PRINCIPLES OF INHERITANCE AND VARIATION



https://www.biography.com/video/gregormendel-mini-biography-35737667892

Quiz:

https://www.youtube.com/watch?v=Hd6Wr8a vQU0

https://www.youtube.com/watch?v=PymZkYID

<u>EeY</u>

https://www.youtube.com/watch?v=yCl-

<u>11Yi68o</u>

https://www.justexam.in/chapter-wisequestion-bank/human-reproduction-12/?&page=3





Reason's for Mendel's success

• Method of working:

- a. focused on one character at a time
- b. Used true breeding pea lines
- c. Avoided cross pollination by undesirable pollen.
- d. Kept accurate records of results obtained.
- Selection of pea plant:
- a. Clear contrasting characters
- b. Produce large number of seeds
- c. Short life span.
- d. Predominantly self -pollinating(true breeding)
- e. Cross pollination produces fertile hybrids.

PRINCIPLES OF INHERITANCE AND VARIATION

- **Genetics:** deals with the inheritance, as well as the variation of characters from parents to offsprings.
- **Inheritance:** is the process by which characters are passed on from parent to progeny.
- Variation: is the degree by which progeny differ from their parents.

MENDEL'S LAWS OF INHERITANCE:

- Gregor Mendel. Conducted hybridization experiments on garden peas for seven years (1856 – 1863) and proposed laws of inheritance.
- Mendel conducted artificial pollination/cross pollination experiments using several true-breeding pea lines.
- A true breeding line is one that, having undergone continuous self-pollination for several generations.
- Mendel selected 14 true-breeding peas' plant varieties, as pair's which were similar except for one character with contrasting traits.

	Height	Seed Shape	Seed Color	Seed Coat Color	Pod Shape	Pod Color	Flower Position
Dominant	and the second s	0	0				
	Tall	Round	Yellow	Green	(full)	Green	Axial
Recessive Trait	State of the second		0	0			
	Short	Wrinkled	Green	White	Constricted (flat)	Yellow	Terminal





Mendel's proposition:

• Mendel proposed that something was being stably passed down, unchanged, from parent to offspring through the gametes, over successive generations. He called these things as **'factors'**.

- •Now a day we call them as **genes.**
- Gene is therefore are the **units of inheritance**.

•Genes which codes of a pair of contrasting traits are known as **alleles**, i.e. they are slightly different forms of the same gene.

Alphabets used:

- Capital letters used for the trait expressed at the F₁ stage.
- Small alphabet for the other trait.
- **'T'** is used for Tall and 't' is used for dwarf.
- **'T'** and 't' are alleles of each other.
- Hence in plants the pair of alleles for height would be TT. Tt. or tt.
- In a true breeding tall or dwarf pea variety the allelic pair of genes for height are **identical** or **homozygous,TT** and **tt** respectively.
- **TT** and **tt** are called the **genotype**.
- **Tt** plant is **heterozygous** for genes controlling one character (height).
- Descriptive terms **tall** and **dwarf** are the **phenotype**.

Test cross:

- When F₁ hybrid is crossed back with the recessive parent, it is known as test cross.
- It is used to know the genotype of the given plant/animal.





Back cross

The cross of the hybrids of F1- generation with either of its parents. Two possibilities:

F1 – hybrid to be crossed with homozygous dominant parent (out cross)F1 hybrids to be crossed with homozygous recessive parent (test cross)







The anthers are cut away on the purple flower. Pollen is obtained from the white flower.



- Pollen is transferred to the purple flower.
- 4. All progeny result in purple flowers.



Law of Dominance:

- Characters are controlled by discrete units called factors.
- Factors occur in pairs.
- In a dissimilar pair of factors one member of the pair dominates (dominant) the other (recessive).

Law of Segregation:

- The alleles do not show any blending and that both the characters are recovered as such in the F₂ generation though one of these is not seen at the F₁ stage.
- The parents contain two alleles during gamete formation; the factors or alleles of a pair segregate or separate from each other such that a gamete receives only one of the two factors.
- Homozygous parent produces all gametes that are similar i.e contain same type of allele.
- Heterozygous parents' produces two kinds of gametes each having one allele with equal proportion.

Deviation from law of dominance

The law of dominance does not occur universally. •Incomplete dominance or blending inheritance or semi-dominance. •Co-dominance.





Incomplete dominance:

- When a cross between two pure breed is done for one contrasting character, the F₁ hybrid phenotype dose not resemble either of the two parents and was in between the two, called incomplete dominance.
- Inheritance of flower color in the dog
 flower (snapdragon or Antirrhinum sp.) is a good
 example of incomplete dominance.
- F₂ generation phenotypic ratio is 1:2:1 in stead of 3:1 as Mendelian monohybrid cross.
- Genotypic ratio of F₂ generation is 1:2:1.













Co – dominance:

- F₁ resembled either of the two parents (complete dominance).
- F₁ offspring was in-between of two parents (incomplete dominance).
- F₁ generation resembles both parents side by side is called **(co-dominance)**.
- Best example of co-dominance is the ABO blood grouping in human.
- ABO blood group is controlled by the **gene** *I*.
- The plasma membrane of the RBC has sugar polymers (antigen) that protrude from its surface and the kind of sugar is controlled by the **gene-***I***.**
- N-acetylgalactoseamine- sugar.
- The gene I has three alleles I^{A} , I^{B} and i.
- The alleles I^{A} and I^{B} produce a slightly different form of sugar while allele i doesn't produce any sugar.
- Each person possesses any two of the three I gene alleles.
- I^{A} and I^{B} are completely dominant over i.
- When *I*^A, and *I*^B present together they both express their own types of sugar; this because of co-dominance. Hence red blood cells have both A and B type sugars.

Co Dominance and multiple allelisim				
Blood group	Possible genotype			
A	IAIA OR IA			
В	I ^B I ^B OR I ^B i			
AB	[A]B			
0	1			

Crosses of blood group (CO DOMINANCE)

Blood group	Possible genotype	Possible phenotype	
AXA	jaja X jaja	A	
	IAIA X IAI	A	
	IAI X IAI	A;O	
ВХВ	IBIBX IBIB	B	
	IBIB X IBI	B	
	IBį X IBį	B; O	
AB X AB	BIALX BIAL	AB: A; B	
0 X 0	ii X ii	0	

Post- Mendelian inheritance – gene intraction

Genes usually function or express themselves individually.
Many cases were seen by geneticists where two genes of the same allelic pair or genes of two or more different allelic pairs influence one another.

• this is called **gene interaction.**

•Bateson introduced factor hypothesis

Inter allelic or intragenic gene interactions Non- allelic or intergenic gene interaction Inter - allelic or intragenic gene interaction

-Genetic interaction between the alleles of a single gene. -i. incomplete dominance -Co- dominance -Multiple allelism - pleiotropic genes

Multiple Alleles:

- Example of ABO blood grouping produces a good example of multiple alleles.
- There are more than two i.e. three allele, governing the same character.

Pleiotropic gene (lethal gene)

Pleio means – many, tropic – affecting Pleotropic gene is the single gene which produces many or moliple unrelated phenotype. eg : lethal gene



Pleiotropy

- Most genes have multiple phenotypic effects, a property called pleiotropy
- For example, pleiotropic alleles are responsible for the multiple symptoms of certain hereditary diseases, such as
 - cystic fibrosis
 - PKU
 - Sickle-cell disease

Non-allelic or intergenic gene interaction

Interaction between genes located at different loci on the same or on different chromosomes for controlling



Fig. 5. 32. Inheritance of purple colour in Sweet Pea (*Lathyrus odoratus*) due to complementary and epistatic interactions of two nonallelic gene pairs.

A single gene product may produce more than one effect:

- Starch synthesis in pea seeds is controlled by one gene.
- It has two alleles **B** and **b**.
- Starch is synthesized effectively by **BB** homozygote and therefore, large starch grains are produced.
- The **'bb'** homozygous has less efficiency hence produce smaller grains.
- After maturation of the seeds, **BB seeds** are **round** and the **bb seeds** are **wrinkle**.
- Heterozygous (Bb) produce round seed and so B seems to be dominant allele, but the starch grains produced are of intermediate size.
- If starch grain size is considered as the phenotype, then from this angle the alleles show incomplete dominance.

INHERITANCE OF TWO GENES:

• Law of independent Assortment:

• When two characters (dihybrid) are combined in a hybrid, segregation of one pair of traits is independent of the other pair of traits.



contrasting traits: seed colour and seed shape

Principle of independent assortment

CHROMOSOMAL THEORY OF INHERITANCE:

- Why Mendel's theory was remained unrecognized?
- **Firstly** communication was not easy in those days and his work could not be widely publicized.
- Secondly his concept of genes (or factors, in Mendel's word) as stable and discrete units that controlled the expression of traits and of the pair of alleles which did not' blend' with each other, was not accepted by his contemporaries as an explanation for the apparently continuous variation seen in nature.
- **Thirdly** Mendel's approach of using mathematics to explain biological phenomena was totally new and unacceptable to many of the biologists of his time.
- **Finally** he could not provide any physical proof for the existence of factors.
CHROMOSOMAL THEORY OF INHERITANCE

Fundamental theory of genetics to identify chromosomes as carriers of genetic materials. - Termed as Boveri- Sutton chromosome theroy Pairing and separation of a pair of chromosomes would lead to the segregation of a pair of Mendelian factor (genes)

Chromosome

¢

Chromatid

Centromere –

Chromatid





Rediscovery of Mendel's result:

 1990 three scientists (deVries, Correns and von Tschermak) independently rediscovered Mendel's result on the inheritance of character

Chromosomal theory of inheritance - verification

Thomas Morgan performed experiments with fruit fly, Drosophila melanogaster

Thomas Morgan

- Father of Experimental Genetics
- Discovery of:
 - Linkage
 - Crossing over
 - Sex linkage
 - Criss-cross inheritance
 - Linkage maps
 - Genes mutations etc..





Why Drosophila?

Complete their life cycle within 2 weeks. Could be easily grown in laboratory. Easily distinguishable male and female flies Large number of contrasting traits Large number of flies are produced in one mating Have 4 pairs of chromosomes.

3 pairs – autosomes

1 - sex chromos

Male – heterogan Female - homogar



Chromosomal theory of inheritance:

- Proposed by Walter Sutton and Theodore Bovery in 1902.
- They work out the chromosome movement during meiosis.
- The behavior of chromosomes was parallel to the behavior of genes and used chromosome movement to explain Mendel's laws.
- Sutton united the knowledge of chromosomal segregation with Mendelian principles and called it the chromosomal theory of inheritance.
 - Chromosome and genes are present in pairs in diploid cells.
 - Homologous chromosomes separate during gamete formation (meiosis)
 - Fertilization restores the chromosome number to diploid condition.
- The chromosomal theory of inheritance claims that, it is the chromosomes that segregate and assort independently.

Morgan's experiment – 1 with Drosophila

Morgan bred many generations of fruit fly spontaneously

Drosopila is normally red eyed flies.

a single white – eyed male fruit fly was seen in the culture.

crossed white eyed male with red eyed female.



Morgan's experiment – 2

Morgan's conclusion after experiment I: -- Mendel's rules for inheritance of dominance and recessive trait could apply. -- the gene for eye colour might have some connection with sex of the fruit fly crossed F1 red eyed female and with white eyed male (parental). -- gene for eye colour is located X chromosome.

-No corresponding allele is present on Y – chromosome.

What is linkage?

Genetic linkage is the tendency of DNA sequences that are close together on a chromosome to be inherited together during the meiosis phase of sexual reproduction.





Brown Body Red Eyes Large Wings Yellow Body White Eyes MIniature Wings

,w,m

MALE



Experimental verification of chromosomal theory:

- Experimental verification of chromosomal theory of inheritance by **Thomas Hunt Morgan** and his colleagues.
- Morgan worked with tiny fruit flies, *Drosophila melanogaster*.
- Why Drosophila?
- Suitable for genetic studies.
- Grown on simple synthetic medium in the laboratory.
- They complete their life cycle in about two weeks.
- A single mating could produce a large number of progeny flies.
- Clear differentiation of male and female flies
- Have many types of hereditary variations that can be seen with low power microscopes.

Linkage and Recombination

- Morgan hybridized yellow bodied, white eyed females to brown-bodied, red eyed male and intercrossed their F1 progeny.
- He observed that the two genes did not segregate independently of each other and the F2 ratio deviated very significantly from 9:3:3:1 ratio (expected when the two genes are independent).
- When two genes in a dihybrid cross were situated on the same chromosome, the proportion of parental gene combinations was much higher than the non-parental type.
- Morgan attributed this due to the physical association or linkage of the two genes and coined the term**linkage.**

What is the difference between Linkage and Recombination?

- Linkage helps to keep certain genes together in the same chromosome whereas, process of recombination mix genes between chromosomes.
- Linkage is a phenomenon that can be seen in any type of cell. However, recombination is a process that occurs during meiosis I.
- Recombination does not occur when there is complete linkage. However, recombination occurs when genes are not completely linked (or when they are incompletely linked).
 - Incompletely linked genes undergo intrachromosomal recombination.

• When recombination occurs in independently assorting genes, recombinants and non-recombinants occur in equal proportions whereas, when recombination occurs in incompletely linked genes recombinant frequency is less than 50% and non-recombinant frequency is more than 50%.

• Both linkage and recombination can be used to build genetic maps/ linkage analysis (maps that show gene locations).

- Linage: physical association of genes on a chromosome.
- **Recombination:** the generation of **non-parental** gene combinations.
- Morgan found that even when genes were grouped on the same chromosome, some genes were very tightly linked (showed very low recombination) while others were loosely linked (showed higher recombination).
- The genes white and yellow were very tightly linked and showed
- 1.3 percent recombination.
- The genes white and miniature wing showed 37.2 percent recombination, hence loosely linked.
- Alfred Sturtevant used the frequency of recombination between gene pairs on the same chromosome as a measure of the distance between genes and **'mapped'** their position on the chromosome.

POLYGENIC INHERITANCE:

- Human have no distinct tall or short instead a whole range of possible heights.
- Such traits are generally controlled by three or more genes and are thus called **polygenic trait**.
- Besides the involvement of multiple genes polygenic inheritance also takes into account the influence of environment.
- Human skin color is another classic example of polygenic inheritance.
- In a polygenic trait the phenotype reflects the contribution of each allele i.e. the effect of each allele is additive.
- Assume that three genes A, B, C control the skin colour in human.
- Dominant forms A, B; AND C responsible for dark skin colour and the recessive forms a, b, c for light color of the skin.
- Genotype with dominant alleles (AABBCC) will have darkest skin color.
- Genotype with recessive alleles (aabbcc) will have lightest skin colour.
- Other combinations always with intermediate colour.

PLEIOTROPY:

- A single gene can exhibit multiple phenotypic expression, such gene is called **pleiotropic gene**.
- The mechanism of pleiotropy in most cases is the effect of a gene on metabolic pathways which contributes towards different phenotypes.
- **Phenylketonuria** a disease in human is an example of pleiotropy.
- This disease is caused due to **mutation** in the gene that code for the enzyme **phenyl alanine hydroxylase.**
- Phenotypic expression characterized by:-
 - Mental retardation
 - Reduction in hairs.
 - Reduction in skin pigmentation.

SEX DETERMINATION:

- Henking (1891) traced specific nuclear structure during spermatogenesis of some insects.
- **50**% of the sperm received these specific structures, whereas 50% sperm did not receive it.
- Henking gave a name to this structure as the **X-body**.
- X-body of Henking was later on named as X-chromosome.

Sex-determination of grass hopper:

- Sex-determination in grasshopper is **XX-XO type.**
- All egg bears one 'X' chromosome along with autosomes.
- Some sperms (50%) bear's one 'X' chromosome and 50% do not.
- Egg fertilized with sperm (with 'X' chromosome) became female (22+XX).
- Egg fertilized with sperm (without 'X' chromosome) became male (22 + X0)

Sex determination in insects and mammals (XX-XY type):

- Bothe male and female has same number of chromosomes.
- Female have autosomes and a pair of X chromosomes. (AA+ XX)
- Male have autosomes and one **large 'X' chromosome** and one very small '**Y-chromosomes. (AA+XY)**
- This is called **male heterogammety** and **female homogamety**.

Sex determination in birds:

- Female birds have two different sex chromosomes designated asZ and W.
- Male birds have two similar sex chromosomes and called **ZZ**.
- Such type of sex determination is called **female heterogammety** and **male homogamety**.

Sex determination in Honey bee:

- Sex determination in honey bee based on the number of sets of chromosomes an individual receives.
- An offspring formed from the fertilization of a sperm and an egg developed into either queen (female) or worker (female).
- An unfertilized egg develops as a male (drone), by means of parthenogenesis.
- The male have half the number of chromosome than that of female.
- The female are diploid having 32 chromosomes and males are haploid i.e. having 16 numbers of chromosomes.
- This is called **haplodiploid sex determination system**.
- Male produce **sperms by mitosis,** they don not have father and thus cannot have sons, but have **grandsons.**

MUTATION:

- Mutation is a phenomenon which results in alteration of DNA sequences and consequently results in changes in the genotype and phenotype of an organism.
- In addition to recombination, mutation is another phenomenon that leads to variation in DNA.
- Loss (deletion) or gain (insertion/duplication) of a segment of DNA results in alteration in chromosomes.
- Since genes are located on the chromosome, alteration in chromosomes results in abnormalities or aberration.
- Chromosomal aberrations are commonly observed in cancerous cells.
- Mutations also arise due to change in a single base pair of DNA. This is known as point mutation. E.g. sickle cell anemia.
- Deletion and insertions of base pairs of DNA causes frame shift mutations.

GENETIC DISORDERS:

- Pedigree Analysis:
- Analysis of traits in several of generations of a family is called the **pedigree analysis.**
- In the pedigree analysis the inheritance of a particular trait is represented in the family tree over generations.



Autosomal Dominant

- Affected individuals have at least one affected parent
- The phenotype generally appears every generation
- Two unaffected parents only have unaffected offspring
- Traits are controlled by dominant genes
- Both males and females are equally affected
- Traits do not skip generations
- e.g. polydactyly, tongue rolling ability etc

Dominant Pedigree



Autosomal recessive:

- Unaffected parents can have affected offspring
- Traits controlled by recessive genes and
- Appear only when homozygous
- Both male and female equally affected
- Traits may skip generations
- 3:1 ratio between normal and affected.
- Appearance of affected children from normal parents (heterozygous)
- All children of affected parents are also affected.
- e.g.- Albinism, sickle cell anaemia etc.



Mendelian Disorder:

- Genetic disorders grouped into two categories
 - Mendelian disorder
 - Chromosomal disorder
- Mendelian disorders are mainly determined by alteration or mutation in the single gene.
- Obey the principle of Mendelian inheritance during transmission from one generation to other.
- Can be expressed in pedigree analysis.
- E.g. Haemophilia, colorblindness, Cystic fibrosis, Sickle cell anemia, Phenylketonuria, Thalasemia etc.

Hemophilia:

- In this disease a single protein that is a part of the cascade of proteins involved in the clotting of blood is affected. Due to this in an affected individual a simple cut will result in non-stop bleeding.
- Sex linked recessive disease.
- The diseases transmitted from unaffected carrier female to some of the male progeny.
- Female becoming hemophilic is extremely rare because mother of such a female at least carrier and the father should be hemophilic.
- Affected transmits the disease only to the son not to the daughter.
- Daughter can receive the disease from both mother and father.

Sickle cell anaemia:

- The defect is caused due to substitution of Glutamic acid (Glu) by Valine (Val) at the sixth position of the beta globin chain of the haemoglobin molecule.
- Substitution of amino acid takes place due to the single base substitution at the sixth codon of the beta globin gene from GAG to GUG.
- The mutant haemoglobin molecule undergoes polymerization under low oxygen tension causing the change in the shape of the RBC from biconcave disc to elongated sickle like structure.
- This is an autosomes linked recessive trait.
- Transmitted from parents to the offspring when both the parents are carrier for the gene (heterozygous).
- This disease is controlled by single pair of allele, HbA, and HbS.
- There are three possible genotypes (HbA HbA, HbA HbS, and HbSHbS.
- Only homozygous individuals for HbS (HbS HbS) show the diseased phenotype.
- Heterozygous (HbA HbS) individuals appear apparently unaffected but they are carrier of the disease as there is 50 percent probability of transmission of the mutant gene to the progeny.

Phenylketonuria:

- Autosomal recessive trait.
- Inborn error of metabolism.
- The affected individual lack one enzyme called phenyl alanine hydroxylase that converts the amino acid phenyl alanine to tyrosine.
- In the absence of the enzyme phenyl alanine accumulated and converted into phenylpyruvic acid and other derivatives.
- Accumulation of these results in mental retardation.
- These derivatives excreted through kidney.

Chromosomal disorders:

- Caused due to absence or excess or abnormal arrangement of one or more chromosome.
- Failure of segregation of chromatids during cell division cycle results in the gain or loss of chromosome(s), called **Aneuploidy.**
- Failure of cytokinesis after telophase stage of cell division results in an increase in a whole set of chromosome in an organism and this phenomenon is called **polyploidy.**
- Trisomy: additional copy of a chromosome may be included in an individual (2n+1).
 Monosomy: an individual may lack one of any one pair of

chromosomes (2n-1)

Down syndrome:

- Caused due to presence of an additional copy of the chromosome number 21 (trisomy of 21).
- This disorder was first described by Langdon Down (1866).
 - Short stature with small round head.
 - Furrowed tongue
 - Partially opened mouth
 - Palm is broad with characteristic palm crease.
 - Physical, psychomotor and mental development is retarded.



Figure 5.16 A representative figure showing an individual inflicted with Down's syndrome and the corresponding chromosomes of the individual
• Klinefelter's syndrome:

- Caused due to the presence of an additional copy of X-chromosome resulting into a karyotype of 47, (44+XXY).
 - Overall masculine development.
 - Also develop feminine character (development of breast i.e. Gynaecomastia)
 - Individuals are sterile.

• Turner's syndrome:

 Caused due to the absence of one of the Xchromosomes i.e. 45 (44 + X0).

• Such females are sterile as ovaries are rudimentary.

• Lack of other secondary sexual characters.



Figure 5.17 Diagrammatic representation of genetic disorders due to sex chromosome composition in humans : (a) Klinefelter Syndrome; (b) Turner's Syndrome What number should replace the question mark? https://www.youtube.com/watch?v=XCa_gQnORTE





What number should replace the question mark?



Ans: 17.

Sol.

It is the sum of the two digits(9 + 8) in the quadrant opposite.

What number should replace the question mark?

			14	
	22			
			34) ;
41				3
		53		?

Ans: 55.

Sol.

Each number indicates its position in the grid. 55 indicates row 5 columns 5.

