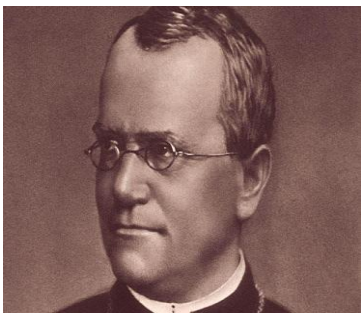
Genetics

Introduction to Genetics



Gregor Mendel(1822-1884)

- The father of genetics is **Gregor Mendel** (**1822-1884**), a late 19th century biologist and Austrian Monk, is known as the father of modern genetics.
- **William Bateson** (1861-1926), British biologist, a proponent of Mendel's work, coined the word *genetics* in 1905, derived from the Greek word *genesis*, meaning "origin". The word *genetics* is used to describe the study of inheritance.
- Mendel observed that organisms inherit traits/characters by way of discrete **"units of inheritance"** – the **genes**.
- **GENETICS** is the branch of biology that deals with heredity and genetic variations.
  Genetics is the study of how genes bring about characteristics, or traits, in living things and how those characteristics are inherited.
- **Heredity** is the transfer of genetically controlled characteristics such as hair color or flower color from one generation to the next in living organisms**.**
- **Inheritance** is the transmission of genetically controlled characteristics or qualities from parent to offspring.

Technical terms used in genetics

a) Chromosomes: Are thread-like structures found in the cell nucleus and they carry the hereditary factors. They are made up of proteins, nucleic acids (DNA and RNA) and genes.

b) Gene: Is the basic unit of inheritance found on the chromosome in the nucleus of a cell. A gene determines the presence or absence of a certain characteristics in an organism. Each gene carries one characteristic. Genes occur in pairs, e.g. TT, AB, OO, Tt and AA.

c) Allele: This is one member of a pair of genes, e.g. T and T are alleles of gene TT.

d) Homozygous: These are genes in which the alleles are identical, e.g. TT, tt, AA, BB.

e) Heterozygous: These are genes in which the alleles are opposite in nature, e.g. Tt, AO.

f) Phenotype: Refers to all characteristics of an organism which are visible or observable and results from the action of the genes, the environment or interaction of both genes and environment.

g) Genotype: This is the nature and arrangement of genes or alleles in an organism's cell. It is the genetic constitution or make up of an organism **e.g. AA, Aa.**

h) Dominant allele:
- An allele which affects the phenotype of a heterozygous organism just as much as when the organism is homozygous for this allele.

- Dominant alleles are always represented by capital letters e.g. T, A, B.

i) Recessive alleles:

- An allele which only affects the phenotype of an organism when the dominant allele is not present.
- An allele whose effect is not seen in the heterozygous organism due to the presence of a dominant allele.
- An allele which affects the phenotype of an organism only in homozygous state.
- Recessive alleles are always represented by small letters e.g. t, b, a, c.

j) Multiple alleles: Three or more different forms of alleles of a gene that determine a particular character, for example, the gene for blood type has three alleles: I_A*(A),* I_B *(B),* and *i (O).*

k) Co-dominance This is where both alleles affect the phenotype of an organism.

l) Cross: Is the interbreeding of two genetically different or similar individuals.

m) Pure breeding /true breeding: An organism which, when crossed with itself or others like itself, always produces offspring like itself.

n) Selfing: Is the crossing an organism with itself or with others like itself.

o) Testcross: Is a genetic procedure used to determine the genetic constitution of an organism by crossing it with another individual of known genetic makeup.

p) Hybridization is the process involving the crossing of two closely related organisms which differ in some way but produce offspring called hybrids with more desirable qualities than either of the parents.

q) Hybrid: Is an organism produced from a cross between two individuals with different genetic constituents.

r) Hybrid vigour Is the situation when the hybrid inherits only the desirable or good characters from the two unrelated parents.

s) Carrier: Is symptomless transmitter of gene or an individual possessing a gene for a particular genetic trait or disorder without being affected by it. In this case, heterozygous state is a carrier.

t) Backcross: Is the cross between an organism, especially a hybrid, with one of its parents or an individual genetically identical to that parent.

u) Parental generation (P) This is the original pure-breeding parent individuals.

v) First filial generation (F_1) The offspring of two pure breeding parents.

w) Second filial generation (F_2) The offspring resulting from selfing the first filial generation (F_1).

Mendel's laws of genetics

Mendel conducted similar experiments with the other pea plant traits. Over many years, he formulated several principles that are known today as Mendel's laws of genetics. His laws include the following:

1. **Mendel's law of dominance:** "*When an organism has two different alleles for a trait, one allele dominates".*
2. **Mendel's law of segregation:** *"A character of an organism is determined by alleles occurring in a pair. Of such a pair, only one allele is carried in a single gamete".*
3. **Mendel's law of independent assortment:** *"Each of the two alleles of one gene may combine randomly with either of the two alleles of another gene".*

Genetic (Mendelian) crosses

An advantage of genetics is that scientists can predict the probability of inherited traits in offspring by performing a genetic cross (*Mendelian cross*). To predict the possibility of an individual trait, the following steps must be observed:

1. A symbol is designated for each allele in the gene pair. The dominant allele is represented by a capital letter and the recessive allele by the corresponding lowercase letter.
2. Determine the phenotypes and genotypes of the parents and the genotype of the gametes and state theme clearly, starting phenotype followed by genotypes.
3. Indicate the action of mating between the phenotypes.
4. Indicate the action meiosis leading to formation of gametes by use of connecting lines.
5. Indicate the action of random fertilization by use of connecting lines or by use of a Punnett square. A *Punnett square* is a boxed figure used to determine the probability of genotypes and phenotypes in the offspring of a genetic cross. *This chart is called **Punnett Square** which was developed by **Reginald C. Punnett,** an English Geneticist.* Write down the possible combination of alleles.
6. From the line cross or Punnett square, the phenotype of each possible genotype and phenotypes can be determined.

Complete Dominance

- This is the inheritance where one of the two alleles in a pair suppresses the other in the heterozygous condition. E.g. a cross of homozygous tall plants and homozygous short plant, all the offspring are tall because the allele for tallness is dominant over the allele for shortness.

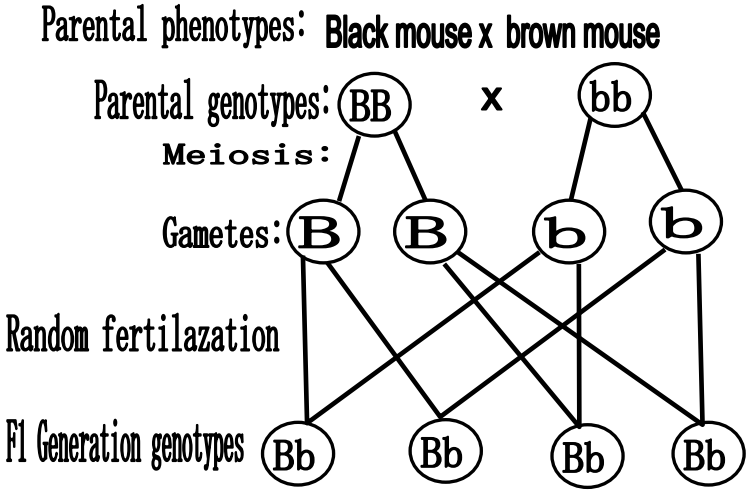
EXAMPLE 1

(a) If a pure breeding (homozygous) black mouse is mated with a pure breeding brown mouse. The gene for black fur is said to be dominant over that for brown fur; carryout a cross to

show the nature of the offspring of $\mathbf{F_1}$ generation. State the phenotype and genotype of the offspring of $\mathbf{F_1}$ generation.

Solution Let **B** represents allele for black fur
b represents allele for brown fur

Genetic cross **or** **Punnett Square**

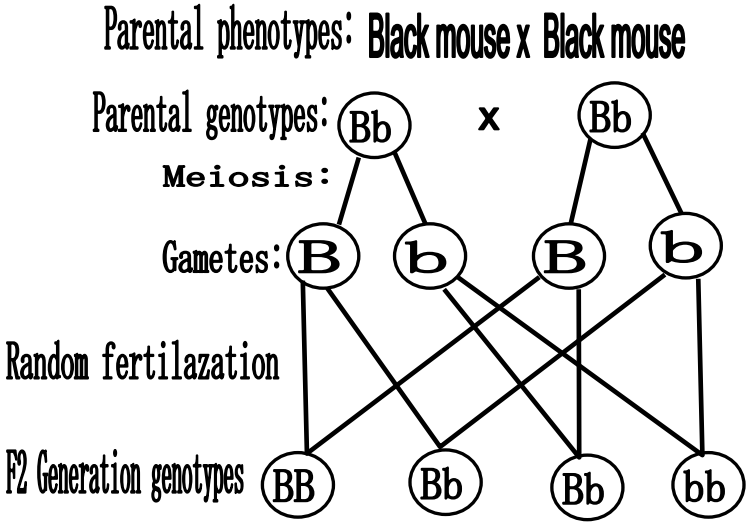


		Black mouse	
	Gametes	**B**	**B**
Brown mouse	**b**	Bb	Bb
	b	Bb	Bb

Phenotype: **all black**
Genotype: **all Bb**

(b) If the F_1 offspring mature and are mated amongst themselves to produce F_2 generation offspring. State the phenotypic ratio and the genotypic ratio of F_2 offspring.

Solution



Genotype: **BB : 2Bb : bb = 1:2:1 BB : Bb : bb**
Phenotype: **3 black : 1 brown = 3 : 1 black to brown**

EXAMPLE 2

A heterozygous tall plant was crossed with a homozygous short plant. Carryout the cross to show the offspring produced.

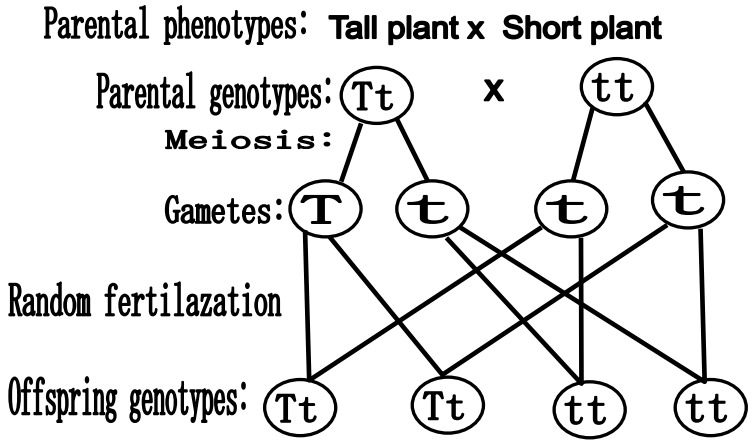
(i) What percentage of the offspring are heterozygous tall?

(ii) What is the probability that one of the plants is short?

Solution

Let **T** represents allele for tallness

t represents allele for shortness



(i) Percentage of heterozygous tall offspring $= \frac{\textit{Number of heterozygous tall plants}}{\textit{Total number of offspring}} \times 100$

$= \frac{2}{4} \times 100$

$= 50\%$

(ii) $\textit{Probability of one plant being short} = \frac{\textit{Number of events}}{\textit{Sample space}}$

$= \frac{1}{2}$

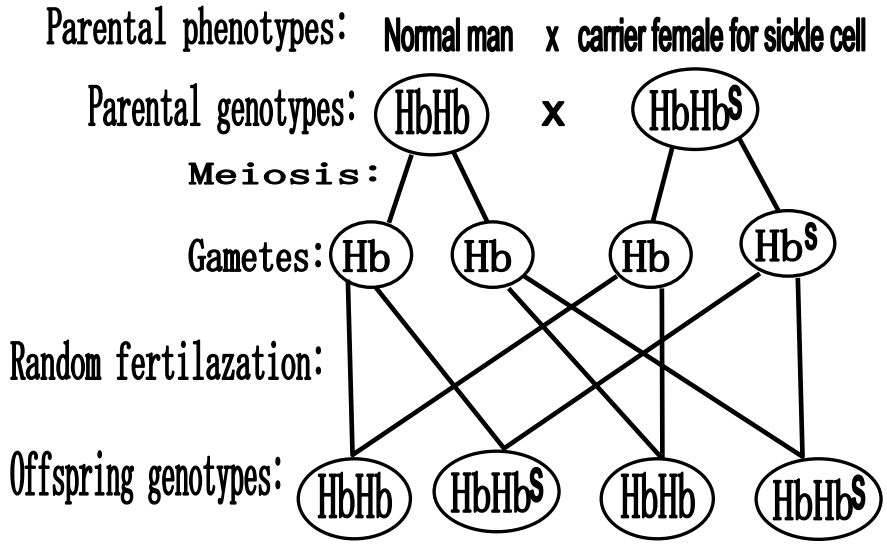
EXAMPLE 3

The gene for normal production of haemoglobin is dominant to the mutant gene which causes sickle cell anaemia. If the female who is heterozygous for the sickle cell anaemia marries a normal man, illustrate, using suitable symbols carry out a genetic cross to determine the genotype and phenotype of the offspring.

Solution

Let **Hb** represents allele for normal production of haemoglobin

Hb^s represents allele for sickle cell anaemia.



- **GENOTYPE:**

2 HbHb and
2 HbHb^{S}

- **PHENOTYPE:**

All normal

ACTIVITY 12.2

1. Two heterozygous tall plants were crossed. Using appropriate genetic symbols carryout the genetic cross.
   (a) Determine the genotypic ratio and phenotypic ratio.
   (b) What percentage of the offspring are:
      (i) Homozygous short plants
      (ii) Heterozygous tall plants
      (iii) Homozygous tall plants

2. Albinism is a condition where melanin fails to develop in the skin and is caused by a recessive alleles.
   (a) Using appropriate genetic symbols, carryout the genetic cross to determine the possible offspring genotypes and phenotypes when a phenotypically normal but carrier male marries an albino female.
   (b) (i) What is the probability that their first child is a carrier?
      (ii) What is the probability that their first two children are albinos?

INCOMPLETE DOMINANCE

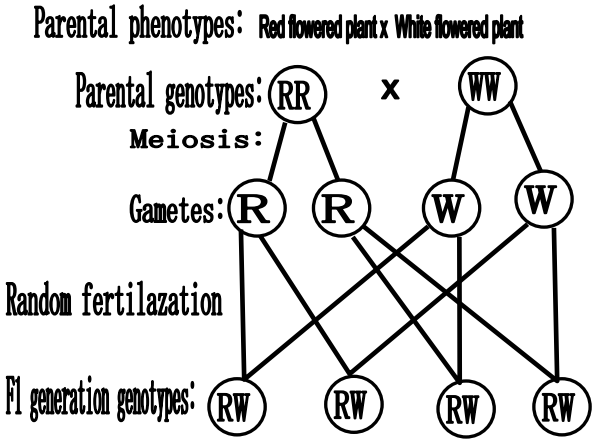
- This is the type of inheritance where one allele does not completely mask or dominate the action of the other and neither allele has dominant control over the traits but an intermediate character/trait is produced.
- In this type of inheritance, the phenotype results from the partial influence of both alleles.
- Examples of incomplete dominance include:
  - ✓ In certain plants, a Cross between red and white–flowered parent plants produce pink–lowered plants. The red and white genes are blended to produce pink.
  - ✓ In certain plants, a cross of rounded–seeded and long–seeded plants produce oval-seeded.

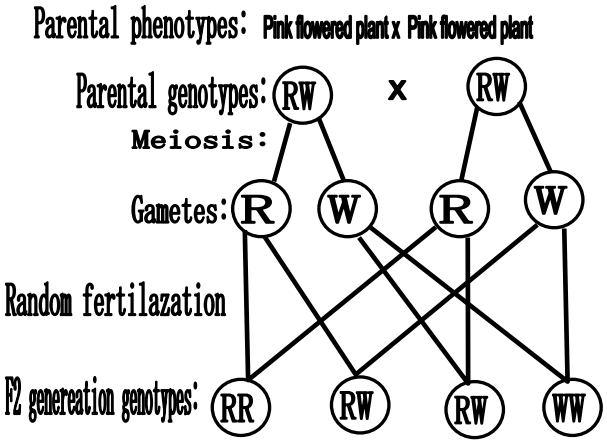
EXAMPLE 4

A pure breeding red-floweredplant was crossed with a pure breeding white-flowered plant and all the resulting F_1 generation had pink flowers.

(a) What percentage of the F_2 plants would have red flowers if the F_1 plants were self pollinated?

Solution Let **R** represents allele for red flower
W represents allele for white flower





Phenotype: All Pink
Genotype: All RW

Phenotype: 1 Red : 2 Pink : 1 white
Genotype: 1 RR : 2 RW : 1 WW

Percentage of Red flowered plants in F_2 $= \frac{Number\ of\ red\ flowered\ plants}{Total\ number\ of\ offspring} \times 100$

$= \frac{1}{4} \times 100 = 25\%$

(b) State the phenotypic and genotypic ratio of the offspring, if pink flowered plants were crosses with red flowered plants.

CO-DOMINANCE

- This is a type of inheritance where each allele/gene, in heterozygous state, has equal effect in making the character of the offspring.
- Both alleles are independently and equally expressed.

- Examples of co-dominance include:
  - ✓ The genes for A and B blood groups are co-dominant and give rise to the AB blood group if they are both inherited. Blood groups is a good example **multiple alleles:**
    - A represents allele for presence of antigen A on red blood cells;
    - B represents allele for presence of antigen B on red blood cells;
    - O represents allele for absence of antigen on red blood cells

N.B Allele A and allele B are dominant and allele O is recessive;

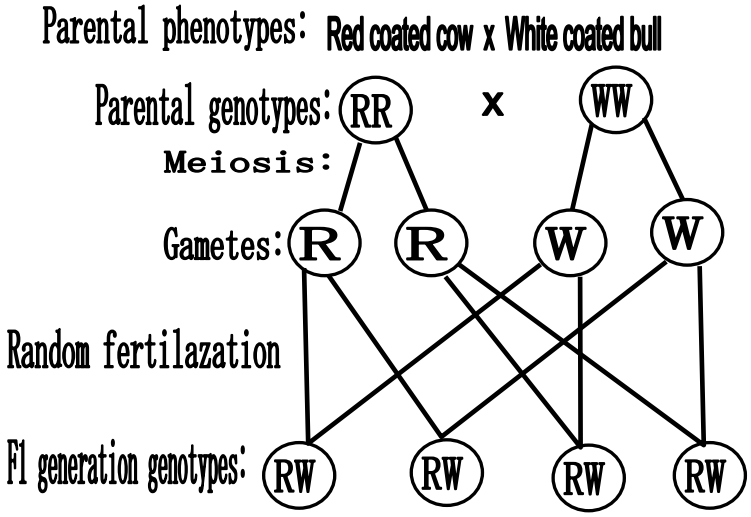
- ✓ When the red fur/coat cow is crossed with a white fur bull, the offspring have coats with a mix of red and white fur/hairs equally and independently expressed; and this is being **Roan** coloured fur. This means that the gene for red coat is co-dominant to the gene for white coat.

EXAMPLE 1

If a certain breed of red cows mate with white bull, the coats of the calves carry both red and white hairs, giving a colour called red roan.

(a) Carryout a cross to give F1 generation offspring.

Solution Let **R** represents allele for red fur
W represents allele for white fur



Genotype: <u>All RW</u>

Phenotype: All red roan

(b) If F1 generation offspring are mated amongst themselves to give F2 generation. Carryout a cross and determine the phenotypic ratio and genotypic ratio.

(c) What is the phenotype and genotype of the offspring if a white bull is mated with a roan cow?

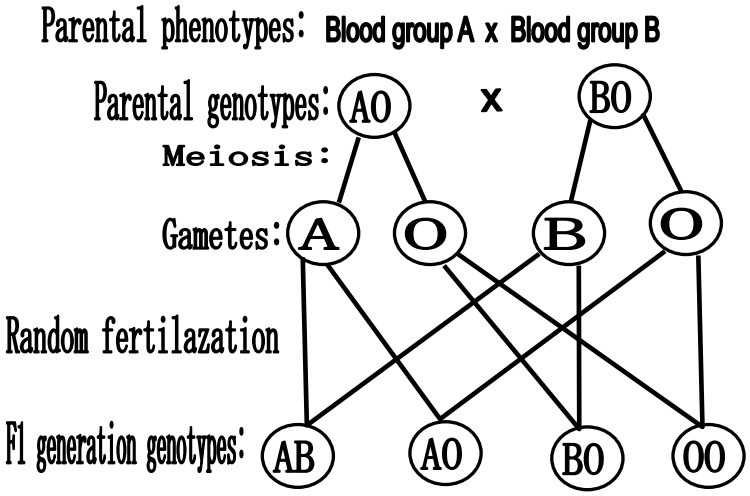
EXAMPLE 2

A man of blood group A heterozygous married a woman of blood group B heterozygous. Carryout a cross and determine the phenotype and genotype of the offspring.

Solution

Let

- **A (I_A)** represents allele for antigen A on red blood cells – *dominant allele*
- B (I_B) represents allele for antigen B on red blood cells – *dominant allele*
- **O (i) or Io** represents allele for absence of antigen on red blood cells – *recessive allele*



Genotype: **1AB, 1AO, 1BO and 1 OO**

Phenotype: **1 blood group AB, 1 blood group A, 1 blood group B and 1 blood group O**

ACTIVITY 3

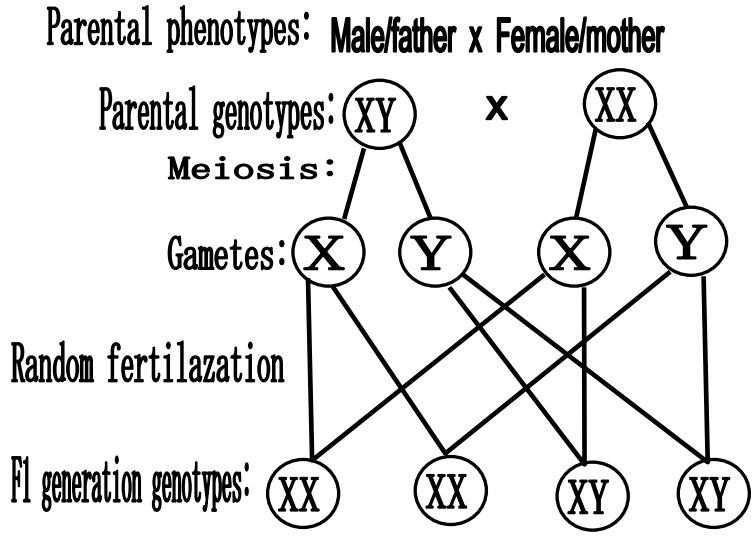
1. A wife is heterozygous for blood group A and the husband has blood O.
   (a) Carryout the genetic cross and determine phenotype and genotype of the offspring
   (b) Determine the probability of:
      (i) One child having blood group O
      (ii) One child having blood group A
      (iii) One child having blood group AB

SEX DETERMINATION IN MAN

Thomas Hunt Morgan

- ★ In **1905** American biologists **Edmund B. Wilson** and **Nettie Stevens** independently discovered and identified the sex chromosomes.
- ★ In **1907**, American biologist and geneticist, **Thomas Hunt Morgan(1866-1945)**, began his own study of chromosomes and genetics in vinegar (fruit) flies, *Drosophila melanogaster*.
- ★ Morgan rapidly discovered something peculiar about the four pairs of chromosomes that each drosophila had.
- ★ In females, the chromosomes of one pair were of the same shape, **rod shaped**, and he called the rod shaped chromosome as **X chromosome**

- ★ But in males, the chromosomes of one pair were not of the same shape: one was **rod shaped** and the other was **hook-shaped**. The hooked chromosome, Morgan called it **Y chromosome**.
- ★ In humanand in fruit fly *Drosophila*, the genotype of the female is **XX,**and the sex is called **homogametic sex;** and that of the male is **XY**and the sex is called **heterogametic sex.**
- ★ However, in humans, the Y chromosome is small/short and hooked and the X chromosome is longer than Y and rod shaped.
- ★ In the case of birds, moths and butterfly the sex genotypes are reversed: the male are XX and the females are XY. In some insects such as the grasshopper, the Y chromosome in male may be absent entirely and so the male has the genotype XO.
- ★ In human body cells there are 46 chromosomes which exist in 23 pairs of homologous chromosomes. Of the 23 pairs one pair is known as sex chromosomes which carry genes determining sex and are called**sex chromosomes**. The remaining 22 pairs of chromosomes are called **autosomes.**
- ★ **A** female child results from fusion of the sperm cell carrying X-chromosome with an ovum carrying X-chromosome**. A** male child results from fusion of the sperm cell carrying Y-chromosome with the ovum carrying X-chromosome. The chance of **a** baby girl or a baby boy to be born is $\frac{1}{2}$as shown below:



GENOTYPE: **2 XX and 2 XY**
PHENOTYPE: **2 girls and 2 boys**

SEX LINKED CHARACTERS/TRAITS

- **Linkage**is the proximity of two or more genes on a chromosome, which tends to cause them to be inherited together.

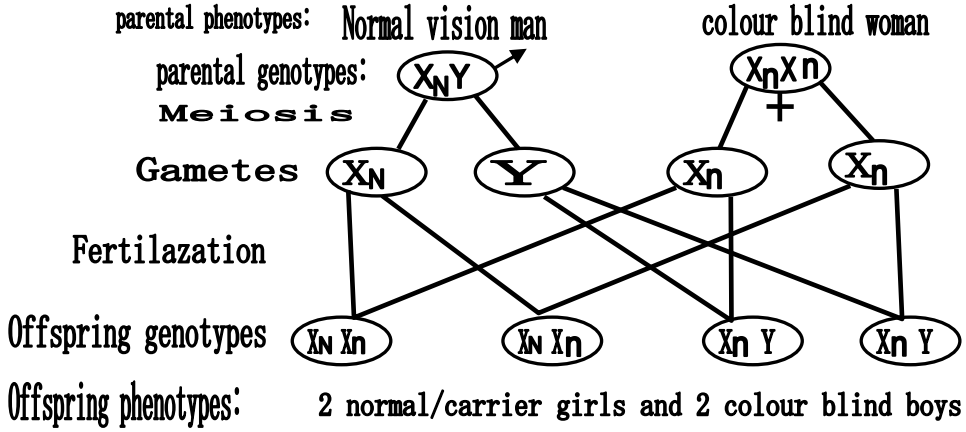
- **Sex-limited traits** refer to the genetically inherited traits or conditions that appear in one sex only, although the genes themselves may be found in either sex.

- **Sex-linked trait** is one whose genes are located on a sex chromosome, typically on the X chromosome.
- Sex-linked traits are common in males because the male has XY sex chromosomes and genes are carried by the X chromosome and Y does not carry genes (or carries very few genes).Therefore, characteristics determined by genes carried on the X chromosome appear in males even if they are recessive.
- The sex-linked genes occur in recessive form and develop as a result of mutation of the normal genes. Most of the sex-linked characters which arise due to sex-linked genes are unfavorable – causing diseases.
- Examples of sex-linked traits include:
  - ✓ **Haemophilia,**
  - ✓ **Duchenne muscular dystrophy**,
  - ✓ **Red-green color blindness in humans** and
  - ✓ **Eye color in fruit flies**:white-eyed male mutant and a wild-type (red-eyed).

EXAMPLE 7

Colour blindness is sex-linked. If a colour blind woman marries a normal man; using suitable symbols, carryout a cross and determine the genotype and phenotype of the offspring.

Solution Let **N** represents allele for normal vision
n represents allele for colour blindness



GENOTYPE: **$2X_NX_n$ and 2 X_nY**

ACTIVITY 12.4

1. If a carrier woman for colour blindness marries a normal man, work out the genotypes and phenotypes of the children.
2. Inheritance of haemophilia is sex-linked. Workout the genotypes and phenotypes of the children, if a normal man married a carrier female for haemophilia.

GENETIC VARIATION

- **Variation** refers to the differences amongst organisms of the same species due to the differences in their genes they inherit and environment they survive in.
- Characteristics such as height, weight, shape of face, knowledge, skills, body scars differ from one person to the next.

Causes/Sources of Variation

- Variation is caused by changes in either the environment or genetic make up of organisms.
- Variations brought about by changes in the environment are not passed from one generation to the next.
- Variations due to the differences in the genes are inherited from their parents.

a. Environmental factors

1. **Diet/nutrients:** organisms with adequate nutrients grow well better than those with little nutrients.
2. **Altitude:** plants at low altitude will grow well and those at high altitude are smaller.
3. **Light intensity**: plants that receive adequate light intensity grow well than those that receive little light intensity.
4. Pathogens and diseases
5. Temperature: the sex of some animals is determined by temperature, e.g. when the temperatures are high the crocodile is male and when the temperatures are low the crocodile is a female.
6. Social function: effect of other organisms and predation.
7. Age
8. Wind
9. Water availability
10. pH (acidity, neutral or alkalinity.
11. Soil type.

b. <u>Genetic factors:</u>

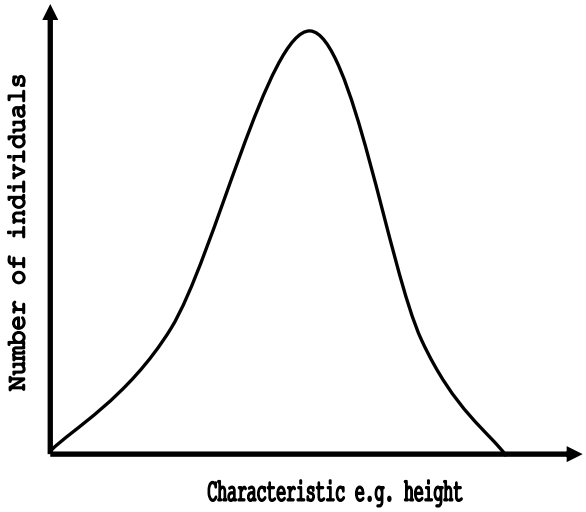
1. Crossing over between homologous chromosomes during prophase I of meiosis.
2. Random fusion of gametes during fertilization.
3. Incomplete dominance in organisms.
4. The nonappearance of a characteristic determined by one gene because it has been suppressed or masked by the activity of another gene – **epistasis**.
5. A random change in a gene or chromosome resulting in a new trait or characteristic that can be inherited – **mutation**.

Types of Variations

1. Continuous variation

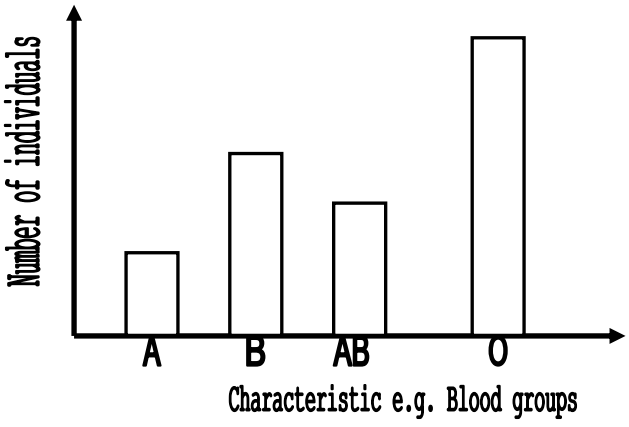
- This is the variation whereby there are no clear-cut and sharp differences amongst organisms of the same species over a given character.

- When a given character is considered, organisms tend to have many intermediate grades of the same character e.g.
  - ✓ height,
  - ✓ weight,
  - ✓ intelligence,
  - ✓ ear length,
  - ✓ fertility,
  - ✓ Cephalic index
  - ✓ yield of milk,
  - ✓ skin colour,
  - ✓ Number of grains on a maize cob.
- Continuous variation can be measured and a mean, mode median obtained.
- Continuous variations are influenced by several genes (polygene) but the features are also influenced by the environment. Thus most continuous variations result from the interaction of the genotype with the environment.
- Continuous variations can be measured and they are quantitative. When we compare many organisms referring to a continuous variation character, a normal distribution curve is obtained.



2. Discontinuous variation

- This is a type of variation which shows clear-cut and sharp differences amongst organisms of the same species over a given character, e.g.
  - ✓ Blood groups: A, B, AB OR O
  - ✓ Sex: male or female
  - ✓ Tongue rolling: able to roll or unable to roll.
  - ✓ Ear lobes: either present or absent
  - ✓ Sickle celled anemia
  - ✓ Haemophilia
  - ✓ Rhesus factor: either positive or negative
- Each discontinuous variation character is controlled by a single gene and the features are clearly genetically determined. That is, they cannot be altered during the lifetime of the individual.
- Discontinuous variations cannot be measured but they are qualitative and no normal distribution curve is obtained.



MUTATION

- Mutation is a random change in a gene or chromosome resulting in a new trait or characteristic that can be inherited.
- Mutation can be a source of beneficial genetic variation, or it can be neutral or harmful in effect.
- Most mutations occur in somatic (body) cells and are not passed from generation to the next.
- Only mutations that occur in the formation of gametes can be inherited.

Types of Mutation

1. **Gene or point mutation:** This occurs at a singles locus on a chromosome. It involves change in the structure of DNA. Examples of gene mutation include:
    - *Sickle cell anaemia*– Abnormal form of hemoglobin and red blood cells are sickle shaped, and soon die causing anaemia. Such individuals die before reproductive age.
    - *Haemophilia* –Failure/ delayed blood clotting due to lack of clotting factor VIII.
    - *Albinism*– Failure of the colour pigment, Melanin, to develop in the skin, hair and eyes. The skin of the albino is very transparent and is quickly damaged by sunlight.
    - *Huntington's disease* (HD), or Huntington's chorea – Hereditary disorder of the nervous system characterized by involuntary twitching movements of the arms, legs, face, and body; and progressive mental deterioration.
    - *Cystic fibrosis* – a hereditary disease starting in infancy that affects various glands and results in secretion of thick mucus that blocks internal passages, including those of the lungs, causing respiratory infections. The pancreas is also affected, resulting in a deficiency of digestive enzymes and impaired nutrition.
2. **Chromosomal mutation:** This affects more than one locus. This may involve changes to the whole sets of chromosomes or change to individual chromosome. Examples include:
    - *Down's syndrome (mongolism):*is Trisomy 21, a defect in which an extra, third copy of chromosome 21 is present in every cell in the body. Consequences: lethal/aborted; flat, broad face; protruding tongue; low intelligence quotient; and short life span.
    - *Klinefelter's syndrome* – there is extra sex chromosome e.g. XXY, XXXY, or XXXXY – phenotypically male but have small testes and no sperms in ejaculation; abnormal breast development and body generally resembles that of females.
    - *Turner's syndrome* – a genetic disorder affecting women in which only one X chromosome per cell is present(XO), instead of two (XX), resulting in underdeveloped ovaries and underdevelopment of the womb, vagina, and breasts.

Causes of Mutations

- ✦ Mutations occur naturally at a low rate. However, chemicals or energy sources called mutagen or mutagenic agents increase the rate of mutation.
- ✦ A **mutagen** is a chemical or energy that induces mutation. These include:

1. High energy (ionizing) radiations radiation, e.g. x-rays, γ(gamma) rays, and ultra-violet rays from the sun.
2. High fluctuations in temperature.
3. Chemicals like nitrous acid, mustard gas, colchicines, asbestos, cigarette, benzene, vinyl chlorides and coal tar.
4. High energy particles such as β-particles, α-particles and neutrons.
5. Viruses and microorganisms

Importance of Mutation

- ✓ Mutations have harmful effects that cause diseases and they are often lethal.
- ✓ Mutations are essential for evolution since they are the ultimate source of genetic variation.
- ✓ Development of new viral strains.
- ✓ Development of new multi-resistant bacteria super bugs.
- ✓ Development of cancer.