BIOLOGY

Chapter 4: Principles of Inheritance and Variation

Principles Of Inheritance and Variation

Introduction:

Genetics is the study of principles and mechanism of heredity and variation. Gregor Johann Mendel is known as 'father of Genetics'. important attributes to the reproductive health of a society.

Inheritance:

Inheritance is the process by which characters are passed on from parent to progeny. It is the basis of heredity.

Variation:

Variation is the degree by which progeny differ from their parents. Variation may be in terms of morphology, physiology, cytology and behavioristic traits of individual belonging to same species.

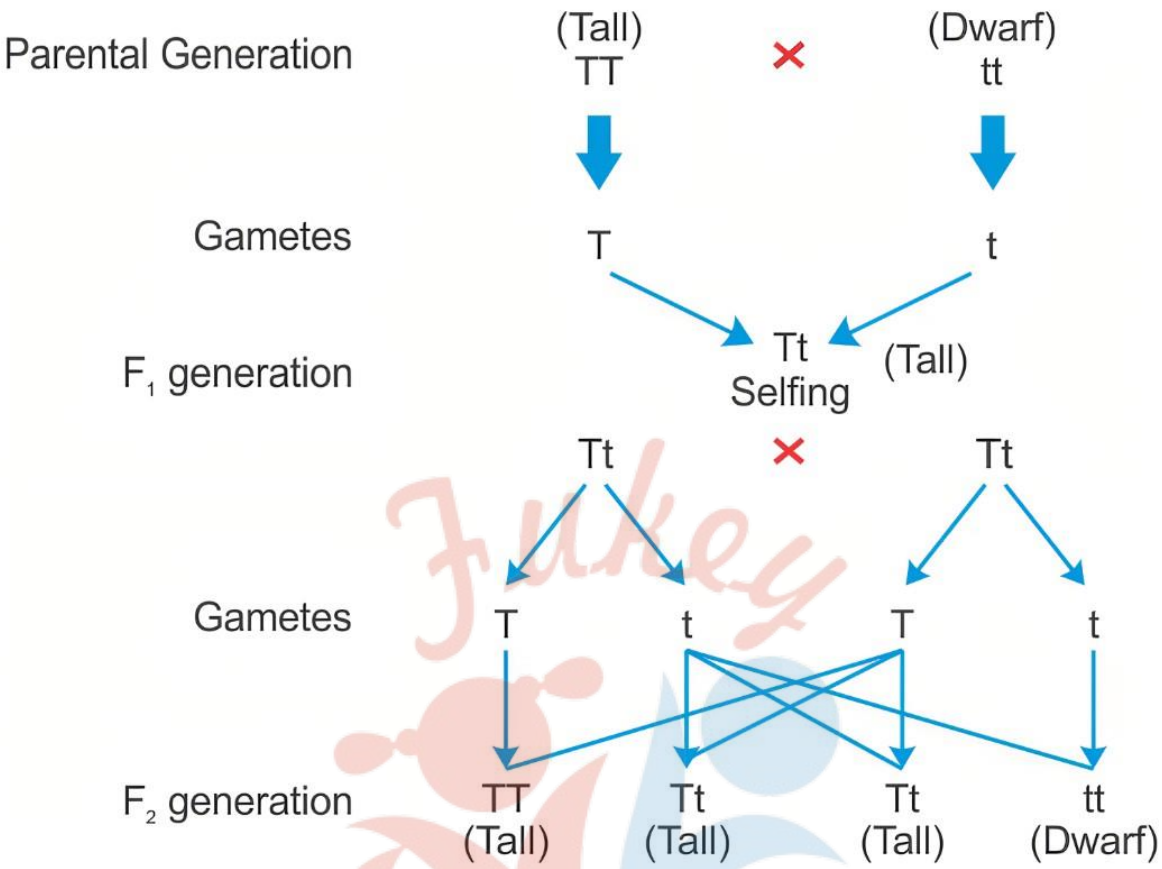
Variation arise due to:

- Reshuffling of gene/ chromosomes.
- Crossing over or recombination
- Mutation and effect of environment.

Inheritance of one gene (Monohybrid cross):

Mendel crossed tall and dwarf pea plant and collected all the seeds obtained from this cross. He grew all the seeds to generate plants of first hybrid generation called F_1 generation. He observed that all the plants are tall. Similar observation was also found in other pair of traits.

Mendel self-pollinated the F_1 plants and found that in F_2 generation some plants are also dwarf. The proportion of dwarf plants is 1/4th and tall plants of 3/4th.



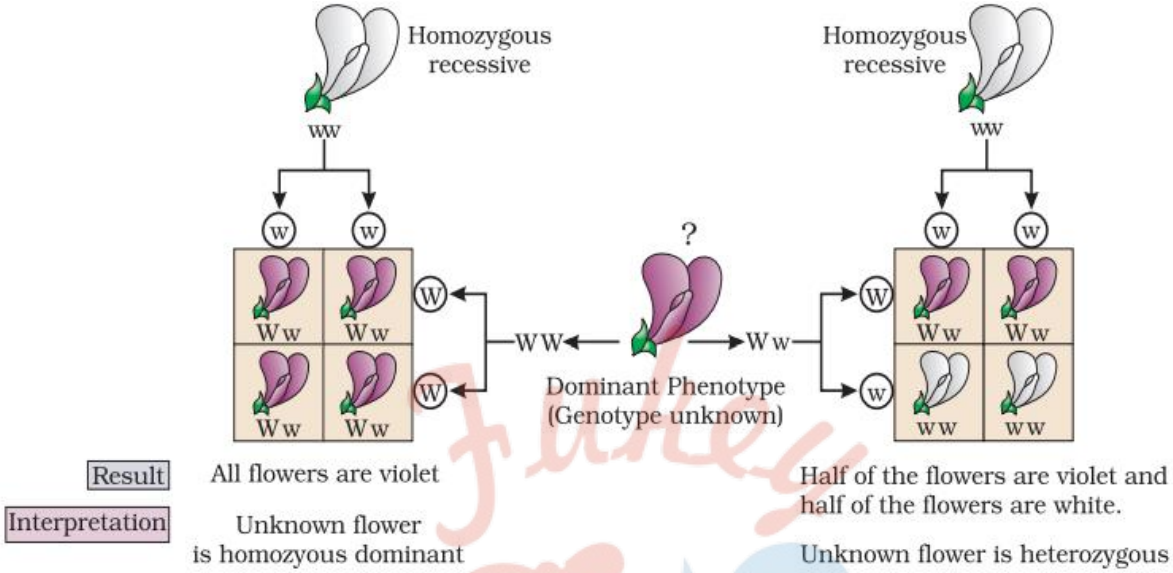
- Mendel called the 'factors' that passes through gametes from one generation to next generation. Now a day it is called as genes (unit of inheritance).
- Genes that code for a pair of contrasting traits are known as alleles.
- Alphabetical symbols are used to represent each gene, capital letter (TT) for gene expressed in F_1 generation and small letter (tt) for other gene.
- Mendel also proposed that in true breeding tall and dwarf variety allelic pair of genes for height is homozygous (TT or tt). TT, Tt or tt are called genotype and tall and dwarf are called phenotype.
- The hybrids which contain alleles which express contrasting traits are called heterozygous (Tt).
- The monohybrid ratio of F_2 hybrid is 3 : 1 (phenotypic) and 1 : 2 : 1 (genotypic).

Dominance: When a factor (allele) expresses itself in the presence or absence of its dominant factor called dominance. It forms a complete functional enzyme that perfectly express it.

Recessive: It can only express itself in the absence of or its recessive factor allele. It forms a incomplete defective enzyme which fails to express itself when present with its dominant allele, i.e., in heterozygous condition.

Test cross:

Test cross is the cross between an individual with dominant trait and a recessive organism in order to know whether the dominant trait is homozygous or heterozygous.



Mendel's Experiment:

Gregor Mendel, after performing his experiments on pea plants, discovered the fundamental laws of inheritance. He proposed three laws of inheritance which we are studying to date. He has chosen pea plants having seven opposite traits of particular characters and conducted his experiment on 14 true-breeding pea plant varieties.

Mendel's Laws:

There were 3 laws that were proposed by Mendel

Law of Dominance: It is explained in this law that all of the traits, or the characters are controlled by the unit called the factors. These factors are found to be in pairs and are called alleles. If they occur in the same pair they are called homozygous, they can be either dominant or recessive and if the alleles occur in a different pair then it is called heterozygous, It will always be dominant. "For example Allele for tallness is dominant over the allele for dwarfism".

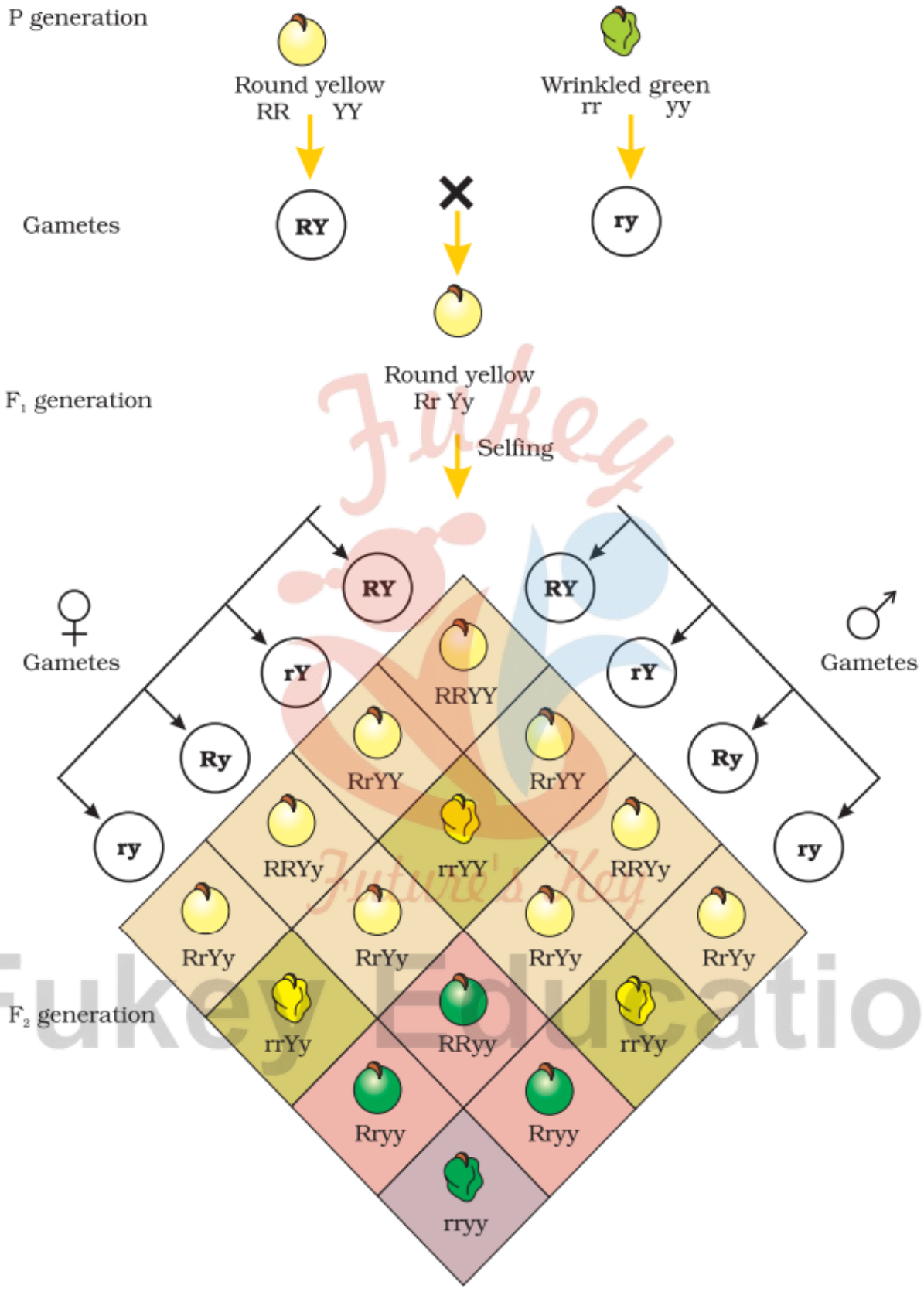
Law of Segregation of Genes: Law of segregation is based on the fact that alleles do not show any blending and that both the characters are recovered as such in the second filial generation though one of these is not seen in the first generation. The segregation of factors or a pair of alleles occurs in such a manner that the gamete receives only one of the two factors from each other. Examples of the law of segregation of alleles. In this R is dominant over r.

Law of Independent Assortment: It states that pairs of traits in the parental generation sort independently from one another when passing from one generation to the next. It is

explained with the help of a dihybrid cross.

Inheritance of Two Genes (Dihybrid Cross):

The inheritance of two genes requires two characters of the same trait. This can be observed with the help of a dihybrid cross. Mendel has chosen two traits that involve the color and the shape of the seed to explain the inheritance of two genes. Y represents the dominant yellow color seed color, y represents a recessive green color while R represents the round shape of the seed, and r represents the wrinkled shape of the seed. The genotype of the parents can then be written as RRYY and rryy. The gametes RY and ry will unite after fertilization and will produce the F1 hybrid RrYy. The dihybrid cross is also useful in the study of the Law of Independent Assortment. After the self-pollination of the F_1 hybrid, the F_2 ratio was found to be 9 : 3 : 3 : 1.

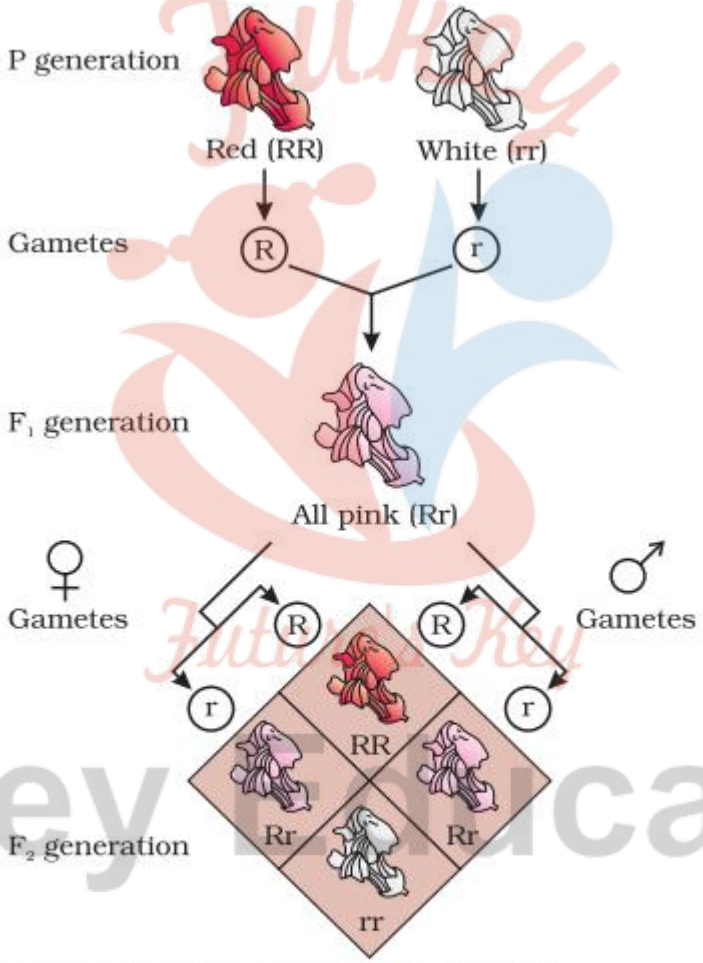


Phenotypic ratio : round yellow : round green : wrinkled yellow : wrinkled green
9 3 3 1

Incomplete Dominance:

Incomplete dominance is a type of inheritance in which one allele for a specific trait is not completely dominant over the other allele i.e. neither allele is dominant over the other in heterozygous organisms. This results in a combined phenotype. Incomplete dominance is also called mosaic or partial dominance. Here new phenotypic characters are expressed entirely.

Mirabilis Jalapa, the marvel of Peru commonly called a 4 o'clock plant. It is a very good example of incomplete dominance because of its Inheritance of flower color. It is shown in the figure given below where red flowers (dominant) were crossed with white flowers (recessive), the F1 generation contains flowers that are pink in color (intermediate). The phenotypic and the genotypic ratio observed will be the same, that is 1 : 2 : 1.



Phenotypic ratio : red : pink : white
1 : 2 : 1

Genotypic ratio : RR : Rr : rr
1 : 2 : 1

Co-dominance:

It is the phenomenon of two alleles lacking dominance-recessive relationship and both expressing themselves in the organism.

Human beings, ABO blood grouping are controlled by gene I. The gene has three alleles I^A, I^B

and i. Any person contains any two of three allele I^A, I^B are dominant over i.

The plasma membrane of the red blood cells has sugar polymers that protrude from its surface and the kind of sugar is controlled by the gene.

When I^A and I^B are present together, both express their own types of sugars because of co-dominance.

Difference between Incomplete Dominance and Co-Dominance:

Incomplete Dominance	Co-Dominance
Effect of one of the two alleles is more conspicuous.	Effect of both the alleles are equally conspicuous.
It produces a mixture of the expression of two alleles.	There is no mixing of the effect of the two alleles.
The F1 does not resemble either of the parents.	The F1 resembles both the parents.
E.g.: Flower colour in dog flower.	E.g.: ABO blood grouping in humans,

Allele from Parent 1	Allele from Parent 2	Genotype of offspring	Blood types of offspring
I^A	I^A	$I^A I^A$	A
I^A	I^B	$I^A I^B$	AB
I^A	i	$I^A i$	A
I^B	I^A	$I^A I^B$	AB
I^B	I^B	$I^B I^B$	B
I^B	i	$I^B i$	B
i	i	$i\,i$	O

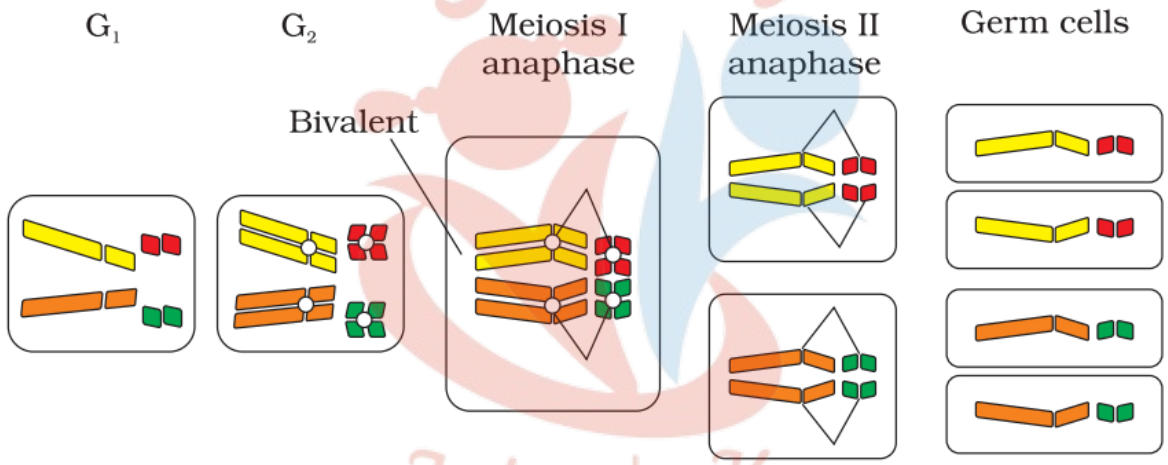
Multiple Allelism or Codominance:

The condition in which three or more alternative forms of alleles present for a single gene on the same chromosome is known as Multiple Allelism and the alleles are known as multiple alleles. For example, Multiple allelism is better understand with the help of the ABO blood group system in humans. The inheritance of the ABO blood group is a gene I (in which I represent isohemagglutinin) that remains in the 3 allelic expressions: IA, IB, and i which are codominant in humans. An individual can possess any two of these alleles. Gene IA is responsible for blood group A and codes for glycoprotein A while gene IB is responsible for blood group B and codes for glycoprotein B.

The gene 'i' does not produce any glycoprotein and so the person who will be having these two alleles together in a homozygous condition will have O group blood. The genes IA and IB are dominant over 'i' but alleles IA and IB are dominant equally and produce both the glycoproteins A and B simultaneously and results in the blood group AB. Such alleles are known as co-dominant alleles.

Chromosomal Theory of Inheritance:

- Chromosome as well as gene both occurs in pair. The two alleles of a gene pair are located on the same locus on homologous chromosomes.
- Sutton and Boveri argued that the pairing and separation of a pair of chromosomes would lead to segregation of a pair of factors (gene) they carried.
- Sutton united the knowledge of chromosomal segregation with mendelian principles and called it the chromosomal theory of inheritance.



Linkage and Recombination:

When two genes in a Dihybrid cross were situated on same chromosome, the proportion of parental gene combination was much higher than the non-parental type. Morgan attributed this due to the physical association or the linkage of the two genes and coined the linkage to describe the physical association of genes on same chromosome.

The generation of non-parental gene combination during Dihybrid cross is called recombination. When genes are located on same chromosome, they are tightly linked and show very low recombination.

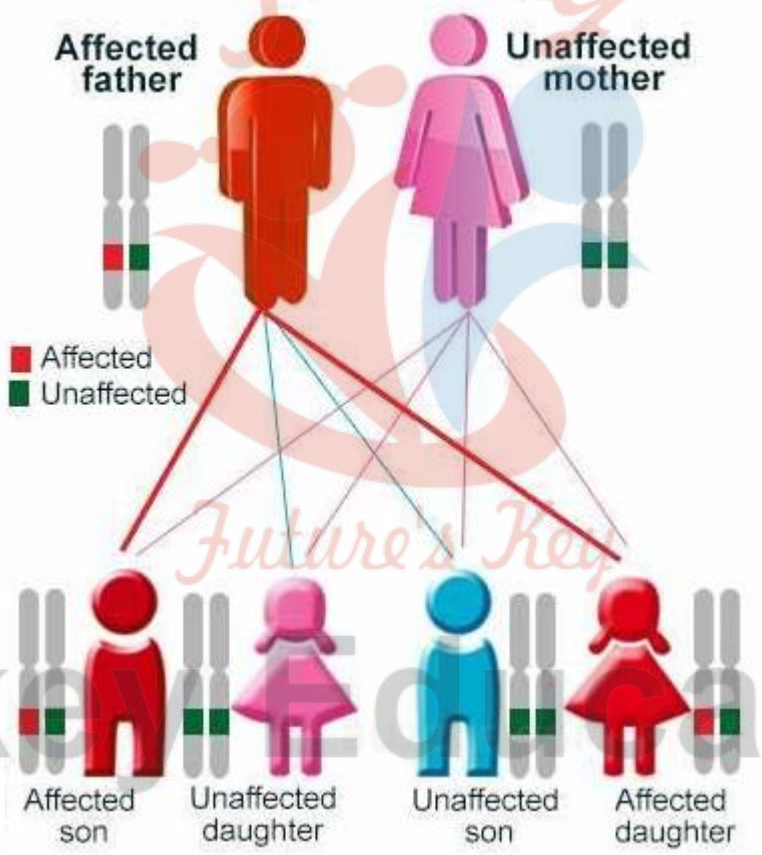
Crossing over:

It leads to separation of linked genes, It involves exchange of segments between non-sister chromatics of homologous chromosomes. The frequency of crossing over can never exceed 50%. It increases variability by forming new gene combinations.

Sex Determination:

Sex determination is the process where the gender of the child can be revealed. Sex chromosomes are responsible for the determination of the sex of a child. In the case of humans, females have XX types of chromosomes while males have one X and one Y type of chromosomes. Thus, when the egg (female gamete) formed will be having identical X-chromosome each but male sperms (male gametes) are not identical as they have one X-chromosome and one Y-chromosome. So it's a matter of chance that which sperm fuses with the egg (X or Y). Thus, the females are said to be homogametic (same type of chromosomes) while males are said to be heterogametic (different types of chromosomes).

In the case of insects, the mechanism of sex determination is of XO type. Here the eggs consist of the X chromosomes while the sperms may have one or none X chromosomes. Thus, the males are said to be homogametic (same type of chromosomes) while females are said to be heterogametic (different types of chromosomes).



Mutation: Mutation is a phenomenon which results in alternation of DNA sequence and consequently results in the change in the genotype and phenotype of an organism. The mutations that arise due to due to change in single base pair of DNA are called point mutation e.g., Sickle cell anaemia.

Pedigree Analysis: The analysis of traits in several of generation of a family is called the pedigree analysis. The inheritance of a particular trait is represented in family tree over several generations. It is used to trace the inheritance of particular trait, abnormality and disease.

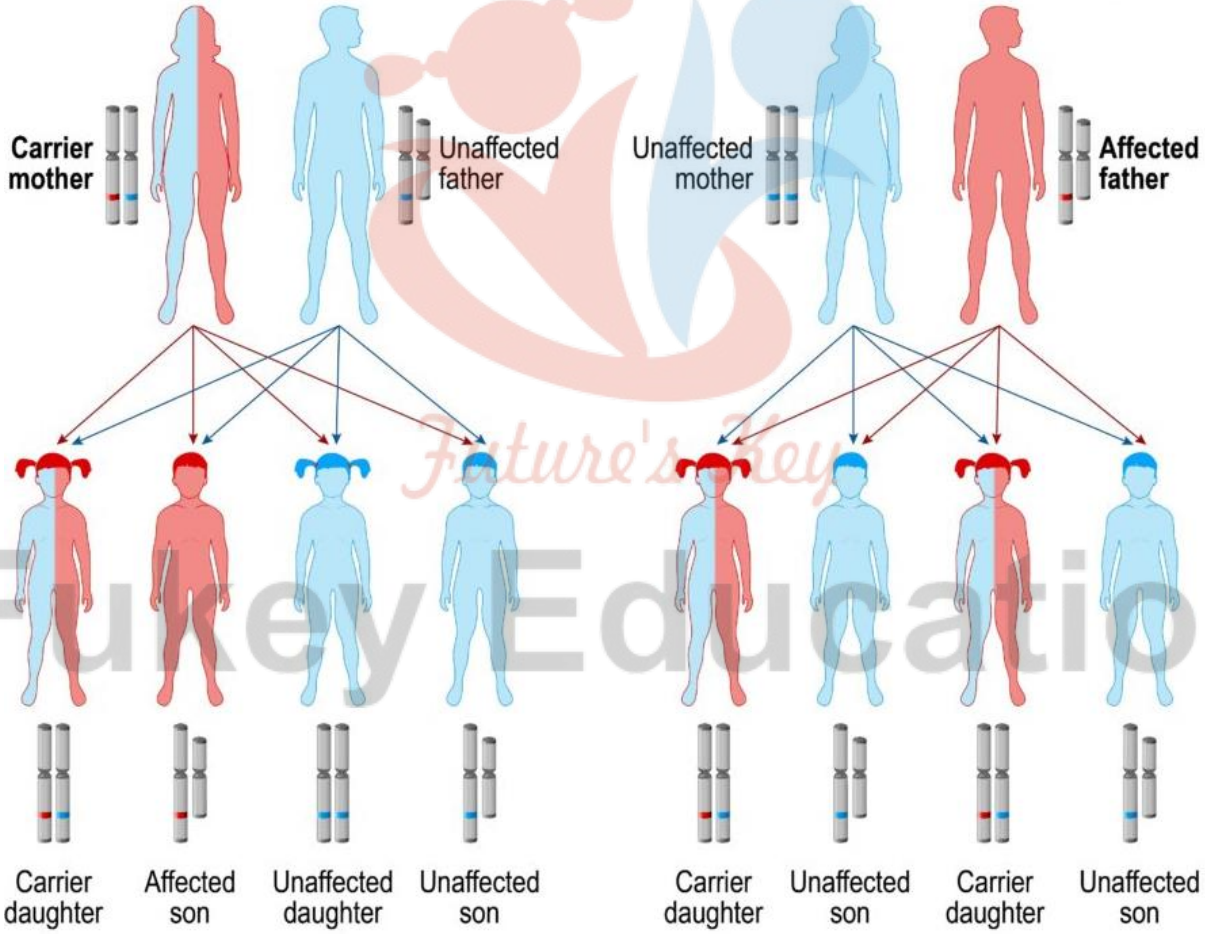
Genetic Disorders: Broadly, genetic disorders may be grouped into two categories Mendelian disorders.

Chromosomal disorders: They are transmitted as the affected individual is sterile. This is always dominant in nature.

Mendelian disorder includes:

Haemophilia:

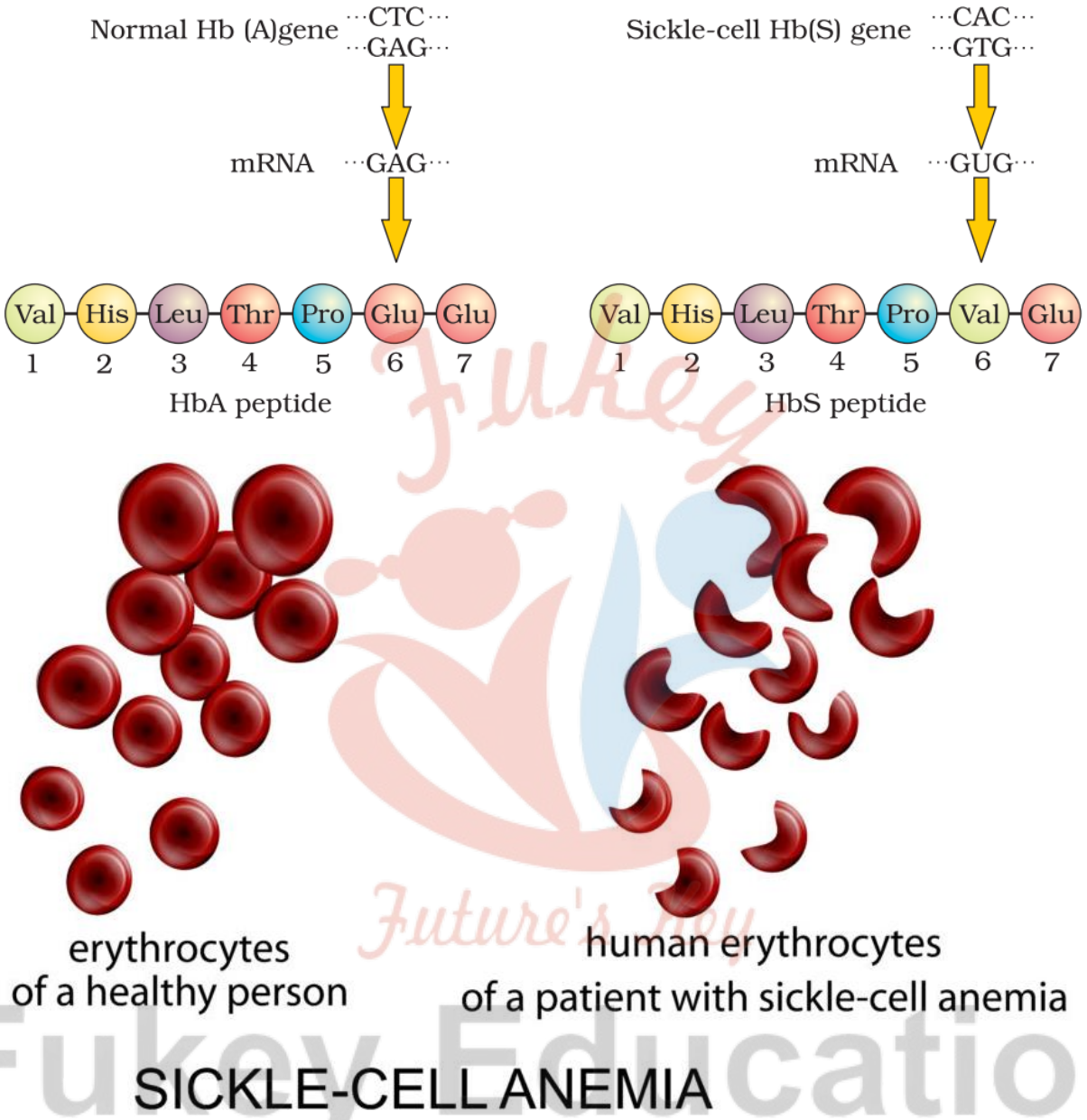
Sex linked recessive disease in which, in an infected individual, a minor cut leads to non-stop bleeding. Heterozygous female (carrier) can transmit the disease to their son. The possibility of a female becoming a haemophilic is extremely rare because mother of such a female has to be at least carrier and the father should be haemophilic (unviable in the later stage of life).



Sickle cell anaemia:

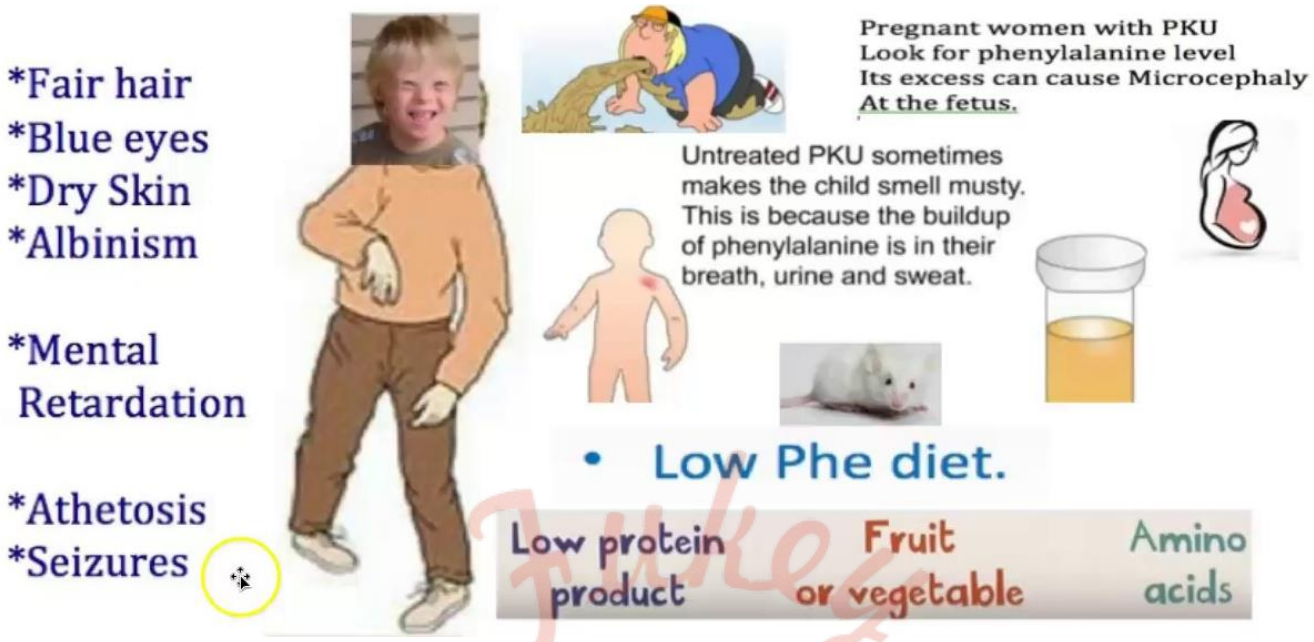
An autosome linked recessive trait in which mutant haemoglobin molecules undergo polymerization under low oxygen tension causing change in shape of the RBC from biconvex disc to elongated sickle like structure. The defect is caused by the substitution of Glutamic acid (Glu) by Valine (Val) at the sixth position of the beta globin chain of the haemoglobin

molecule. The substitution of amino acid in the globin protein results due to the single base substitution at the sixth codon of the beta globin gene from GAG to GUG.



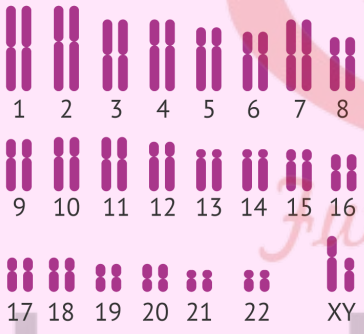
Phenylketonuria:

Inborn error of metabolism inherited as autosomal recessive trait. The affected individual lacks an enzyme that converts the amino acids phenylalanine to tyrosine. As a result of this phenylalanine is accumulated and converted into phenyl pyruvic acid and other derivatives that results into mental retardation.

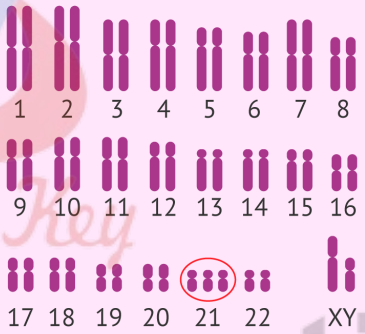


Aneuploidy:

It is chromosomal disorder Failure of segregation of chromatids during cell division results in loss or gain of chromosome called aneuploidy.



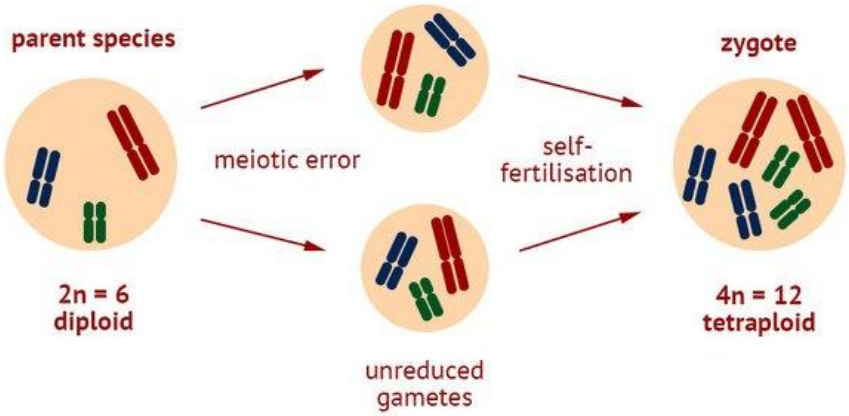
Karyotype of a healthy embryo



Karyotype of an embryo with aneuploidy

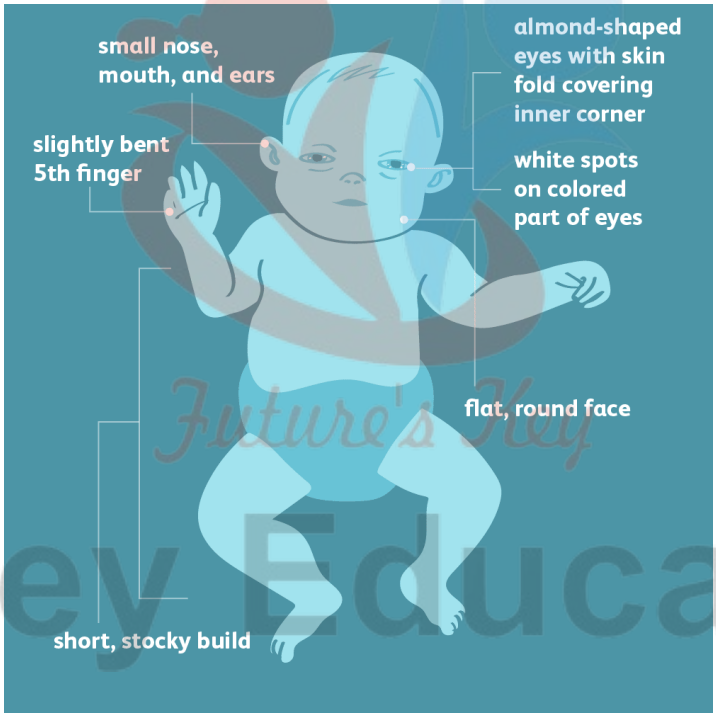
Polyploidy:

The failure of cytokinesis leads to two sets of chromosome called polyploidy.



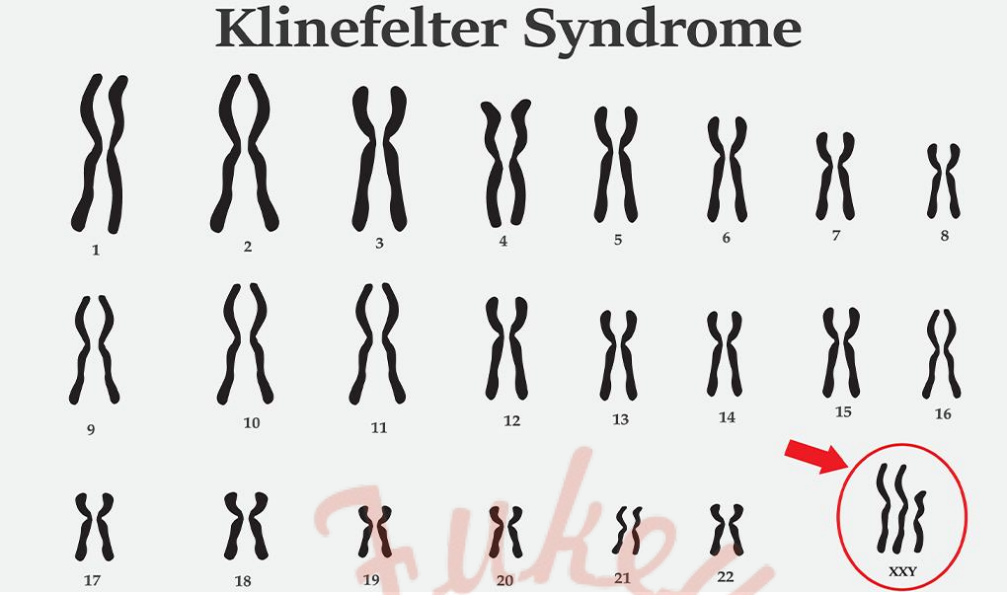
Down's Syndrome:

Down's Syndrome is due to presence of additional copy of the chromosome number 21. The affected individual is short statured with small, rounded head, furrowed tongue and partially opened mouth. Mental development is retarded.



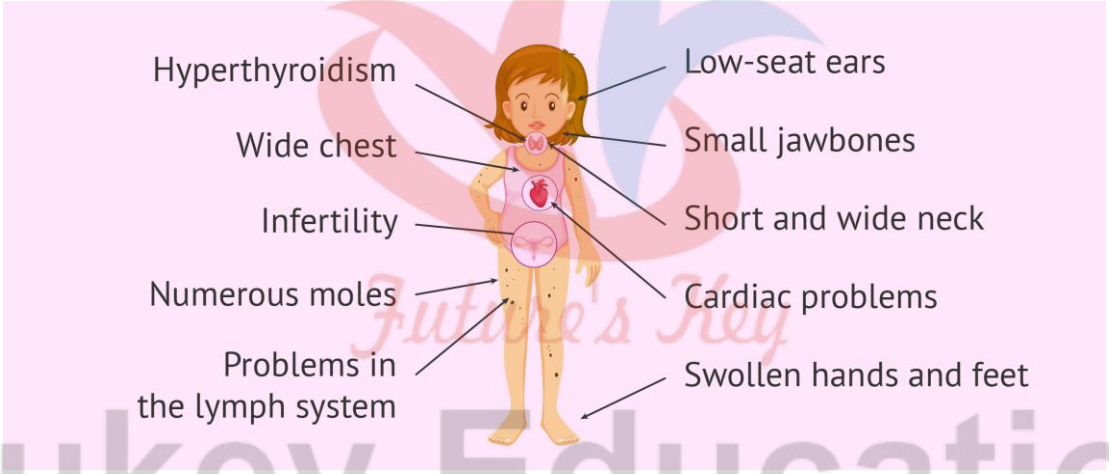
Klineflener's Syndrome:

Klineflener's Syndrome due to presence of an additional copy of X-chromosome (XXY). Such persons have overall masculine development however, the feminine development (development of breast, i.e., Gynaecomastia) is also expressed. They are sterile.



Turner's Syndrome:

Turner's Syndrome caused due to the absence of one of the X chromosomes. 45 with XO, such females are sterile as ovaries are rudimentary. They lack secondary sexual characters.



Class : 12th Biology
Chapter- 5 : Principles of Inheritance and Variation

Principles of Inheritance and Variation

S.No.	Characters	Contrasting Traits
1.	Stem height	Tall/dwarf
2.	Flower colour	Violet/white
3.	Flower position	Axial/terminal
4.	Pod shape	Inflated/constricted
5.	Pod colour	Green/yellow
6.	Seed shape	Round/wrinkled
7.	Seed colour	Yellow/green

Studied 7 Pairs of contrasting characters of pea plant

Gregor Mendel

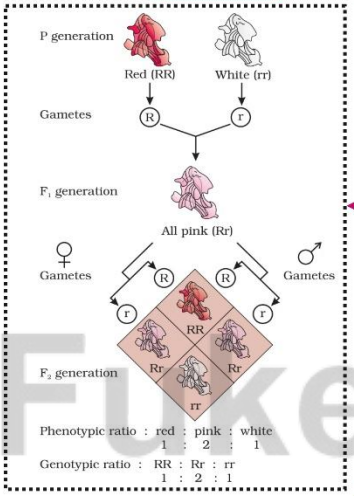
Proposed three laws

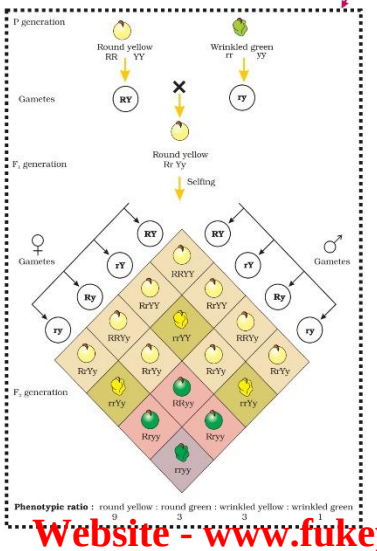
- Law of Dominance
- Law of Segregation
- Law of Independent assortment

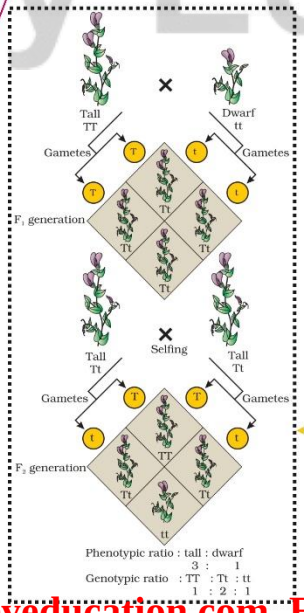
In heterozygous condition only one member of a pair expresses its effect in the hybrid and is called as dominant while the manifestation of the other is masked and is known as recessive

During gamete formation, the factors (alleles) of a character pair present in parents segregate from each other such that a gamete receives only one of the two factors

When two pairs of traits are combined in a hybrid, segregation of one pair of characteristics is independent of the other pair of characters.







- Autosomal dominant: E.g. Muscular dystrophy.
- Autosomal recessive: E.g. Sickle cell anaemia, Albinism
- Sex linked: E.g. Haemophilia

- Example, Down's syndrom (trisomy of 21).
- Klinefelter's Syndrome (XXY in male).
- Turner's syndrome (XO in female).

Mendelian Disorders

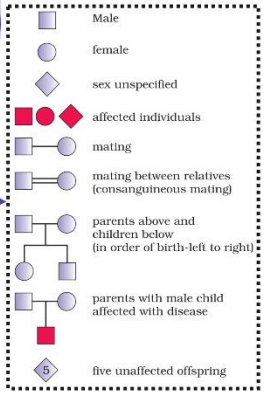
Chromosomal Disorders

Genetic Disorder

Pedigree Anlysis

- Definition
- Symbol used
- Importance

Record of occurrence ofatrait in several generations of a family



Pedigree study is utilized to trace the inheritance of a specific trait, abnormality or disease

Principles of Inheritance and Variation

Mutation

- Sudden heritable change in DNA sequence, which results in changes in the genotype and the phenotype of an organism.
- Leads to variation in DNA

- Frameshift Mutation
- Point Mutation

Sex Determination

- ZZ—ZW mechanism Example, Birds
- XX—XO mechanism Example, grasshopper
- XX – XY mechanism Example, Human being

Chromosomal Theory of inheritance

- Proposed by Walter Sutton and Theodore Boveri in 1902.
- Thomas Hunt Morgan formulated chromosomal theory of inheritance using fruit flies (Drosophila melanogaster).
- Morgan coined the term Linkage.

Non-Mendeliam Inheritance

- Incomplete Dominance
- Co-Dominance
- Pleiotropy
- Multiple Allels
- Incomplete Dominance

- Two alleles of a gene are equally dominant and express themselves even when they are together.
- E.g. ABO blood grouping in human

A gene exists in more than two allelic forms E.g. AB , O blood grouping

One of the two alleles of a gene is incompletely sp dominant over the other allele. E.g., Flower colour in Antirrhinum sp. and Mirabilis jalapa.

Ability of a gene to have multiple phenotypic effects as it influences a number of characters simultaneously

Important Questions

➢ Multiple Choice Questions:

1. All genes located on the same chromosome:
   (a) form different groups depending upon their relative distance
   (b) form one linkage group
   (c) will not form any linkage groups
   (d) form interactive groups that affect the phenotype
2. Conditions of a karyotype 2n + 1 and 2n ± 2 are called:
   (a) Aneuploidy
   (b) Polyploidy
   (c) Allopolyploidy
   (d) Monosomy.
3. Distance between the genes and advantage of recombination shows:
   (a) a direct relationship
   (b) an inverse relationship
   (c) a parallel relationship
   (d) no relationship.
4. If a genetic disease is transferred from a phenotypically normal but carrier female to only some of the male progeny, the disease is:
   (a) Autosomal dominant
   (b) Autosomal recessive
   (c) Sex-linked dominant
   (d) Sex-linked recessive.
5. In sickle cell anaemia glutamic acid is replaced by valine. Which one of the following triplets codes for valine?
   (a) GGG
   (b) AAG
   (c) GAA
   (d) GUG.
6. Person having genotype lA lB would show the blood group as AB. This is because of:
   (a) Pleiotropy
   (b) Co-dominance
   (c) Segregation
   (d) Incomplete dominance.

7. ZZ / ZW type of sex determination is seen in:
   (a) Platypus
   (b) Snails
   (c) Cockroach
   (d) Peacock.
8. A cross between two tall plants resulted in offspring having a few dwarf plants. What would be the genotypes of both the parents?
   (a) TT and Tt
   (b) Tt and Tt
   (c) TT and TT
   (d) Tt and tt.
9. In a dihybrid cross, if you get 9 : 3 : 3 : 1 ratio it denotes that:
   (a) The alleles of two genes are interacting with each other
   (b) It is a multigenic inheritance
   (c) It is a case of multiple alleles m
   (d) The alleles of two genes are segregating independently.
10. Which of the following will not result in variations among siblings?
   (a) Independent assortment of genes
   (b) Crossing over
   (c) Linkage
   (d) Mutation.
11. Mendel's law of independent assortment holds good for genes situated on the:
   (a) non-homologous chromosomes
   (b) homologous chromosomes
   (c) extra nuclear genetic element
   (d) same chromosome.
12. Occasionally, a single gene may express more than one effect. The phenomenon is called:
   (a) multiple allelism
   (b) mosaicism
   (c) pleiotropy
   (d) polygeny.
13. In a certain taxon of insects some have 17 chromosomes and the others have 18 chromosomes. The 17 and 18 chromosome-bearing organisms are:
   (a) males and females, respectively
   (b) females and males, respectively

(c) all males
(d) all females.

14. The inheritance pattern of a gene over generations among humans is studied by the pedigree analysis. Character studied in the pedigree analysis is equivalent to:
(a) quantitative trait
(b) Mendelian trait
(c) polygenic trait
(d) maternal trait.

15. It is said that Mendel proposed that the factor controlling any character is discrete and independent. This proposition was based on the:
(a) results of F3 generation of a cross.
(b) observations that the offspring of a cross made between the plants having two contrasting characters shows only one character without any blending.
(c) self-pollination of F1 offsprings
(d) cross-pollination of parental generations.

➢ Very Short Question:

1. Name any one plant that shows the phenomenon of incomplete dominance during the inheritance of its flower colour.

2. Name the base change and the amino acid change, responsible for sickle cell anaemia.

3. Name the disorder with the following chromosome complement.
(i) 22 pairs of autosomes + X X Y
(ii) 22 pairs of autosomes + 21st chromosome + XY.

4. A haemophilic man marries a normal homozygous woman. What is the probability that their daughter will be haemophilic?

5. A test is performed to know whether the given plant is homozygous dominant or heterozygous. Name the test and phenotypic ratio of this test for a monohybrid cross.

6. Name the phenomena that occur when homologous chromosomes do not separate during meiosis.

7. Name one trait each in humans & in drosophila whose genes are located on sex chromosome.

8. What is meant by aneuploidy?

9. What genetic principle could be derived from a monohybrid cross?

10. Which one change is the cause of sickle – cell anaemia?

➢ Short Questions:

1. Mention two differences between Turner ' s syndrome and Klinefelter's syndome.
2. The human male never passes on the gene for haemophilia to his son. Why is it so?
3. Mention four reasons why Drosophila was chosen by Morgan for his experiments in genetics.
4. Differentiate between point mutation and frameshift mutations.
5. Give any two similarities between behavior of genes (Mendel's factor) during inheritance & chromosomes during cell division.
6. Which law of Mendel is universally accepted? State the law?
7. Why do sons of haemophilic father never suffer from this trait?
8. How is the child affected if it has grown from the zygote formed by an XX-egg fertilized by Y-carrying sperm? What do you call this abnormality?

➢ Long Questions:

1. Study the given pedigree chart and answer the questions that follow:

   (a) Is the trait recessive or dominant?

   (b) Is the trait sex-linked or autosomal?

   (c) Give the genotypes of the parents shown in generation I and their third child is shown in generation II and the first grandchild shown in generation III.

2. Mention the advantages of selecting a pea plant for the experiment by Mendel.
3. What is recombination? Discuss the applications of recombination from the point of view of genetic engineering.

➢ Assertion and Reason Questions:

1. For question two statements are given-one labelled Assertion and the other labelled Reason. Select the correct answer to these questions from the codes (a), (b), (c) and (d) as given below.

a) Both assertion and reason are true and reason is the correct explanation of assertion.
b) Both assertion and reason are true, but reason is not the correct explanation of assertion.
c) Assertion is true, but reason is false.
d) Both assertion and reason are false.

Assertion: Number of chromosomes in one genome is equal to number of linkage groups.
Reason: Two homologous chromosomes form a linkage group.

2. For question two statements are given-one labelled Assertion and the other labelled Reason. Select the correct answer to these questions from the codes (a), (b), (c) and (d) as given below.

a) Both assertion and reason are true and reason is the correct explanation of assertion.
b) Both assertion and reason are true but reason is not the correct explanation of assertion.
c) Assertion is true but reason is false.
d) Both assertion and reason are false.

Assertion: Linked gene show dihybrid ratio of 9 : 3 : 3 : 1.
Reason: Linked gene undergo independent assortment.

➢ Case Study Questions:

1. Read the following and answer any four questions from (i) to (v) given below:

While studying inheritance of characters, a teacher gave the example of inheritance of attached earlobe and hypertrichosis of the ear to her students. A man with attached earlobes and extensive hair on pinna married a woman having free earlobes. The couple had four children, one son with attached earlobes and hairy pinna, one son with a free earlobes and hairy pinna and two daughters with attached earlobes. One of the daughters married a man with free earlobes and sparse hair on pinna. Teacher said if this couple would have sons there would be equal chances for both having free or attached earlobes and sparse hair on pinnae.

(i) Attached and free earlobe are respective example of?

a) Dominant and recessive traits.
b) Recessive and recessive traits.
c) Recessive and dominant traits.
d) Dominant and dominant traits.

(ii) Hypertrichosis of the ear is.

a) X linked trait
b) Y linked trait
c) Autosomal dominant trait
d) Autosomal recessive trait.

(iii) If a female with attached earlobe married a male homozygous for free earlobe sparse hair on pinna then what would be the chances of daughter to have attached earlobe?

a) 0%
b) 100%
c) 25%
d) 75%

(iv) If a man with attached earlobe and hairy pinna married a woman with attached earlobe then what would be the chances of son to have hairy pinna?

a) 50%
b) 100%
c) 75%
d) 0%

(v) A male with attached earlobe, sparse hair on pinna married a female with attached earlobe. Which of the following is correct regarding their progenies?

a) All sons have a free earlobe with hairy pinna.
b) All daughters have an attached earlobe.
c) 50% daughters have an attached earlobe whereas 50% daughters have a free earlobe.
d) 50% sons have attached earlobe with hairy pinna and 50% sons have a free earlobe

2. Read the following and answer any four questions from (i) to (v) given below:

According to Mendel, one gene control the expression of one character only. The ability of a gene to have multiple phenotypic effect because it influences a number of characters is an exception. The gene having a multiple phenotypic effect because of its ability to control of two or more characters can be seen in cotton. In cotton, a gene for the lint also influences the height of plant, size of the ball, number of ovules and viability of seeds.

(i) Genes with multiple phenotypic effects are known as?

a) Hydrostatic genes.
b) Duplicate genes.
c) Pleiotropic genes.
d) Complimentary genes.

(ii) Which of the following disorder is an example of genes with multiple phenotypic effects?

a) Phenylketonuria.
b) Haemophilia.
c) Sickle cell anaemia.
d) Both (a) and (c)

(iii) Which of the following is an example of gene with multiple phenotypic effect?

a) Drosophila white eye mutation.
b) Kernel colour in wheat.
c) Height in human beings.
d) Skin colour in human beings.

(iv) Which of the following statements is not correct regarding genes with multiple phenotypic effect?

a) It is not essential that all the traits are equally influenced.
b) Occasionally a number of related changes are caused by a gene.

c) It occurs due to effect of the gene on two or more inter-related metabolic pathways.
d) None of these.

(v) Assertion: In garden pea, the gene which controls the flower colour also controls the colour of the seed coat and presence of red spots in the leaf axils.

Reason: A pleiotropic gene influences more than one trait.

a) Both assertion and reason are true and reason is the correct explanation of assertion.
b) Both assertion and reason are true but reason is not the correct explanation of assertion.
c) Assertion is true but reason is false.
d) Both assertion and reason are false.

✓ Answer Key-

➢ Multiple Choice Answers:

1. (b) form one linkage group
2. (a) Aneuploidy
3. (a) a direct relationship
4. (d) Sex-linked recessive.
5. (d) GUG.
6. (b) Co-dominance
7. (d) Peacock.
8. (b) Tt and Tt
9. (d) The alleles of two genes are segregating independently.
10. (c) Linkage
11. (a) non-homologous chromosomes
12. (c) pleiotropy
13. (a) males and females, respectively
14. (b) Mendelian trait
15. (b) observations that the offspring of a cross made between the plants having two contrasting characters shows only one character without any blending.

➢ Very Short Answers:

1. Dog flower (Snapdragon or Antirrhinum sp.)
2. GAG changes as GUG, Glutamic acid is substituted by valine.
3. Ans.(i) Klinefelter' s Syndrome (ii) Downs syndrome

4. Their daughter can never be haemophilic. (0%).
5. Test cross 1 : 1.
6. Non – disjunction.
7. Humans – Colorblindness

   Drosophila – Eye colour
8. Aneuploidy is the phenomena of gain or loss of one or more chromosomes that results due to failure of separation of members of homologous pair of chromosomes during meioses.
9. Law of dominance.
10. It is caused due to a point mutation at 6th position in B-chain of hemoglobin in which glutamic acid is replaced by valine.

➢ Short Answer:

1. Turners Syndrome : The individual is female and it has 45 chromosomes

   i.e., one X chromosome is less.

   Klinefelters Syndome : The individual is male and has 47 chromosomes

   i.e., one extra X chromosome.
2. The gene for haemophilia is present on X chromosome. A male has only one X chromosome which he receives from his mother and Y chromosome from father. The human male passes the X chromosome to his daughters but not to the male progeny (sons).
3.

   i. Very short life cycle (2-weeks)

   ii. Can be grown easily in laboratory

   iii. In single mating produce a large no. of flies.

   iv. Male and female show many hereditary variations

   v. It has only 4 pairs of chromosomes which are distinct in size and Shape.
4. Point Mutations : Arises due to change in a single base pair of DNA e.g., sickle cell anaemia. Frame shift mutations : Deletion or insertion/duplication/addition of one or two bases in DNA.
5. (i). In diploid cells, the chromosomes are found in pairs just like that of mendelian factors.

   (ii). During meiosis, the chromosomes of different homologous pairs are assorted independently into gametes at random showing parallelism with mendelian factors.
6. Mendel's law of segregation is universally accepted It states that – "the two alleles of a gene remain separate & do not contaminate each other in F1 or the hybrid. At the time of gamete formation two alleles separate & pars into deferent gametes.

7. Since haemophilic is a sex – linked character, it shows criss – cross inheritance i-e from father to his daughter therefore son of haemopilic father is never haemophilic.

8. If a child has grown from the zygote formed by XX-egg fertilized by Y-sperm, the child will suffer from klinefiter syndrome & will have XXY genotype. It is characterized by prominent feminine characters e.g. tall stature with feminised physique, Breast development pubic hair pattern, poor beard growth & sterility.

➢ Long Answer:

1. (a) Dominant.

(b) Autosomal.

(c) The genotype of parents in generation I – Female: aa and Male: Aa

The genotype of a third child in generation II-Aa Genotype of the first grandchild in generation III – Aa

2. Advantages of selecting pea plant as experimental material:

Mendel selected pea plant (Pisum sativum) because:

i. Many varieties were available with observable alternative forms for a trait or a characteristic.

ii. Peas normally self-pollinate; as their corolla completely encloses the reproductive organs until pollination is complete.

iii. It was easily available.

iv. It has pure lines for experimental purpose, i.e. they always breed true.

v. It has contrasting characters. The traits were seed colour, pod colour, pod shape, flower shape, the position of flower, seed shape and plant height.

vi. Its life cycle was short and produced a large number of offsprings.

vii. The plant can be grown easily and does not require care except at the time of pollination.

3. Answer: Recombination refers to the generation of a new combination of genes which is different from the parental types. It is produced due to crossing over that occurs during meiosis prior to gamete formation.

Applications of recombination:

i. It is a means of introducing new combinations of genes and hence new traits.

ii. It increases variability which is useful for natural selection under changing environment.

iii. It is used for preparing linkage chromosome maps.

iv. It has proved that genes lie in a linear fashion in the chromosome.

v. Breeders have to select small or large population for obtaining the required cross-overs.

For obtaining cross-overs between closely linked genes, a very large population is required.

vi. Useful recombinations produced by crossing over are picked up by breeders to produce useful new varieties of crop plants and animals. Green revolution and white revolution were implemented using the selective recombination technique.

➢ Assertion and Reason Answers:

1. (b) Both assertion and reason are true, but reason is not the correct explanation of assertion.

Explanation:

Linkage group is linearly arranged groups of genes which fail to show independent assortment as these are present on the same chromosomes.

2. (d) Both assertion and reason are false.

Explanation:

Linked genes are those genes which occurs on the same chromosome while unlinked genes are the ones found on different chromosomes. Linked gene show dihybrid ratio of 3 : 1. Linked genes do not show independent assortment.

➢ Case Study Answers:

1.

(i) (c) Recessive and dominant traits.

Explanation:

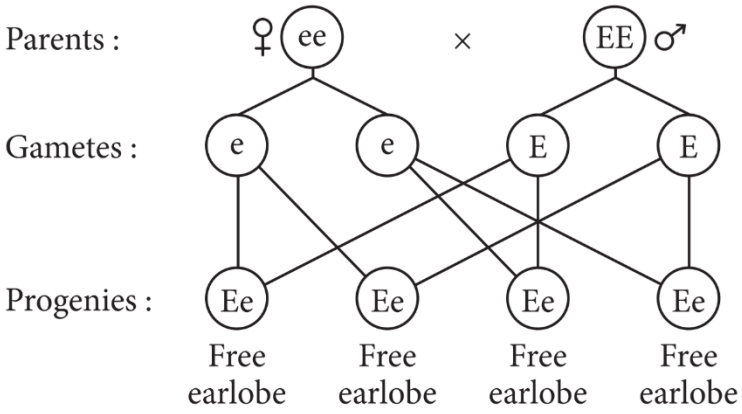
In humans, free earlobes is dominant over attached earlobes

(ii) (b) Y linked trait

(iii) (a) 0%

Explanation:

If a female with attached earlobes (ee) married a male with free lobe (EE) and sparse hair on pinna then chance of any progeny to have attached ear lobe is zero. It can be depicted as follows



(iv) (b) 100%

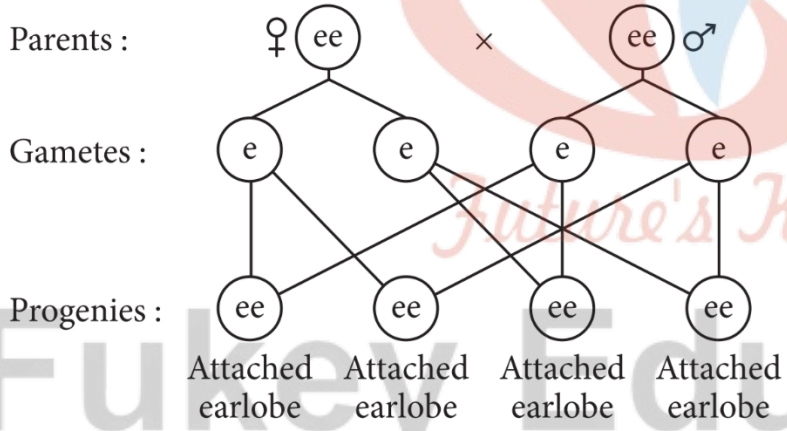
Explanation:

If a man with attached earlobe (ee) and hairy pinna married a woman with attached earlobes (ee) than 100% chances of sons to have hairy pinna as hypertrichosis or hairy pinna is Y linked feature.

(v) (b) All daughters have an attached earlobe.

Explanation:

If a male with attached earlobe sparse hair on pinna married a female with attached earlobe then all daughters have an attached earlobe.



2.

(i) (c) Pleiotropic genes.

(ii) (d) Both (a) and (c)

Explanation:

The ability of a gene to have multiple phenotypic effects because it influences a number of characters simultaneously is known as pleiotropy. In human beings pleiotropy is exhibited by syndromes, i.e., sickle cell anaemia and phenylketonuria.

(iii) (a) Drosophila white eye mutation.

Explanation:

Kemel colour in wheat, height in human beings and skin colour in human beings are examples of polygenic inheritance, i.e., inheritance controlled by three or more genes. ln Drosophila, white eye mutation pleiotropic effect, it causes depigmentation in many part of the body.

(iv) (d) None of these.

(v) (a) Both assertion and reason are true and reason is the correct explanation of assertion.