a cell from the mammary glands of another sheep. The ovum was implanted into the sheep's uterus.
- ✓ The lamb, Dolly was a clone from the sheep which donated the mammary gland cell.
- ✓ Cloning of plants is more common and successful. It has resulted in tissue culture techniques through which new varieties of crops such as pyrethrum and bananas have been produced.
- ✓ Designs to produce human-like creatures (human cloning) to be reared as a source of human spare parts in surgeries and transplants.
- ✓ **Human genome**
- ✓ Genome is the total genetic content of any cell in an organism. It comprises of all genes on all the chromosomes. In human there are up to 100,000 genes. The human genome project aims at;
- ✓ **-Gene mapping**
- ✓ This is identifying specific positions occupied by specific genes on a chromosome e.g. Haemophilia is located on the X chromosome.
- ✓ **-Sequencing of gene**
- ✓ It involves analyzing DNA to reveal the order of bases in all chromosomes
- ✓ Importance of human genome project
- ✓ Identification of defective genes hence facilitating their correction.
- ✓ Identification of genes that is susceptible to certain diseases so that individuals can take preventive measures