3 Genetics — Some Basic Fundamentals

***Syllabus** : Genetics : Mendel's laws of inheritance and sex-linked inheritance of diseases.*

Scope of Syllabus : Monohybrid cross, dihybrid cross. The following terms to be covered : gene, allele, heterozygous, homozygous, dominant, recessive, mutation, variation, phenotype, genotype. Sex determination in human beings.

Sex-linked inheritance of diseases to include haemophilia and colour blindness (only criss cross inheritance).

Whenever a child is born the family members usually start comparing his appearance as to whom he/she resembles more. All this, although a matter of chance, is governed by certain laws. This chapter presents some fundamentals of Genetics including Mendel's laws of inheritance as envisaged in the syllabus. In order to properly understand Mendel's laws of inheritance (page 29) and the sex-linked inheritance (page 26), it is essential to know beforehand, some basic aspects of genetics. You will learn about the chromosomes, the genes and some very common hereditary traits in humans. You can do several investigatory projects, if desired.

3.1 WHAT IS GENETICS ?

Genetics is the study of transmission of body features (both similarities and differences) from parents to offspring and the laws relating to such transmission.

GREGOR MENDEL — An Austrian Monk and Father of Genetics

Gregor Mendel (1822 – 84) was born in a peasant family. He had his early education in a monastery and later he studied Science and Mathematics at University of Vienna. *He wanted to be a teacher but luck did not favour him, and he failed in the examination of teaching certificate. But what he discovered later as a craze, has made him a* ***"teacher of teachers"***. He had returned to his monastery and spent the rest of his life as an abbot. The monastery had a lovely garden which satisfied his urge to understand some secrets of life regarding the inheritance of certain features in garden pea. His findings are now called Mendel's laws of inheritance.

Mendel — "Father of Genetics"

Two modern applications of genetics

Genetic engineering is the technique in which the ***genetic constitution of an organism*** (bacterium) ***is altered by introducing new genes into its chromosomes***. The genetically modified organism (GMO) thus produced is grown to multiply fast and the gene product is obtained in large quantities. The hormone ***insulin*** was the first such product. The insulin producing gene of mammals has been successfully introduced in certain bacteria which have been made to produce it.

Genetic counselling is yet another practical aspect. Newly married couples are advised to consult a specialist regarding the possibilities of any undesirable trait which their children might inherit. Diseases like ***haemophilia*** (bleeder's disease with a tendency to bleed freely from even a slight wound), ***thalassaemia*** and ***sickle cell anaemia*** with defective haemoglobin are examples of genetic diseases which can be prevented to some extent by proper genetic matching of the prospective parents.

Heredity

The term heredity may be defined as "transmission of genetically based characteristics from parents to offspring."

The term heredity is rather old. For centuries, man has known that many characteristics are inherited in animals (domesticated ones) as well as in humans, and that he could breed varieties of animals and plants with desirable qualities.

Like begets like
It means that young ones look like their parents

- Cats produce cats and not dogs.
- A mango seed germinates into a mango tree.
- Humans give birth only to humans and not to apes.
- Even the curd bacteria which grow in milk undergo hundreds of generations each day, and continue to produce the same type of bacteria and not of any different type.

Like Begets Like and Yet the variations

All organisms —whether animals, plants or microorganisms, produce their **own** kind through reproduction. But the offspring are never identical to their parents; some difference, how-so-ever small it may be, is found in them.

3.2 VARIATIONS IN POPULATION

Human beings as a species share many main characters or traits among themselves which identify the **species** *Homo sapiens*. Yet the various **races** or tribes look different in several features. Even within the same race or tribe, the individual members in the **population** show differences. Further, the smallest unit of population is the **family** and there too, members show differences in body features. *These small differences among individuals are called* ***variations***.

The same is true for animals also. We keep pets, like dog or cat and domesticate animals, like cow or buffalo. There are so many breeds of each one of them and even in the same breed, there are minor differences, and variations among individuals.

CHARACTER AND TRAITS

Any inheritable feature is a character. The alternative forms of a character are called traits.

INHERITANCE IN HUMANS

We inherit thousands of **characters** from our parents (father and mother) who in turn, had inherited them from their parents. Thus the family members— brothers, sisters, cousins—tend to resemble one another. Here is a list of some such **characters** and their **traits** :-

1. Colour of the eyes—brown or blue
2. Hair shape—Curly or straight
3. Eyebrows—Heavy bushy or thin
4. Hair on the middle joint of fingers — Growth or no growth
5. Colour vision—Normal or red-green colour blindness
6. Tongue rolling—Rolling of tongue into U-shape when extended out from the mouth or no rolling
7. Hand use—Right-handedness or left-handedness
8. Skin colour—Albinism (total absence of pigment in skin) or normal (light or dark) pigment.
9. Ear lobe—Free or attached
10. Lips—Thick or thin
11. Rh Blood group—Rh positive or Rh negative.

PROGRESS CHECK

1. Mention if the following statements are ***True*** (T) or ***False*** (F) :
   (i) Genetics and heredity are the same thing. T/F
   (ii) "Like begets like", this applies only to animals. T/F
   (iii) The entire human population shows variations. T/F
2. Which of the following in humans are established genetic traits ? (Tick-mark the correct ones) in the box provided.
   (i) Capacity to be a good cricketer ☐
   (ii) Curly hair ☐
   (iii) Left-handedness ☐
   (iv) Quality of voice ☐
   (v) Red-green colour blindness ☐

3.3 CHROMOSOMES—THE CARRIERS OF HEREDITY

Chromosomes are only visible when a cell nucleus is about to divide. Photographs of the dividing cell nucleus can be taken through a high powered light microscope. These photographs are used for artificially arranging the chromosomes according to their size and shape on a chart (**karyotype**). One such human karyotype is shown below in Fig. 3.1

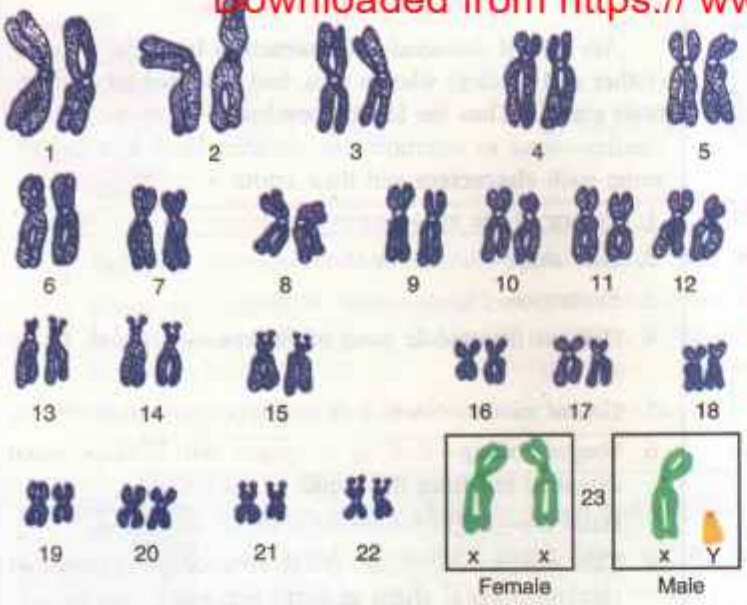


Fig. 3.1 : The human chromosomes arranged in pairs in order of their size and shape (karyotype). Chromosomes become distinctly visible only during cell division. Each chromosome is split into two chromatids joined by a small circular dot-like centromere

Chromosome number

The chromosome number is constant for the individuals of a species, and each body cell has the **same** number. Humans have 46 chromosomes.

Chromosome numbers of some other common animals and plants are as follows :

Ascaris	.....	2
Onion	.....	16
Maize	.....	20
Lion	.....	38
Tiger	.....	38
Domestic cat	.....	38
Mouse	.....	40
Humans	.....	46
Gorilla	.....	48
Potato	.....	48
Monkey	.....	54
Dog	.....	78
Chicken	.....	78
Crayfish	.....	200
Some insects	.....	More than 1000

Chromosomes in Homologous Pairs

Once again look at the above list carefully. In each organism, the chromosomes occur in even numbers. This is so because they always occur *in pairs*. Thus in humans, the 46 chromosomes are present in 23 pairs (Fig. 3.1). The two chromosomes of each pair are similar in size and shape and are derived as one each from the two parents. These identical chromosome pairs are called homologous chromosomes.

Homologous chromosomes

A pair of corresponding chromosomes of the same shape and size, one from each parent.

3.4 THE TWO MAIN CATEGORIES — AUTOSOMES AND SEX CHROMOSOMES

In Fig. 3.1, you would notice that each one of the chromosome pairs numbered 1-22, has identical chromosomes and these are categorised as **autosomes**. But the 23rd pair is different and its chromosomes are called **sex chromosomes** which are designated as X and Y. The XX pair with similar partners is found in females whereas the XY pair with dissimilar partners is found in males. The Y chromosome of males is much smaller than the X chromosome (Figs 3.1 & 3.2).

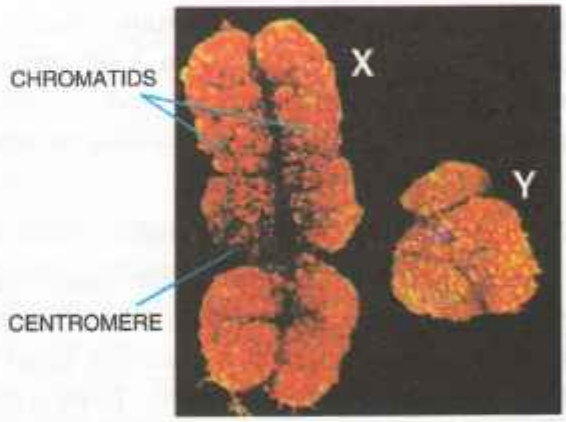


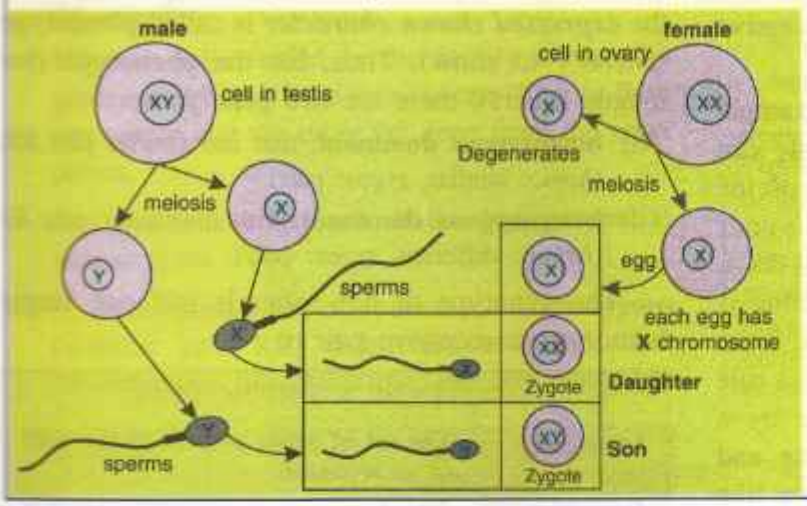
Fig. 3.2 : Two human male sex chromosomes (X and Y) showing the chromatids and centromere in each

3.5 SEX DETERMINATION — SON OR DAUGHTER

The sex of the child depends upon the kind of sperm that fertilises the egg. The egg contains only one X chromosome, but half of the sperms released into the female are X-bearing and the remaining half are Y-bearing. It is simply a matter of chance as to which category of sperm fuses with the ovum :-

– If the egg (X) is fused by X-bearing sperm, the resulting combination is XX, *i.e.* female type and the child produced is female **(Daughter)**.

Son or Daughter?
(So far only a matter of chance)



All eggs are alike (each with one X-chromosome), but sperms are either with X- or with Y-chromosome (50% of each kind). **Which sperm fertilises the egg determines whether the child will be male or female.**

Successful trials have been reported of isolating the two kinds of sperms and getting the ovum fertilised by the particular kind for the desired sex of the unborn.

– If the egg (X) is fused by Y-bearing sperm, the resulting combination is XY, *i.e.* male constitution and the child produced is a male **(Son)**.

PROGRESS CHECK

1. Mention the following :
   (i) Total number of **pairs** of chromosomes in each body cell in humans .....
   (ii) Any two animals having **19 pairs** of chromosomes .....
   (iii) Number of **pairs** of autosomes in humans. .....
   .....
2. A certain couple got only four daughters in a row and no son. Does it mean that the husband does not produce Y-bearing sperms? Explain .....

3.6 CHROMOSOMES — CARRIERS OF GENES

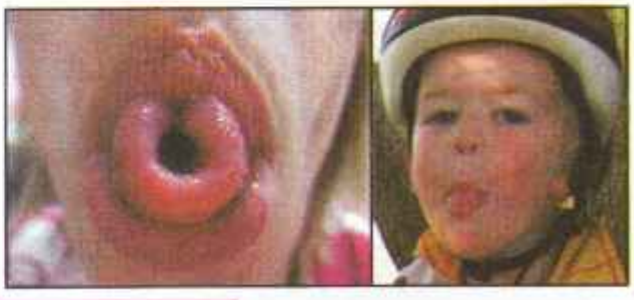
All species have a fixed number of chromosomes. However, the characteristics of species including physical appearance, body functions, behaviour, etc., are not simply the outcome of chromosome number, but these are the result of the units called **genes** which the chromosomes carry. The lion and the cat have the same number of chromosomes (38). Yet one is distinct from the other in body size, appearance, colour, behaviour, etc. All such characteristics of an organism are the result of the genes located on the chromosomes.

GENES AND GENOME

- **GENES : Genes are the specific parts (DNA segments) of a chromosome, which determine the hereditary characteristics.**
- **The Number of Genes in Humans : According to the latest findings there are nearly 30,000 genes in humans. Chromosome No. 1 has the largest number of genes (2968) and chromosome Y has the fewest (231).**
- **GENOME is the full complement of DNA (including all genes and the intergenic regions) of an organism.**

3.7 GENES AND THEIR ALLELES

Normally, every gene has two alternative forms for a character producing different effects. These alternative forms are called the **alleles**. *For example*, in the character of tongue rolling (projecting the tongue out of the mouth in the form of a tube), there are two possibilities — *either* **one can roll the tongue** *or* **one cannot**. Thus there are two alleles of this gene — one for rolling and the other for non-rolling.



The year 2009 — Hundredth birthday of the term "gene". The word was coined by geneticist Wilhelm Johanssen in 1909 to simply describe what parents passed to their offspring, the detailed DNA structure came to knowledge much later.

DOMINANT AND RECESSIVE ALLELES

Out of the two alleles of a gene, one is **dominant** (superruling) and the other is **recessive** (subordinate or submissive).

To understand this, again take the same example of **tongue rolling**. A large number of people can roll their tongue into a tube projecting out of the mouth. Try whether you can do it. Ask your classmates to try the same. Most of them can do it, but a few may not. The gene for tongue rolling is located on a particular pair of chromosomes. Thus every individual has a pair of this gene. It is a rule in genetics to represent the dominant gene by a capital letter, so here **"R"** for tongue rolling and the recessive gene by the same but small letter like **"r"** for non-rolling. Thus, a recessive gene is one which, in the presence of the contrasting (dominant) gene, is not expressed.

ALLELES : Alternative forms of a gene, occupying the same position (locus) on homologous chromosomes and affecting the same characteristic but in different ways.

A pair of chromosomes showing four imaginary genes A, B, C, and D. The situations depicted here are:

(i) AA is homozygous (similar) dominant pair,
(ii) bb is homozygous (similar) recessive pair and
(iii) Cc and dD are heterozygous (dissimilar) pairs.

Remember – *The chromosome as a whole can neither be dominant nor recessive.*

3.8 GENOTYPE AND PHENOTYPE

The three situations pertaining to any pair of genes, as for example in tongue rolling, can be as follows :

(i) RR (both **dominant**)
..... TONGUE ROLLER

(ii) Rr (one **dominant**, one recessive)
..... TONGUE ROLLER

(iii) rr (both **recessive**)
..... NON-ROLLER

You can see from the above that the tongue rollers have two kinds of genetic constitutions (RR or Rr) whereas non-rollers have only one genetic constitution (rr).

The ***genetic constitutions*** (pertaining to the kinds of genes possessed) are called **genotype** and the ***expressed shown character*** is called **phenotype** (*phene* : to show). Thus, for the phenotype (for tongue rollers), there are two genotypes:

(i) **homozygous dominant**, that has similar pair RR (*homo*: similar, *zygos*: pair)

(ii) **heterozygous dominant** with dissimilar pair Rr (*hetero:* different, *zygos:* pair)

The genotype of non-roller is just one single homozygous recessive pair rr.

GENOTYPE – PHENOTYPE

Genotype – The set of genes present in the cells of an organism

Phenotype – The observable characteristic which is genetically controlled.

3.9 FROM PARENTS TO CHILDREN — TONGUE ROLLING — AN EXAMPLE OF INHERITANCE

Fig. 3.3 illustrates a family chart of two parents and their three children, again illustrating the trait of **tongue rolling**.

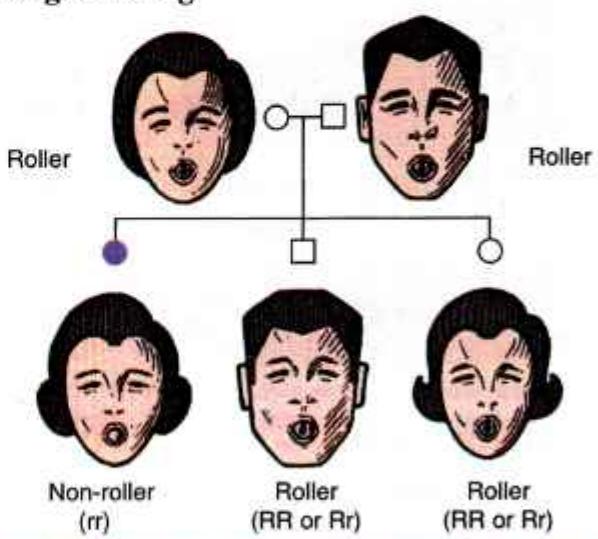


Fig. 3.3 A pedigree (family) chart showing inheritance of the ability to roll the tongue. (Symbols : O = female, □ = male; solid symbol indicates the character being traced)

Observe the following points in the above pedigree (family) chart :

— **Males** are shown by squares and **females** by circles.

— Both father and mother are tongue rollers (**hollow symbols** represent the usual expressed character).

— Of the three children born, two can roll (hollow symbols) and one cannot (**solid symbol**).

— The recessive trait (rr) of non-rolling in one of the children could have come from nowhere else but the parents.

— The non-roller child in the family chart with genotype "rr" must have received one "r" gene from one parent and the other "r" gene from the other parent.

— Conclusion is that each of the two parents is heterozygous (Rr).

— If, on the other hand, one parent was **homozygous** (similar pair RR) dominant and the other **heterozygous** (dissimilar pair Rr) dominant, then every child would have got at least one dominant gene from the homozygous dominant parent through the sex cells. The situation can be schematically explained as given in a Punnett square (Fig. 3.4).

Punnett square is a simple diagram in which the different types of gametes (sex cells with the concerned trait) of one (female) parent are placed along one side of the square and those of the other parent (male) are placed along the other side. Then, the possible combinations (genotypes) of the opposite gametes are given in the sub-squares. The resulting phenotypes can be written under the genotypes.

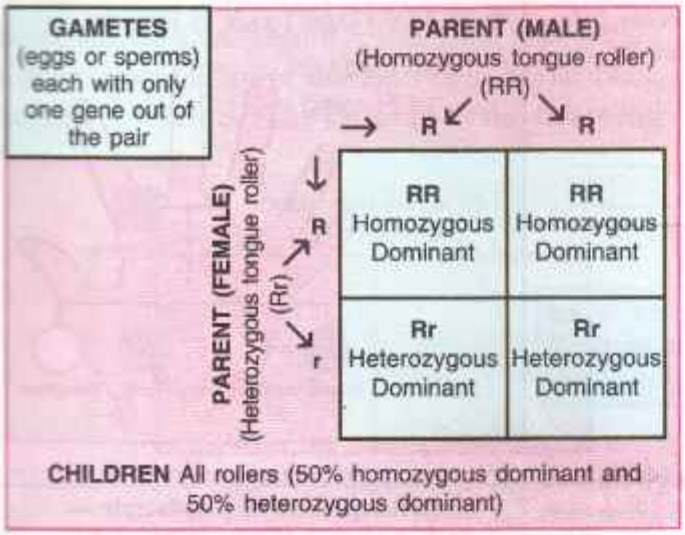


Fig. 3.4 : A possible family where both parents are phenotypically similar (tongue rollers) but genotypically dissimilar (one homozygous dominant and "RR" the other heterozygous "Rr"). All their children will be tongue rollers

Dominant and recessive forms of some common Hereditary Traits in Humans

Character	Dominant trait	Recessive trait
Eye colour	Brown	Blue
Tongue	Rolling	Non-rolling
Hand	Right-handedness	Left-handedness
Ear lobe	Free	Attached
Lips	Thick	Thin
Rh blood group	Positive	Negative
Colour vision (red-green)	Normal	Colour blind
Cheek	Dimpled	Normal

Some rather uncommon hereditary traits

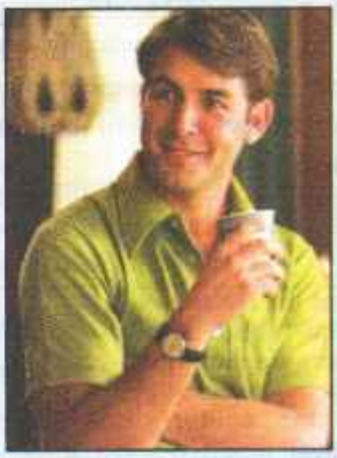
Polydactyly (Extra fingers & toes) = Dominant
Albinism (Total absence of skin pigment) = Recessive
Camptodactyly (Inability to straighten the little finger) = Dominant

SOME BIOLOGY IN DESIGNING THE WRIST WATCHES !

Most people wear the wrist watch on left hand and only *very few* on right hand. Is it ***true*** ?

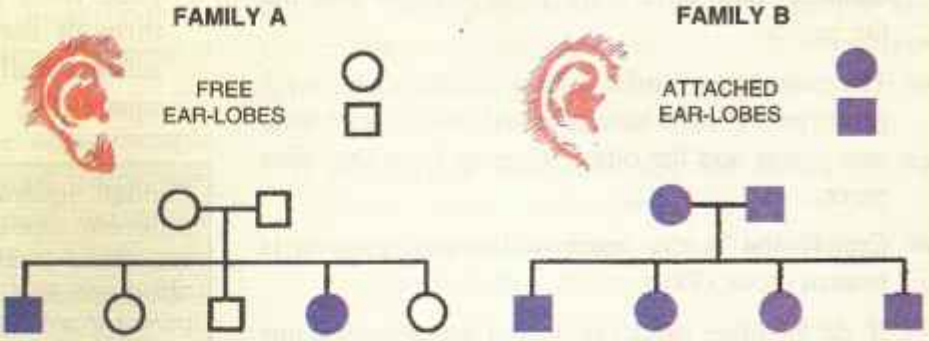
Something to do with human genetics.

(**Hint** : Interpret the position of the knob of the wrist watch and the related human genetic trait).



AN INTERESTING EXERCISE TO TRY

Can you try to explain the inheritance of the character of **Free** or **Attached Ear lobes**. Given alongside are pedigree charts of two families A and B. In family A, both parents have free ear lobes and those of family B, have attached ear lobes (Recollect from previous example what the symbols of circle and square, indicate).



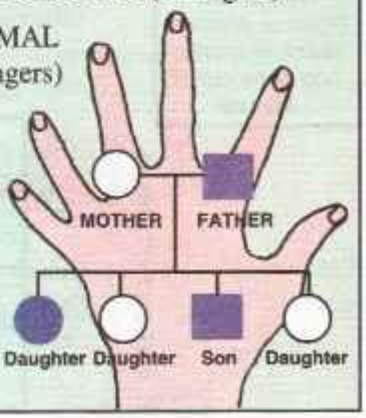
Pedigree charts of two families A & B showing the inheritance of attached ear lobes

Can you find answers to the following ?

1. Which trait — free ear lobe or the attached ear lobe is dominant ?
2. Family B has all children of one type only whereas family A has both types. Parents in which family A or B, are heterozygous for the character ?
3. What is the difference indicated by
   (i) Squares and circles ?.....
   (ii) Solid symbols and hollow symbols ? .....

Can you describe the situation here too?

● ■ POLYDACTYLY (6 Fingers)
○ □ NORMAL (5 Fingers)



3.10 SEX-LINKED INHERITANCE

Sex-linked inheritance is the appearance of a trait which is due to the presence of an allele exclusively either on the X chromosome or on the Y chromosome.

'X' linked inheritance :

Certain disorders caused due to heredity such as **haemophilia and colour-blindness** are more common in males than in females. Such defects are due to **recessive genes**, which occur on the **'X' chromosome**. (You can check if you are normal or colour blind by reading the numbers given in the three coloured circles on page 126. If you can see them, you are normal otherwise colour blind as in fourth circle).

Haemophilia is a genetic disorder in which the sufferers (homozygous recessive female and the recessive X-bearing male) are at a risk of bleeding to death because the blood fails to clot in them. Rare cases of haemophiliac males do occur but practically, none of haemophiliac females.

The following cases explain the sex-linked (X-chromosome linked) **inheritance of colour-blindness** (or **haemophilia**) **in humans.**

CASE 1

Normal mother (XX) × Colour-blind father (X°Y)

Colour-blind father (X°Y)
Sperms (two types)

Normal mother (XX) Eggs (all similar)	X°	Y
X	XX°	XY
X	XX°	XY

[None of the children is colour-blind but daughters are carriers (XX°) of the defective allele for colour-blindness]

XX° : Daughters - heterozygous dominant, normal vision
XY : Normal sons

An extreme case of polydactyly — THE LATEST WORLD RECORD.

15 Month old **"Akshat"** from *Bareilly, Uttar Pradesh*, with 7 fingers in each hand and 10 toes in each foot (thus 34 digits in all). He was successfully operated at AIIMS, New Delhi in June/July 2011; to remove the extra digits and reshaping a few of them both in hands and feet.

CASE 2

Carrier mother (not colour-blind) (XX°) and (XY) Normal father

Normal father (XY)
Sperms (two types)

Carrier mother (X°X) (not colour-blind) Eggs (two types)	X	Y
X°	X°X Carrier daughter	X°Y Colour-blind son
X	XX Normal daughter	XY Normal son

CASE 3

Think of a possibility of a marriage between carrier woman (XX°) and a colour blind man (X°Y). Is there a possibility of the birth of a colour blind daughter ? **Work out the progeny in the following Punnett square.**

Colour-blind man (X°Y)
sperms (two types)

Carrier woman (X°X) Eggs (two types)		

CRISS-CROSS INHERITANCE

(Mother to Son and Father to daughter)

Inheritance of X-linked genes as in colour blindness and haemophilia is also called **criss-cross inheritance**. This is because the son (male sex) may get it from the otherwise normal but carrier mother (as in case 2) and a colour blind father may pass it on to the daughter making her colour-blind if the mother is a carrier. (Refer to case 3, if you have solved it.)

EXTRA **Not in syllabus**

'Y'-linked inheritance :

The Y–chromosome linked traits (characters) occur in males but not in females. *For example*, traits such as **hypertrichosis of ears** (hair growing out of ears), and **pattern baldness** are found in men only. This is because the dominant genes of such traits are found on 'Y' chromosome which determines the male sex.

PROGRESS CHECK

1. Approximately, how many genes have been discovered in humans ?
2. Define the following terms :-
   (i) Allele .....
   (ii) Dominant gene .....
   (iii) Genotype .....
   (iv) Phenotype .....
   (v) Heterozygous dominant .....
3. Can there be a heterozygous recessive ? Explain.
   .....
   .....
4. List any four traits in humans which you can easily study just by observing and making family charts.
   (i) .....
   (ii) .....
   (iii) .....
   (iv) .....

3.11 MENDEL'S EXPERIMENTS ON INHERITANCE

The basic principles of genetics were discovered for the first time by Gregor John Mendel in the mid-nineteenth century. Mendel was an Austrian monk and he conducted breeding experiments on garden pea (*Pisum sativum*) out of sheer interest. His findings became a milestone in biology.

Mendel had selected garden pea for three reasons:

1. **Many varieties** were available in **alternative forms** of a character.
2. Varieties were available in **pure forms that bred true**, i.e. produced the same type generation after generation.
3. Peas are **normally self-pollinated** but self-pollination could be prevented by removing corresponding reproductive parts (male part stamens and the female part carpels) of the flower and could as well be **cross-pollinated artificially**.

Mendel took varieties of this plant showing seven pairs of contrasting features as shown ahead in Fig. 3.5.

Mendel crossed pure breeding varieties, first, by taking only one feature at a time **(monohybrid cross)** and then, by taking two features together

CHARACTER	DOMINANT vs. RECESSIVE TRAIT	
Flower **colour**	Purple	White
Seed **colour**	Yellow	Green
Seed **shape**	Round	Wrinkled
Pod **colour**	Green	Yellow
Pod **shape**	Inflated	Constricted
Flower **position**	Axillary	Terminal
Plant **height**	Tall	Dwarf

Fig. 3.5 : Seven pairs of contrasting features of garden pea studied by Mendel

(dihybrid cross). He tried with all the seven features and his observations were similar for all.

CASE 1 : One of the monohybrid crosses is shown below in Fig. 3.6. A pure breeding plant bearing **terminal flowers** was cross-pollinated with a pure breeding plant having **axillary flowers** (flowers borne in the axil of leaves).

The resulting seeds after sowing produced all plants with axial flowers only (none bearing terminal flowers). When these hybrid plants (F_1 generation*) were self-pollinated, they produced in F_2 generation plants with axillary flowers and plants with terminal

* F_1= First filial generation. **Filial generation** means a cross between offsprings. The first, second and third generation of the offspring are known as first filial, second filial and the third filial generation and are denoted by the symbols F_1, F_2 and F_3 respectively.

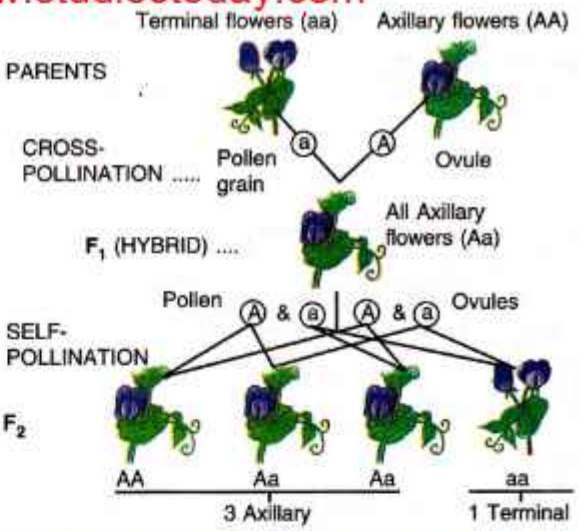


Fig. 3.6 : Crossing of garden pea bearing terminal flowers with pea bearing axillary flowers through two generations (F_1 and F_2)

flowers in the ratio of 3 : 1. These were the visible forms which we call the phenotypes. Out of these, the one with terminal flowers (aa) in subsequent (self-pollinated) generation produced all plants with terminal flowers only. Out of the other remaining three with axillary flowers :

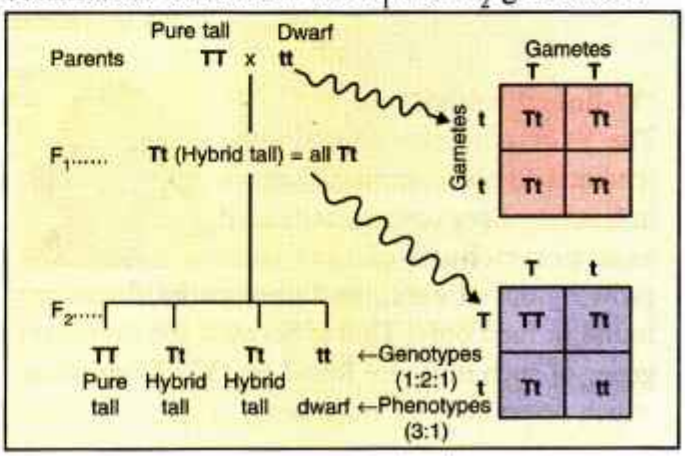
- One (AA) produced all plants with axillary flowers only
- Two (Aa) again produced in the same ratio 3 : 1 as was in F_2 generation.

This kind of ratio obtained by crossing for two ***different traits*** of a ***single character*** is known as **monohybrid ratio**. This consisted of the following:

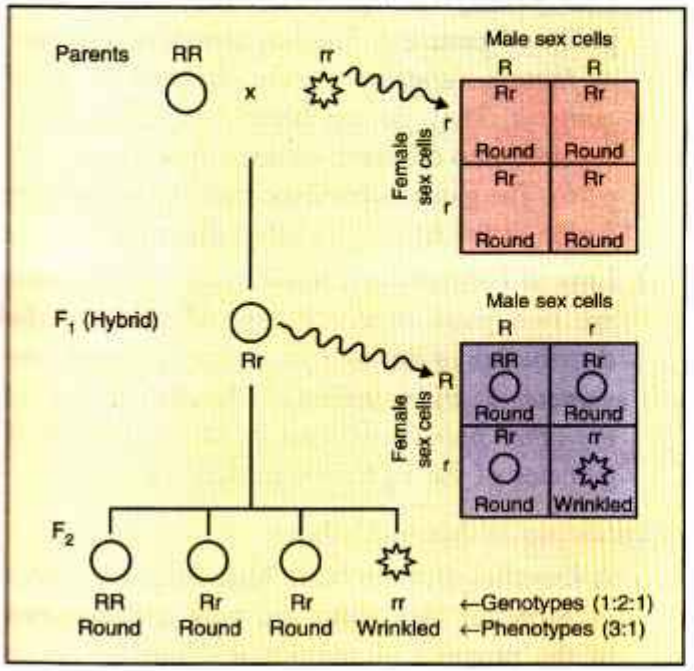
Phenotypic (visible feature) ratio = **3 : 1** (three axillary and one terminal)

Genotypic (gene feature) ratio = **1 : 2 : 1** (one AA, two Aa and one aa)

CASE 2 : Pure **tall** (TT) pea plants were crossed with **dwarf** (tt) plants and the progenies were obtained as follows in the F_1 and F_2 generation.



CASE 3 : Similarly, on crossing the plants grown from pure **round(RR)** seeds with plants grown from pure **wrinkled(rr)** seeds, the results were as given below :



In all the above three cases, the monohybrid ratios are same:

Phenotypic ratio 3 : 1;

Genotypic ratio 1 : 2 : 1

Some of the generalized principles based on the above breeding experiments were as follows :

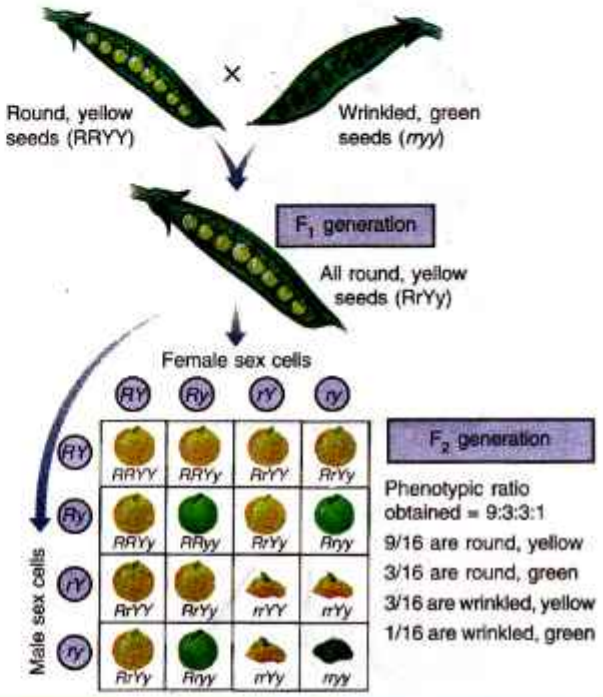
1. Each pair of contrasting characters depends on a pair of genes.
2. Each individual carries such genes in duplicate.
3. An individual produces gametes (sex cells) which have only one member of a pair of genes.
4. Fertilization of the gametes restores the duplicate condition of the genes.
5. Sex cells with respect to the genes they contain, get fertilised at random.

Dihybrid ratio (breeding results with two pairs of contrasting characters).

Mendel tried several combinations. One such combination was the crossing of a variety with **round and yellow** seeds with another variety having **wrinkled and green** seeds.

The ratio is 9 : 3 : 3 : 1.

The above results from parents to F_1 and then to F_2 can be easily understood from the following :



Thus, the two kinds of ratios in the two kinds of hybridisations are as follows :

Monohybrid ratios in F_2 generation

Phenotypic — 3 : 1

Genotypic — 1 : 2 : 1

Dihybrid ratios in F_2 generation

Phenotypic — 9 : 3 : 3 : 1

Genotypic — (very complex)

3.12 MENDEL'S LAWS OF INHERITANCE

Mendel's generalizations of the results of breeding experiments are summarised under three laws:

1. **Law of Dominance.** *Out of a pair of contrasting characters present together, only one is able to express itself while the other remains suppressed.* The one that expresses is the **dominant** character and the one

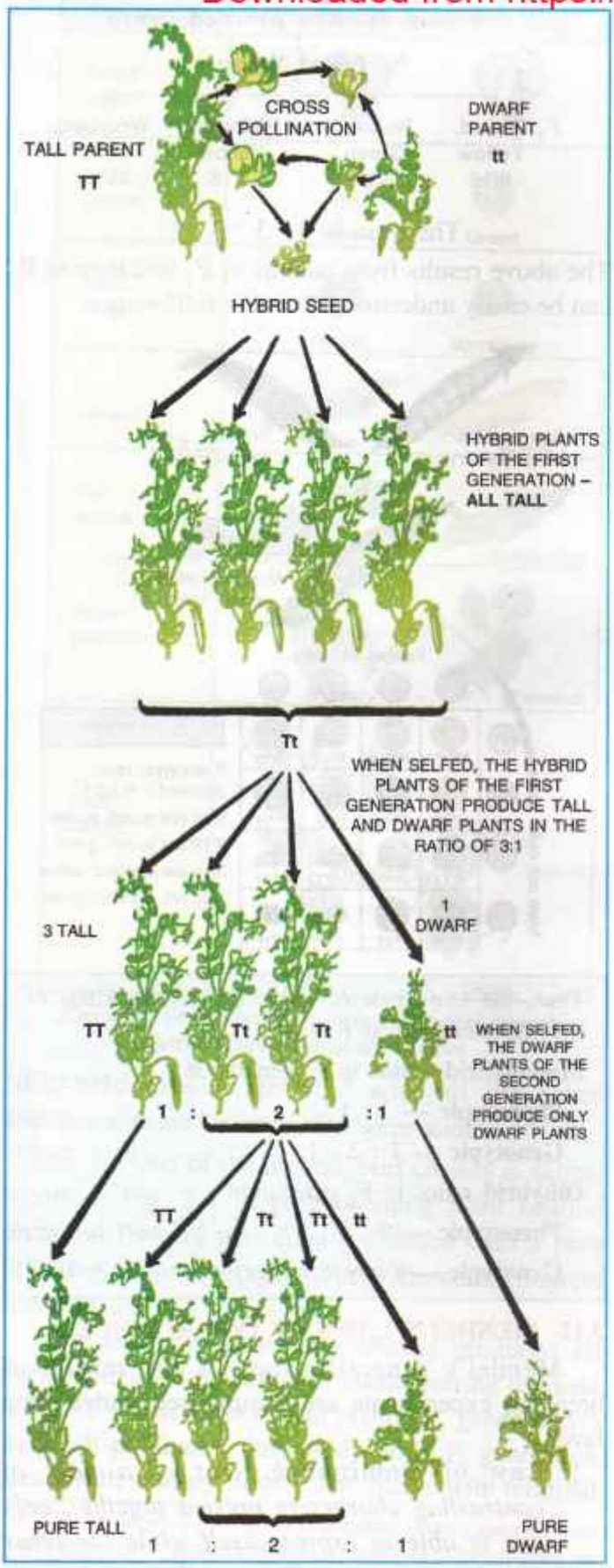


Fig. 3.7 : Diagrammatic representation of Mendel's results obtained by crossing tall plants with dwarfs of garden pea

unexpressed is the **recessive**. The recessive character can express only when the pair consists of both recessives (homozygous recessive).

2. **Law of Segregation** (also called the law of purity of gametes). The *two members of a pair of factors separate during the formation of gametes*. They do not blend but segregate or separate into different gametes (see Fig. 2.11A p.16). The gametes combine together by random fusion at the time of zygote formation.

3 **Law of Independent Assortment**. When there are two pairs of contrasting characters, *the distribution of the members of one pair into the gametes is independent of the distribution of the other pair* (as is seen in the production of gametes of the F_1 hybrid in dihybrid cross).

Application of Mendel's laws

(i) A knowledge of the basic Mendelian principles gives us an idea about the new combinations in the progeny of hybrids and enables us to predict their frequency.

(ii) Such information is of great importance to both plant and animal breeders for producing better breeds.

(iii) New types of plants with new combinations of useful characters can be produced by hybridisation.

3.13 MUTATION

Mutation is a sudden change in one or more genes, or in the number or in the structure of chromosomes.

Mutation alters the hereditary material of an organism's cells and results in change in certain characters or traits. *For example* :

(i) **Sickle cell anaemia.** It is a blood disease caused by a gene mutation. The mutation causes change in the DNA resulting in the production of sickle shaped RBCs.

(ii) **Radioactive radiations** also alter the gene structure and their effects can be seen generation after generation. An atomic explosion which had occurred during World War-II 1945 in Japan (Hiroshima, Nagasaki),

had led to a number of deformities in the body of plants and animals which are still persisting.

PROGRESS CHECK

1. Who discovered for the first time the basic principles of genetics? .....
2. Give the common and scientific names of the organism on which Gregor Mendel had worked.
   Common name .....
   Scientific name .....
3. Define the following terms:
   (i) Monohybrid .....
   (ii) Dihybrid .....
   (iii) Filial generation .....
4. List any four traits in humans which you can easily study just by observing and making family charts.
   (i) ..... (ii) .....
   (iii) ..... (iv) .....

REMEMBER

Monohybrid cross : A cross between two pure breeding different varieties of organism taking the alternative traits of *one* single character. ***Example*** **:** A cross between pure tall (TT) and dwarf (tt) pea plants.

Monohybrid ratio : The kind of ratio obtained in the progeny in F_2 generation, by crossing for *two* different traits of a single character.

Dihybrid cross : A cross between two parents taking into consideration alternative traits of *two* different characters. ***Example*** **:** A cross between tall, purple flower bearing pea plant with dwarf and white flower bearing pea plant.

Dihybrid ratio : The kind of ratio obtained in the progeny in F_2 generation by crossing for contrasting the traits of *two* different characters.

Gene : Basic unit of heredity. It is a specific part (DNA segment) of a chromosome which controls the expression of a character. ***Example*** **:** The gene for free ear lobe in humans.

Allele : The alternative form of a gene occupying the same position on a chromosome and affecting the same characteristic but in two alternative ways. ***Example*** **:** The free and attached ear lobe alleles of the ear lobe character.

Dominant allele : The super-ruling allele of a gene. ***Example*** **:** The allele for tallness of pea plant.

Recessive allele : The suppressed allele of a gene. ***Example*** **:** The allele for dwarfness of pea plant.

Homozygous : The condition in which a pair of chromosomes carries similar alleles of a particular character. ***Example*** **:** A colour blind daughter will have both the X chromosomes with defective alleles.

Heterozygous : The condition in which a pair of chromosomes carries dissimilar alleles for a particular character. ***Example*** **:** A daughter (XX^c) from a normal homozygous mother (XX) for colour vision and a colour blind father (X^cY) has one normal and one defective allele.

Genotype : The combination of genes in an organism. ***Example*** **:** TT (homozygous dominant tall), Tt (heterozygous tall) and tt (homozygous recessive dwarf) pea plants.

Phenotype : The expressed character which is genetically controlled. ***Example*** **:** **Tall** pea plants (genotypes TT or Tt), **dwarf** pea plants (genotype tt).

Variation : Small differences between individuals due to inheritance.

Mutation : Sudden change in one or more genes or in the number and structure of chromosomes in the progeny, which normally may not have existed in the parents, grand parents or even great grandparents. ***Example*** **:** Albinism (total loss of skin pigment).

Homologous chromosomes : A pair of corresponding chromosomes of the same size and shape, one from each parent.

REVIEW QUESTIONS

A. MULTIPLE CHOICE TYPE

(Select the most appropriate option)

1. Which one of the following has the smallest number of chromosomes ?
   (a) Onion (b) Mouse
   (c) Monkey (d) *Ascaris*
2. Which one of the following is the phenotypic monohybrid ratio in F_2 generation ?
   (a) 3 : 1 (b) 1 : 2 : 1
   (c) 2 : 2 (d) 1 : 3

B. VERY SHORT ANSWER TYPE

1. **Match** the terms in column I with their explanations in column II.

Column I (Term)	Column II (Explanation)
a. Genetics	(i) Chromosomes similar in size and shape
b. Autosomes	(ii) The alternative forms of a gene
c. Recessive gene	(iii) Study of laws of inheritance of characters
d. Allele	(iv) A gene that can express only when in a similar pair
e. Homologous chromosomes	(v) Chromosomes other than the pair of sex chromosomes.

2. **Name** two animals which have nineteen pairs of chromosomes.
3. **Name** any two genetic diseases in humans.
4. Which one of the following genotypes is homozygous dominant and which one homozygous recessive in regard to tongue rolling :
   Rr, rr, RR.

C. SHORT ANSWER TYPE

1. **Differentiate** between :
   (a) genotype and phenotype.
   (b) character and trait.
   (c) monohybrid and dihybrid cross (phenotypic ratio).
2. Among lion, tiger and domestic cat, all the three have the same number of 38 chromosomes, yet they have different appearances. **How** do you account for such differences ?
3. **List** any three features of garden pea with their dominant and recessive traits.
4. **Explain** why generally only the male child suffers from colour blindness and not the female?
5. In a certain species of animals, black fur (B) is dominant over brown fur (b). **Show** the possible ratio of genotypes and phenotypes of the offspring of the pure breeding different coloured parents.

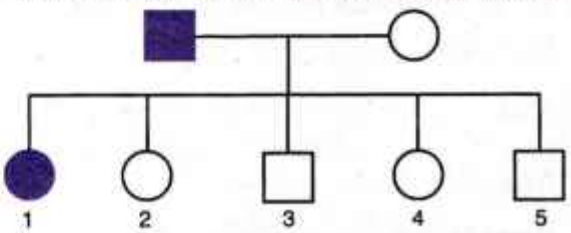
D. LONG ANSWER TYPE

1. **Explain** the following terms :
   (a) Heterozygous (b) Homozygous
   (c) Pedigree chart.
2. **State** the three Mendel's laws of inheritance.
3. Does the sex of the child depend on the father or it is just a matter of chance? **Discuss.**

E. STRUCTURED/APPLICATION AND SKILL TYPE

1. In a certain species of animals, black fur (B) is dominant over brown fur (b). **Predict** the genotype and phenotype of the offspring when both parents are 'Bb' or have heterozygous black fur.
2. Two pairs (A & B) of rabbits were crossed as given below:

   (a) Can you tell which coat colour (black or white) is dominant?.....
   (b) Is the coat colour sex-linked?.....
3. **Make a Punnett square** for finding out the proportion of different genotypes in the progeny of a genetic cross between
   (a) A **pure tall** (TT) pea plant with a **pure dwarf** (tt) pea plant.
   (b) Red flower variety of pea (RR) with white flower variety of pea (rr).
4. A family consists of two parents and their five children and the pedigree chart below shows the inheritance of the trait colour blindness in them.



   (a) Who is colour blind in the parents — the Father or the Mother ?

(b) How many daughters and how many sons have been born in the family ?

(c) What does the child 1 indicate about this trait ?

(d) On which chromosome is the gene of this trait located ?

(e) Name one other trait in humans which follows the similar pattern of inheritance.

"REBIRTH" ? AS "CLAIMED BY THE PARENTS"

As reported in newspapers, can we call it a case of "rebirth" of their son who had died exactly 11 months before, or is it simply **an excellent example of hereditary similarity**?

(Died May 3, 2005)

(Born April 12, 2006)

"HEBRA"

A cross between Horse & Zebra

HEBRA ? 'Eclyse', a zebra-horse crossbreed, in Schloss Holte, Germany. The father of 'Eclyse' was **a horse from Italy** and the mother was **a zebra from the Safari Park.**

Similarly —

Tigon	—	**Tiger × Lioness**
Zorse	—	**Zebra × Horse**
Biffalo	—	**Bison × Buffalo**
Zenkey	—	**Zebra × Donkey**
Liger	—	**Lion × Tigress**
Geep	—	**Goat × Sheep**
Pomato	—	**Potato × Tomato**

A case of one Black and one White twins born to a **mixed race darkish mother** and a **white father** (Reported in Times of India, 24th October, 2006).

- Both are twin sisters, one black and one white.
- Both are dizygotic (fraternal) twins produced from two fertilized eggs.
- One of the **genetic probabilities** leading to this type of inheritance could be as follows.

Skin colour, as we know today, is determined by 3 genes — let us suppose A, B and C.

Mother (Darkish) — A a, Bb, Cc

Father (Almost white) — aa, Bb, cc

Two of the several combinations of these genes through the egg and sperm could be as follows :

Darkish (Darker than the mother) — A a, BB, Cc

White — aa, bb, cc