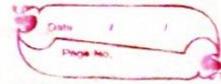


## Chapter-3

### Notes

### Genetics



- \* Law in biology was given by Mendel in 1865. → also known as father of genetics
- \* Based on set of observations that are supported by experimental data and the generalized form is accepted as law.
- \* In biology data or observations are collected from various elements found in existing nature.
- \* A generalized pattern based on data is searched, observations are confirmed experimentally & repeated several times for authentication of data.
- \* Logical form of pattern exhibited in the observations & data is expressed as law.
- \* Basic studies and experiments related to the inheritance of characters from one generation to the other has been the basis of many theories of modern genetics.
- \* ~~the~~ Gregor Mendel (1865) proposed the basic rules of heredity. The conclusion were presented on paper titled "Experiments on Plant hybrids".
- \* His idea was ignored and 16 years later after death of Mendel 3 scientists Hugo De Vries, Karl Correns & Erich Tschermak reached the same conclusion.
- \* Later his work was rediscovered in 1900 and his original work was republished in *Flora*, (1901). Bateson also supported Mendel's work while working on I. hybridization.

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## Notes Mendel's observation -

He selected Garden Pea as the experimental plant for hybridization (crossing) exp.

\* His observations were as follow -

1) Plants or organism chosen for experiments must possess distinct & detectable differences - Only one character must be considered at one time.

2) Chosen plant must be bisexual and must possess undergo sexual reproduction, so that inheritance of diff. characters from male & female parents can be studied.  
*sexually attracted to both  
males & females.*

3) Controlled and self fertilization must be possible for the chosen organisms.

4) Life cycle of the experimental plant must be small so that a number of generations of that plant can be studied.

5) Handling and performing experiments must be easy on subject - (Plant)

## Notes Terms in Genetics

- 1) Genes - Units of inheritance and contains information which is required to express a particular character in an organism.
- 2) Alleles - Genetic information for any character present in the form of a pair of contrasting form.   
*thing in order to show diff. to compare people.*
- 3) Parent Generation - true breeding plants produced by self fertilization.
- 4) F<sub>1</sub> Generation - First filial generation obtained by performing cross between two parents.
- 5) Phenotype - refers to physical appearance or physical structure of an individual.
- 6) Genotype - Genetic composition of individual.
- 7) Homozygous - Gene pair for character in which both maternal & paternal genes are identical.
- 8) Heterozygous - Gene pair that differs in allelic forms for a particular character.
- 9) Dominant allele - allele that is present in F<sub>1</sub> progeny (heterozygous form) and masks the expression of other allele present along with it is called a dominant allele.

## Notes

\*) Recessive allele = whose expression is masked by other allele present along with it

### Mendel's law of inheritance

\* He performed number of crosses between Parents (P) which differs in single character and obtain  $F_1$  generation

\* All offspring (Child) of  $F_1$  generation resembles one of the parents appearance.

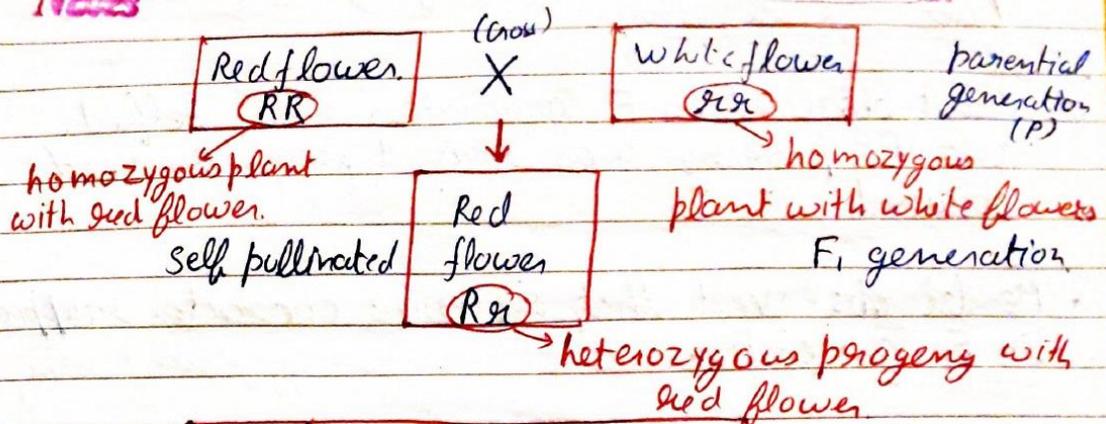
eg → red flower (parent) crossed with white flower (parent)  
all the progeny <sup>→ someones children</sup> had red flowers.

The character thus observed in  $F_1$  generation is called dominant (i.e. red colour of flower) whereas the character that remains unexpressed (white colour of flower) is called recessive.

Monohybrid <sup>cross</sup> → Type of cross between plants differing in a single pair contrasting characteristics

F

Notes



		♂ Pollen		
		R	r	
♀ Ovules	R	Red flower $RR$	Red flower $Rr$	
	r	Red flower $Rr$	White flower $rr$	

F<sub>2</sub> generation

\* Mendel states that when pure or homozygous plants for contrasting characteristics for a pair are crossed, only one of the two alleles is seen in F<sub>1</sub> generation.

Zygote - a cell that starts the process of forming a baby person or animal formed by joining together of a male's sperm and a female's egg.  
Notes  
Law of Segregation -

- \* Plant obtained in  $F_1$  generation were selfed and  $F_2$  progeny was raised from the seed obtained.
- \* Mendel observed that recessive character reappeared in  $F_2$  generation.
- \* 25% of  $F_2$  progeny exhibited the recessive character, 75% expressed dominant trait.

### Results obtained by Mendel -

- \* All characters are controlled by factors genes
- \* A diploid organism contains two alternative forms of the genes. These forms are called alleles.
- \* During formation of gametes, a male or female cell that joins with a cell of opposite sex to form a zygote.
- \* On fertilization reunion of two alleles takes place resulting in a diploid generation.
- \* In  $F_2$  generation the dominant and recessive character or alleles segregate and the gametes thus

Law of segregation / Law of purity of gametes -

States that the heterozygotes of  $F_1$  generation have alleles of two contrasting characters

## Notes

- \* Each gamete receives only one allele of a character either dominant or recessive.

### Principle of Independent Assortment-

- \* He considered two pairs of contrasting characters for study during cross. Such crosses were called the dihybrid crosses.

- \* He crossed a homozygous pea plant having a yellow round seeds with another homozygous pea plant having green wrinkled seeds.

- \* All  $F_1$  generation had yellow round seeds. When these plants of  $F_1$  progeny were crossed 4 types of plants were obtained in the ratio.

yellow round seeds - 9

yellow wrinkled seeds - 3

Green round seeds - 3

Green wrinkled seeds - 1

- \* It states that when parents differ from each other in one or more contrasting factors then the inheritance of one pair or factor is independent to that of other pair.

## Notes

### Chromosomal Theory of Inheritance

Drawbacks of Mendel's work:

- \* Acc. to Mendel certain discrete units or factors controlled the transfer of character from one generation to the other. These units were stable & did not mix with each other.
- \* He explained biological phenomenon using mathematics. This approach.
- \* He could not give any proof for the existence of factors.

Mendelian work was rediscovered by de Vries, Correns and Tschermak. It was discovered that certain structure in the nucleus doubled and undergo cell division.

This structure is called chromosomes.

- \* Sutton & Boveri (1902) emphasized on the fact each somatic cell contains diploid number of chromosomes.   
 → a part of cell in living thing that carries the set, shape & characters.

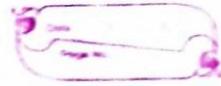
- \* It has two sets of morphologically similar chromosomes called homologous chromosomes.   
 → pair of chromosomes, approximately same length, position (centromere), pattern.

Similar chromosomes inherited from father and mother.

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• stage of Mitotic division

• stage of Meiotic division  
**Notes**



\* Sutton considered genes to be components of chromosomes and justified Mendel's work on the basis.

Sutton & Boveri

Similarities between behaviours of chromosomes & genes

\* Chromosomes occur in pairs - similar to alleles of a gene.

\* Meiosis homologous chromosomes separate like alleles of a gene that separate during gamete formation.

\* Similar behaviour is observed in meiosis for assortment and separation of chromosomes.

## Gene Interaction -

Determination of a single <sup>observable characteristics of an individual.</sup> phenotypic (traits) character of an organism, interaction of two alleles takes place in various ways and can be termed as genetic interaction.

① Intra Allelic Genetic Interactions - occurs between two alleles of the same gene

a) Incomplete Dominance - when none of two alleles for trait shows complete dominance over the other.

These express a blended or mixed character of both alleles and phenomenon is called incomplete dominance  
eg → inheritance of flower colour in 4 o'clock plant

Antibodies  $\rightarrow$  fight against foreign substances

Antigens  $\rightarrow$  that stimulates the immune system to produce antibodies. <sup>can be bacteria, viruses or fungi that cause infection</sup>

Notes

RW  $\rightarrow$  pink colored flower.

RR  $\rightarrow$  Red colored  
40' clock

White color  
40' clock

WW

	RR	R
W	RW $r_n$	RW $r_n$
W	RW $r_n$	RW $r_n$

### (b) Co-dominance $\rightarrow$

Both alleles are expressed equally at the same time.

eg  $\rightarrow$  ABO Blood groups in human beings.

AB blood group produces antibodies against B antigen and also against A antigen. This shows both A & B both do not dominate each other but dominate over 'o'. Hence called co-dominant.

AB O

### (c) Multiple Alleles $\rightarrow$

Some factors or genes exist in more than two allelic form.

### (2) Inter Allelic Genetic Interaction $\rightarrow$

$\rightarrow$  This type of interaction is seen between the genes located on the same or different chromosomes which interact with each other for the expression

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Phenotypic trait: - result of mutual action of more than one gene and constant interaction of their product with the environmental factors like temp, light etc.

of a single phenotypic trait of an organism.

@ Epistasis - Interaction between two different genes where one gene masks the expression of the other gene. The gene which suppresses the expression of the other gene is called epistatic gene while the gene whose expression is masked is called hypostatic.

① Dominant epistasis -

② Recessive epistasis

③ Duplicate Genes with cumulative effects.

④ Duplicate Recessive epistasis

⑥ Polygenic Inheritance -

Single Gene disorders in Humans -

genetic

Diseases are caused due to single gene disorders and can be categorised as dominant or recessive or X-linked disorders.

Common disorders caused due to mutation in single gene are

① Huntington's Disorder -

causing or ending in death - fatal disorder caused by an autosomal dominant single gene disorder. <sup>rather than sex chromosome.</sup>

## Notes

### Characteristics

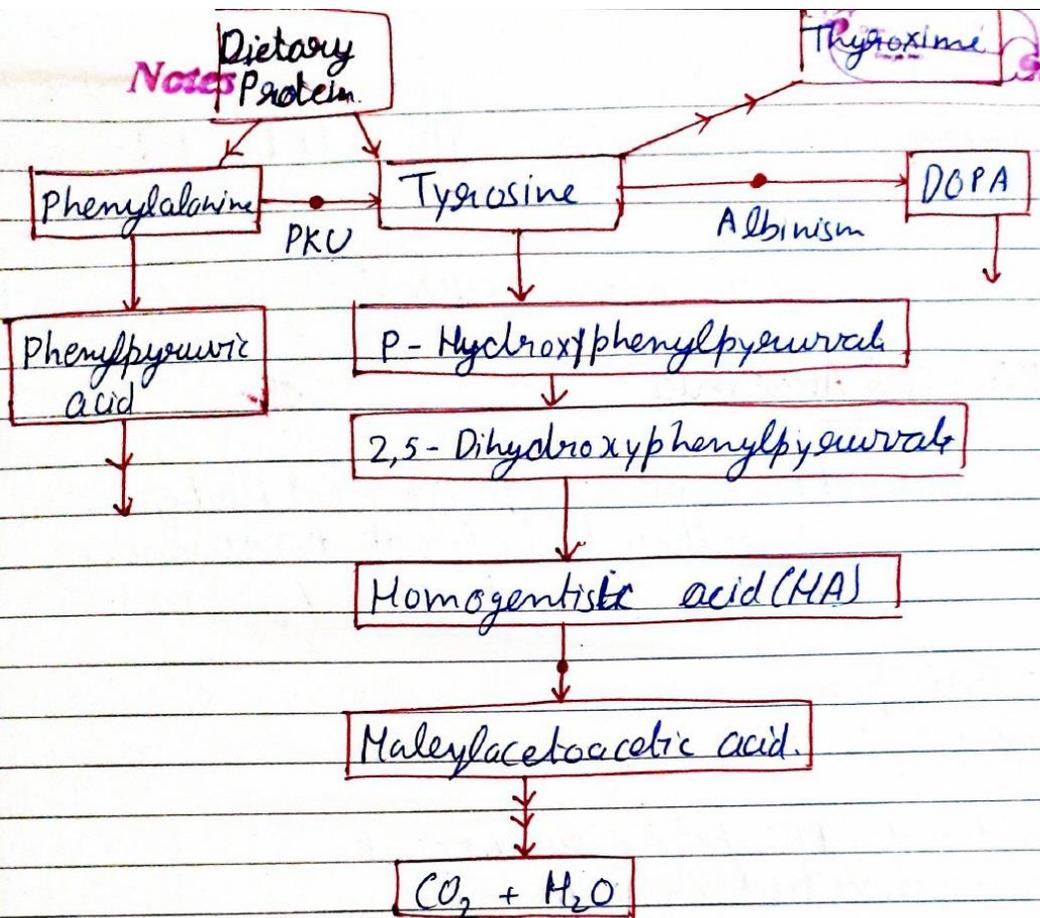
- \* Uncontrolled jerking of body (involuntary twitching of voluntary muscles)
- \* Progressive degeneration of central nervous system along with gradual deterioration in mental & physical health.

### ② Phenylketonuria (PKU)

- \* Recessive autosomal disorder is found in persons with homozygous genotype with mutated gene.
- \* It is passed by two carriers, each having a mutated gene and a normal gene.
- \* It is basically a disorder in the gene (PAH)
- \* When a mutant defective version of this gene is transferred from both parents to the child, the child would express the disorder but if the altered gene from only one parent is passed to the child, the disease is not exhibited by the child but behaves as a carrier of the mutant gene.

### Symptoms

- \* Darkening of cartilaginous regions e.g. ear pinna
- \* Person shows proneness to arthritis <sup>made of cartilage (bone)</sup>
- \* Since normal oxidation of alkapton (homogentisic acid or dihydroxyacetic acid) into acetoacetic acid does not take place, large amount <sup>liberated from</sup>



### Disorder in phenylalanine Metabolism

of this compound is accumulated in blood and is excreted through urine.

③ Albinism: includes individuals with very little or no production of melanin pigments.

melanin - compound that determines the colour of skin, hair & eyes in human beings.

Symptoms -

→ very light coloured skin & white hair.

## Notes

- \* Eyelashes and eyebrows pale with light blue to brown colour.
- \* Easily produce sun burns on skin.

### ④ Sickle-cell Anaemia

- \* Anaemia - RBC have a lifespan of about 120 days... but in the individuals with this disorder RBCs are destroyed in 10-20 days.
- \* Periodic pain.
- \* Delayed growth.
- \* In this disorder RBCs become rigid & sticky.
- \* It is a recessive autosomal disorder.

### Gene Mapping -

which helps to determine the linear order of genes on genetic units where these are arranged according to the genetic order.

- \* It helps to find out the distance between the genetic units.

### Two basic genetic principles -

- \* genes are arranged in a linear order/sequence in the chromosome.
- \* freq. of cross over between two genes is directly prop. to distance between them in the chromosome.

## Notes

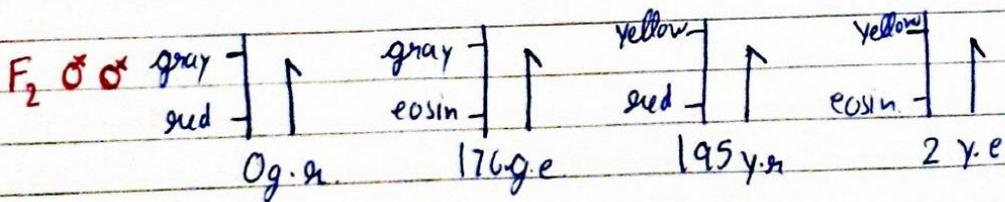
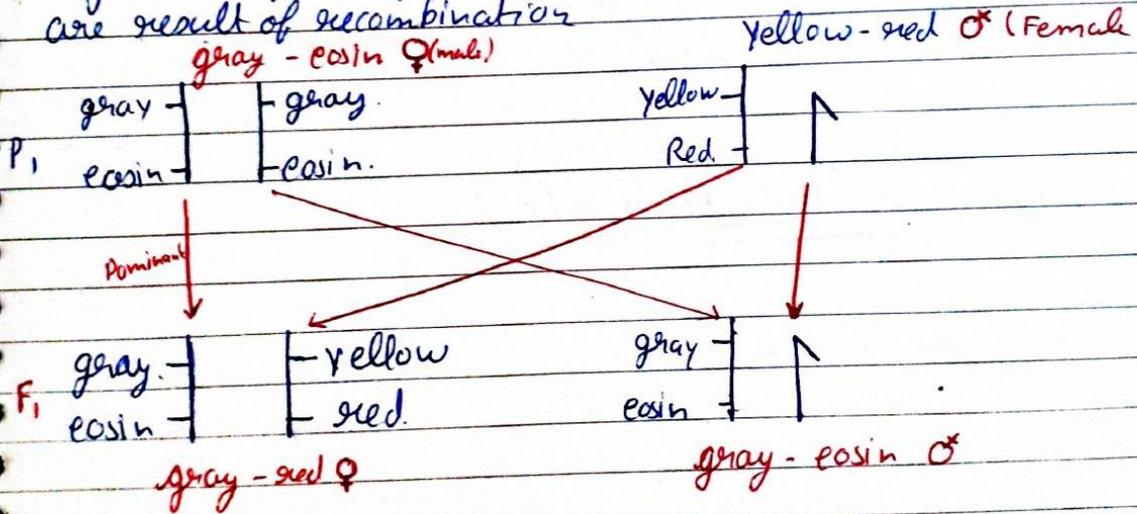
### i) Genetic Mapping - (linkage mapping)

$$\text{recombination of freq.} = \frac{\text{No. of recombinant progeny} \times 100}{\text{Total no. of progeny}}$$

example:

gray - eosin flies and yellow - red flies are the parent flies and all the alleles related to this characteristic are present on X chromosome.

Any male offspring with gray - red or yellow - eosin combination are result of recombination



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## Notes

### 2) Physical Mapping =

- \* It is the mapping of the genome by alignment of restriction maps of specific DNA obtained by fragmentation of the genome.   
 → complete set of genes in a cell or living thing.

#### a) Restriction fragment length polymorphism (RFLP)

a labelled DNA sequence that hybridizes with one or more fragments of digested DNA sample after they are separated by gel electrophoresis (which helps in separation of the DNA fragments obtained). Thus revealing a unique blotting pattern characteristic to a specific genotype at specific locus.

#### b) Polymerase Chain Reaction (PCR)