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# CONCEPT TUTORIALS



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Ch - 5

Heredity : Transfer of information (characters) from individual of one generation to next generation is called heredity.

Father of genetics : Gregor Johann Mendel.

Mendel's Experimental Plant : Mendel worked on the garden Pea plant (*Pisum Sativum*).

Why Mendel selected Pea Plant for his experiment:

1. Pea Plant shows true or pure traits. The trait of each kind of pea plant were preserved in generations after generation.
2. Pea Plant had bisexual flowers.
3. Cross pollination could be easily done.
4. Pea plant, being annual, had a short life cycle.
5. Pea plant produced many seeds in one generation.
6. It could be raised, maintained and handled easily.
7. Pea plants having each of the seven characters were easily available.

Mendel selected seven characters of pea plant

Dominant

Recessive

Form of seed	- Round	wrinkled
colour of cotyledons	- yellow	green
colour of flower	- yellow	white
Shape of pod	- Inflated	Constricted
colour of pod	- Green	yellow
Position of flower	- Axillary	Terminal
Height of plants	- Tall	Dwarf

Mendel's law

1. Law of Dominance :- This law states that one factor in a pair may express itself and prevent expression of the other.

2. Law of Reciprocity :- This law states that one factor in a pair may not express itself and the other factor express itself.

3. Law of Reciprocal :- This law states that same traits involved but carried by sexes opposite to those in the original cross.

4. Law of Segregation :- The factors of each character segregate (separate) during gamete formation so that each gamete receives only one factor for each character and is always pure. This is the third law of Mendel.

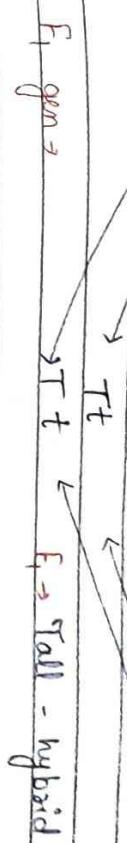
5. Traits do not blend :- Mendel noted that none of the plants in  $F_1$  gen. was intermediate in size between the two parents. But in  $F_2$  gen., the dominant and recessive traits express in the ratio of 1:2:1 or 3:1

1 - Tall ♀ - Hybrid (tall)

This ratio is also called Mendel's Monohybrid phenotypic ratio.

The Punnett Sq. of law of dominance, recessive, reciprocal, segregation is same. i.e monohybrid reciprocal, segregation is same. i.e monohybrid

Pea Plant ( $M$ )	Phenotype	Tall (Dominant)	Genotype	$TT$
				$tt$



$F_2$  gen. → Selfing of  $F_1$  generation

T	TT	Tt
t	Tt	tt

$F_2$  gen. →  $TT$ ,  $Tt$ ,  $tt$

Tall	hybrid	Dwarf
(Pure)	(Tall)	(Pure)

or

3:1 Tall : Dwarf

Mendel's Second Law :- Law of Independent Assortment

This law states that the alleles of different characters located in different pairs of homologous chromosomes are independent of one another in their segregation during gamete formation and is coming together into the offspring by fertilization, both processes occurring randomly.

## Punnett Square of Law of Independent Assortment

Phenotype Round yellow

Genotype RY

RRYY

Wrinkled green

rryy

Smooth green

rryy

Smooth yellow

RRyy

Wrinkled yellow

rrYY

Wrinkled green

rryy

Smooth green

RRyy

Smooth yellow

rrYY

Wrinkled yellow

rrYY

Wrinkled green

rryy

Smooth yellow

RRyy

Smooth green

rrYY

Wrinkled yellow

rrYY

Wrinkled green

rryy

Smooth yellow

RRyy

Smooth green

rrYY

Wrinkled yellow

rrYY

Wrinkled green

rryy

Smooth yellow

RRyy

Smooth green

rrYY

Wrinkled yellow

rrYY

Wrinkled green

rryy

Mendelism

The study of Mendel's laws of heredity

is called Mendelism.

## Important Definitions

Gene :- A gene may be defined as the unit of inheritance which is carried from the parent by a gamete in a chromosome and controls the expression of a character in the young one. A gene is a linear segment of DNA.

Symbols used for genes :- TT, Tt, tt.

Alleles :- A pair of genes that control the two alternative expressions of the same character and have the same loci (sites) in the homologous chromosomes are called alleles.

Example :- T and t for tallness and dwarfness are alleles for height.

Gene locus :- A specific region of a chromosome representing a single gene or allele is known as gene locus.

Dominant and Recessive Allele :- The allele which always expresses itself even in the presence of contrasting allele is known as the dominant allele.

The allele which fails to express itself in the presence of its contrasting allele is known as recessive allele.

Wild and Mutant Allele :- An original allele, dominant in expression and wide spread in the population is called wild gene.

An allele formed by mutation in the wild allele, recessive in expression and less common

in the population is termed as mutant allele.

(5) **Homozygous** and **Heterozygous Organisms** :-

**Homozygous** organism:- An organism in which both the alleles of a character at the corresponding loci in homologous chromosomes are identical e.g. TT (Homozygous dominant) tt (homozygous recessive)

**Heterozygous** organisms:- An organism in which the two alleles of a character at the corresponding loci in homologous chromosomes are unlike.

e.g. (Tt) Hybrid

(7) **Genotype and Phenotype** :-

**Genotype**:- It refers to the sum total of genes inherited from both the parents whether they are expressed or not.

**Phenotype**:- It refers to the expressed or observable structural and functional traits by the interaction of genes and environment.

### Test Cross

A cross between an individual genotype and an individual which is homozygous for recessive characters is known as a test cross.

**Pea Plant** : Pea Plant  
**Phenotype** : Tall (Dominant)  
**Genotype** : TT  
**Pea Plant** : Dwarf (Recessive)  
**Genotype** : tt

**F<sub>1</sub> gen.** : Tt  
**Tt** × **tt**

**Back Test Cross** :- F<sub>1</sub> × Recurrent Parent

**Tt** × **tt**

**F<sub>2</sub> gen.** : T T t t

t	Tt	tt
t	Tt	tt

Ratio 2:2  
Tall : Dwarf

(hybrid) (pure)

**Organism** and one of the original parental types is called a back cross.

**Pea Plant** : Pea Plant

**Phenotype** : Tall (Dominant)

**Genotype** : TT

**Pea Plant** : Dwarf (Recessive)  
**Genotype** : tt

Tt

Tt

Tt

**Back Cross** :- F<sub>1</sub> × Recurrent Parent

**F<sub>1</sub> gen.** : Tt  
**Tt** × **TT**

**Cross** :- All the genotypes of all the individuals in an inbreeding population make up the gene pool

F<sub>1</sub> gen | T | t

T | TT | Tt | TT : Tt

T | TT | Tt | (Pure) : Tall  
(Tall) (Hybrid)

F<sub>1</sub> gen - Hybrid pink (RW)

R | RR | RW | W

R | RR | RW | WW

RR : RW : WW  
(Pure red) (Pink) (Pure white)

1 : 2 : 1 = F<sub>2</sub> gen.

Chromosomal Theory of inheritance :-  
This Theory suggested by Sutton and Boveri  
Character of this Theory :-

1. Male and female parents pass on to the young
2. One only a sperm and egg respectively
3. The species provides only the nucleus to the

Zygote  
The nucleus contains chromosomes, which

4. Genes located on chromosomes are in linear
5. Order.

6. The chromosomes of each kind separate in meiosis and a gamete receives only one chromosome of each type.

7. Zygote formed by fertilization, contains pairs of chromosome
8. Each chromosome must carry several genes

incomplete or partial or blended or intermediate inheritance.

Phenotype Pure red flower Pure white flower  
Genotype RR WW

RW RW

Co-dominance :- When the two alleles neither show dominant-recessive relationship nor show intermediate condition, but both of them express themselves simultaneously, then this condition is known as co-dominance.

Example:- Human blood

Three blood groups in human - A, B and O  
But four phenotypes are produced  $\rightarrow$  A, B, AB, O  
These phenotypes are produced by three different alleles  $\rightarrow$  I<sup>A</sup>, I<sup>B</sup> and i<sup>O</sup>

I stands of glycoprotein

A, B, O  $\rightarrow$  Antigens present on glycoproteins

## Human Blood Groups

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Genotypes	Blood Groups	Phenotypes (Blood Groups)
$I^A I^A, I^A i^o$	A	
$I^B I^B, I^B i^o$	B	
$I^A I^B$	AB	
$i^o i^o$	O	

Allele from Parent I	Allele from Parent II	Genotype of Offspring	Blood Groups
$I^A$	$I^A$	$I^A I^A$	A
$I^A$	$I^B$	$I^A I^B$	AB
$I^A$	$i$	$I^A i$	A
$I^B$	$I^A$	$I^B I^A$	AB
$I^B$	$I^B$	$I^B I^B$	B
$I^B$	$i$	$I^B i$	B
$i$	$i$	$i i$	O

Phenotype      White flowers  
Genotype       $ccEE$        $CCee$   
 ~~$ccEe$~~        ~~$CCee$~~   
 ~~$ce$~~        ~~$Ce$~~

Gametes       $CE$        $ce$   
 $cE$        $CE$        $ce$   
 $CE$        $ce$

F<sub>1</sub> gen:       $CcEe$        $CcEe$   
 Purple      Purple

Multiple allele :  $\rightarrow$  When there are more than two alleles occupying the same locus on a chromosome are known as multiple allele.

The four phenotypes are:- A, AB, B and O

multiple forms (alleles) of a gene in a population occupying the same locus on a chromosome are known as multiple allele.

characters :-  $\rightarrow$  These are more than two alleles of the same gene.

$F_2$ gen $\rightarrow$	$CE$	$CE$	$ce$	$ce$
$CE$	$CCEE$	$CCee$	$ccEE$	$ccEe$
$CE$	$CCeE$	$CCee$	$ccEe$	$ccEE$
$ce$	$ccEE$	$ccEe$	$ccee$	$CCee$

$F_2$ gen $\rightarrow$	$CE$	$CE$	$ce$	$ce$
$CE$	$CCEE$	$CCee$	$ccEE$	$ccEe$
$CE$	$CCeE$	$CCee$	$ccEe$	$ccEE$
$ce$	$ccEE$	$ccEe$	$ccee$	$CCee$

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E<sub>2</sub> ratio : 9 : 7  
Purple : white

Purple : white

Supplementary or modifying genes →

Two independent pairs of genes, which interact to produce a new trait together, but each dominant gene alone produces its own trait.

Phenotype	Pea Comb	×	Rose Comb
Genotypes	PPRr	×	ppRR
Chamfer	P <sub>r</sub>	×	pR
F <sub>1</sub> gen	PpRr	×	PpRr
	Walnut	×	walnut

F<sub>2</sub> gene PR, P<sub>r</sub>, pR, p<sub>r</sub>

PR	P <sub>r</sub>	pR	p <sub>r</sub>
PPRR	PPR <sub>r</sub>	PpRR	PpR <sub>r</sub>
Walnut	W	W	W
P <sub>r</sub>	PPR <sub>r</sub>	PpR <sub>r</sub>	PpR <sub>r</sub>
W	Pea	W	P
pR	PpRR	PpR <sub>r</sub>	PpR <sub>r</sub>
W	W	Rose	Rose
pR	PpR <sub>r</sub>	PpR <sub>r</sub>	PpR <sub>r</sub>
W	W	Rose	Rose

②  Qualitative inheritance

Qualitative Traits : These traits may pertain to shape, size, colour, structure and so on. The qualitative traits being controlled by a single pair of genes (monogenes) are also termed monogenic traits. This inheritance of qualitative traits is known as qualitative or monogenic inheritance.

Quantitative Traits : These are measurable phenotypic traits like height, weight, milk yield, seed number etc. They are usually controlled by more than one pair of genes. So, they are called polygenic traits.

Punnett Sq. of Polygenic traits :

Genotype aabbcc

Phenotypic (very light) pure white

AABBCC

Negro (very dark)

F<sub>1</sub> gen AaBbCc × AaBbCc

F<sub>2</sub> gen  $\frac{1}{64} : \frac{6}{64} : \frac{15}{64} : \frac{20}{64} : \frac{15}{64} : \frac{6}{64} : \frac{1}{64}$

Pure white	Very light brown	Light brown	Mulatto	Dark Brown	Very dark brown	Black

F<sub>2</sub> ratio Walnut : Pea : Rose : Single

9 : 3 : 3 : 1

Polygenic Traits - A single phenotypic trait governed by more than one pair of genes is called a polygenic trait.

Phenotypic trait

Qualitative → Quantitative

Pleiotropy → The multiple effect of a gene is called Pleiotropy and a gene having multiple phenotypic effects is called a pleiotrophic gene. For example → The genes that control the flower colour in sweet pea also control the colour of the seed coats and red spots in axils of the leaves.

Lethal genes → A mutation in any genes may cause the synthesis of a non-functional or non-functional protein. These genes are called lethal genes. There are of two types :- (1) Dominant lethal genes (2) Recessive lethal genes.

LINKAGE → Linkage is a genetic tendency of an allele located close to each other on a chromosome that is inherited during meiotic cell division in sexual reproduction.

The genes which shows linkage are called linked genes.

Types :- Linkage is of two types :-  
 (a) Complete linkage (b) Incomplete genes.

Complete gene linkage → When the progeny (offspring) are same as that of parents. There is no recombination. F1 generation is called complete linkage.

Parents      Female      Male

Phenotype    Red eyes, normal wings      Purple eyes, vestigial wings

Genotype

RW | RW

RW | RW

F1 gen. → RW | RW

Red eyes, normal

wings

Recombinants = nil

Ratio 2 : 2 [Red eyes, normal wings] : [Purple eyes, vestigial wings]

Test Cross		F1 gen × Recombinant parents	
F2 gen →	RW   rw	RW   rw	X
	RW   rw	RW   rw	X
	RW   rw	RW   rw	X
	RW   rw	RW   rw	X
Ratio	2 : 2	2 : 2	2 : 2
Recombinants = nil			

Incomplete linkage	
when the progeny (offspring)	are like their parents but new recombinations are formed
Parents	Female
Phenotype	Blue flower, long Pollen
Genotype	BBLL
Chromosomes	BL BL
F1 gen	BBL LBLL
	BL BL

F1 generation		F2 gen × bl	
F2 gen :-	BBLL		
	X		

F2 gen :- Blue flower : Blue flower : Red flower : Red flower  
 long pollen      Round pollen      Long pollen      Round pollen

Ratio →

4 : 1 : 1 : 1

: 1 : 1 : 4

: 1 : 1 : 4

: 1 : 1 : 4

: 1 : 1 : 4

: 1 : 1 : 4

: 1 : 1 : 4

Factors affecting Linkage :-

1. Age :- Increase in age, increasing the strength of linkage.

2. Temp :- Rise in temp. ~~increase~~ decrease the linkage

X-rays :- Exposure to X-rays reduces the strength of linkage

## Sex Linkage :- The sex chromosomes ( $X$ and $Y$ )<sup>20</sup>

carrying the sex-determining genes, also carry several other characters. These genes travel along with the sex-controlling genes from generation to generation are called sex-linked genes.

The character controlled by the sex-linked genes are called sex-linked characters. Their location in the sex chromosomes is called sex linkage.

### [1] Inheritance of X-linked eye colour in Drosophila

Phenotype      Pure red eyed (female)      White eyed (male)

Genotype

$X^R X^R$        $X^R X^r$        $X^r X^r$

$X^R Y$        $X^r Y$

F <sub>2</sub> gen	$X^R$	$X^r$
$X^R$	$X^R X^R$ , $X^R X^r$	$X^R Y$
$X^r$	$X^r X^r$	$X^r Y$

$X^R X^R$ ,  $X^R X^r$ ,  $X^R Y$ ,  $X^r Y$

Red eyed      White Red eyed      White eyed (M)

Ratio of

F<sub>2</sub> gen

1 : 1 : 1 : 1

### Character of X-linkage or Sex linkage

- It is cross-cross. Father passes X-linked allele to the daughter and mother passes to her son as well as daughters.

- Only homozygous females express an X-linked recessive trait.

- Heterozygous females are carriers of X-linked recessive trait.

- Many recessive alleles are harmful.

- Most of the sex linked traits are recessive.

Crossing over :- It is the mutual exchange of the corresponding segments of the adjacent paternal and

maternal chromosomes of the synapsed homologous chromosomes in the pachytene of meiosis-I, produce new combinations of genes.

Mechanism → Crossing over is also known as breakage and reunion theory.

It comprises four steps :-

(1) Synapsis (2) Tetrad formation (3) exchange of chromatide (4) disjunction.

Synapsis - The homologous chromosomes come to lie close together in pairs in the zygote stage of the prophase of meiosis-I. This pairing is called synapsis. This pairing is quite brief and point to point along their lengths.

A complex structure called synaptonemal complex is formed. Under electron microscope this complex reveals three parallel dense lines placed in the same plane. This synaptonemal complex is made up of DNA and protein.

The paired homologous chromosomes are called Bivalents.

Tetrad formation - The chromatids of each synapsed chromosome slightly separate and become visible in the pachytene substage of the prophase of meiosis.

A group of four homologous chromatids is called a tetrad.

Crossing over - Crossing over also occurs in the pachytene substage. The adjacent non-sister chromatids break at homologous sites, mutually exchange small corresponding segments and rejoin. This process leads to physical exchange of segments and is so exact that neither chromatide gains or loses

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any gene.

The points of exchange are termed as chiasmata. Breakage of chromatids is brought about by enzymes endonuclease and repairing of broken segment is done by enzyme ligase.

Disjunction Or Terminalization :- The force of repulsion is now set up between the chromosomes. After the completion of crossing over, the synaptic forces end and the homologous chromosomes move apart.

Factors affecting Crossing over :- The distance between the frequency of crossing over of genes is directly proportion to the distance between them in the chromosome. This is also called Morgan and Sturtevant's hypothesis.

The more distant two genes are in a chromosome, the greater opportunity they have for crossing over.

- 1) Age :- Increase in age, decrease in crossing over.
- 2) X-rays :- Crossing over increases in X-ray.
- 3) Sex :- No effect of sexes.
- 4) Temp :- Increase in temp., increases crossing over.

- 5) Chemicals :- Many chemicals found to alter crossing over.
- 6) Trisomy :- One cross decreases the chances of another crossing over near it.

Types of Crossing over :-

Crossing over may be single, double or multiple.

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It may be reciprocal or complementary

### Reciprocal variation

(a)  $\underline{\underline{XX}}$  and  $\underline{\underline{XY}}$  are known as sex-chromosomes. Female is heteromorphic as it carries  $\underline{\underline{XX}}$ . Male is heteromorphic as it carries  $\underline{\underline{XY}}$ . X-chromosome is larger one and Y-chromosome is also some is smaller. Y-chromosome is also called androdine.

(b)  $\underline{\underline{XX}} - \underline{\underline{XO}}$  :- In some species, male has only one sex chromosome. The condition in the male  $\underline{\underline{XO}}$  (o man absent) and in female it is  $\underline{\underline{XX}}$ . e.g. Cockscomb and Siamese.

(c)  $\underline{\underline{ZW}} - \underline{\underline{ZZ}}$  :- In some species, female is heteromorphic and male is homomorphic. Female is  $\underline{\underline{ZW}}$  and male is  $\underline{\underline{ZZ}}$  e.g. fishes, reptiles, birds.

(d)  $\underline{\underline{Zo}} - \underline{\underline{Zz}}$  :- In some species, the female lacking one chromosome i.e.  $\underline{\underline{Zo}}$  and the male is  $\underline{\underline{Zz}}$  e.g. in some butterflies and moths.

Variation :- All organisms show variations.

Variations are of two types :-

1) Somatic or somatogenic  
2) Germinal or blastogenic

Somatic variations :- It affects the somatic cells of an organism. This is also called the acquired variation.

Somatic variations is produced by three types of

factors (1) environment (2) use and disuse of organs (3) conscious effects.

Germinal Variations :- It affects the germ cells of an organism.

Germinal variations are of two types :-

### Continuous

### Discontinuous

- |                                                                           |                                                                  |
|---------------------------------------------------------------------------|------------------------------------------------------------------|
| 1. It refers to small, indistinct differences from the average condition. | 1. It refers to large, conspicuous differences from the parents. |
| 2. There is an intermediate stage.                                        | 2. These are no intermediate stage                               |
| 3. It can be represented by a smooth curve                                | 3. It cannot be represented by a curve.                          |

- |                                  |                                        |
|----------------------------------|----------------------------------------|
| 4. It is very common             | 4. It is not common                    |
| 5. There is no evolution         | 5. There is evolution                  |
| 6. It is also called fluctuation | 6. It is also called sport fluctuation |

Mutation :- The term mutation was introduced

by "Hugo De Vries" in 1901. This is also called mutation theory.

If mutation occurs in germ cells, it is called germinal mutations.  
If mutation occurs in somatic cells, it is called somatic mutation.

## Chromosomal Mutations :-

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It is also called chromosomal Aberration. Chromosomal mutations are of two types:-

- 1) Intrachromosomal Modifications
- 2) Interchromosomal Modifications

[1] Intrachromosomal Modifications :- These changes affect a single chromosome. They occur in two ways:- deletion and inversion.

Deletion :- Deletion is lacking of segment of chromosome. Deletions may be terminal or intercalary. Due to deletion affected chromosome loses some genes and become shorter than normal.

Terminal deletion means deficiency of genes from tip of the chromosome.

Intercalary deletion means deficiency of genes from centre of the chromosome.

Example :- Cri-du-chat syndrome

Inversion :- A segment of a chromosome separates and rejoins it in an inverted position. Inversion may also be terminal or intercalary.

Intercalary inversion may be pericentric (if it includes centromere) and may be paracentric (if it does not include centromere).

## [2] Interchromosomal Modifications :-

These changes affect two chromosomes. They occur in two ways:- translocation and duplication.

Translocation :- A segment of a chromosome breaks off and joins a non homologous chromosome. Both the affected chromosomes get modified. The donor

suffers deletion and becomes shorter than normal. The other chromosome gets extra set of genes and becomes longer than normal.

Duplication :- It is the occurrence of a chromosome segment more than once in a chromosome. Duplication may be in tandem sequence or in reverse order.

### Gene Mutations :-

These are sudden, stable changes in the genes. Due to mutation in genes, there is change in phenotype of an organism.

Mutation :- The smallest portion of a gene that can mutate is called a muton.

Types of Gene Mutations :- There are 3 types of gene mutations.

- (1) Substitution
- (2) deletion
- (3) insertion

Substitution :- In this one or more nitrogenous bases are changed with others. Substitutions are of 2 types:- (a) Transitions (b) Transversions.

Transitions :- In this purine is replaced by purine (A and G) and pyrimidine is replaced by another pyrimidine (T and C).

Transversions :- In this purine are replaced by pyrimidine or vice versa.

- (1) Deletion :- These are gene mutations in which one or

more nitrogenous bases are lost from a segment of DNA.

Insertion (Additions) :- In this one or more nitrogenous bases are added or introduced into a segment of DNA that constitutes a gene.

## Frameshift mutations :- Mutations involving insertion or deletion of a base can result in an effect if there is any mutations on deleting or inserting base, it changes whole of the frame.

This is called frameshift mutations.  
TAC CAT TAG ATT

Deletion of T  
in place of C  
TAC CCA TAA GAT T      TAC CAT <sup>T</sup> AAG ATT

## Non-sense, Miss-Sense and Sense-Sense Mutation

Non-sense: Which terminate (stop) the process

They are 3 in number UGA, UAG, UAA.

Miss-Sense: It changes a codon which alters a specific amino acid in polypeptide often making the latter non-functional

Sense-Sense: It changes a codon which does not alter the aminoacid of a polypeptide.

Point and Cross Mutation: When there is a mutation in gene at single nitrogenous base is called point mutation.

The gene mutations which involve more than one nitrogen bases or the entire gene are termed as gross mutation.

## Origin of Mutation Or How mutations takes place:

Mutations occurs due to two reasons:-

- Spontaneous mutations
- Induced mutations

Spontaneous Mutations: These occur at random and their frequency is low. They may arise by errors in the process of replication and from failure to repair damaged DNA properly.

Some genes increase the mutation rate, it is known as mutator or mutator genes.

Some genes decrease the mutation rate, these are known as suppressor genes.

## Induced Mutations

Biological Mutations: These occurs due to three types:-

Physical Mutations: These occurs due to radiations and temp

Radiations: X-rays, gamma rays, alpha, beta rays, UV rays are the mutagenic agents.

Temp: Increase in temp, breaks the two strands of DNA, Hence acts as mutagen.

Chemicals: Some chemicals such as mustard gas, nitroso acid, formaldehyde etc act as mutagen.

## Importance of Mutations

Adaptation: It provides variability for organisms to adapt to new environment.

Organic evolution: Mutations contribute to the evolution of the species.

Agriculture: (1) Mutation have produced seedless fruits (grapes, banana, oranges)

(2) Improved varieties of ornamental plants such as roses, dahlia, etc.

(3) Mutations have produced new vegetables like cabbage

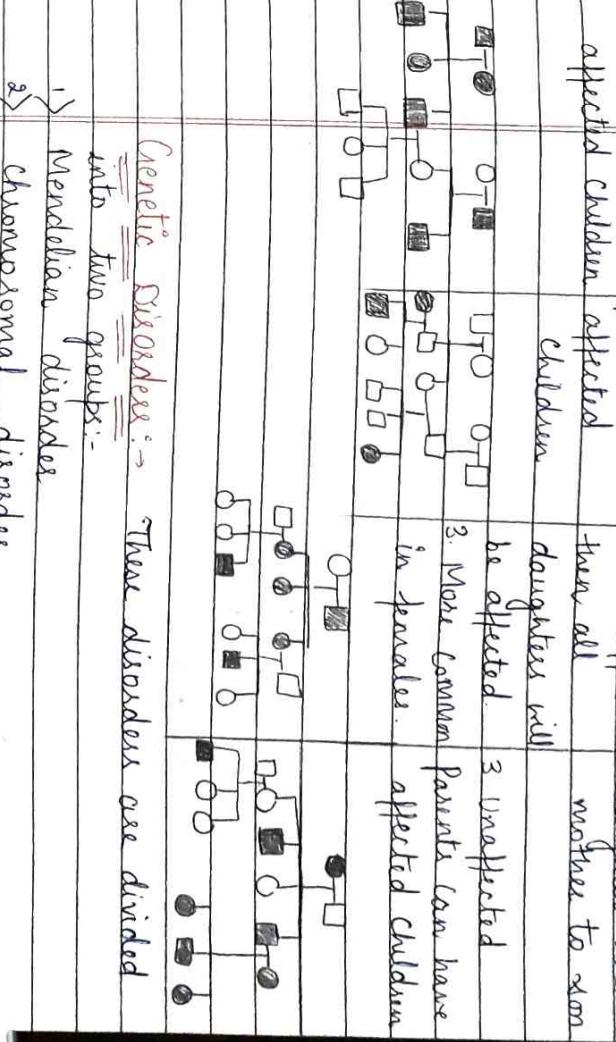
(4) Animal husbandry: Mutations have increased yield of milk, eggs, meat and wool in animals.

(5) Industry: Many useful products formed from microorganisms which has been mutate. Like antibiotic.

Pedigree Analysis → Pedigree analysis is a kind of genetic analysis in which a trait is traced through several generations of a family to determine how the trait is inherited.

Problems in the study of Human Inheritance

1. Human traits are often not controlled by a single pair of genes.
2. Humans have a mixed ancestry.
3. A person can survive only 3 or 4 human generations in his lifetime.
4. The generation time is long, often 20 yrs or more.
5. Environment has a powerful influence on human traits.
6. Chromosome number in humans is large (46).



Genetic Disorders → These disorders are divided into two groups:-

1) Mendelian disorders

2) Chromosomal disorders

Mendelian disorders → These occurs mainly due to alternation and mutation in the single gene.

(a) Hemophilia (Bleeder's disease) → Hemophilia is a defect of blood which prevents its clotting. A seriously affected person may bleed to death after even minor skin cut.

Cause - Hemophilia is a sex-linked trait caused by a recessive gene, located on X-chromosome. This gene controls the synthesis of a protein.

Signs used in Pedigree analysis :-

◻ → Male ◻ - Sex unexpected

● - Affected female

○ → female ◻ → Affected male ◻ → Mating

needed for the clotting of blood.

women are always carriers because she has two X chromosomes. If only one woman has a gene for haemophilia, it's dominant because a gene for haemophilia in the other X chromosome will check the expression of the recessive gene.

### Punnett Sq. for haemophilia

Pheotype	Normal female	X	Haemophilic Male
Genotype	XX	X <sup>h</sup>	X <sup>h</sup> Y
F <sub>1</sub> gen.			

F <sub>2</sub> gen.	X	X <sup>h</sup>	X <sup>h</sup>	X <sup>h</sup> Y
	XX	X <sup>h</sup> X		X <sup>h</sup> Y

Cause - The liver enzyme haemogenitase oxidase

catalyzes the breakdown of homogenetic acid to carbon dioxide and water with the help of kick. Hence, homogenetic acid is excreted in urine.

### Phenylketonuria (PKU)

Symptoms :- It causes extreme mental retardation in children. It causes hypopigmentation of skin. Causes :- The diseased person lacks enzyme phenylalanine hydroxylase needed to change one amino acid, phenylalanine, to another i.e. tyrosine. Lack of enzyme causes damage to brain.

(4) Albinism :- Symptoms :- It is characterized by lack of dark pigment melanin in the skin, hair and iris. Cause :- It is caused by the absence of the enzyme tyrosinase which is necessary for the synthesis of the pigment melanin.

### Sickle-cell Anaemia

Sickle-cell Anaemia :- It is an autosomal hereditary disease. In this disease, the shape of red blood corpuscles is changed. The R.B.C.'s becomes sickle shaped or crescentic moon shaped. Change in the shape of red blood corpuscles is due to the presence of defective type of haemoglobin called sickle cell haemoglobin or haemoglobin S.

Haemoglobin of

normal Person = Val-His-Leu-Thr-Pro-Cys-Cys  
1 2 3 4 5 6 7

Haemoglobin chain  
1 2 3 4  
Val - His - Leu - Thr - Pro  
5 6  
Vol. 1

### of sickle-cell anaemic person

In defected person, the  $\beta$ -chain of haemoglobin at 6<sup>th</sup> position amino acid glutamic acid (Glu) is substituted by amino acid (val). This change in haemoglobin molecule results in the change in the shape of RBC.

Symptoms :- Due to change in the shape of RBC's get stuck in the small capillaries and reduce circulation.

The sickle cells also break down more rapidly, decreasing the number of red blood corpuscles leading to anaemia.

Poor circulation and anaemia effects deprive the tissues of oxygen. This also produces a variety of other symptoms such as tiredness, headache, fever, muscle cramps, poor growth, jaundice, low resistance to infection and failure of kidneys and heart.

### (2) Burkitt Square:

Phenotype Normal female  
Genotype  $Hb^A Hb^A$

Phenotype Abnormal male  
Genotype  $Hb^S Hb^S$

Fig:

F <sub>1</sub> gen	$Hb^A$	$Hb^A$	$Hb^S$
$Hb^S$	$Hb^A Hb^A$	$Hb^S Hb^A$	
		$Hb^A Hb^S$	$Hb^S Hb^S$

Ratio 1 : 2 : 1  
Normal : Carrier : Diseased

### (6) Red green colour blindness :- It is a sex linked recessive trait

Symptoms:- Red green colour blindness is the inability of certain human beings to distinguish red from green colour.

Cause:- It is produced due to malfunctioning of the retinal rods. Males suffered from this disease and females are always carrier.

### (7)

Muscular Dystrophy :- In this disease, the X-chromosome fails to produce a protein called dystrophin.

This protein is thought to relay the nerve's signal to the calcium storage sacs in the muscle cells. As a result, calcium is not released. Muscle contraction is halted at the very first step.

F<sub>2</sub> gen :-  $Hb^A$   $Hb^S$

$Hb^S$   $Hb^A Hb^S$   $Hb^S Hb^S$  Ratio 2 : 2

### (8)

Cystic Fibrosis :- This genetic disorder is caused by single autosomal recessive gene. There is lack of an enzyme which controls glycoprotein. Due

to excess amount of glycoprotein, excising gland like sweat glands of skin, lungs, liver and pancreas start functioning abnormally.

(9) **Thalassemia** :- It is a blood disorder passed down through inheritance in which the body makes an abnormal form or inadequate amount of haemoglobin. Haemoglobin is the protein in red blood cells that carries oxygen. The disorder results in large numbers of red blood cells being destroyed which leads to anaemia.

Symptoms :- Feeling tired, pale skin, yellowish skin and dark coloured urine.

### AUTOSOMAL CHROMOSOMAL DISORDERS

(1) **Down's Syndrome** :- It is also called Mongolism. Causes :- Down's syndrome is an aneuploid, trisomic disease. It is caused by the presence of an extra chromosome No. 21. Both the chromosomes of the pair 21 pass into a single egg due to non-disjunction during oogenesis in the mother ovary. Thus, the egg has 24 chromosomes and the offspring has 47 chromosomes ( $45 + X\text{Y}$  in male +  $45 + XX$  in female).

The presence of three chromosomes of the same kind is known as trisomy. Down's syndrome is also known as ~~trisomy~~ trisomy. The women above 40 years of age are more likely to produce children having Down's syndrome.

Symptoms :- It is characterized by rounded face,

broad forehead, a mongolian type of eyelid fold, flattened nasal bridge, permanently open mouth, projecting lower lip, protruding tongue, short neck, flat hands and stubby fingers. Heart and other organs may have deformities. Uterus and genitalia are under developed. There is little intelligence due to malformation of brain.

Klinefelter's Syndrome :- It is an aneuploid condition with 3 sex chromosomes (trisomy). Causes :- This syndrome is caused by  $XYY$  genotype. This genotype results from the union of a non-disjunct  $XX$  egg and a normal  $Y$  sperm or normal  $X$  sperm and abnormal  $XY$  sperm. The individual have 47 chromosomes ( $2n+1$ ). Symptoms :- The person having Klinefelter's syndrome is a sterile male with small testes, usually long legs, obesity, sparse body hair, with many facial like characters such as breasts. The person usually has normal intelligence. The more the  $X$ -chromosome is the greater is the mental defect.

Turner's syndrome :- Turner's syndrome is an aneuploid condition with a single sex chromosome (monosomy). Causes :- Turner's syndrome is caused by  $X0$ -genotype. This genotype results from the union of the abnormal  $O$  egg with a normal  $X$ -sperm or a normal  $X$  egg and abnormal  $O$ -sperm. The individual have 45 chromosome ( $2n-1$ ). Symptoms :- The individual having Turner's syndrome

is a sterile female with underdeveloped breast, reduced ovaries, small uterus, loose skin of neck, mostly normal intelligence, short stature with many male characters like heavy neck muscles, narrow hips. She may not menstruate or ovulate.

Cri-du-chat Syndrome : It is also known as Cat cry syndrome. It is a genetic condition present from birth. It is caused by the deletion of genetic material on the small arm of the chromosome 5. Infants with this condition often have a high pitched cry that sounds like that of a cat.

Patau Syndrome : This syndrome is caused by a chromosomal abnormality. There is due to extra chromosomal material on chromosome 13. The person have heart defects, brain or spinal cord abnormalities, very small or poorly developed eyes, extra fingers on toes, an opening in the lip (cleft lip), cleft palate and weak muscle tone.

Hypoploidy : Loss of chromosomes. If there is loss of one chromosome, it is called Monosomy. (2n-1) If there is loss of one pair of chromosome, it is called Nullisomy (2n-2)

Separate in Meiosis-II. This is called non-disjunction. Its result is that one gamete receives both the chromosomes of a pair and the other gamete is left without their particular chromosome. An egg with two homologous chromosomes (XX) fertilized by a sperm with one homologous chromosome (Y or X) will form an individual with three homologous chromosomes (XXX or XXY).

The XYY is sterile male with many female characters.

Aneuploidy is of 2 types :-(a) Hypoploidy  
(b) Hyperploidy

Hypoploidy : Addition of chromosomes

If one chromosome add, it is Trisomy (2n+1)  
If one pair of chromosome add, it is Tetrasomy (2n+2)

Polyploidy : It is the addition of one or more complete sets of chromosomes. It is also called genetic mutation. Sometimes in Meiosis-I, all the synapsed chromosomes fails to

Aneuploidy AND Polyploidy

Aneuploidy : It involves the addition or deletion of one or more chromosomes from the usual diploid set of chromosomes. Some times, the two homologous chromosomes of a particular pair fails to separate in meiosis. In two sister chromatids fail to

## Important Differences

separate and pass to one pole of the spindle. This produces a diploid gamete.

A diploid gamete on being fertilized by a haploid gamete will give a triploid offspring having three sets of chromosomes and on being fertilized by a diploid gamete will produce a tetraploid.

**Polyplody** is of two types ① **Autopolyploidy** ② **Allotetraploidy**

**Autopolyploidy**: In this, there is increase in the number of chromosomes sets of same species.

It is also called intra-specific polyplodity.

e.g.: Seedless watermelon, seedless grapes.

**Allotetraploidy**: It develops in a hybrid between two species by doubling of chromosome sets. e.g. AAB to AABB. It is also called inter-specific polyploidy e.g. hexaploid wheat, tetraploid American cotton.

Incomplete Dominance	Co-dominance
1. It is the phenomenon where dominant alleles do not express completely in the F <sub>1</sub> gen.	1. It is the phenomenon seen in the heterozygous characters of both dominant and recessive alleles.
2. Effect of one of the two alleles is more clearly visible.	2. Effect of both the alleles is clearly visible.
3. The effect in hybrid is intermediate of two alleles.	3. Effect of both the alleles are separate and independent.
4. It expresses a fine mixture of expressions of both the alleles.	4. There is no any such mixing of the expressions of both the alleles.
5. This phenomenon shows quantitative effect.	5. The quantitative effect is absent.

**Briefly Mention the contribution of T. H. Morgan in genetics**

**Ans:-** T. H. Morgan, an American Zoologist

- He proved that genes are located on chromosomes with his experiments on *Drosophila*.
- Morgan discovered cross-cross inheritance.
- He also discussed mutability of genes.
- He described linkage, crossing over, chromosome mapping and also proposed chromosomal theory of linkage.

He was awarded Nobel Prize in 1933.

Phenotype

1. The external appearance of an organism is its phenotype.
2. It depends on genetic make up of an organism that transfers the character into their progeny.

Monohybrid

- (1) It is the cross made between two pure individuals of a species so as to study the inheritance of single pair of allele.
- (2) In  $F_1$  gen. the genotypic ratio is 1:2:1
- (3) In  $F_2$  gen. the phenotypic ratio is 3:1
- (4) The ratio that the test cross produces is 1:1

Dihybrid

- (1) It is the cross made between two pure organisms of a species so as to study the inheritance of two pairs of alleles of two different characters.
- (2) In  $F_2$  gen. the genotypic ratio is 1:2:1:2:4:2:1:2:1
- (3) In  $F_2$  gen. the phenotypic ratio is 9:3:3:1
- (4) The ratio of test cross is 1:1:1:1

Dominance

- (1) A dominant allele express itself in presence or absence of recessive factor.
- (2) Tallness, Round seed, violet coloration in flower are dominant characters seen.

Recessive

- (1) A recessive allele express itself only in the absence of dominant factor.
- (2) Dwarfism, wrinkled seed and white coloration in flower are recessive characters seen.

Homozygous

- (1) Two similar alleles are present for a single trait.

- (2) Homozygous genotype is either dominant or recessive i.e. RR or rr

- (3) It produces one type of gamete only

- (4) It produces two different types of gametes.

Heterozygous

- (1) Two different alleles are present for a single individual trait.

- (2) Heterozygous genotype is dominant and recessive

- (3) It produces two different types of gametes.

Aneuploidy

- (1) It occurs due to alteration in number of set of a chromosome.

- (2) This is rare in humans

- (3) This is common in plants

Polyplody

- (1) It occurs due to alteration in number of particular chromosome or part such as  $2n-1$ .

- (2) This is common in humans

- (3) This is rare in plants

## Very Short Question Answer

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(1) Q:- Who rediscovered Mendel's law of heredity ?

Ans:- De Vries, Coel Correns and Tschermak.

(2) Q:- On which plant did Mendel work ?

Ans:- Garden pea

(3) Q:- How many contrasting traits Mendel noted in garden pea ?

Ans:- Seven

(4) Q:- Give one example of complementary genes.

Ans:- Flower colour in sweet pea.

(5) Q:- Write the genotype of man with blood group 'A'.

Ans:-  $T^A T^A$ ,  $T^A t^A$ ,  $t^A t^A$

(6) Q:- What is the tendency of the genes located in the same chromosome to stay together in heredity called ?

Ans:- Linkage

(7) Q:- What is a locus ?

Ans:- Site of a gene in a chromosome.

(8) Q:- How many chromatids are involved in crossing over at one chiasma ?

Ans:- Two

(9) Q:- How many linkage groups are found in man ?

Ans:- 23

(10) Q:- Who started the scientific study of mutations ?

Ans:- Morgan

(11) Q:- Mention the methods which cause morphological modifications of chromosomes.

Ans:- Deletion, inversion, translocations and duplication

(12) Q:- Name three kinds of mutagens. What are mutagens ?

Ans:- Radiation, heat and chemical Agents that cause mutations

(13) Q:- What is the location of the gene for haemophiliac ?

Ans:- X-chromosome

X - chromosome.

(14) Name the phenomenon that occurs when homologous chromosomes do not separate during meiosis

Ans:- Non-disjunction

(15) Which disorder is caused in man by the presence of one extra sex chromosome ?

Ans:- Klinefelter's syndrome

(16) How many chromosomes a person with Turner's syndrome has ?

Ans:- Forty-five

(17) Which extra chromosome causes Klinefelter's syndrome

Ans:- Sex chromosome X.

(18) Name the genetic disorder caused due to the presence of extra chromosome No. 21

Ans:- Down's syndrome.

(19) Who proposed the chromosome theory of inheritance ?

Ans:- Sutton and Boveri

(20) Name the process antagonistic (opposite) to linkage.

Ans:- Crossing over.

(21) Name the expression where F<sub>1</sub> generation resembles both the parents is called ?

Ans:- Co-dominance

(22) The condition of having an additional pair of chromosomes is genetically referred as:-

Ans:- Trisomy

(23) Name the stage of cell division where separation of an independent pair of chromosomes occurs

Ans:- Anaphase of Meiosis I

(24) X-chromosome was earlier termed by Hunking as:-

Ans:- X Body

(25) Eggs that are fertilized by sperms having X-chromosome

Ans:- Female.

IMPORTANT PUNNETT SEQ:-

- i) A mother with blood group O has a foetus with blood group B. Will there be any problem in the mother or foetus? If so, specify the problem.

Ans: No, RBC's of mother with blood group O lack antigens A and B. Therefore, the plasma factor of a foetus with blood group B will not cause any reaction.

- (2) A man with blood group A married a woman with B group. They have a son with AB group and a daughter with blood group O. Work out the cross and show the possibility of such inheritance.

<u>Parents</u>	<u>Man</u>	<u>Woman</u>
<u>= =</u>	$I^A I^O$	$I^B I^O$
<u>Gametes</u>	$I^A$ $I^O$	$I^B$ $I^O$

<u>F<sub>1</sub> gen :-</u>	$I^A$	$I^O$	$I^A I^B$ = AB
$I^B$	$I^A I^B$	$I^B I^O$	$I^B I^O$ = B
$I^O$	$I^A I^O$	$I^O I^O$	$I^A I^O$ = A

In this type of cross the possibility of occurrence of AB, A, B and O groups in F<sub>1</sub> gen.

- (3) Q:- A woman with blood group O married with a man with AB group. Show possible blood groups of offsprings.

<u>Parent</u>	<u>Man</u>	<u>Woman</u>
<u>Generation</u>	$I^A I^B$	$I^O I^O$
<u>Gametes</u>	$I^A I^B$	$I^O I^O$
<u>Offsprings</u>	$I^A$ $I^B$ $I^A I^O$ $I^B I^O$ $I^A I^O$ $I^B I^O$	$50\% I^A I^O = A \text{ Blood group}$ $50\% I^B I^O = B \text{ Blood group}$