A TEXT BOOK OF GENETICS

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PREFACE

We are gland to present the book, Genetics in the hands of undergraduate students. The present book is being humbly offered to satisfy the demand for better, exhaustive and authoritative coverage on various aspects of food and nutrition.

We have written this text book, Food and Nutrition with full academic interest and by adding the essence of devotion, dedication and determination and urge to provide updated information to the students. The aim of this text book is to provide full details and the basic knowledge of the subject in the most effective and positive manner. The knowledge level of the students in the subject is always given top priority and hence the complicated concepts are simplified. The whole text book is readable and the same time made very informative. Exhaustive exercise is given for each topic to prepare the students for examination.

The very special features of this book are, adequate and accurate illustrations, the whole matter is fertilised with good, simple information. Each and every topic in fully illustrated, diagrams are necessary to understand the matter correctly.

We are extremely grateful to our Chairman, Principal of the respective college and authorities of BOS in Zoology, Kavayitri Bahinabai Chaudhari North Maharashtra University Jalgaon.

All comments and suggestion for the improvement of book will be thankfully accepted.

- **Authors**

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Unit - 1

Introduction to Genetics

Terminology:

1) Alleles or Allelomorphs : A pair of contrasting characters or related factors controlling a single trait. Exa. – Height – Tall (T) and Dwarf (t), Colour – Red (R) and White (r).
2) Factor or Determiner : It is a functional unit of heredity present in the gametes (gene) which determines the character of the organism.
3) Gametes : Haploid sex cells formed by segregation of the organisms.
4) Parents : A diploid individual formed by the fusion of two gametes. Exa – TT, Tt, tt.
5) Offsprings : These are individuals produced by the parents.
6) Phenotype : External appearance of an individual. Exa.- Tall, Dwarf, Red, White.
7) Genotype : Genetic or internal constitution of an organism. Exa. – TT (Pure tall), Tt (Hybrid tall), tt (Pure white).
8) Homozygous : An organism with identical (similar) determiners or genes. Exa.- TT, rr.
9) Heterozygous : An organism with dissimilar determiners or genes. Exa. – Tt, Rr.
10) Hybrid : A heterozygous individual formed by the parents having contrasting characters. Exa.- Tt – Hybrid Tall, Rr - Hybrid Red.
11) F-1generation or First filial generation : It is the first generation of hybrid individuals obtained by crossing parents with contrasting characters.
12) F-2 generation or Second filial generation : It is the generation resulting by selfing or self crossing of F-1 hybrids.
13) Inbreeding : It is crossing between closely related individuals.
14) Pure breed : A homozygous individual formed by the parents and with identical characters and breed true to the species atleast three consecutive generation.
    Exa. – TT- Pure Tall, RR- Pure Red, tt- Pure Dwarf, rr- Pure White.
15) Dominant : The character or allele that expressed in F-1 generation is called as Dominant. Exa. – TT, RR, Tt, Rr.
16) Recessive : The character or allele that suppressed in F-1 generation is called as Recessive. Exa. – tt, rr.

A TEXT BOOK OF GENETICS

Introduction, Scope and Significance of Genetics:

The word genetics is derived from the Greek word 'gen' which means 'to become or to grow into'. Genetics is the branch of biology which deals with the laws or principles of heredity and variation in plants and animals.

Heredity means the transfer of similar characters from one generation to the next generation i.e. from parents to their offsprings.

The offsprings are similar to their parents but they are differing from them to some extent. This is called as variation. The variations are leading to the process of evolution.

Scope and Significance of Genetics:

Genetics is a branch of science that deals on scientific examination of genes, heredity and variations in organisms. The use of genetic knowledge can be traced to early civilizations when people relied on genetic information to improve the productivity of domesticated species of plants used for food such as corn, wheat and rice, and the species of domesticated animals.

The genetic influences were applied not only to increase the yields of crops. The agriculturist of the old time used genetic knowledge to alter species and produce traits such as being resistant to diseases and pest, while producing nutritious and healthful harvest.

Even in the modern time, the use of genetics is helpful in areas such as medical care and human health. Scientists today consider the role of genes, including the defective genes, in their reaction towards drugs, with hope of providing the best and safest dose and type of drugs for illnesses, based on the patient's genetic makeup.

The pharmaceuticals also have benefited from it since the inception of genetics. Around the world today, the people who are afflicted with serious genetic illnesses reach 5%. A few of the diseases which are believed to have genetic component are diabetes, hypertension, cardiovascular, asthma and hypertension. This means, that offspring can have the likelihood of contracting such diseases. Advances in molecular genetics allowed scientists to earn insights into the nature of serious illnesses like cancer. Likewise, they were able to come up with tools to fight many of these illnesses, and provide additional diagnostic tests.

In addition, through the knowledge on DNA and genetics, scientists are able to treat and prevent recurrence of Down's syndrome and PKU, able to provide more effective antibiotics especially the antibiotic-resistant bacteria.

The field of bioengineering has been employing genetic knowledge to directly modify the genetic material of an organism through procuring new genes and new genetic traits. This is done by permitting the segments of DNA to be transferred to different locations or be removed from the DNA molecules. It is also examining the human growth hormone for treatment of dwarfism. Insulin, too, is a product of genetics through modified bacteria. In order to produce foods and

drugs, they are synthesized from fungi and bacteria which have been manipulated genetically.

The human gene directs the construction and constitution of the cells. The genes are found on the DNA. One strand of DNA has thousands of genes which are in control of the human body and how what body parts are created. The DNA decides the color of the person's skin, eyes, hair. It is also responsible for the physical and mental attributes of the person. A person is what he is based on his DNA. Even the sex is determined by the genes. And each part of the human body contains its own body pattern, the outcome of which is due to the genes termed as the homeotic.

Genetics is among the frontiers of science in the modern era. Understanding its importance can lead to understanding of the organisms surrounding us, including the mankind. It also applicable in various fields like in agriculture, medical field, medicine, plant improvement, animal breeding, nursing etc.

Important Questions

Q.1 Multiple choice questions (1 marks each)

1) The innate tendency of offspring to resemble their parents is called

a) Variation **b) Heredity** c) Inheritance d) Resemblance

2) Who is regarded as the father of genetics?

a) Bateson b) Morgan **c) Mendel** d) Watson

3) The chemical factors that determine traits are called

a) Alleles. b) Traits. **c) Genes**. d) Characters.

4) Genetics is:

a) The study of genes b) Study of traits

c) The study of heredity d) Study of Gregor Mendel

5) The location on a chromosome where a particular gene is located is known as the:

a) Alleles. b) Dihybrid **c) Locus** d) Diploid

6) The alternate forms of a gene on homologous chromosomes are called

a) Loci **b) Alleles** c) Homozygotes d) Tetrads.

7) "Phenotype" is based on the Greek root words for

a) Appearance and shape. b) Hereditary and image.

c) Mathematical and form. d) Different and image.

8) Hereditary information is found in a cell's

a) Chloroplasts **b) Chromosomes** c) Cytoplasm d) Membranes

9) The scientist who gave the chromosomal theory of the inheritance.

a) Bateson and De-varies **b) Sutton and Boveri**

c) Mendel and Morgan d) Watson and crick

Q.2 Define /explain /comment (2 marks each)

1) Dominant
2) Allele
3) Recessive
4) Gene
5) Heredity
6) Genetics
7) Trait
8) Homozygous
9) Heterozygous
10) Homologous.

Q.3 Question for (3 mark each)

1) Give an account of scientific application of genetics.
2) Scope of genetics.
3) Significance of genetics.

Unit – 2

Mendelian Genetics

Mendelian Laws:

The modern science of genetics starts with the experiments of the Australian monk Gregor Johann Mendel, who was the first man to conduct decisive experiments in heredity and to formulate the basic laws of genetics. Hence, he is called as the Father of Genetics.

Mendel concluded from his breeding experiments by deriving four laws known as Mendel's law of inheritance. These are,

1) Law of Dominance,
2) Law of segregation or Purity of gametes,
3) Law of Independent Assortment
4) Law of Recombination

Monohybrid Cross: (3 : 1 Ratio)

A cross is made by considering only one contrasting character is called as Monohybrid Cross. The phenotypic ratio obtained in F-2 generation is called as Monohybrid Ratio, which is 3 : 1.

A cross between two parents that differ by one pair of alleles is called as Monohybrid cross. The phenotypic ratio obtained in F-2 generation is called as Monohybrid Ratio, which is 3 : 1.

Exa.– Pea plant :*Pisum sativum* – Length of stem.

Parent (P-1): **Tall** **X** **Dwarf**

Genotype: TT tt

Gamete: T t

F-1 Hybrid: Tt

Tall

Parent (P-2): **Tall** **X** **Tall**

Genotype: Tt Tt

Gamete: T t T t

F-2 Generation:

Gametes	T	t
T	TT Tall	Tt Tall
t	Tt Tall	Tt Dwarf

Genotypic Ratio: 1 : 2 : 1

Phenotypic Ratio: 3 : 1

Mendel crossed two pure breed pea plants, one with a Tall stem and the other with a Dwarf or short stem. The F-1hybrids were all Tall plants, indicating that the Tall is dominant character as only dominant character appears in F-1 generation.

When F-1 plants (hybrids) were cross themselves, then in the F-2 generation Tall and Dwarf pea plants appear in the ratio of 3 : 1.

Law of Segregation or Purity of Gametes:

The Law of segregation states, When a pair of contrasting factors or genes or allelomorphs are brought together in a heterozygote or hybrid, the two members of the allelic pair remain together without mixing and when gametes are formed the two separate out, so that only one enters each gamete.

In the cross between Tall and Dwarf plants, F-1 generation plants were heterozygous (hybrid) containing both factors or genes for tallness and dwarfness. It had been observed that in F-2 generation dwarf plants again appeared. This clearly indicated that this character of dwarfness has been transmitted through the tall plant of F-1 generation. Though the factors or genes remained in close association in F-1 generation, there was no mixing or blending and they remained unaffected with each other. At the time of gamete formation in F-1 plants, both the allelic factors segregated from each other. Therefore, one gamete received a factor for tallness and the other gamete received factor for dwarfness. Thus gametes are pure for any gene. A gamete has one or other of a pair of allelomorphs and is never hybrid with reference to any single character. In this way a gamete may convey only the factor for tallness or dwarfness, but it can't carry both.

This observation led Mendel to formulate the law of Segregation. It states that, when hybrid forms gamete, the contrasting factors or alleles segregate and enter into different gametes. The gametes formed are pure and never hybrid. Hence, it is also called as law of Purity of gametes.

Law of Dominance and Recessive:

The law of dominance and recessiveness states, When two homozygous individuals with one or more sets of contrasting characters are crossed, the characters that appear in the F-1 hybrid are dominant and those that do not appear in F-1 are recessive characters

In various experiments, Mendel found that only one of each pairs of contrasting characters was present in the hybrids i.e. in F-1 generation.

In the cross between Tall and Dwarf plants, all plants were Tall in F-1 generation but in F-2 generation they were Tall as well as Dwarf. In short, only tallness was expressed in F-1 generation, as it has not allowed the expression of dwarfness.

Mendel called the character that appeared in F-1 generation as Dominant and the character that was suppressed as Recessive. Based on this Mendel formulated the Law of Dominance. It states that, crossing between organisms for contrasting character of a pair, only one character of a pair appears in the F-1 generation i.e. one character (dominant) prevents the expression of the other character (recessive).

Law of Independent Assortment:

Law of independent assortment states, In case of inheritance of two or more pairs of characters simultaneously, the factors or genes of one pair assort out independently of the other pairs.

Mendel gave this law based on his dihybrid cross experiment. Here the total number of individuals in F-2 will be sixteen which occur in a ratio of 9:3:3:1 where two parental classes and two new combinations will be produced.

Dihybrid Cross: (9 : 3 : 3 : 1 Ratio)

A cross is made by considering two pairs contrasting characters, called as Dihybrid Cross. The phenotypic ratio obtained in F-2 generation is called as Dihybrid Ratio, which is 9 : 3 : 3 : 1.

Exa. – Pea plant:*Pisum sativum* – Seed – Colour and Shape

Parent (P-1): **Yellow Round** **X** **Green Wrinkled**

Genotype: YYRR yyrr

Gamete: YR yr

F-1 Hybrid: YyRr

Yellow Round

Parent (P-2): **Yellow Round** **X** **Yellow Round**

Genotype: YyRr YyRr

Gamete: YR Yr yR yr YR Yr yR yr

F-2 Generation:

0 / 0	YR	Yr	yR	yr
YR	YYRR Yellow Round	YYRr Yellow Round	YyRR Yellow Round	YyRr Yellow Round
Yr	YYRr Yellow Round	YYrr Yellow Wrinkled	YyRr Yellow Round	Yyrr Yellow Wrinkled
yR	YyRR Yellow Round	YyRr Yellow Round	yyRR Green Round	yyRr Green Round
yr	YyRr Yellow Round	Yyrr Yellow Wrinkled	yyRr Green Round	yyrr Green Wrinkled

Ratio: Yellow Round **:** Yellow Wrinkled **:** Green Round **:** Green Wrinkled

9 : 3 : 3 : 1

Mendel crossed a pure breed of Yellow Round seed with another pure breed of Green Wrinkled seed pea plants. The F-1 hybrids were all Yellow Round. This indicating that, Yellow and Round are dominant characters as only Dominant character appears in F-1 generation.

When F-1 plants (hybrids) were self crossed (inbreed), they gave Four types of plants in F-2 generation as Yellow Round,Yellow Wrinkled,Green Round andGreen Wrinkled in the ratio of 9:3:3:1.

Out of these four, two showed the same combination of characters as the parents and the two new combinations were produced. The appearance of four different phenotypes clearly indicates that the characters of colour of the seed and shape of the seed segregate independently.

Back and Test Cross:

When F-1 individual (hybrid) cross with one of the two parents from which they were derived, then such a cross is called as Back cross.

In back cross when F-1 hybrid back cross to the parent with dominant character, no recessive individuals are obtained in the progeny i.e. all dominant individuals.

On the other hand, when F-1 individuals are cross with recessive character parent, both phenotypes appear in the progeny.

The back cross with recessive parent is known as Test cross. Because it is used to test whether individual is homozygous (pure) or heterozygous (hybrid). For monohybrid test cross ratio is 1 : 1 and for dihybrid test cross ratio is 1 : 1 : 1 : 1.

Monohybrid Back and Test Cross:

Exa.– Pea plant:*Pisum sativum* – Length of stem.

Parent (P-1): Homozygous Tall X Homozygous Dwarf

Genotype: TT tt

Gamete: T t

F-1 Hybrid: Tt

Heterozygous Tall

Back Cross

Parent (P-2):F-1 Hybrid **X** Dominant ParentF-1 Hybrid **X**Recessive Parent

Hetero.Tall Homo.Tall Hetero.Tall Homo. Dwarf

Genotype: Tt TT Tt tt

Gamete: T t T T T t t t

F-2 Generation:TT TT Tt Tt Tt Tt tt tt

Homo.Homo.Hetero.Hetero.Hetero.Hetero.Homo.Homo.

Tall Tall Tall Tall Tall Tall Dwarf Dwarf

All Dominant 1 : 1 Test Cross

In a monohybrid cross, when homozygous Tall (TT) and homozygous Dwarf (tt) pea plant cross, in F-1 generation heterozygous Tall (Tt) plant appears.

When F-1 heterozygous Tall (Tt) is back crossed with dominant parent (TT), in F-2 generation all dominant character plant produced.

While when F-1 heterozygous Tall (Tt) is back crossed with recessive parent (tt), in F-2 generation both character plants produced in 1: 1 ratio,so called as Test cross.

Dihybrid Back and Test Cross:

In a dihybrid back cross, when homozygous Yellow Round (YYRR) and homozygous Green Wrinkled (yyrr) pea plant crossed, in F-1 generation heterozygous Yellow Round (YyRr) plant appears.

When F-1 heterozygous Yellow Round (YyRr) is back crossed with Dominant parent (YYRR), in F-2 generation all dominant character plants produced.

While, when F-1 heterozygous Yellow Round (YyRr) is back crossed with Recessive parent (yyrr), in F-2 generation four types of plant produced in 1: 1: 1: 1 ratio. So it is Test cross.

Exa. – Pea plant:*Pisum sativum* – Seed – Colour and Shape

Parent (P-1): Homozygous Yellow Round **X** Homozygous Green Wrinkled

Genotype: YYRR yyrr

Gamete: YR yr

F-1 Hybrid: YyRr

HeterozygousYellow Round

Back Cross

Parent (P-2): F-1 Hybrid **X** Dominant Parent		F-1 Hybrid **X** Recessive Parent	
Heterozygous Yellow Round	Homozygous Yellow Round	Heterozygous Yellow Round	Homozygous Green Wrinkled
Genotype : YyRr	YYRR	YyRr	yyrr
Gamete : YR Yr yR yr	YR	YR Yr yR yr	yr
F-2 Generation: YYRR YYRr YyRR YyRr	YyRr YyRr	yyrr	yyrr
Yellow Round Yellow Round Yellow Round Yellow Round	Yellow Wrinkled Yellow Round	Green Wrinkled	Green Wrinkled
All Dominant		1:1:1:1 Test Cross	

Difference Between Genotype and Phenotype:

Term genotype and phenotype were introduced by Johanseen in 1911.

SN	Genotype	Phenotype
1	The genetic make-up or the gene content of a plant or animal is called as Genotype.	External appearance of plant or animal is called as Phenotype.
2	Genotype is the gene composition of organism which determines characters	Phenotype refers to the visible character of an organism.
3	Genotype can be ascertained from the ancestry of the individuals	Phenotype can be read out from the individual by direct observation.
4	Individual having genotype usually have the same phenotype	Individuals having identical phenotype may not have same genotype.
5	In the F-2 generation of Mendel's expt., in monohybrid cross genotypic ratio is 1:2:1.	In the F-2 generation of Mendel's expt., in monohybrid cross phenotypic ratio is 3:1.

Incomplete Dominance:

The law of dominance does not occur universally. In certain cases, the dominant and recessive alleles are capable of some degree of expression when in the heterozygous condition. The F-1 heterozygote does not resemble any of the true breeding parents and usually an intermediate in character between them. The two alleles maintain their individual identities and segregate from each other at the time of gamete formation in F-1 and both the parent types are recovered in F-2 generation.

Exa.– 4 O'clockplant:*Mirabilis jalapa* – Flower colour.

Parent (P-1):	**Red**			**X**		**White**	
Genotype :	RR					rr	
Gamete :	R					r	
F-1 Hybrid :				Rr			
				Pink			
Parent (P-2) :	**Pink**			**X**		**Pink**	
Genotype :	Rr					Rr	
Gamete :	R	r		R		r	
F-2 Generation :	RR	Rr		Rr		rr	
	Red	Pink		Pink		White	
Ratio :	1	:	2		:	1	

When a homozygous Red flowered 4 O'clock plant (RR) is crossed with homozygous White flowered 4 O'clock plant (rr) then they produced heterozygous Pink flowered plant in F-1 generation. That is intermediate between the two parents instead of Red or White as expected form Mendelism. This is because neither Red nor White allele is dominant but each allele has its influence in colour development and hybrid appears Pink.

If the F-1 Pink flower plants are self pollinated, the F-2 progeny shows Red, Pink and White flowered plants in the ratio of 1: 2: 1.

The appearance of the intermediate character in F-1 generation is known as Incomplete Dominance.

When a dominant allele does not mask completely the phenotypic expression of the recessive allele in a heterozygote, then a blending of both dominant and recessive traits takes place in the F-1 and F-2 heterozygotes. This phenomenon is known as Incomplete or Partial dominance.

Co-dominance:

Co-dominance is the equal expression of both alleles in heterozygotes. Both alleles are fully expressed and there is no compromise as found in partial dominance. One homozygote shows one trait and the other homozygote shows a different trait. The heterozygote shows both traits, fully expresses with the two co-dominant alleles acting in different ways.

In the phenomenon of co-dominance, both dominant and recessive alleles lack their dominant and recessive relationships and both have capacity to express them phenotypically, in the heterozygous condition.

Exa.: ABO blood group in Man.

Parent	**:**	**Male**			**Female**	
		Homozygous Blood Group		**X**	Homozygous Blood Group	
		A			**B**	
Genotype	:	I^AI^A			I^BI^B	
Gamete	:	I^A	I^A		I^B	I^B
Offsprings	:	I^AI^B	I^AI^B		I^AI^B	I^AI^B
Blood Group:		**AB**	**AB**		**AB**	**AB**

If a person having homozygous for A blood group and a lady have homozygous for B blood group, in the next generation their offsprings blood group will have a blood type AB, because both the A and B alleles areco-dominant with each other.

There are certain similarities and differences between co-dominance and incomplete dominance.

i) In both, F-2 phenotypic and genotypic ratios of monohybrid crosses are the same (1 : 2 : 1).

ii) In both, the heterozygote is phenotypically distinguishable from the homozygote.

iii) In incomplete dominance both alleles express themselves in the hybrid, but the effect of one is greater than that of other. This produces an Intermediate or Blending effect in heterozygotes.

iv) In co-dominance, both alleles are equally and fully expressed in the heterozygote. There is distinct evidence of gene products of both alleles. The term codominance implies that is a definite product of each allele can be identified.

v) In co-dominance, therefore, there is distinct expression of both alleles, with two distinct gene products. Since both alleles contribute to the phenotypes, it has thefeatures of both strains.

Important Questions

Q.1 Multiple choice questions (1 marks each)

1) In a Mendelian monohybrid cross, which generation is always completely heterozygous?
a) F1 generation b) F2 generation c) F3 generation d) P generation

2) If a pea plant shows a recessive phenotype,
a) it can be either TT or Tt. c) it can be either Tt or tt.
b) it can be only TT. **d) it can be only tt**.

3) The symbol "F" in the results of a testcross stands for
a) dominant. c) first trait to show up.
b) "faulty" or unexpected results. **d) filial**.

4) In which kind of cross would you expect to find a ratio of 3:1 among the F2 offspring?
a) monohybrid cross b) dihybrid cross c) testcross d) a poloygenic cross

5) If individuals exhibiting a dominant phenotype are crossed and produce only offspring with the dominant phenotype, what would be the logical genotype of the parents?
a) homozygous recessive b) heterozygous dominant
c) homozygous dominant d) heterozygous recessive

6) Which of the following refers to a cross in which traits are considered simultaneously? Is it:
a) dihybrid cross b) filial cross c) punnett cross d) none of the above

7) What are Mendel's factors called today?
a) alleles **b) genes** c) traits d) characters

8) A male and female bison that are both heterozygous for normal skin pigmentation(*Aa*) produce an albino offspring (*aa*). Which of Mendel's principles applies?
a) dominance only **c) dominance and segregation**
b) independent assortment only d) segregation only

9) A cross of a black chicken (*BB*) with a white chicken (*WW*) produces all speckled offspring (*BBWW*). This type of inheritance is known as
a) incomplete dominance **b) codominance** c) polygenic inheritance d) multiple alleles

10) A red flowered plant (RR) is crossed with white flowered plant (rr) and offspringsare pink (Rr), this shows that gene R is –
a) Mutant b) Recessive c) hybrid **d) Incomplete dominant**

11) A cross between F1 and recessive parent is
a)Reciprocal cross b) Monohybrid cross **b) Test cross** d) Dihybrid cross

12) How many types of gametes will be formed by the genotype individual AaBbCc?

a) 16 **b) 8** c) 4 d) 3

13) In the F1 generation of a monohybrid cross, the phenotypic ratio would be:

a) 3:1 b) 1:2:1 c) 2:1:1 d) 1:1:2

14) The phenonmenon of 'independent assortment' is based on

a) Monohybrid cross **b) Dihybrid cross** c) Trihybrid cross d) Back cross

Q.2 Define /explain /comment (2 marks each)

1) Phenotype 2) Genotype 3) Back-cross 4) Segregation 5) Monohybrid

6) Dihybrid 7) Co-dominance 8) Incomplete dominance.

Q.3 and Q. 4: Short Notes / Attempt (04 marks each)

1) Give an account on Mendel's law of inheritance with example.

2) Difference between genotype and phenotype.

3) Write the genotypic and phenotypic ratios of F2 generation in a dihybrid crossexperiment.

4) Explain incomplete dominance with an example

5) What is the genotypic ratio of F2 generation in monohybrid cross?

6) State and explain with suitable example Mendel's law of segregation or purity ofgametes.

7) How can the law of segregation be confirmed cytologically?

8) State the law of segregation. Explain monohybrid cross experiment in pea plant.

9) Which is more important? The principle of segregation or the principle of dominance?Why?

10)State and explain with a suitable example Mendel's law of dominance. Do we nowaccept it as a law? Why? Discuss its merits and Demerits.

11)State and explain with a suitable example Mendel's law of independent assortment.Do we now accept it as a law? Why?

12) What is Mendel's law of combination? Explain its significance.

13) Explain the significance of Mendel's laws. Discuss their merits and demerits. what arethe objections to Mendel's principle?

Q.5 Problems for (4 marks each).

1) Let the allele for tall be represented by T and the allele for dwarfness by t. whatwill be the gametes produced by the parents and the height of the offspring (tallor dwarf) from each of the following crosses: a) Tt x tt b) TT x Tt c) Tt x Tt

2) In a certain species of animals black fur (B) is dominant over brown fur (b).Predict a genotype and phenotype of the offspring's

3) When both parents are Bb or have heterozygous black fur.

4) State the law of independent assortment. Describe the di-hybrid cross experimentwhen homozygous dominant round and yellow seeded plant is crossed withhomozygous recessive wrinkled and green seeded plant.
5) Write the genotypic and phenotypic ratios of F2 generation in a dihybrid crossexperiment with suitable example.
6) A black mouse mates with a brown mouse, and all the offspring's are black. whyare no brown offspring's produced? Explain your answer fully.
7) A farmer believes that some of his rose combed Wyandotte fowl may carry afactor for single comb. Can you suggest a method for finding out which fowls areheterozygous? In poultry, rose comb is dominant over single comb.
8) When a wild type female *Drosoplila, VVSS*, with normal long wings and normalred eyes is mated to a mutant type male fly *vvss*, with (short) vestigial wings and(brown) sepia-eye colour, the F1 females of this cross to an F1 male and counted223 wild-type flies with normal long wings and red eyes, 70 flies with normal redeyes and vestigial wing, 71 flies with normal long wings and sepia eyes, and 24flies with vestigial wings and sepia eyes. Explain the genetics of this cross withemphasis on the free assortment of genes.

Unit - 3

Gene Interaction

Definition:

From Mendelian monohybrid and dihybrid crosses, it can be noted that, each character (trait) is controlled by a pair of factors or genes. But later on Bateson and Punnet proved that in many cases the expression of a single character is controlled by the interaction of more than one pair of genes. This is called as Interaction of Genes or Factor.

Bateson and Punnet states that, the several pair of genes located in different pairs of chromosomes are interacts to each other and affect the production of a single character.

Exa. – 1) In fruit fly *Drosophila melanogaster*, eye pigment is controlled by more than 20 different genes. 2) Coat colour of Mammals is the result of combine action of several genes.

Concept of Gene:

Mendel assumed that the appearance of characters were due to something. He called Factor. In 1902, Sutton and Boveri showed the relationship between genes and chromosomes and concluded that chromosomes are the carrier of hereditary particles and that is Mendelian factor (element), are located on chromosomes. Later on 1909, Johansson applied the term 'gene' to these hereditary factors. The another concept of gene are as follows –

1) Genes are situated in chromosomes.
2) There are several genes in each chromosome.
3) Each gene occupies a fixed position in the chromosome. The position of a gene in a chromosome is known as its Locus.
4) A single gene may occur in several different forms or states called as Alleles.
5) The two alleles of a gene may related to each other as Dominant and Recessive.
6) Some genes have more than two alleles. This is known as Multiple alleles.
7) A gene may show sudden change from one form or state to another. Such a change in state is called as Mutation and new allele formed is known as Mutant. A mutant gene causing death is called as Lethal gene.
8) Genes on one chromosome may be transferred to another. This is due to crossing over in meiosis or due to translocations.
9) Duplication of each chromosome during mitosis is preceded by self duplication of genes of that chromosome. Self duplication of genes is called as Replication.
10) Inheritance involves transmission of genes from parents or offsprings.
11) Genes express themselves through the production of chemical substance (Protein enzyme) which control cell metabolism.

Modifications in Mendelian Phenotypic Ratio:

The gene interaction may occur in between genes located in the same chromosome or different chromosomes. This type of gene interaction is known as Non-allelic gene interaction. The gene interaction may also occur in between the two alleles of a single type of gene. Such type of gene interaction is known as Allelic gene interaction and often leads to modification of the basic Mendelian ratios.

Some of the important forms of modifications due to gene interactions are as follows,

Epistasis:

Epistasis is the interaction between the genes at two or more loci. When two gene pairs which are non-allelic (i.e. located at different loci) may be concerned with development of same character and one of these gene pairs inhibits or suppresses the expression of the other gene. The gene which suppresses the expression of the other non-allelic gene is called as Epistatic over the other and the one whose expression is suppressed by the presence of the other gene is called as Hypostatic. This phenomenon of inhibition of expression of one by the other non-allelic gene is called as Epistasis. Epistasis is of two types – Recessiveand Dominant epistasis.

1) Recessive Epistasis (9 : 3 : 4 Ratio):

When one allelic pair in homozygous recessive condition suppresses or is epistatic over the expression of the other allele is called as Recessive Epistasis. OR The recessive alleles of one gene locus masked the action (phenotypic expression) of alleles of another gene locus. This type of epistasis is called Recessive Epistasis.

This is the condition where the epistatic gene is recessive to its own allele. Thus in this case the epistatic gene can have its inhibition influence or effects only when it is in homozygous condition. In mice, grey (agouti) coat colour is dominant over black. Thus grey mice are either homozygous (BB) or heterozygous (Bb) for the dominant gene, while black mice are always homozygous (bb) for the recessive gene. But a recessive Epistatic gene ‘a’ prevents any type of pigment formation while its dominant allele ‘A’ permits pigment formation. As a result all mice homozygous for gene ‘a’ are albinos. This can be seen in a typical Dihybrid cross.

When a cross is made between a Grey mouse homozygous for both the dominant genes B and A (BBAA) and an Albino mouse homozygous for both recessive genes (bbaa). All F-1 hybrids are Grey (BbAa). On mating two F-1 hybrids, in F-2 generation they produce 9 Grey (Agouti) – due to at least one ‘B’ and one ‘A’, 3 Black – due to homozygous recessive ‘b’ and 4 Albino mouse – due to homozygous for the gene ‘a’ in the ratio of 9 : 3 : 4. This is modified ratio of Mendelian Dihybrid ratio.

Exa. – Coat colour in Mice:

Parents (P-1) : **Male** **X** **Female**

Agouti Albino

Genotype : BBAA bbaa

Gametes : BA ba

F-1 Generation : BbAa

(Hybrid) Agouti (Grey)

Parents (P-2) : **Agouti** **X** **Agouti**

Genotype : BbAa BbAa

Gametes : BA Ba bA ba BA Ba bA ba

F-2 Generation :

O/O	BA	Ba	bA	ba
BA	BBAA Agouti	BBAa Agouti	BbAA Agouti	BbAa Agouti
Ba	BBAa Agouti	BBaa Albino	BbAa Agouti	Bbaa Albino
bA	BbAA Agouti	BbAa Agouti	bbAA Black	bbAa Black
ba	BbAa Agouti	Bbaa Albino	bbAa Black	bbaa Albino

Ratio : Agouti : Black : Albino

9 : 3 : 4

2) Dominant Epistasis (13 : 3 Ratio):

When out of two genes, the dominant allele of one gene masked the activity of alleles of another gene and expressed itself phenotypically is called as Dominant epistasis. It is effective even in heterozygous condition.

In White leghorn chickens a dominant gene 'C' is responsible for producing the dominant colour but dominant Epistatic gene 'I' prevents expression of a dominant colour.

In a typical Dihybrid cross between a White Leghorn (CCII) and a White Plymoth Rock (ccii), all F-1 hybrids are white (CcIi). But in the F-2 generation, the typical Dihybrid ratio is modified to 13 : 3 with 13 White and 3 Coloured chickens.

The coloured chickens are those which are homozygous for recessive allele 'i' of the Epistatic gene 'I' and have atleast one dominant gene 'C' for pigmentation.

Exa. – Feather colour in Fowl

Parents (P-1) : **Male** **X** **Female**

White Leghorn Plymoth Rock (White plumage)

Genotype : CCII ccii

Gametes : CI ci

F-1 Generation : CcIi

(Hybrid) White Plumage

Parents (P-2) : **White Plumage** **X** **White Plumage**

Genotype : CcIi CcIi

Gametes : CI Ci cI ci CI Ci cI ci

F-2 Generation :

O/O	CI	Ci	cI	ci
CI	CCII White	CCIi White	CcII White	CcIi White
Ci	CCIi White	CCii Coloured	CcIi White	Ccii Coloured
cI	CcII White	CcIi White	ccII White	ccIi White
ci	CcIi White	Ccii Coloured	ccIi White	ccii White

Ratio : White : Coloured

13 : 3

Supplementary Factor (9 : 3 : 4 Ratio):

Supplementary genes are defined as two independent pairs of genes interacting in such a manner that, one dominant gene produce its effect whether the other is present or not. While the second one produces its effect only in the presence of the first.

Exa. – Guinea pig : Coat - colour.

Parents (P-1) : **Male** **X** **Female**

Black Albino

Genotype : CCaa ccAA

Gametes : Ca cA

F-1 Generation : CcAa

(Hybrid) Agouti (Grey)

Parents (P-2) : **Agouti** **X** **Agouti**

Genotype : CcAa CcAa

Gametes : CA Ca cA ca CA Ca cA ca

F-2 Generation :

O/O	CA	Ca	cA	ca
CA	CCAA Agouti	CCAa Agouti	CcAA Agouti	CcAa Agouti
Ca	CCAa Agouti	CCaa Black	CcAa Agouti	Ccaa Black
cA	CcAA Agouti	CcAa Agouti	ccAA Albino	ccAa Albino
ca	CcAa Agouti	Ccaa Black	ccAa Albino	ccaa Albino

Ratio: Agouti : Black : Albino

9 : 3 : 4

In 1909, Castle studies supplementary gene or factor in Coat colour of Mice and Guinea pig.In Guinea pig, Black colour of the coat is due to a gene 'C', is dominant over Albino (c). Besides this, there is a Wild type variety called Agouti, in which the coat colour is more or less Grayish.

Agouti coat colour is due to the presence of Dominant gene 'A'. This gene when present either in single or double dose, Black fur turns into Agouti (Gray).

Therefore, Black coat colour is always homozygous for the recessive gene 'a' in addition to possessing at least one Dominant gene 'C'. In the absence of 'C' the Dominant gene 'A' or its recessive 'a' has no effect. Therefore, the Albino may or may not possess the gene 'A'.

The phenotypes of three kinds of Guinea pig are as follows –

Pure Black – CCaa, Ccaa, Pure Albino – ccAA and Agouti (Grey) – CcAa.

Agouti (Grey) : Hairs are black at base and tip with yellow band in between. As a result natural grey colour is produced.

The Castle crossed a Homozygous Black Guinea pig (CCaa) with a Homozygous Albino (ccAA). F-1 offsprings are Agouti (CcAa) type. The Agouti factor came from the Albino parent in which the Agouti factor produces no effect.

When F-1 hybrid (Agouti) are inbreed, in F-2 generation they produces 9 Agouti - due to at least one 'A' and one 'C' factor, 3 Black – because there is at least one 'C' but no 'A' factor and 4 Albino – due to recessive 'c' factor but other factor 'A' may be or may not.

Thus 9 : 3 : 4 ratio obtained for coat colour in Guinea pig due to the interaction of genes, which is modified ratio of Mendelian Dihybrid ratio.

Complementary Factor (9 : 7 Ratio):

Exa. – Sweat Pea Plant :*Lathyrus odoratus* – Flower colour.

Parents (P-1) : White Flower X White Flower

Genotype : CCrr ccRR

Gametes : Cr cR

F-1 Generation : CcRr

(Hybrid) Red Flower

Parents (P-2) : Red Flower X Red Flower

Genotype : CcRr CcRr

Gametes : CR Cr cR cr CR Cr cR cr

F-2 Generation :

O/O	CR	Cr	cR	cr
CR	CCRR Red	CCRr Red	CcRR Red	CcRr Red
Cr	CCRr Red	CCrr White	CcRr Red	Ccrr White
cR	CcRR Red	CcRr Red	ccRR White	ccRr White
cr	CcRr Red	Ccrr White	ccRr White	ccrr White

Ratio: Red : 9

White : 7

Bateson and Punnet demonstrated that, sometimes two or more dominant genes occurring in different loci of the same chromosome or separate chromosome interact with one another to produce a trait (character), one complementing the other. But neither of them produces that character in the absence of other. Such genes are called as Complementary genes. Such case one pair acts as the actual Determiner and the other as Supporter or Initiator.

Bateson and Punnet (1905) crossed two White flower varieties of Sweat Pea, *Lathyrus odoratus*. All the plants of F-1 generation produces Red flowered plants.

When the F-1 plants are self pollinated, the F-2 generation consist of Red and White flowered plants in the ratio of 9 : 7. This is modification of the normal Dihybrid ratio.

The Red colour is due to the presence of a pigment called Anthocyanin. The anthocyanin is produced from a colourless substance called Chromogen by the action of an enzyme or activator. The chromogen cannot convert into anthocyanin in the absence of an enzyme. Thus for the production of Red colour, both chromogen and enzyme should be present in the plant. In the absence of any one, Red colour cannot be produced.

Here, in Sweat pea, the gene 'C' is responsible to produce a colour base (chromogen). While the gene 'R' produces an enzyme which activates the colour base to produce the colour.

One of the white flower plants has the genotype 'CCrr' and the other 'ccRR'. The offsprings of the cross 'CcRr' will develop the Red colour because the Dominant 'C' and 'R' are present together. In the crossing of F-1 hybrids, in F-2 generation 9–Red colour flower plants and 7–White colour flower plants appear.

When any one 'C' or 'R' gene come across homozygous recessive i.e. cc or rr, there is no formation of Red colour. Hence, the flowers are White coloured.

Inhibitory factor (13:3 Ratio):

An Inhibitory factor is one which by itself has no phenotypic effect but when present in the dominant form prevents or inhibits the expression of another independent dominant gene present in another chromosome.

Exa. – Feather colour in Fowl:

Parents (P-1) : **Male** **X** **Female**

White Leghorn White Wyandotte

Genotype : CCII ccii

Gametes : CI ci

F-1 Generation : CcIi

(Hybrid) White

Parents (P-2) : **White** **X** **White**

Genotype : CcIi CcIi

Gametes : CI Ci cI ci CI Ci cI ci

F-2 Generation:

O/O	CI	Ci	cI	ci
CI	CCII White	CCIi White	CcII White	CcIi White
Ci	CCIi White	CCii Coloured	CcIi White	Ccii Coloured
cI	CcII White	CcIi White	ccII White	ccIi White
ci	CcIi White	Ccii Coloured	ccIi White	ccii White

Ratio: White : Coloured

13 : 3

In White leghorn chickens a dominant gene 'C' is responsible for producing the dominant colour but dominant Inhibitor gene 'I' prevents expression of a colour.

In a typical Dihybrid cross between a White Leghorn (CCII) and a White Wyandotte (ccii), all F-1 hybrids are white (CcIi) in colour. But when the White birds of F-1 generation are crossed among themselves, in the F-2 generation, 13 White and 3 Coloured chickens produced in the ratio of 13:3. This is modified Dihybrid ratio.

The White Leghorns possess a colour gene 'C' which is prevented from being expressed by an Inhibiting gene 'I'. Thus they are unable to develop their colour. The Wyandotte birds were white because of the absence of the colour gene as well as inhibitory gene.

The coloured chickens are those which are homozygous for recessive allele 'i' of the Inhibitor gene (I) and have atleast one dominant gene 'C' for pigmentation.

Penetrance:

The percentage of individuals expressing the character for a particular genotype is called penetrance.

If all individuals express the character for particular genotype, the penetrance is called Complete penetrance. In complete penetrance, the character is expressed in 100% individuals. Exa. – Mendel's Tall and Dwarf plants.

In Mendel's pea plants, all the plants containing the genotype TT produce Tall character. Similarly all the plants containing Tt produce Tall character and tt produce Dwarf character. Thus, in pea plants there is 100% penetrance.

If a few individuals do not express the character even though they contain the necessary genes, the penetrance is called **Incomplete penetrance**. Exa. – Blue eyes.

The gene for blue eye BB produce blue eyes only in 90% human beings. About 10% people have white eyes even though they contain the genes BB for blue eyes. So eye colour in man has only 90% penetrance.

Penetrance is influenced by environmental factors such as food, light, temperature etc.

Expressivity :

The variation in the degree of expression of a particular gene is called as Expressivity.

A particular gen may produce varying degree of expression in different individuals. Exa. - In man the polydactylous condition may be penetrant in the left hand (6 fingers) and not in the right (5 fingers) or it may be penetrant in the feet and not in the hands.

Expressivity is due to influence of environmental factors on the genes.

Pleiotropism:

We have observed that a specific gene has a specific effect upon a specific phenotypic character or trait i.e. one gene (allele) control one phenotypic character. But, this is not found everywhere. A single gene may influence more than one phenotypic character.

Exa - In Drosophila, the recessive gene for vestigial wings causes vestigial wings in homozygous condition. However, it affects or influences the expression of balancer behind the wings (halter),

certain bristles, the structure of reproductive organs, egg production is lowered and longevity is reduced.

The production of many characters by a single pair of genes is called as Pleiotropism.

Important Questions

Q.1 Multiple choice questions (1 marks each)

1) When a single gene influences more than one trait, it is called

a)Epistasis b) Pseudodominance **c) Pleiotropy** d) None of these

2) Complementary interaction of gene gives the ratio

a)13:3 b) 15:1 **c) 9:7** d) 1:1

3) The penetrance is

a)insertion of gene b) elimination of gene

c) ability of a gene to express d) incomplete expression of gene

4) Epistasis is …… type of interaction

a) Inter allelic b) Intra allelic c) Allelic genetic d) Both a and c

5) Variation in human skin color is an example of

a) Incomplete dominance. **b) Polygenic traits**. c) Codominance. d) Multiple alleles.

6) Epistasis is

a) Masking effect of one allele over other **b) Masking effect of one gene over other**

c) Masking effect of one plant over other d) Masking of protein

7) Colour blindness in man is

a) Due to deficiency of vitamin A b) Due to absence of visual pupil in retina

c) Due to absence of rods in retina d) A sex-linked abnormality

8) Sickle cell Anemia is caused by

a) Pleiotropic gene **b) Penetrant gene** c) Multiple alleles d) Lethal gene

9) Polydactyly refres to

a)Five fingers b) Less than five fingers

c) Occurance of extra fingers d) Six fingers

10) If a woman heterozygous for colour blindness marries a colour blind man, what is theprobability that their first child will be colour blind daughter?

a) 50% **b) 25 %** c) 75% d) 100%

11) Hemophilia is a sex-linked recessive trait in humans. If a father and a son are bothhemophiliacs, but the mother is normal, her genotype must be:

a) $X^{h}X^{h}$ **b) $X^{H}X^{h}$** c) $X^{H}X^{H}$ d) $X^{h}Y$

12) When a color blind man marries a woman pure for normal color vision, it is probablethat one of the following situations may result. Is it probable that

a) all the children will be color blind
b) all the grandchildren will be color blind
c) only the sons will be colorblind
d) half the grandsons will be color blind

13) Carriers of the colour-blindness trait include:

a) Men who are heterozygous for the trait.
b) Men who are homozygous for the trait.
c) **Women who are heterozygous for the trait**.
d) Women who are homozygous for the trait.

Q.2 Define /explain /comment (2 marks each)

1) Gene interaction
2) Epistasis gene
3) Hypostatic gene
4) Duplicate gene
5) Expressivity
6) Penetrance

Q. 3Question for (4mark each)

1) Gene interaction
2) Concept of gene
3) Epitasis
4) Dominant epitasis
5) Recessive epitasis
6) Supplementary factors
7) Complementary factors
8) Inhibitory factors
9) Pleiotropic gene

Q.4 Question for (6 mark each)/ Describe in detail.

1) With a suitable example explain the concept of incomplete dominance (or nodominance).
2) Distinguish between epitasis and dominance. What does gene interaction mean?
3) Explain Polygenic Inheritance with suitable examples.

Q. 5 Distinguish between (4 marks)

1) Epistasis and dominance

Q.6 Problems for (4 marks each).

1) Two unlinked loci effect mouse hair color. AA or Aa mice are agouti (brown). Micewith genotype aa are albino because all pigment production is blocked, regardless ofthe phenotype at the second locus. At the second locus, the B allele (agouti coat) isdominant to the b allele (black coat). What would be the result of a cross between twoagouti mice of the genotype AaBb? (Ans- 9 Agouti: 3 Black: 4Albino)
2) Coat colors of dogs depend upon the action of at least two genes. At one locus adominant epistatic inhibitor of coat color pigment (*I*-) prevents the expression of coloralleles at another independently assorting locus, producing white coat color. When therecessive condition exists at the inhibitor locus (*ii)*, the alleles of the hypostatic locusmay be expressed, *iiB*- producing black and *iibb* producing brown. When dihybridwhite dogs are mated together, determine the phenotypic proportions expected in theprogeny?
3) Matings between black rats of identical genotype produced offspring as follows: 14cream-colored, 47 black, and 19 albino. (*a*)What epistatic ratio is approximated bythese offspring? (*b*)What are the genotypes of the parents and the offspring (use yourown symbols)?

Unit - 4

Chromosomes

Introduction to Morphology and Composition:

The nucleus of a cell contains a darkly staining material called Chromatin. In an interphase cell the chromatin material is organised into a number of long, loosely coiled, irregular strands or threads which together convey the impression of a network, called as Chromatin reticulum. When the cell begins to divide, the chromatin bodies condense to form shorter and thicker threads, which are called as Chromosomes.

In 1988 the term chromosome was first coined by W. Waldever for darkly stained bodies due to affinity for basic dyes.

Chromosomes are the carriers of hereditary characters, which are passed from one generation to the next. The genetic information is stored in the chromosomes. In prokaryotes there is a single chromosome, which is circular molecule of genetic materials is either DNA or RNA. But in eukaryotic cells, this genetic material is in the form of chromatins or chromosomes, present in nucleus of the cell,

The chromosomes and chromatin are interchangeable form of genetic material at different stages of the cell cycle. In non-dividing eukaryotic cells it is chromatin, which is amorphous and randomly dispersed within the nucleus. As and when cells prepare for mitosis or meiosis, the chromatin becomes highly condensed and organised in to the species-specific number of chromosomes.

Morphology of Chromosomes:

The chromosomes exhibit considerable size, shape and number. It can be best to studied at the metaphase or anaphase or cell division when they are present as definite organelles, being most condensed or coil.

Number:

The chromosome number for any given species of plant of animal is constant. Therefore, these are importance in the determination of phylogeny and taxonomy of the species. The somatic cell of the organism contains two set of chromosomes, forming homologous pairs and called as Diploid (2n), while their gametes have only one set of chromosome and are haploid (n). This haploid set of chromosome is known as Genome.

The species-specific chromosome number varies greatly in the plant and animal kingdoms. The number of chromosomes is variable from one to several hundred among different species. The horse roundworm, *Parascaris equorum*and *Ascaris megalocephalus univalens*has only two

chromosomes in its body cells, while some secretory cells in some insects possess as many as 20,000 or even upto 1,00,000 chromosomes.

Common Name	Species	Chromosomes	Common Name	Species	Chromosomes
Animals					
Man	*Homo sapiens*	23 pairs	Pigeon	*Columbia livia*	40 pairs
Chimpanzee	*Pan troglodytes*	24 pairs	Chicken	*Gallus domesticus*	39 pairs
Rhesus monkey	*Macaca mulatta*	21pairs	Frog	*Rana pipens*	13 pairs
Cattle	*Bos taurus*	30 pairs	Toad	*Bufo americanus*	11 pairs
Dog	*Canis familiaris*	39 pairs	Fish	*Cyprinus carpio*	52 pairs
Cat	*Felis domesticus*	19 pairs	Roman snail	*Helix pomatia*	27 pairs
Horse	*Equus calibus*	32 pairs	Starfish	*Asterias forbesi*	18 pairs
Donkey	*Equus asinus*	31 pairs	House fly	*Musca domestica*	6 pairs
Rat	*Rattus norvegicus*	21 pairs	Fruit fly	*Drosophila melanogaster*	4 pairs
Mouse	*Mus musculus*	20 pairs	Mosquito	*Culex pipiens*	3 pairs
Rabbit	*Oryctolagus cuniculus*	22 pairs	Honey bee	*Apis mellifera*	16 pairs
Guinea pig	*Cavia cobaya*	32 pairs	Spanish butterfly	*Lysandra nivescens*	190 pairs
Plants					
Field bean	*Vivia faba*	3 pairs	Banana	*Musa paradisiaca*	11 pairs
Garden pea	*Pisum sativum*	7 pairs	Yellow pine	*Pinus ponderosa*	12 pairs
Onion	*Allium cepa*	8 pairs	Sunflower	*Halianthus annus*	17 pairs
Radish	*Raphanus sativus*	9 pairs	Coffee	*Coffea Arabica*	22 pairs
Orange	*Citrus sinensis*	9 pairs	Upland cotton	*Gossypium hirsutum*	26 pairs
Corn	*Zea mays*	10 pairs	Cane sugar	*Saccarum officinarum*	40 pairs

Table : Diploid chromosome number in some Plants and Animals.

Size:

The size of chromosome is measured at mitotic metaphase and may be short as 0.25 um in fungi and birds or as long as 30 um in some plants such as Trillium. However, most metaphase

chromosomes fall within a range of 5 um in man, 8 um to 12 um in maize and 3.5 um in drosophila.

The organisms with less number of chromosomes contain comparatively large sized chromosomes than the chromosomes of the organisms having many chromosomes. Plant cell possesses larger chromosome than the animal cells. The grasshopper, crickets, mantids, newts and salamanders have larger chromosomes. Variation in size of the chromosomes can be induced by number of environmental factors.

The chromosomes in a cell are never alike in size, some may be large and others may be too small. The largest chromosomes are Lampbrush chromosome of certain vertebrate oocytes and Polytene chromosome of certain dipteran insects.

Shape:

The shape of the chromosome is changeable from phase to phase in the continuous process of the cell growth and cell division. In the resting phase or interphase stage of the cell, the chromosomes occur in the form of thin, coiled, elastic and contractile, thread like stainable structure and the chromatin threads. In the metaphase and anaphase, the chromosomes become thick and filamentous.

Each chromosome contains a clear zone, known as Centromere or Kinetochore along their length. The centromere divides the chromosome into two parts. Each part is called as Arm. The position of centromere varies from chromosome to chromosome and it provides different shapes.

Classification of Chromosomes based on the Centromeric position :

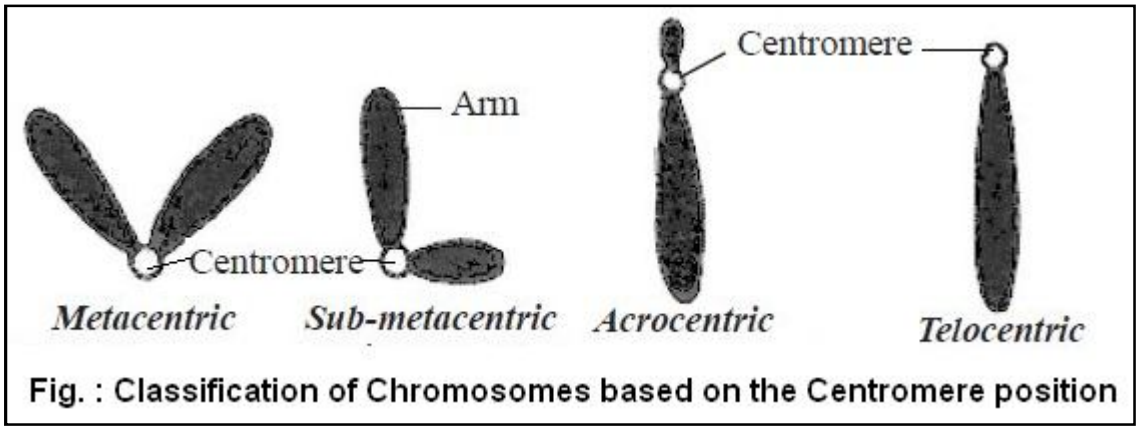


Fig. : Classification of Chromosomes based on the Centromere position

Based upon the position of centromere, chromosomes are classified into four types, Metacentric, Submetacentric, Acrocentric and Telocentric.

1. Metacentric:

The Centromere occupies a middle position with reference to the length of the chromosome. The two arms thus resulted are equal in length. They appear 'V' shaped during anaphasic movement in cell division. Exa – Trillium, Tradescantia, Amphibia etc.

2. Sub metacentric:

When the centromere is located some distance away from the middle region of the chromosome, the position is said to be sub-median and the chromosome will be shorter than the other. They appear 'L' shaped during anaphasic movement in cell division.Exa - Human beings.

3. Acrocentric:

The centromere is located a little away from the end of the chromosome. As a result, one arm of the chromosome will be extremely short and the other very long. The centromere is sub-terminal in position. They appear 'J' shaped during anaphasic movement in cell division. Exa - Grass hoppers.

4. Telocentric:

When the centromere is located at the tip of the chromosome is called as Telocentric chromosome. The centromere is terminal in position. They will have only one long arm. They appear 'I' shaped during anaphasic movement in cell division. Telocentric chromosomes are very rare. Truly telocentric chromosomes have been identified by Marks (1957) in certain plants, protozoa and certain mammals.

Structure of Chromosome:

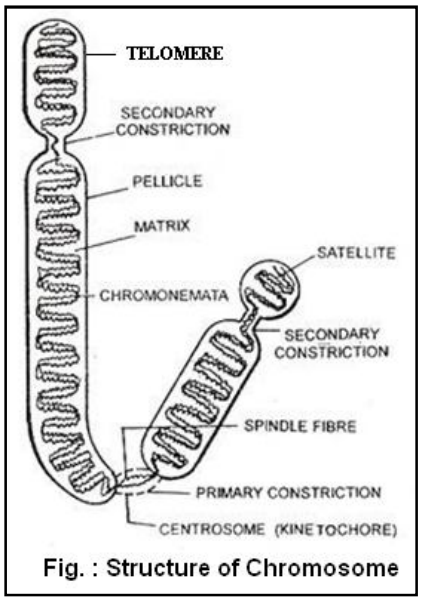


Fig. : Structure of Chromosome

a) Pellicle: It is the outer envelope around the substance of chromosome. It is very thin and is formed of achromatic substances.

b) Matrix: It is the ground substance of chromosome which contains the chromonemata. It is also formed of nongenic materials.

c) Chromonemata: In the matrix of each chromosome two identical, spirally coiled threads are embedded called as chromonemata. The two chromonemata are also tightly coiled together that they appear as single thread of about 800A thickness. Each chromonemata consists of about 8 microfibrils and which is formed of a double helix of DNA.

d) Chromomeres: At regular intervals on the chromonemata small dense masses are observed called as Chromomeres.These are more distinct in the prophase stage when chromonemata are less coiled and most clearly visible during leptotene and zygotene stages of meiotic prophase.

Chromomeres are regions of tightly folded DNA and are believed to correspond to the units of genetic function in the chromosomes.

e) Chromatid: At mitotic metaphase each chromosome consists of two symmetrical structures called chromatids. Each chromatid contains a single DNA molecule. Both chromatids are attached to each other only by the centromere and become separated at the beginning of anaphase, when the sister chromatids of a chromosome migrate to the opposite poles.

f) Centromere: It is a small structure in the chromonema and is marked by a constriction. At this point the two chromonemata are joined together. This is known as centromere or kinetochore or primary constriction. Centromere divides the chromosome into two sections or arms. The short arm of the chromosome is labeled the "p" arm. The long arm of the chromosome is labeled the "q" arm. Its position is constant for a given type of chromosome and forms a feature of identification.

Depending upon the number of centromeres, chromosomes are classified into Monocentric, Dicentric, Polycentric, Acentric and Diffused chromosomes.

Based upon the location of centromere the chromosomes are classified into Metacentric, Submetacentric, Acrocentric and Telocentric.

g) Secondary Constriction or Nucleolar Organiser: The chromosome besides having the primary constriction or the centromere possesses secondary constriction at any point of the chromosome. Constant in their position and extent, these constrictions are useful in identifying particular chromosomes in a set.

Secondary constrictions can be distinguished from primary constriction or centromere, because chromosome bends only at the position of centromere during anaphase. The chromosome region distal to the secondary constriction i.e. the region between the secondary constriction and the nearest telomere is known as satellite.

Therefore, chromosomes having secondary constrictions are called satellite chromosomes or sat-chromosomes. The number of sat-chromosomes in the genome varies from one species to the other.

Nucleolus is always associated with the secondary constriction of sat-chromosomes. Therefore, secondary constrictions are also called nucleolus organiser region (NOR) and sat-chromosomes are often referred to as nucleolus organiser chromosomes. NOR of each sat-chromosome contains several hundred copies of the gene coding for ribosomal RNA (rRNA).

h) Telomeres:

These are specialized ends of a chromosome which exhibits physiological differentiation and polarity. Each extremity of the chromosome due to its polarity prevents other chromosomal segments to be fused with it. The chromosomal ends are known as the telomeres. If a chromosome breaks, the broken ends can fuse with each other due to lack of telomeres.

Types of Chromosome:

There are two types of chromosomes autosomes and allosomes or sex chromosomes.

Autosomes or Somatic Chromosomes: The chromosomes which have no relation with sex and contain a genes which determines the somatic character of the individual are called as Autosomes or Somatic characters.

Humans have 23 pairs of chromosomes. There are 22 pairs of autosomes in humans. Each autosome contains a large number of genes arranges in a definite sequence. In these homologous pairs, the two chromosomes are of the same length. The position of the centromere is the same. Mitosis is the process by which all these chromosomes duplicate and give one copy of each chromosome to each of the daughter cells.

Allosomes or Sex chromosomes: The chromosomes which are responsible for the determination of sex of the individual are called as Allosomes or Sex chromosomes.

Humans have 23 pairs of chromosomes. There are 22 pairs of autosomes and 01 pair of heteromorphic or dissimilarsex chromosomes i.e one X and one Y chromosome. The human female has 44 autosomes and one pair of homomorphic or similar sex chromosomes designated as XX. While human male has 44 autosomes and one pair heteromorphic or dissimilar sex chromosomes designated as XY

Material of the Chromosomes:

The material of the chromosomes is the chromatin. Depending on their staining properties with basic dyes particularly the Feulgen reagent, the chromatin material is divided into two types,

Euchromatin: Portions of chromosomes that stain lightly or only partially condensed, this chromatin is termed Euchromatin. Euchromatin contains structural genes which replicate and transcribe during G_1 and S phase of interphase. It is considered genetically active chromatin, since it has a role in the phenotype expression of the genes. In euchromatin, DNA is found packed in 3 to 8 nm fibre.

Heterochromatin: Portions of chromosomes that stain dark and remains in the condensed state, is called Heterochromatin. In 1928, Heitz defined it as those regions of the chromosome that remain condensed during interphase and early prophase and form the chromocentre is called as Heterochromatin.

Heterochromatin is characterized by its high content of repetitive DNA sequences and very few structural genes. It is late replicating and is not transcribed. It is thought that in heterochromatin the DNA is tightly packed in the 30 nm fibre. It is established now that genes in heterochromatic region are inactive.

Chromosomal Banding Pattern:

Chromosomes display a banded pattern when treated with some stains. Bands are alternating light and dark stripes that appear along the lengths of chromosomes.

Unique banding patterns are used to identify chromosomes and to diagnose chromosomal aberrations, including chromosome breakage, loss, duplication, translocation or inverted segments.

In 1969, T.C. Hsu and others introduced new methods for staining chromosomes by which distinct patterns of stained bands and lightly stained inter-bands became evident.

In recent years a number of chromosome banding techniques have been developed that employ molecular cytogenetic techniques, for example fluorescence in situ hybridization (FISH).

A range of different chromosome treatments produce a different banding patterns like G-bands, Q-bands, C-bands, R-bands, NOR-bands and T-bands.

Chromosome Banding:

1. G-Banding:

The most useful chromosome banding method is G-banding. This technique was developed by Hsu and Arrighi. It is observed that when the chromosomes are incubated in saliva are stained with Giemsa stain or treated with urea or detergents. G-bands appear in the areas which are S-rich proteins. Giemsa stained preparations are more permanent and require ordinary microscope optics and illumination.

2. Q-Banding:

This technique was developed by Casperson. It is observed, when the chromosomes are stained with quinacrine mustard and observed through fluorescencemicroscope, the regions of chromosomes rich in adenine and thymine get stained intensely.

The guanine-cytosine regions remain unstained. These regions are called Q-bands. The defect of this staining is that, the stains fade after a short-time, moreover, special microscopic optics plus ultraviolet illumination are needed to see these bands.

3. C. Banding:

This technique was developed by Pardue and Gall. The chromosomes are treated with strong sodium hydroxide followed by warm saline and then stained with Giemsa stain. C-bands are especially evident around the centromere and in other chromosomes that contain substential amounts of highly repetitive constitutive heterochromatin.

4. R. Banding:

These bands appear when chromosomes are incubated in a buffer at high temperature and stained with Giemsa stain. R-bands correspond to the regions on charomosomes having proteins lacking sulphur. These are reciprocal of G bands.

5. NOR-banding:

NOR-banding involves silver staining (silver nitrate solution) of the "nucleolar organizing region", which contains rRNA genes.

6. T-Banding:

T-banding involves the staining of telomeric regions of chromosomes using either Giemsa or acridine orange after controlled thermal denaturation. T bands apparently represent a subset of the R bands because they are smaller that the corresponding R bands and are more strictly telomeric.

7. DAPI/Distamycin A Staining:

DAPI/distamycin A fluorescent staining technique is a method for labelling a specific subset of C bands. DAPI/Distamycin A staining is useful in identifying peri-centromeric breakpoints in chromosomal rearrangements and in identifying chromosomes that are too small for standard banding techniques.

Human Karyotypes:

A complete set of the entire metaphase chromosome in a somatic cell is called as its karyotype.

In 1960, human metaphase chromosomes (23 pairs) were first classified in a conference of cytogeneticists at Denver, Colorado, so known as Denver classification. Subsequent classifications are mainly based upon this classification.

Arranging chromosomes of a species according to their shape, size and structure is called as Karyotype. It helps to identify a particular chromosome.

A representation of the chromosomes of a species in the form of a diagram is called Idiogram. It is the enlargement of photographs of stained chromosomes. In idiograms, the chromosomes are arranged in a series of decreasing size.

In 1961, Mooehead et. al. developed the new technique for making chromosome spreads from cultured peripheral blood. It allowed identification of individual chromosome. It was helpful to recognize the chromosomal abnormalities.

To analyze human karyotype a sample of peripheral blood is collected from an individual into a syringe containing heparin (anticoagulant). Then leucocytes are separated by centrifugation and cultured for about three days in culturing medium, supplemented by phytohaemoglutinin (PHA) as it stimulates growth and cell division. The dividing lymphocytes are arrested at metaphase stage at $37^{o}C$, by exposing them to cholchicine for one hour.

The cells are then harvested and slides prepared. The metaphase spreads are photographed chromosome cut-out re arranged according to size and location of centromeres. The study of complete chromosome complement in this manner is called as Karyotype analysis.

The normal human karyotype contains 22 pairs of autosomal chromosomes and one pair of sex chromosomes. In female both sex chromosomes are morphologically similar and denoted as XX. But in male sex chromosomes are dissimilar in shape and hence denoted as X and Y.

Patan divided the human chromosomes into seven groups and designated A to G.

Group – A : Includes chromosome pairs 1, 2 and 3. These are largest in size and all are metacentric.

Group – B : This group involve chromosome pair 4 and 5. These are smaller than group A chromosomes and are submetacentric.

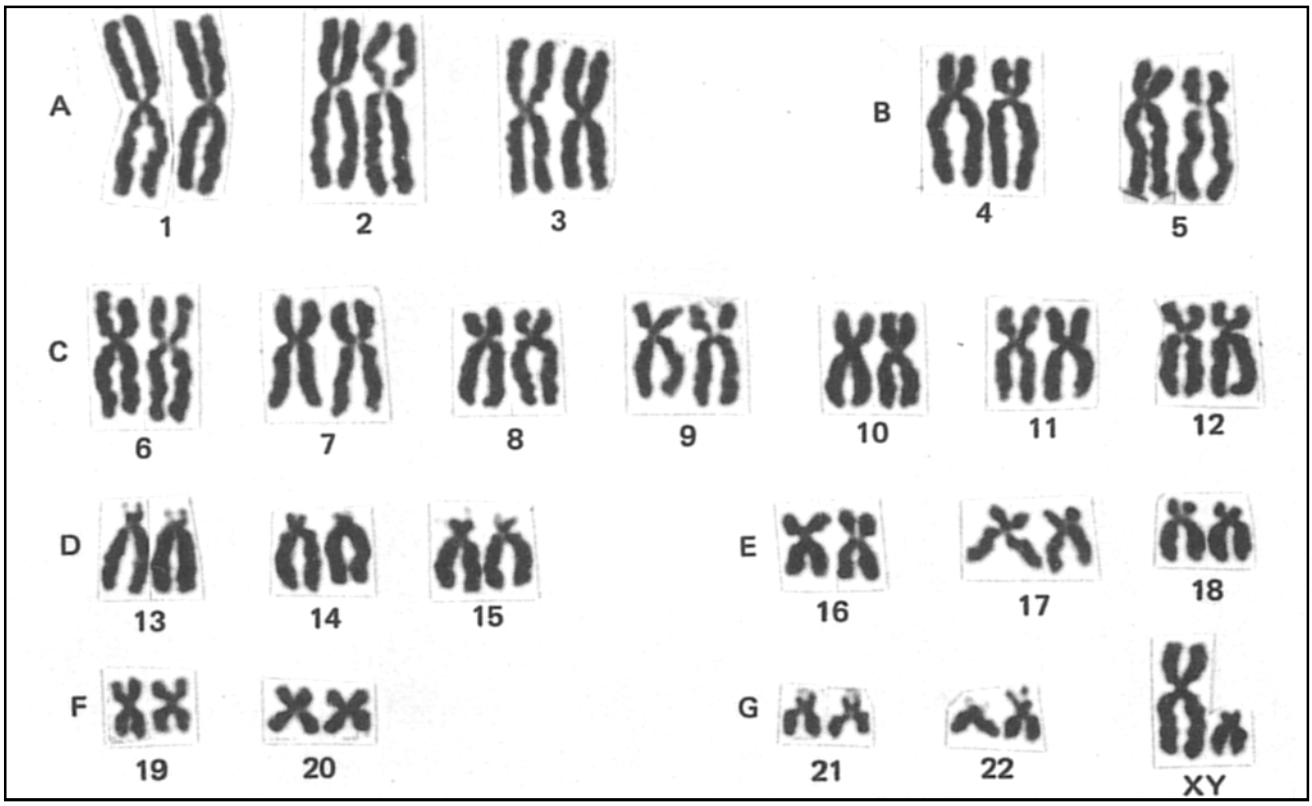
Group – C : In this includes chromosome pairs 6 to 12 and X chromosomes, are all medium in length and submetacentric but smaller than group B.

Group – D : It includes medium sized acrocentric chromosome 13, 14 and 15 plus satellites and one very small and one very large arm and morphologically similar chromosomes.

Group – E : This includes two morphologically distinguishable categories, chromosome number 16 is short and metacentric whereas 17 and 18 are submetacentric chromosomes.

Group – F : This group includes two small metacentric chromosomes 19 and 20.

Group – G : This group includes very small, acrocentric with satellites chromosomes 21, 22 and it also includes a variable Y chromosome.



Molecular Organization of Chromosome:

Chromosome is composed of DNA, RNA and protein. The protein of chromosome is of two types, the histones and the non-histones. Purified chromosome isolated from interphase nuclei has 30-40% DNA, 50-65% protein and 0.5-10% RNA, but there is a considerable variation due to species and tissues of the same species.

DNA:

The amount of DNA present in normal somatic cells of a species is constant for that species. Any variation in DNA from this value is correlated with a variation at the chromosome level. Gametes of a species contain only half of the amount of DNA present in its somatic cells. The amount of DNA present in somatic cells also depends on the phase of cell cycle.

Protein:

Proteins associated with chromosomes may be classified into two groups: basic proteins or histones and non-histone proteins.

Histones: It constitute about 80% of the total chromosomal protein. They are present in an almost 1:1 ratio with DNA (weight/weight). Their molecular weight ranges from 10,000-30,000 and they are completely devoid of tryptophan. Histones are a highly heterogenous class of proteins separable in 5 different fractions designated as H_1 H_2a, H_2b, H_3 and H_4.

Fraction H_1 is lysine rich, H_2a and H_2b are slightly lysine rich, while $H_3$and H_4 are arginine rich. These five fractions are present in all cell types of eukaryotes, except in the sperm of some animal species where they are replaced by another class of smaller molecule basic proteins called protamines.

Histones play a primary function in chromosome organisation where H_2a, H_2b, H_3 and H_4 are involved in the structural organization of chromatin fibres, while fraction H_1 holds together the folded chromatin fibres of chromosomes.

Non-histone: The non-histone proteins make up about 20% of the total chromosome mass, but their amount is variable and there is no definite ratio between the amounts of DNA and non-histones present in chromosomes.

There may be 12 to more than 20 different types of non-histone proteins which show variation from one species to the other and even in different tissues of the same organism. This class of proteins includes many important enzymes, such as DNA and RNA polymerases etc.

Special Types of Chromosomes:

Some tissues of certain organisms contain chromosomes which differ significantly from normal ones in terms of either morphology or function; such chromosomes are referred to as special chromosomes. These are Polytene chromosome, Lampbrush chromosomes and Accessory or B chromosomes.

Polytene chromosomes:

It was first discovered by Balbiani in 1881. It was found is salivary gland cells of chironomous larva. Hence it is also called as salivary gland chromosome. This kind of chromosome are found in gut epithelium, Malphigian tubules and some Diptera like Drosophila, Chironomous, Sciara, Rhyncosciara etc. These chromosomes are very long and very thick, hence they are known as giant chromosomes. In Drosophila melanogaster it is 1000 times larger than somatic chromosome. The larger size of the chromosome is due to the presence many longitudinal strands called Chromonemata (many stranded). The many strands of chromosome are due to repeated division of the chromosome without the cytoplasmic division. This type of division is called as Endomitosis.

Structure:

They are many times larger than the normal chromosomes, reaching a length of 2000 um and are visible even under a compound microscope. The polytene chromosome consists of five long and one short arm radiating from a central point called chromocentre. It is formed by the fusion of centromeres of all the eight chromosomes found in the cell. Of the 6 arms, the short arm represents the fused IV chromosome and the longest represents the fused sex chromosomes.

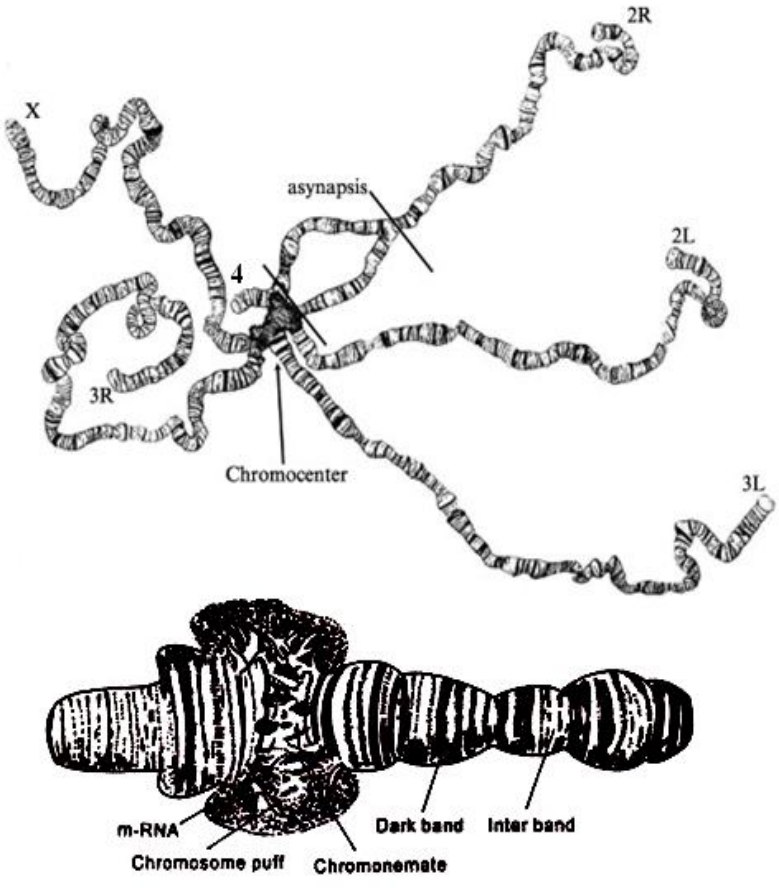
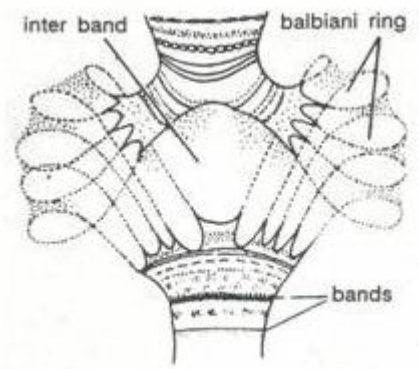


Fig. : Polytene chromosome

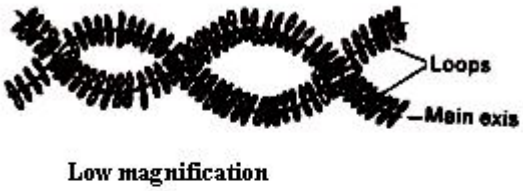


Balbiani ring

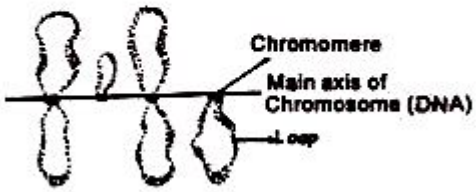
These arms contain numerous chromonemata resulting from repeated replication of DNA, without separation into daughter chromosomes. The arms show characteristic dark bands and light bands. The dark bands are euchromatic regions. Some of the dark bands temporarily swell up and form enlargements called chromosomal puffs or Balbiani rings. These regions contain actively transcribing DNA involved in the synthesis of RNA types.

Lampbrush chromosomes:

Lamp brush chromosomes were first observed by Flemming in 1882 in sections of Salamander oocytes and later described by Ruckert in the year 1892. They appeared like brushes used for cleaning lamps, hence the name lampbrush chromosome. They are transitory structures and can be observed during the diplotene stage of prophase I in meiosis in the oocytes of all animal species both vertebrates and invertebrates like, Sepia (Mollusca), Echinaster (Echinodermata) and in several species of insects, shark, amphibians, reptiles, birds and mammals (humans). Lampbrush chromosomes have also been found in spermatocytes of several species, giant nucleus of Acetabularia and even in plants. Generally they are smaller in invertebrates than vertebrates. They are observed in oocytes because oocytes are high in DNA content. They measure about 1500 to 2000 um in length.

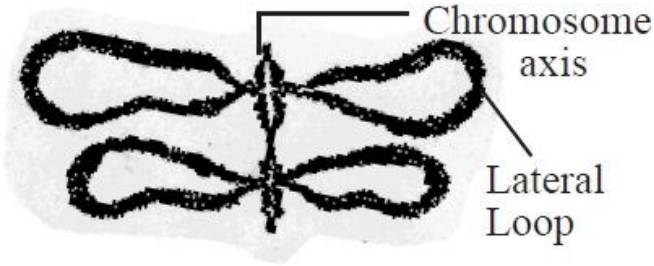


Low magnification



High magnification

Fig. : Lamp brush chromosome



A lampbrush chromosome consists of a main axis and many lateral loops extend in opposite directions. The main axis of each chromosome is formed of 4 chromatids. The axis consists of a series of thickenings called chromomere and inter-chromomere regions. From each chromomere a pair of lateral loops one on each side. Each loop has an axial fibre. The axial fibre is the continuation of the chromonema of the main axis. It contains DNA. The axial fibre of loop is surrounded by a matrix. The matrix is formed of RNA and proteins. The synthesis of proteins and yolk takes place in the lateral loops.

Important Questions

Q.1 Multiple choice questions (1 marks each)

1) The number of autosomes found in a diploid human cell is
   a)48 **b) 44** c) 23 d) 40
2) The specialized structures located at the ends of eukaryotic chromosomes are called
   a)Terminators **b) Telomeres** c) Centromeres d) Kinetochores
3) Haploids have
   a) Single set of genome b) Double set of genome
   c) Multiple set of genome d) No genome
4) The functional unit of gene is
   a) Muton b) Recon **c) Cistron** d) Retron
5) In which of the following chromosome centromere is present at the tip -
   a) Acrocentric **b) Telocentric** c)Metacentric d) Sub metacentric
6) Polytene chromosomes are found in–
   a) Amphibian oocyte b) Fish oocyte **c) Dipteran insects** d) Lepidopteran insects

Q.2 Define /explain /comment (2 marks each)

1) Karyotype 2) Salivary gland chromosome 3) Muton
4) Homologous chromosomes. 5) Down's syndrome 6) Cistron 7) Recon

Q. 3 Question for (4mark each)

1) Nucleosome 2) Types of chromosome 3) Human Karyotypes
4) Molecular organization of chromosome 5) Autosomes
6) Polytene chromosome 7) Lambrush chromosomes

Q. 4 Question for (8mark each)

1) Give an account of the morphology, ultrastructure of chromosome.
2) Detail note on chromosomes.
3) Briefly describe the structure of Polytene chromosome
4) Draw well labeled diagram of Lampbrush chromosome
5) Describe the Classification based on the centromeric position
6) Types of chromosome (autosomes and sex chromosome)
7) Chromosomal banding pattern
8) Human Karyotypes
9) Molecular organization of chromosome.
10) Special types of chromosomes - Polytene chromosomes
11) Lambrush chromosome
12) What are chromosomal disorders?
13) What are cistron, recon and muton? How are these related to the term 'gene'?
14)State and explain in brief chromosomal theory of inheritance. What is the presentconcept of gene?

Q. 5 Distinguish between (4 marks)

1) Diploid and haploid
2) Chromatid and chromosome
3) Euchromatin and heterochromatin
4) Chromosome and gene

Unit - 5

Lethal Genes

Concepts and Consequences:

The genes which reduce the viability of individual or the genes which causes the death of the individual carrying them or the gene which affect viability as well as visible trait (character) of the organism is called as Lethal gene.

Most lethal genes are recessive and cause death only in homozygous condition. Different lethal genes differ in the time at which they bring about their lethal effect. The time depends upon the development and functioning of the structures, abnormalities of which cause death.

Lethal Genes in Man:

In man, lethal gene cause certain heart defects cause death in early embryonic stages, as the heart is indispensible for the developing embryo.

But lethal genes which cause defects of digestive organs, lungs or kidneys do not cause death before birth as these structures do not function during embryonic stages.

Exa. - In man, lethal gene causing internal adhesion of lungs. A child with such a homozygous gene pair might be able to survive during embryonic development, but at birth, when it suddenly becomes dependent upon its lungs for its oxygen supply, it would die, because its lungs could not expand properly.

Thus, the penetrance of lethal gene is variable. The lethal effect is expressed at different ages in different individuals. Similarly intensity of lethal expression is also variable i.e. some show mild effect, some show intense effect and some show intermediate effect.

Congenital Ichthyosis:Autosomal recessive congenital ichthyosis is one of the rare types of ichthyosis. It is an instance of homozygous recessive fatal gene in individuals.Children with this disease are born with crusted leathery skin with deep splits. These splits lead to bleeding, infection and death.

Juvenile Amaurotic Idiocy: The infants of this disease lose eyesight between the age of 4 and 7. Mental and physical powers deteriorate and they become die before adolescence. This disease is due to a recessive gene in homozygous condition.

Infantile Amaurotic Idiocy:A hereditary disorder of lipid metabolism occurring most frequently in individuals of Jewish descent in eastern Europe.Accumulation of lipids in nervous tissue results in death in early childhood.

Cooley's anemia (Thalassemia):

Thalassemia is an inherited disorder that affects the production of normal hemoglobin. Thalassemia includes a number of different forms of anemia, which are classified as alpha

thalassemia or beta thalassemia. The severity and type of anemia depend on the number of genes that are affected.

Beta thalassemia major (Cooley's anemia): Beta thalassemia is caused by abnormal or missing genes that affect the beta chain of the hemoglobin molecule. There is one beta chain gene on each #11 chromosome and a total of two #11 chromosomes per person, one inherited from each parent. Both #11 chromosomes that an affected person has inherited are abnormal. Both of the chromosomes have abnormal genes that do not direct the body to make normal beta chains or normal amounts of beta chains. Inheriting two abnormal genes causes the most severe type of beta thalassemia. Thalassemia major patients need frequent blood transfusions and may not survive a normal lifespan. During the first one to two years of life, they can be pale, fussy, have a poor appetite and have many infections. Other signs or symptoms may include growth retardation, abdominal swelling and jaundice. Without treatment, the spleen, liver and heart become enlarged and bones can become thin, brittle and deformed. A major problem is the buildup of iron from blood transfusions in the heart and other organs, resulting in heart failure for some patients in their teens or early 20s.

Beta thalassemia minor or thalassemia trait: Only one gene has an abnormality, resulting in less severe anemia. Thalassemia minor is further divided into categories,

Thalassemia minima **-** A person has few or no symptoms.

Thalassemia intermedia **-** A person has moderate to severe anemia.

Lethal alleles in Animals:

In Mice:

In the house mouse, *Mus musculus*, Yellow coat colour is due to gene 'Y' which is dominant to all genes for other coat colour like black, brown, white etc.

Dr. Cuenot observed that, the mating between two black rats always resulted in a black progeny. But when two Yellow mice were crossed, they always resulted in litters of yellow and black mice in the ratio of 2:1.

The yellow colour is due to heterozygous condition 'Yy'. The recessive homozygous condition 'yy' produces black mice, but the dominant homozygous condition 'YY' would be lethal. Thus, it was reducing the ratio 1:2:1 into 2:1.

Thus the homozygous dominant yellow individuals (YY) die in the embryonic stages. The dominant gene in mouse is lethal, causing death in homozygous condition.

Exa.– House mouse, *Mus musculus* - *Coat colour*.

Parent :	**Yellow**		**X**	**Yellow**	
Genotype :	Yy			Yy	
Gamete :	Y	y		Y	y
Offsprings :	YY	Yy		Yy	yy
	Yellow Inviable (Dies)	Yellow		Yellow	Black
Ratio :		2	:	1	

In Chickens:

In some cases, the lethal gene is not completely recessive, but intermediate, showing a partial effect in the heterozygous condition.

In chickens the dominant gene 'C' is responsible for a modified character known as Creeper. Such chicken possess short, crooked legs and cannot walk normally, they creep about without raising their bodies off the ground.

When two creepers were crossed a ratio of 2 creepers to 1 normal instead of 3:1 appears. The homozygous recessive creepers have such a great abnormalities that they die.

Important Questions

Q.1 Multiple choice questions (1 marks each)

1) Lethal genes can

a) Kill their bearers b) Handicap their bearers

c) Kill the insects on plants d) Kill the weeds

2) Lethal genes (genes that result in the failure to develop a vital organ or metabolic pathway are nearly always recessive. Animal breeders who discover a unique trait andcross-breed to increase the occurrence of that trait often encounter a noticeable increasein lethal genes. Why?

a) The lethal recessive gene may be linked to the desire trait gene.

b) Spreading the gene among offspring of both sexes will increase the likelihood itwill be sex-linked and expressed.

c) The cross-mating of closely related individuals, or inbreeding, increases chances the two recessive genes will "meet" in offspring.

d) "Pleiotropy"- the gene that is being selected for may have the second effect ofbeing lethal

3) Lethal genes are

a) recessive homozygous b) recessive heterozygous

c) dominant heterozygous d) codominant

4) A cross between two heterozygotes for one trait yields a phenotypic ratio of 2: 1. What isthe best explanation?

a) The dominant trait is lethal in its homozygous form.

b) Either the dominant or the recessive allele in its homozygous form is lethal.

c) The trait causes semisterility in one of the parents.

d) The recessive allele for the trait is lethal in its homozygous form.

5) Who first discovered the lethal gene?

a) Cuenot b) Mendel c) De-varies d) Morgon

6) All of the following about Huntington's disease is true except

a) It is an autosomal dominant disease.

b) It is caused by a delayed-action gene that is expressed when the affected individualis about 40.

c) The offspring of homozygotes are more severely affected than

d) The offspring of an affected parent have a 50% chance of inheriting the lethalgene.

Q.2 Define /explain /comment / attempt (2 marks each)

1) Lethal genes

2) Congenital ichthyosis

3) Infantile amaurotic idiocy

4) Cooley's anemia(Thalassemia)

5) Lethal alleles in animals

Unit – 6

Multiple Alleles

Concept, Characteristics and Importance of Multiples Alleles:

Many genes occur in two alternative forms or status, both influencing the same trait and occupying the same locus in homologous chromosomes. Such different forms of the same gene are called as Alleles or Allelic to each other. As we observed or seen in Mendelian laws.

However, many and possibly all genes can change or mutate in several or many different ways, giving rise to several alternative forms or alleles, which are collectively called as Multiple alleles.

Multiple alleles are defined as, three or more number of mutant forms or alleles of the same gene occupying the same locus on homologous chromosome. All these alleles present a set or series of multiple alleles.

Characters of Multiple Alleles:

1) Multiple alleles occupy the same locus in the homologous chromosome.
2) There is no crossing over within multiple allelic series.
3) Multiple alleles always influence (affect) or represent or regulate the same character.
4) The wild type allele is almost always Dominant to all others at the same locus. The other mutant alleles may be either dominant or intermediate among themselves when two different alleles are brought together in the same genotype.
5) When two mutant alleles are crossed, the phenotype of the progeny is always a mutant type and not the wild type.
6) The best example of multiple alleles is ABO - blood group in Man, Coat colour in Rabbit, Guinea pigs, Mice and Wings of Drosophila.

ABO blood group in Man :

In 1900, Karl Landsteiner discovered four blood groups in Man and won the Noble prize for this. He showed that, the blood of all persons is not alike (similar). When the blood of individuals mixed, in some cases RBCs get clumped or clot or agglutinated. This led to the discovery of four blood groups in Man. The four blood groups are called A, B, AB and O.

Blood is typed according to the presence or absence of Antigens and Antibodies.

Antigens: Antigens are proteins and are capable of stimulating the production of specific antibodies.

Antibodies: Antibodies are substance produced by animals in response to contact with foreign antigens and react specifically to particular antigens.

Blood Types (ABO Blood Groups):

According to Landsteiner the RBCs of Man contain two naturally occurring Antigens – antigen – A and antigen – B. Depending upon the types of antigens blood is classified into four types or groups.

Similarly, the blood plasma contains Antibodies – anti-A and anti-B, which causes clumping or agglutination of RBCs containing corresponding antigens. No person could carry an antibody which would affect the antigens in his own RBCs. But the blood of other person may contain such antibodies. Therefore, very accurate knowledge of antigens and antibodies is essential in blood transfusion.

Antibodies related to blood group are called as Agglutinins as they bring about agglutination reaction and the blood group antigens on which antibodies have their effect are called as Agglutinogens.

	Blood Group	Antigens	Antibody
1.	A	A	anti-B
2.	B	B	anti-A
3.	AB	A and B	No antibody
4.	O	No antigen	anti-A and anti-B

Inheritance of ABO Blood Group:

The ABO blood group system of man gives a best example of multiple alleles. In man production of the blood types (groups) antigen is due to a gene 'I' (I stands for Isohaemogglutinogen). Gene-I occur in three possible allelic forms-I^A, I^B and I^O or i.

1) I^A represents the gene (allele) which produces antigen – A.
2) I^B represents the gene which produces antigen – B.
3) Person homozygous for these two genes (I^A I^B) will have antigen – A and B in their blood.
4) I^O or i represents the gene which produce none of these antigens.

The blood group character is controlled by a set of these three alleles. Which obviously compose of a series of multiple alleles i.e. I^A, I^B and I^O or i.

Gene I^A and I^B are both dominant over gene I^O or i ($I^A > I^B$, $I^B > I^O$). But not over each other ($I^A = I^B$). As I^O is recessive to both I^A as well as I^B the type O individuals are homozygous I^O I^O or ii.

The genotype of four blood groups is as follows –

	Blood group	Genotype
1.	A	I^AI^A, I^AI^O
2.	B	I^BI^B, I^BI^O
3.	AB	I^AI^B
4.	O	I^OI^O or ii

These genotypes indicate that the blood groups are also inherited in the simple Mendelian fashion. Offsprings with all four kinds of blood groups are possible in a cross between two persons heterozygous for blood group A and B.

Parent	:	**Male**			**Female**	
		Heterozygous Blood Group		**X**	Heterozygous Blood Group	
		A			**B**	
Genotype	:	I^AI^O			I^BI^O	
Gamete	:	I^A	I^O		I^B	I^O
Offsprings	:	I^AI^B	I^AI^O		I^BI^O	I^OI^O
Blood Group:		**AB**	**A**		**B**	**O**

Rh – Blood Group System/Rh – Factor:

In 1940, Karl Landsteiner, A.S.Weiner, Mourant, Levine and others discovered another proteinous substance in the red blood cells of Rhesus monkey called Rh-antigen.

When the blood from a Rhesus monkey was injected into Guinea pigs, it was found (observed) that Guinea pigs produced antibodies which agglutinate the RBCs of Rhesus monkey. This indicates that Rhesus monkey contains a particular antigens and it was named as Rh-Factor.

When human blood was tested with serum of Guinea pig which containing antibodies against antigen –Rh. It was found that, the cells of some person clump, whereas the blood of other persons is not clumped or affected. Therefore, he was concluded that some persons have the same antigen-Rh, as found in Rhesus monkey while others do not have it. The person with this antigen is called as Rh+ve and those without it called as Rh-ve.

Anti-Rh antibodies do not occur naturally, neither Rh +ve person nor Rh-ve person. They possess anti-Rh antibodies in the blood at the time of birth. However, Rh -ve persons can develop these antibodies if exposed to Rh antigen.

The gene ‘R’ for Rh +ve condition is Dominant over ‘r’ for Rh –ve condition. Therefore Rh +ve individuals are may be homozygous or heterozygous. But Rh –ve individuals are always homozygous.

The ABO and Rh blood group systems are two independent blood group systems. A donar selected for blood transfusion may be suitable for one system but may not be suitable for other system. Therefore it is necessary to make sure that blood of the donor matches with that of the recipient for both systems. Rh –ve recipient should always given blood if Rh –ve only.

Erythroblastosis Foetalis:

If Rh –ve women marries with Rh +ve man. The Rh -ve women will sensitize by Rh+ve blood may give birth to abnormal child. Her children may inherit Rh +ve antigen from their father. Antibodies of such mother may pass through the placenta and cause damage to the RBCs of the child in the uterus. This causes the disease Erythroblastosis foetalis.

This disease is a kind of Anemia due to haemolysis of RBCs in the foetus which leads to Jaundice. As the blood vessels in the liver become clogged or blocked with broken cells (RBCs) and bile is absorbed by the blood. This disease may cause death of the foetus.

Rh -ve women may become sensitized by carrying a Rh +ve child in her uterus. Some of the cells (RBCs) having Rh+ve antigen from embryo (foetus) may enter into the blood stream of mother and gets sensitized.

During the first pregnancy large amount of antibodies are not forms before the birth of first child. Hence may not cause great damage to the child. But the subsequent positive children (Rh+ve) may be seriously affected due to high level of antibodies in the mother blood, child may die.

Therefore, during second pregnancy Doctor tests blood of such mother for the level of antibodies in advance and take necessary precautions to prevent death of the child. In such cases, sometimes the new born baby is given complete change of the blood and to replace the damaged cells.

Significance of Blood Group:

1) Knowledge of blood group is important is blood transfusion. Cross matching of blood is done before blood transfusion. When there is no clumping, then blood can be given to patient.

2) In medico-legal application it helps to determine parenthood of babies which may have been accidentally exchanged in hospital.

3) The blood group knowledge is useful for investigation of criminal cases by examining blood strains.

Coat Colour in Rabbit:

In Rabbit coat colour is due to the gene 'C', which occurs in four different forms or alleles. Which compose a series of multiple alleles

i)Agouti or Full colour : The coat of the wild type rabbit is "agouti" or full colour, in which individuals have banded hairs, the portion nearest the skin being gray, succeeded by a yellow band and finally a black or brown tip. The allele for full colour may be represented by capital letter C^{+}.

ii) Chinchilla: In some individuals, the coat lacking the yellow pigment and due to the optical effect of black and gray hairs, have the appearance of silvery-gray, called Chinchilla. The allele for chinchilla is represented as C^{ch}.

iii) Himalayan (Russian): The Himalayan type coat is white except for black extremities (nose, ears, feet and tail). The condition in which black pigmentation is confined to the ears, tips of limbs and tail, is called coloured. In Himalayan rabbits eyes remain pigmented. The allele for Himalayan coat is represented by C^{h}.

iv) Albino: The albino coat totally lacks in pigmentation and the eyes of an albino also remain pink due to lack of pigment in iris of eye. The allele for albino is represented by c.

Crosses of homozygous agouti (C^+C^+) and albino (cc) individuals produce a uniform Agouti F_1. Interbreeding of the F_1 produces 3 Agouti : 1 Albino in an F_2 generation. Two third of F_2 Agouti are found to be heterozygous by testcrosses. Thus, it is a case of monohybrid inheritance, with Agouti completely dominant to Albino.

Likewise, crosses between Chinchilla and Agouti produce all Agouti individuals in the F_1 and a 3 Agouti : 1 Chinchilla in the F_2. Such complete dominance of agouti also occurs on Himalayan. Further crosses, reveal that C^{ch} allele for Chinchilla, though is recessive to C^+ allele for Agouti coat or skin is incompletely dominant over Himalayan (C^h) and albino (c) alleles.

Likewise, C^h allele for Himalayan coat is recessive to C^+ (Agouti) and C^{ch} (Chinchilla) but dominates over Albino. The results of all these crosses exhibit that C^+ (Agouti), C^{ch} (Chinchilla), C^h (Himalayan) and c (Albino) are allelic to each other and the alleles of this multiple allelic series have dominance hierarchy is - $C^+ > C^{ch} > C^h > c$.

The possible phenotypes and their associated genotypes of this multiple allelic series are -

P_1:	Agouti	X	Albino	P_1:	Agouti	X	Chinchilla
	C^+C^+	↓	cc		C^+C^+	↓	$C^{ch}C^{ch}$
		Agouti				Agouti	
F_1:		C^+c		F_1:		C^+C^{ch}	
F_2:	$1C^+C^+ : 2\ C^+c : 1cc$			F_2:	$1C^+C^+ : 2\ C^+C^{ch} : 1\ C^{ch}C^{ch}$		
	3 Agouti : 1 Albino				3 Agouti : 1 Chinchilla		
P_1:	Agouti	X	Himalayan	P_1:	Chinchilla	X	Himalayan
	C^+C^+	↓	C^hC^h		$C^{ch}C^{ch}$	↓	C^hC^h
F_1:		Agouti		F_1:		Light gray	
		C^+C^h				$C^{ch}C^h$	
F_2:	$1\ C^+C^+ : 2\ C^+C^h : 1\ C^hC^h$			F_2:	$1C^{ch}C^{ch} :\ 2\ C^{ch}C^h : 1\ C^hC^h$		
	3 Agouti : 1 Himalayan				1 Chinchilla : 2 Light gray : 1 Himalayan		
P_1:	Chinchilla	X	Albino	P_1:	Himalayan	X	Albino
	$C^{ch}C^{ch}$	↓	cc		C^hC^h	↓	cc
F_1:		Light gray		F_1:		Himalayan	
		$C^{ch}c$				C^hc	
F_2:	$1C\ ^{ch}C^{ch} : 2\ C^{ch}c : 1cc$			F_2:	$1\ C^hC^h : 2\ C^hc : 1\ cc$		
	1 Chinchilla : 2 Light gray : 1Abino				3 Himalayan : 1 Albino		

Concept of Polygenic Inheritance with reference to Skin Colour in Man:

Mendel's laws of heredity provide a simple and adequate explanation of clear cut differences like Tall vs Dwarf, Yellow vs Green cotyledons. These are sharply contrasting alternate character of individual plants, are known as Qualitative character and their inheritance is called as Qualitative Inheritance.

However there are many characters like height, weight or intelligence in man, the amount of fruits, seeds, milks or meat produced by plants or animals. These characters do not show clear cut differences between individuals and all gradations occur between the two extremes, such characters are called as Quantitative characters and shows a continuous variation and their inheritance is called as Quantitative Inheritance.

The quantitative inheritance is controlled by more than two genes located at different loci. These genes are called as Multiple genes or Polygenes.

The multiple or polygenes are defined as, a group of genes located at different loci of different chromosomes and involved in the expression of single character. The inheritance of such genes is termed as Multiple factor inheritance or Polygenic inheritance.

The inheritance of quantitative characters was studied by H. Nilson Elhe (1908) in Sweden and E. M. East (1910) in Harvard University, U.S.A. They showed that a continuously varying character is due to the combined action of several or many genes. Each of which has a small effect on the same trait or character and such genes are called as Cumulative genes or polygenes and its inheritance is called as Polygenic Inheritance.

Characters:

1) Heterozygous is intermediate between homozygous dominant and homozygous recessive.
2) A particular quantitative character may be controlled by several genes.
3) The environment influences such characters to a much higher degree.

Skin Colour in Man:

1913, C. B. Davenport studied inheritance of skin colour in Negro and White populations in U.S.A. In U.S.A. the populations derived from marriages between Negro and White individuals are known as Mulattoes.

In man, black skin is due to the presence of melanin pigment and white skin is due to the absence of melanin pigment in a person. Its activation or expression is depends upon genes and amount of sunlight the skin receives. The Negroes race has a heavy deposit of this pigment because they are native of North and South Africa.

Exa. – Skin Colour in Man

Parents (P-1) : **Negro** **X** **White**

Genotype : AABB aabb

Gametes : AB ab

F-1 Generation : AaBb

(Hybrid) Mulattoes

Intermediate skin colour

Parents (P-2) : **Mulattoes** **X** **Mulattoes**

Genotype : AaBb AaBb

Gametes : AB Ab aB ab AB Ab aB ab

F-2 Generation :

O/O	AB	Ab	aB	ab
AB	AABB Like Negro	AABb Darker than Mulattoes	AaBB Darker than Mulattoes	AaBb Like Mulattoes
Ab	AABb Darker than Mulattoes	AAbb Like Mulattoes	AaBb Like Mulattoes	Aabb Lighter than Mulattoes
aB	AaBB Darker than Mulattoes	AaBb Like Mulattoes	aaBB Like Mulattoes	aaBb Lighter than Mulattoes
ab	AaBb Like Mulattoes	Aabb Lighter than Mulattoes	aaBb Lighter than Mulattoes	aabb Like White

Ratio :

Negro Skin Colour	Colour between Mulattoes and Negro (Darker)	Colour of Mulattoes	Colour between Mulattoes and White (Lighter)	White Skin Colour
1	4	6	4	1

Negroes have four colour genes – 'AABB' and White having no colour genes i.e. 'aabb'. When a Negro and White races were crossed, they produced a hybrid of genotype 'AaBb', having an intermediate colour Mulattoes just between the Black and White parents. The Mulattoes contain only two dominant genes – A and B with cumulative effects and produced only 50 % pigments.

When two Mulattoes marry, they have children showing different degree of pigmentation from pure Black to White in the ratio of 1:4:6:4:1.

i.e. One – Negro – having 4 colour genes

Four – Darker than Mulattoes – having 3 colour genes

Six – Mulattoes – having 2 colour genes

Four – Lighter than Mulattoes – having 1 colour gene

One – White – having no colour genes.

Important Questions

Q.1 Multiple choice questions (1 marks each)

1) Blood group B will have alleles

a) ii b) $I_A I_A$ c)**$I_B I_B$** d) ii or $I_B I_B$

2) Father with blood type A and Mother with blood type O produce a child. The child:

a) must have blood type A b) must have blood type O

c) may have blood type A or O d) may have blood type B,

3) Which blood type would not be possible for children of a type AB mother and a typeA father?

a) O b) A c) B d) AB

4) *Erythroblastosis foetalis* is caused when fertilization takes place between gametes of

a) Rh- female and Rh+ male b) Rh+ female and Rh- male

c) Rh+ female and Rh+ male d) Rh~ female and Rh- male.

5) Which of the following is controlled by multiple alleles

a) Colour blindness b) Sickle cell anaemia c) Phenylketouria **d) Blood group**

6) Coat color in a certain species of animal is governed by multiple alleles. The order ofdecreasing dominance for these alleles is as follows: black (B), buff, (f), brown (r) andalbino (b). What phenotypic ratios are observed from a Bb X rb cross?

a) 1 black: 1 buff b) all black **c) 2 black: 1 brown: 1 albino**d) 1 buff: 1 brown

Q.2 Define /explain /comment (2 marks each)

1) Universal donar 2) Multiple alleles 3) Rh factor 4) Universal recipient

Q.3 Question for (4mark each)

1) Rh blood group 2) Bombay blood group 3) Blood transfusion

4) Multiple alleles 5) Compatibility 6) ABO blood group

7) Medico-legal significance of blood groups. 8) Erythroblastosis foetalis

Q. 4 Question for (8mark each)

1) Write note on Multiple Alleles and its inheritance.

2) What are multiple alleles? Give a brief account of multiple allelism.

3) Give an account of multiple alleles in the light of ABO blood group.

4) Explain Rh factors on the basis of multiple alleles.

5) What are multiple alleles? Explain with reference coat colour in rabbit.

6) What are applications of blood groups?

7) Brief note on multiple alleles with suitable examples.

Q. 5 Distinguish between (4 marks)

1) Polygene and modified gene

Q.6 **Problem solving. (4 marks)**

1) A woman of blood group 'A' marries a man of blood group 'O'. There are threechildren in the family. The children's blood types O, A and AB. Which child wasdefinitely adopted?
2) If a person of blood group AB marries one belonging to group O, what will be theblood groups of their children's? Determine the genotype of parents.
3) What genotypes and their proportions' would be produced by the following crosses.

   a) IA x ii b) IAIB x IAi c) IBIB x IAi d) IAIB x IAIB
4) In rabbits full colour (C) , Chinchilla (cch), Himalayan (ch), and Albinism (ca) formthe series of multiple alleles with dominance in order given. What will be theappearance of the offspring's from the following crosses in rabbits (whereas cch ch isand cch ca is light grey:

   a) C cch x C ca b) cch ch x ch ch c) ch ca x cch cch d) C ch x cch ca
5) In rabbits full colour (C), Chinchilla (cch), Himalayan (ch), and Albinism (ca) form theseries of multiple alleles with dominance in order given. Is it possible to cross twoagouti rabbits and produce both Chinchilla and Himalayan progeny?

Unit – 7

Linkage and Crossing over

In 1910, T. H. Morgan discovered that, the genes present in the same homologous pair of chromosomes do not undergo random assortment but are inherited together. This is known as phenomenon of Linkage.The genes are situated in the same chromosome are fairly close to each other, they tend to be inherited together from one generation to the next. Such genes are called as Linked genes.

The tendency of genes to remain together during the process of inheritance is called as Linkage.

T.H. Morgan and his co-workers found two types of linkage - Complete Linkage and Incomplete Linkage.

1) Complete Linkage:

When linked characters or genes are inherited together through two or more generation is called as Complete Linkage.

Complete linkage is a phenomenon in which parental combinations of characters appear together for two or more generation in continuous and regular fashion.

In this type of linkage, genes are closely associated and tend to transmit together. They do not undergo any breakage by accident or during gametogenesis. This phenomenon is rare and is found in some insects like Drosophila – Male.

Exa.: Fruit fly – Drosophila melanogaster – Body colour and Wing size.

Parents (P-1) :	**Male**			**Female**
	Grey Body Long Wing		**X**	**Black body Vestigeal Wing**
Genotype :	GL/GL			bv/bv
Gametes :	GL			bv
F-1 Generation :		GL/bv		
(Hybrid)		Grey Body Long Wing		
		Test Cross		
Parents (P-2) :	**F-1 Hybrid**		**X**	**Double Recessive Parent**
	Grey Body Long Wing			**Black Body Vestigeal Wing**
Genotype :	GL/bv			bv/bv
Gametes :	GL	bv	bv	bv
F-2 Generation :	GL/bv	GL/bv	bv/bv	bv/bv
	Grey Long Body Wing	Grey Long Body Wing	Black Vestigeal Body Wing	Black Vestigeal Body Wing
Ratio :		**50%**	**:**	**50%**

Wild Drosophila having Grey Body (G) and Long Wings (L) were crossed with Black Body (b) and Vestigeal Wings (v). All the F-1 hybrids are Grey Body and Long Wings (GL/bv). Because the gene 'G' and 'L' are Dominant over 'b' and 'v' genes.

A test cross between a F-1 Male and Double Recessive Female parent, in the F-2 generation 50% Grey Body and 50% Black Body and Vestigeal Wings.

It is seen that, the Grey body is always inherited together with long Wings. Because the characters or genes are linked.

Similarly, black body and Vestigeal Wings are linked character and inherited together.

2) Incomplete Linkage:

The linked genes do not always stay together because homologous non-sister chromatids may exchange segments of varying or distant length with one another during meiotic prophase.

The exchange of chromosomal segments in between homologous chromosomes is known as Crossing over.

The linked genes are widely or distantly located in chromosomes and have chances of separation by crossing over are called as Incomplete linked genes and the phenomenon of their inheritance is called as Incomplete Linkage.

Incomplete linkage found in Female Drosophila, Tomato, Maize, Pea, Poultry, Mice, Man etc.

Exa. : Maize : Seed – Colour and Endosperm

Parents (P-1) :	**Coloured Full**		**X**	**Colourless Shrunken**
Genotype :	CS/CS			cs/cs
Gametes :	CS			cs
F-1 Generation : (Hybrid)	CS/cs Coloured Full			

Test Cross

Parents (P-2) :	**F-1 Hybrid Coloured Full**		**X**	**Double Recessive Parent Colourless Shrunken**
Genotype :	CS/cs			cs/cs
Gametes :	CS Cs cS cs			cs
F-2 Generation :	CS/cs Coloured Full	Cs/cs Coloured Shrunken	cS/cs Colourless Full	cs/cs Colourless Shrunken
Ratio :	**48%**	**2%**	**2%**	**48%**

Hutchinson studied the phenomenon of linkage in Maize. He crossed two varieties of Maize, One having Coloured and Full Endosperm seeds (CS/CS) and another variety with Colourless and Shrunken Endosperm seeds (cs/cs).

The colour gene 'C' is Dominant over the Colourless gene 'c' and Normal or Full endosperm gene 'S' is Dominant over the Shrunken endosperm gene 's'.

In the F-1 generation all heterozygotes are with Coloured and Full Endosperm (CS/cs). When F-1 hybrid (CS/cs) is test crossed with Double Recessive Parent (cs/cs), four types of seed plants are obtained instead of two in F-2 generation.

Test cross results clearly shows that the Parental combination of alleles (CS/CS and cs/cs) are those expected from complete linkage, appears in 96% cases. The other two are new combinations (Cs/cs and cS/cs), appear in 4% cases.

Thus, in 4% of the cases crossing over is takes place between linked genes. So, it is called as Incomplete Linkage.

Significance of Linkage:

i) Linkage does not permit the breeders to bring the desirable characters in one variety.

ii) Linked characters are maintained for generations because linkage prevents the incidence of recombination.

Crossing Over:

Crossing over is defined as, an interchange of corresponding chromosomal parts between chromatids of homologous chromosomes of a pair, resulting in recombination of genes and producing individuals with new combinations of characters.

Crossing over takes place during prophase - I of meiosis. Crossing over is another name for recombination or physical exchange of equal pieces of adjacent non-sister chromatids.

Mechanism of Crossing Over:

Crossing over takes place in 4 steps – Synopsis, Duplication of chromosomes, Crossing over and Terminalization.

1) **Synopsis:** During the Zygotene stage of Prophase – I of meiosis homologous chromosomes pair up, they come in close contact due to attraction between identical regions of chromosomes. This is the two-strand bivalent (Diad) stage.
2) **Duplication of chromosomes:** Each bivalent chromosome of the pair splits into sister chromatids attached to the unsplit centromeres. DNA molecules along the chromosomal portions also become duplicated.
3) **Crossing Over:** Non-sister chromatids of the homologous pairs twist each other, due to enzyme known as Endonuclease. The chromatid threads are connected with each other at a point known as Chiasma. A segment of one side fuses with a segment of the opposite side due to an enzyme known as Ligase.

The number of times crossing over and chiasmata formation takes place is proportional to the length of chromosome. More number of chiasmata is formed between distant genes and less number of chiasmata formed between closer genes.

4) **Terminalization:** The non-sister chromatids repel each other due to lack of attraction between them. The repulsion or separation of chromatids starts from the centromere and proceeds towards the end just like a zipper. The twisting chromatids separate and then the chromosomes separate completely.

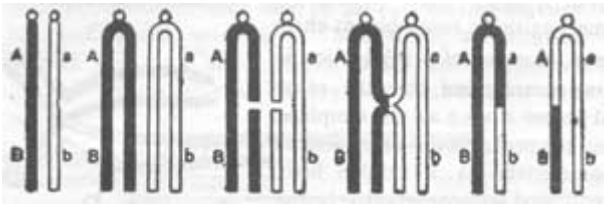


Fig. : Mechanism of Crossing Over.

Types of Crossing Over:

Crossing over is of two types –

1) Types of Crossing Over according to the Cell Type:

a) Somatic or Mitotic Crossing Over: It takes placein somatic cells. It is rare in organisms. Exa. – Drosophila, Aspergillus.

b) Germinal or Meiotic Crossing Over: It takes place in germinal cells during gametogenesis. It takes place in many plants and animals.

2) Types of Crossing Over according to the Nature of Chiasma:

a) Single Crossing Over: When the chiasma formation takes place at a single point of the chromosome pair this type of crossing over is known as single crossing over. In this types two crossed over chromatids and two non crossed over chromatids are formed.

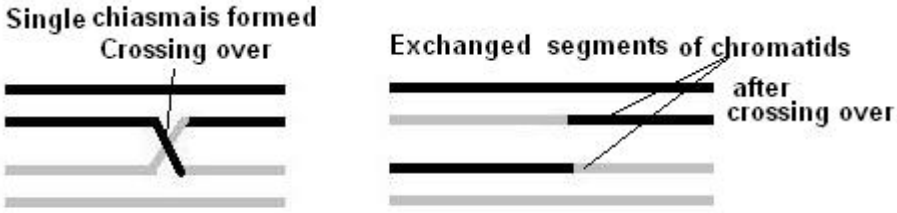


Fig - Single crossing over

b) Double crossing over: When the chiasmata occur at two places in the same chromosomes known as double crossing over In the double crossing over formation of each chiasma is independent of the other and in it four types of recombination is possible.

Two types of chiasma may be formed in double cross over :-

i) **Reciprocal Chiasma:** In this type both the chiasma are formed on two same chromatids. So, the second chiasma restores the order which was changed by the first Chiasma and as a result two non-cross over chromatids are formed.
In this type, out of four chromatids only two are involved in the double crossing over.

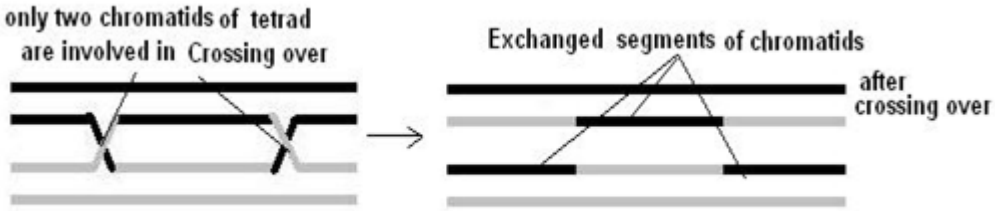
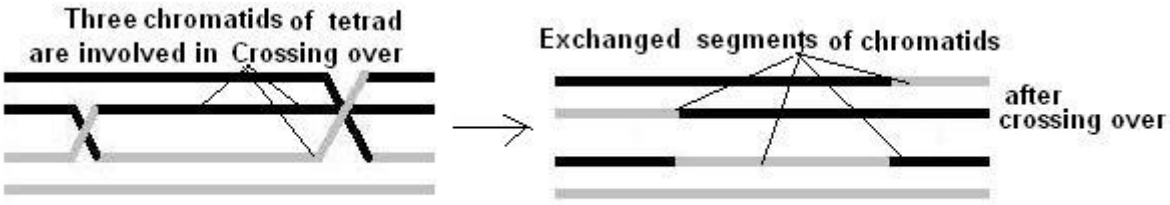


Fig – Double crossing over (Reciprocal chiasma)

ii) **Complimentary Chiasma:** When both the chromatids taking part in the second chiasma are different from those chromatids involved in the first. In this type, four single cross over are produced but no non cross over. Complimentary chiasma occurs when three or four chromatids of tetrad undergo crossing over.



Fig· Double crossing over (Complimentary chiasma)

c) **Multiple Crossing Over:** When crossing over takes place at more than two points in the same chromosome pair it is known as multiple crossing over. It occurs rarely.

Significance of Crossing Over:

i) Crossing over provides direct evidence that the genes are linearly arranged on the chromosome.

ii) The frequency of crossing over is helpful in the mapping of chromosomes.

iii) Crossing over provides origin for new characters due to change of segment from one chromosome to another and thus it is a source of genetic variations.

iv) Linkage groups and linear order of the genes throw much light on the mechanism and nature of genes.

v) Crossing over gives an operational definition of a gene – the smallest section or unit of a chromosome within which crossover does not take place. It means that the minimum chromosome unit which is able to cross over is one whole gene, not a fractional part of a gene.

vi) Crossing over has great importance in the field of breeding to improve the varieties of plants and animals.

Important Questions

Q.1Multiple choice questions (1 marks each)

1) Synapsis takes place at –

a) Zygotene b) Diplotene c) Pachytene d) Diakinesis

2) Frequency of crossing over between two genes is directly proportionate to

a) Distance between genes b) Distance between cells

c) Distance between chromosomes d) Distance between cellular organelles

3) A Crossing over in meiosis is an exchange of genetic material between

a) sister chromatids of the same chromosome.

b) sister chromatids of homologous chromosomes.

c) sister chromatids of non-homologous chromosomes.

d) non-sister chromatids of homologous chromosomes

4) Chromosome theory of linkage was proposed by

a) Beadle and Tatum b) Bateson and Punnett**c) Morgan and Castle** d) Sutton and Bovery

5) Crossing over taking place at

a) mitosis b) meiosis II **c) meiosis I** d)all of the above

6) What is the occurrence of two or more hereditary unit on the same chromosomecalled?

a) (Gene) Linkage b) Crossing c) both a and b d) None of these

Q.2 Define /explain /comment (2 marks each)

1) Linkage 2) Incomplete linkage 3) Chiasma

4) Pachytene 5) Daikinesis 6) Zygotene

7) Leptotene

Q.3 Question for (4mark each)

1) Write short note on Incomplete linkage

2) Write short note onComplete linkage

3)Write short note onSignificance of linkage.

4) Write short note onCrossing over at four strand stage.

5) Explain the concept of Linkage and Crossing Over.

Q. 4 Question for (6 mark each)

1) What is the significance of crossing over (which leads to genetic recombination) tothe process of evolution?

2) Describe the cytological observation that suggests that crossing over occurs during thefirst meiotic prophase.

3) Briefly describe chiasma formation. Discuss whether chiasmata are the cause or theconsequences of crossing over.

4) Describe the phenomenon of linkage by giving suitable examples. Why is linkage anexception to Mendel's second law?

5) Linkage studies provided the first proof that a gene occupies a fixed locus on aspecific chromosome, Discuss.

Unit - 8

Sex Determination

In the sexually dimorphic (dioceous) organisms, besides morphological and behavioural differences between the sexes, sexual diversity also occurs in the chromosomes. The chromosomal differences between the sexes are first discovered by German zoologist, Henking (1891) in male plant bug, *Pyrrhocorus apterus*. He noticed that, half of spermatozoa contain extra chromosomes. As its function is unknown he called as X – body or unknown body.

In 1902, C. E. Mc Clung observed gametogenesis in grasshopper and suggested that, the X – body was concerned with sex determination. In 1905, Stevens and Wilson studied gametogenesis in several insects. He showed that X – body was a chromosome and X – body is known as X – chromosome or sex chromosome, which is responsible for determination of sex of individual.

Chromosome Theory in Sex Determination:

The chromosomal theory of sex determination was determine by Miss. Stevens (1905) and is supported by Bridges (1922) and Goldschmidt (1938). According to this theory, chromosome is the main factor to determine the sex.There are two types of sex chromosomes X and Y. They differ in size and genetic composition.

The X chromosome is larger in size and straight. It contains a large amount of euchromatin and small amount of heterochromatin. The euchromatin is rich in DNA.

The Y chromosome is smaller in size. It contains a small amount of euchromatin and large amount of heterochromatin. Hence the amount of DNA in Y chromosome is less.

The chromosomal theory is subdivided into various theories,

1) Theory of Heterogenesis:

In 1906, Correns proposed the theory of heterogenesis. According to this theory, one sex produces two types of gametes and each type of gamete determines a different sex on fertilization. Such individual is called as Heterogametic and other sex produces same type of gametes. Such individuals are called as Homogametic. This is called as theory of heterogenesis.

a) XX – XY Method:

In this method one sex is heterogametic and has XY sex chromosome while other sex is homogametic and has XX sex chromosome. This type of sex determination is found in Human being and fruit fly, *Drosophila melanogaster*.

Exa. : Man

Parents :	Male	X	Female	
	(44 + XY)		(44 + XX)	
Gametes :	(22 + X)	(22 + Y)	(22 + X)	(22 + X)
	Sperms		Eggs	
Offsprings :	(44 + XX)		(44 + XY)	
	Female		Male	

In human being there are 23 pairs of chromosomes in somatic cells (body cells) except gametes. Out of these chromosomes, 22 pairs are autosomes which are same in both the sexes and the remaining two are sex chromosomes either with XX or XY combination. That is female has 44 autosomes and two sex chromosomes – XX and the male has 44 autosomes and two sex chromosomes – XY.

Female individual produces only X type of egg while male produces sperms of two types i.e. X type and Y type sperm. The female is thus homogametic and male is heterogametic.

Sex of the offspring depends on the heterogametic individual i.e. egg (X type) fertilized by sperm (X type) will result in XX i.e. female. While egg (X type) fertilized by sperm (Y type) will result XY i.e. male offspring.

b) XO – XX Method:

In this method, the females of many species possess autosomes + two X chromosomes while the males possess autosomes + one X chromosome i.e. in males the sex chromosome X is without a homologue. This type of sex determination is observed in many insects like Cockroach, Grasshopper, Bugs, Lepidopterans etc.

Exa. : Grasshopper

Parents :	Male		X	Female
	(22 + XO)			(22 + XX)
Gametes :	(11 + X)	(11 + O)		(11 + X) (11 + X)
	Sperms			Eggs
Offsprings :	(22 + XX)			(22 + XO)
	Female			Male

In 1902, C. E. McClung noted the absence of chromosome from Grasshopper testes during meiosis. He found 11 pairs of chromosomes with one odd chromosome (23 chromosomes) in the male germ cells. While in female germ cells, the number of chromosomes are 12 pairs (24 chromosomes). He considered the odd chromosome is an X chromosome, as it involves in the determination of sex of the offsprings.

Thus, the female is homogametic (22 + XX) and produces one or similar king of egg with 11 autosomes and one X chromosome (11 + X). The male is heterogametic (22 + XO) and produces two types of sperms, one with 11 autosomes and X chromosome (11 + X) and other with 11 autosomes only (11 + O).

Thus, sex determination in the Grasshopper shows different pattern. When an egg is fertilized by X bearing sperm (11 + X), it will produce a female grasshopper. While an egg is fertilized by no sex chromosome (11 + O), will result in male grasshopper.

The XO method of sex determination is differs from the XY method only in the absence of Y chromosome. The letter O means zero and indicates absence of sex chromosome in one sex (male). Hence, called as XO method.

In some Lepidopterans, female is heterogametic i.e. female produces two types of ova (egg), one with X chromosome and other type without X chromosome. While male is homogametic i.e. male produces all similar sperms, each carry X chromosome.

c) ZZ – ZW Method:

In this method the males are homogametic while females are heterogametic. Just reverse the XY and XO method. Therefore to avoid or eliminate confusion, commonly Z and W letters are used instead of X and Y respectively. Thus, males are ZZ and females are ZW in genotype.

Exa. : Birds – In Fowl

Parents :	Male	X	Female	
	Rooster		Hen	
	(16 + ZZ)		(16 + ZW)	
Gametes :	(8 + Z) (8 + Z)		(8 + Z)	(8 + W)
	Sperms		Eggs	
Offsprings :	(16 + ZZ)		(16 + ZW)	
	Male		Female	

The male (Rooster) having 8 pair of autosomes and 1 pair of similar sex chromosomes (16 + ZZ), while in female (Hen) 8 pair of autosomes and 1 pair of dissimilar sex chromosomes (16 + ZW).

Thus, male produces similar type of sperms after spermatogenesis i.e. (8 + Z), while female produces two dissimilar ovum after oogenesis i.e. (8 + Z) and (8 + W).

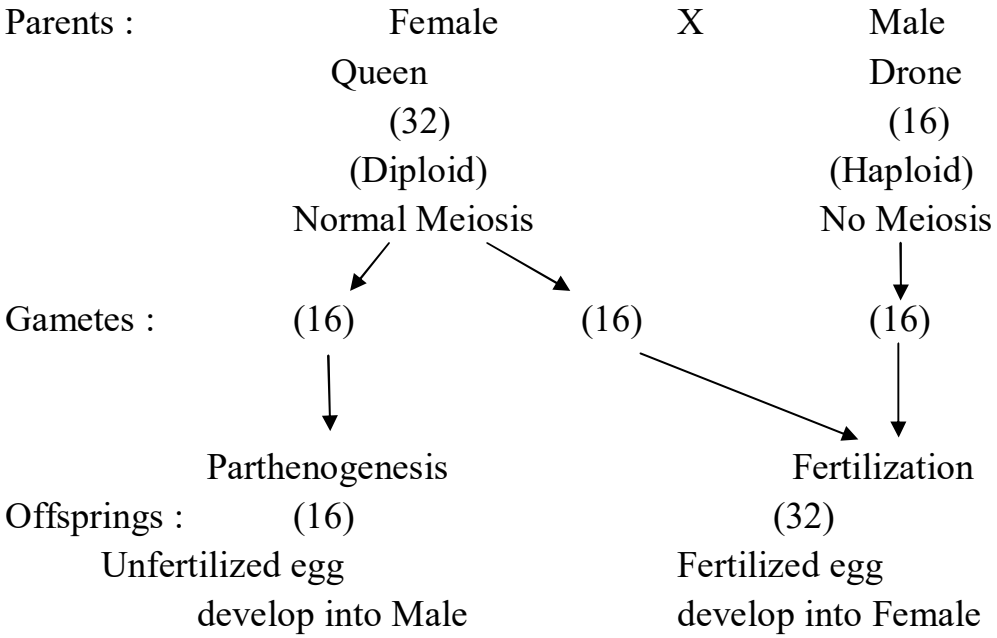
When the sperm having Z chromosome (8 + Z) fertilizes with ovum containing Z chromosome (8 + Z), then zygote develop into male (Rooster), while sperm having Z chromosome (8 + Z) fertilizes with ovum containing W chromosome (8 + W), then zygote develops into female (Hen).

2) Haploid – Diploid Mechanism of Sex Determination or Honey bee Method:

This type of sex determination is found in hymenopterans like honey bee, wasp, ants etc. These insects can determine the sex of their progeny at their own will. This is a universal example wherein mother can decide the sex of her child.

In honey bee (Apis), there are three forms or caste, queen, worker and drones. Out of these drones are males and are haploid (16) while other two, queen and workers are diploid females (32). The haploid males are produced from unfertilized eggs through Parthenogenesis but diploid females are produced after normal fertilization i.e. from fertilized eggs.

Exa. : Honey bee



In a bee hive, there are two types of cells, one is smaller in size used for development of workers and another is larger in size for drones. During nuptial flight, the sperms are inseminated in queen's body by the drone and are stored in a seminal receptacle and are always available for fertilizing eggs produced throughout the remainder of her life. When she lays an egg in a worker cell, sperms are ejected from the seminal receptacle fertilizing the egg, which after fertilization will develop into a female bee. Thus, the diploid females are produced.

If queen lays an egg in drone's cell, some sort of pressure is exerted on the duct of the seminal receptacle so that, the sperms are unable to pass out and fertilize the egg. Thus, unfertilized eggs are laid in the cell. Here special type of development takes place and the unfertilized eggs developed into a male (haploid).

3) Gynandromorphs: (Gr. Gyno – Woman, Andro – Male, Morphe – Form)

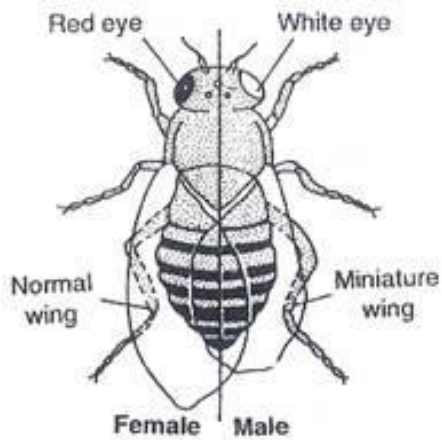
Gynandromorphs are the organisms in which the body is comprised of both male and female parts. In such animal one part of the body has female characteristics and other part shows the male characters. Exa. - Drosophila, Silk moth, beetles and bees.

T. H. Morgan, C. B. Bridges, Goldschmidt observed gynandromorphs in Drosophila and silk worms. Depending upon the relative position of the two phenotypes on the body, three types of Gynandromorphs are recognized.

a) Bilateral Gynandromorphs:

In this, half of the body shows male characters and other half of the body shows female characters or vice-versa. Exa. - Drosophila. Silk worm (Silk moth).

Ingynandromorph of drosophila, the right half of the fly showed male character like shorter wing, black tripped abdomen and a sex comb on the first leg. The left side showed contrasting female characters. The right eye was white and left one red.



b) Antero-postero Gynandromorphs:

In this anterior part of the body shows the character of male sex or female sex and the posterior part shows the characters of the opposite sex. Exa. – Beetle.

c) Sex Piebalds:

In this case, the female sex has scattered spots of male tissue in an irregular manner or vice versa**.** It is also called as Sex mosaics. Exa. – Moths.

GenicBalance Theory of Sex Determination:

The genic balance theory was proposed by Calvin Bridges (1926). He states that instead of XY chromosomes, sex is determined by the genic balance or ratio between X-chromosomes and autosome genomes.

The theory is basically applicable to Drosophila melanogaster over which Bridges worked. He found that the genic ratio X /A of 1.0 produce fertile females whether the flies have XX + 2A or XXX + 3A chromosome complement. A genic ratio (X /A) of 0.5 forms a male fruitfully. This occurs in XY + 2A as well as X0 + 2A. It means that expression of maleness is not controlled by Y- chromosome but is instead localized on autosomes.

This theory states that genes for maleness are located on autosomes and for femaleness on X-chromosomes in Drosophila.

The X-chromosomes, however, carry female determining genes like Sxl. Bridges further proposed that a genic ratio of less than 0.5 (e.g., XY + 3A or X/3A or 0.33) produced infertile meta-males (super males) while a genic ratio between 0.5 and 1.0 produces intersexes with a lot of morphological and sexual abnormalities.

Sterile meta-females (super females) are produced with the genic ratio of 1.5 (3X/2A). The sterile meta-males and meta-females have been called glamour boys and girls of fly world by Dodson.

Chromosome Complement	X / A Ratio	Sexual Morphology
X X X + 2A	3/2 or 1.5	Metafemale
X X X + 3A	3/3 or 1.0	Female
XX + 2A	2/2 or 1.0	Female
X X + 3A	2/3 or 0.67	Inter sex
X X X + 4A	3/4 or 0.75	Inter sex
XO + 2A	1/2 or 0.5	Male
XY + 2A	1/2 or 0.5	Male
XY + 3A	1/3 or 0.33	Metamale

Sex Linked Inheritance:

Sex linked inheritance was discovered by T.H. Morgan in 1910. The transmission of body characters from parents to offspring along with sex is calledsex linked inheritance.It is also called sex Linkage.

The genes controlling body characters located on the sex chromosomes are called sex linked genes. The body characters (other than sex characters) controlled by genes located on the sex chromosomes are calledsex linkedcharacters. Exa.Colour blindness, Hemophilia, Eye colour in Drosophila, Hypertrichosis (Hair in the ear pinna), *Ichthyosis hystrix*.

The sex linked genes are located on X chromosome or Y chromosome or both X and Y chromosome.

The genes, controlling body characters, located on X chromosome are called X-linked genes.The inheritance of X-linked genes is calledX- linked inheritance.The characters controlled by X-linked genes are called X- linked characters.

Exa. - Haemophilia, colour blindness, eye colour in Drosophila

The genes controlling body characters located on Y chromosome are called Y- linked genes.The inheritance of the inheritance of linked genes is called linked genes is called Y- linked inheritance.The characters controlled by Y- linked genes are called Y- linked characters.

Exa. - Hypertrichosis (hair in the pinna), Ichthyosis hystrix (scales on the body).

The genes controlling body characters located on both X and Y chromosomes are called XY linked genes**.** The inheritance of XY linked genes is calledXY linked inheritance.The characters controlled by XY linked genes are calledXY linked inheritance.

Exa. - *Xeroderma pigmentosum*, *Retinitis pigmentosa*, nephritis, etc.

Most of the sex linked characters are recessive. They are more common in man than in woman.

Most of the sex linked genes follow criss-cross inheritance (zig-zag inheritance).

The inheritance of a character from the father to his grandson through his daughter is called criss-cross inheritance.That is, the sex linked character appears only in alternate generations.

Exa. – Fruit fly - *Drosophila melanogaster* – Eye colour.

Parent (P-1) : Female X Male Female X Male

Phenotype : **Red Eye White eye White Eye Red Eye**

Genotype : X^RX^R X^wY X^wX^w X^RY

Gamete : X^R X^R X^w Y X^w X^w X^R Y

Offsprings:

0 / 0	X^w	Y		0 / 0	X^R	Y
X^R	$X^R X^w$ Red Eye	$X^R Y$ Red Eye		X^w	$X^R X^w$ Red Eye	$X^w Y$ White Eye
X^R	$X^R X^w$ Red Eye	$X^R Y$ Red Eye		X^w	$X^R X^w$ Red Eye	$X^w Y$ White Eye

In *Drosophila* the gene for eye colour is located on the X chromosome. The allele for red eye colour which is normal in wild flies is dominant to the mutant allele for white eyes.

As females have two chromosomes X (with a locus for eye color), they might behomozygous or heterozygous for either allele.

Males, who carry only one X chromosome, are always hemizygous. They carry only one X chromosome inherited from their mother and it determines their eye color.

In the left hand example, homozygous red eyed females (**RR**) mate with hemizygous white eyed males (**w-**). In the offspring, all the daughters are red eyed heterozygotes (**Rw)** and all sons are red eyed hemizygotes (**R-**).

In the right hand, homozygous white eyed females (**ww**) mate with hemizygous red eyed males (**R-**).

In the offspring, all the daughters are red eyed heterozygotes (**Rw**) and all sons are white eyed hemizygotes (**w-**).

Important Questions

Q.1 Multiple choice questions (1 marks each)

1) Eye colour in drosophila is an example of

a) Sex linked inheritance b) sex limited inheritance

c) Sex influenced inheritance d) incomplete dominance

2) A genetic pedigree showing that only males are affected by a certain disorder isevidence of what type of inheritance?

a) dominant **b) sex-linked** c) recessive d) passive

3) Sexual identity is determined by the sex chromosome make-up of an individual only. True or false?

a) True **b) False** c) Variable d) Basis is unknown

4) Sex chromosomes of a normal male are

a) XX b) YY **c) XY** d) Any of these, depending on the father

5) Sexual identity is determined by the sex chromosome make-up of an individualonly. True or false?

a) True b**) False** **c**) Variable d) Basis is unknown

6) In birds with a ZW sex determining mechanisms, males are

a) cytogametic **b) homogametic** c) heterogametic d) pleiogametic

7) In humans, the genetic basis for determining the sex "male" is accomplished by

a) an autosome

b) the number of X chromosomes

c) a balance between the number of X chromosomes and the number of haploidsets of autosomes

d) none of the above

Q.2 Define /explain /comment (2 marks each)

1) Autosome 2) Supersexes.

Q.3 Question for (4mark each)

1) Explain Y-inked inheritance.
2) Give an example for X-linkage.
3) Give an example for Y-linkage
4) Sex linkage in Drosophila
5) Hemophilia
6) Colour blindness
7) Sex determination
8) Triploid intersexes
9) Gynandromorphs in *Drosophila.*

Q. 4 Question for (4 mark each)

1) Write any four characteristic features of sex-linkage.
2) Explain with suitable example the principle of inheritance of sex linked charactors.
3) A colour blind man has a normal brother and a colour blind sister. give genotypes ofthe parents
4) Describe Chromosome theory in sex determination
5) Describe genic balance theory of sex determination
6) What is sex determination? Describe various example of sex chromosomalmechanism of sex determination.
7) How is the sex determine in *Drosophila.*
8) What is the role of Y chromosome in determining of sex?
9) What is the significance of XO methods of determination?

Q.5 Distinguish between (4 marks)

a) Autosome and sex chromosome

b) Compare and contrast of XY and XO methods of sex determination.

Unit – 9

Mutation

Mutation is a permanent change of the nucleotide sequence of the genome of an organism, virus or extra chromosomal DNA or other genetic elements.

A mutation is any change in the DNA sequence. Mutations can lead to genetic disorders or disease. Most mutations are recognized because the phenotype, that is the characteristics displayed by an organism, has changed. There are many different types of mutation.

They can occur on a macroscopic level in the form of chromosomal mutations or they may be the result of a single base pair change in the DNA sequence. Mutations can occur within a gene preventing the synthesis of the correct protein, they may occur in gene promoter regions or in DNA regulatory regions changing the expression levels of the protein.

Mutations are rare events occurring at a rate of 1 in every 50 million bases added to the nucleotide chain. Most of the changes that happen are quickly repaired by our very efficient and accurate DNA repair system. However, this repair mechanism is not foolproof. Failure to repair all mutations has led to the introduction of some mutations that have made organisms fitter and better able to adapt to their environments. However, many mutations have detrimental effects for an organism and it is these mutations that are the basis of many human genetic disorders and disease.

Gene mutation:

Genes are segments of DNA located on chromosomes . A gene mutation is defined as an alteration in the sequence of nucleotides in DNA. This change can affect a single nucleotide pair or larger gene segments of a chromosome. Mutations cause changes in the genetic code that lead to genetic variation and the potential to develop disease.

Gene mutations can be classified into three types :

1) **Substitutions:**These are gene mutations where one or more base pairs are substituted or replaced by others. These are further divided into two types –
   a) **Transitions:** In these one purine is replaced by another purine or one pyrimidine is replaced by another pyrimidine.
   b) **Transversions:** In these a purine is replaced by a pyrimidine or vice versa. Substitutions are also referred as point mutations.
2) **Insertions:**These are gene mutations where one or more additional bases (nucleotides) are inserted or incorporated into the DNA molecules.
3) **Deletions:**These are mutations where one or more bases (nucleotides) are lost from the DNA molecule.

Spontaneous and Induced Mutation:

A) Natural or Spontaneous Mutations:

Spontaneous mutations occur suddenly in the nature and their origin is unknown. They are also called as Background mutation. Exa. – Oenothera, maize, bread molds, microorganisms (bacteria and viruses), Drosophila, mice, man etc.

Some spontaneous mutations arise by the action of mutagens present in the environment. These are cosmic radiation, radioactive compounds, heat and naturally occurring base analogues like caffeine.

B) Induced Mutations:

Mutations can be induced artificially in the living organisms by exposing them to abnormal environment such as radiation, certain physical conditions (temperature) and chemicals are called as Induced mutation. The substances or agents which induce artificial mutations are called as Mutagenes or Mutagenic agents.

Chromosomal Aberration or Chromosomal Mutations:

The morphological character (structure) of an animal is constant i.e. all rats or cats look similar. This similarity in character depends upon genes present in a chromosome. Normally the structure and number of chromosome and hence the genes are constant. But sometimes due to accidents or irregularity, particularly at the time of cell division, the structure or number of chromosome may change. This change in chromosomes results in the change of phenotypic characters of an animal or plant.

This change in the organization of chromosomes or alteration in chromosomal structure is known as Chromosomal Aberration or Mutation, resulting in a sudden change in the character of an organism.

There are two types of Chromosomal aberrations –variations in chromosome structure or morphology and numerical changes in chromosomes.

A) Variations in Chromosome Structure or Morphology:

Generally chromosomes segregate themselves in an orderly manner during cell division and produce haploid cells or gametes containing ‘n’ chromosomes which possess genes in a definite linear arrangement. Each gene is present at a fixed locus or position. However, sometimes this definite arrangement of the gene breaks up, producing change or mutation. Such change in the arrangement or structure of genes on a chromosome is called as Chromosomal aberrations. Such changes result in the alteration of phenotypic characters of the individuals.

There are various types of structural changes in a chromosome. They are,

1) Deletion or Deficiency:

When some part of a chromosome is lost, this is known as **Deletion or Deficiency**. The chromosome in this case breaks up at two places, the broken part gets separated while the two ends of the chromosome join together and give rise to a mutated one.

Deficiencies have an effect on inheritance also. In presence of a deficiency, a recessive allele will behave like a dominant allele (pseudo-dominance). This principle of pseudo-dominance exhibited by deficiency heterozygotes has been utilized for location of genes on specific chromosomes in Drosophila, maize and other organisms.

In human beings deletion in the 5^{th} pair of homologous chromosomes results in a condition known as Cri-du-chat syndrome. This syndrome results from the loss of the short arm of the V chromosome. The person is physically retarded and produces a sound like the cry of a cat, hence the name cat's cry or Cri-du-chat.

Experimentally deletions are useful in studying the locus of genes on a chromosome.

2) Duplication or Addition:

Sometimes the deleted portion of a chromosome becomes attached to another chromosome at the centromere, thus there is a double or duplicated part in a chromosome. This type of aberration is called as Duplication or Addition.

There is a repetition of a chromosomal segment, when this extra part gets attached to the centromere, and it behaves like an independent chromosome. A germ cell which gets such a duplicated chromosome receives extra genes in duplicated form, resulting in a new species and it is important in evolution.

The bar eye of Drosophila is duplication of small segment (16 A region) of the X chromosome.

3) Inversion:

During inversion the chromosome breaks at two points and the central piece gets detached. This broken piece then gets reattached in its original position with the two ends reversed. This is due to formation of loop.

Inversion occurs mostly in meiosis and causes a chromosome with some of the genes in the reverse order.

There are two types of inversions –

a) Pericentric Inversion: When centromere is present in the region of inversion, it is called as Pericentric inversion. Such inversions result in a dicentric chromosome. Exa. Maize.

b) Paracentric Inversion: When centromere is not involved in inversion, it is called as Paracentric inversion.

4) Translocation:

These are change in the arrangement of genes in the chromosome. As a result the quality and quantity of the genes does not change and hence translocations are also known as chromosomal arrangements.

The phenotypic characters of individuals having translocations in their chromosomes are normal except that the posisition of their genes are changed. There is shifting of a part of one chromosome to another non-homologous chromosome. When parts of two non-homologous chromosomes breaf off and exchange places with each other it is known as Reciprocal translocation.

There are two types of translocation –

a) **Homozygous Translocations:**In which, the linkage groups of the genes change and their positions (loci) change with their homologous counterparts.

b) **Heterozygous Translocations:** In which, there is a change in meiotic division in the prophase, which produces a cross shaped pairing configuration. Such configuration is observed in polytene chromosome of maiza and salivary gland chromosome of Drosophila.

In Oenothera, translocation produces new speceis. Thus, addtion or substraction of genes changes their linkages groups and it plays an important role in evolution.

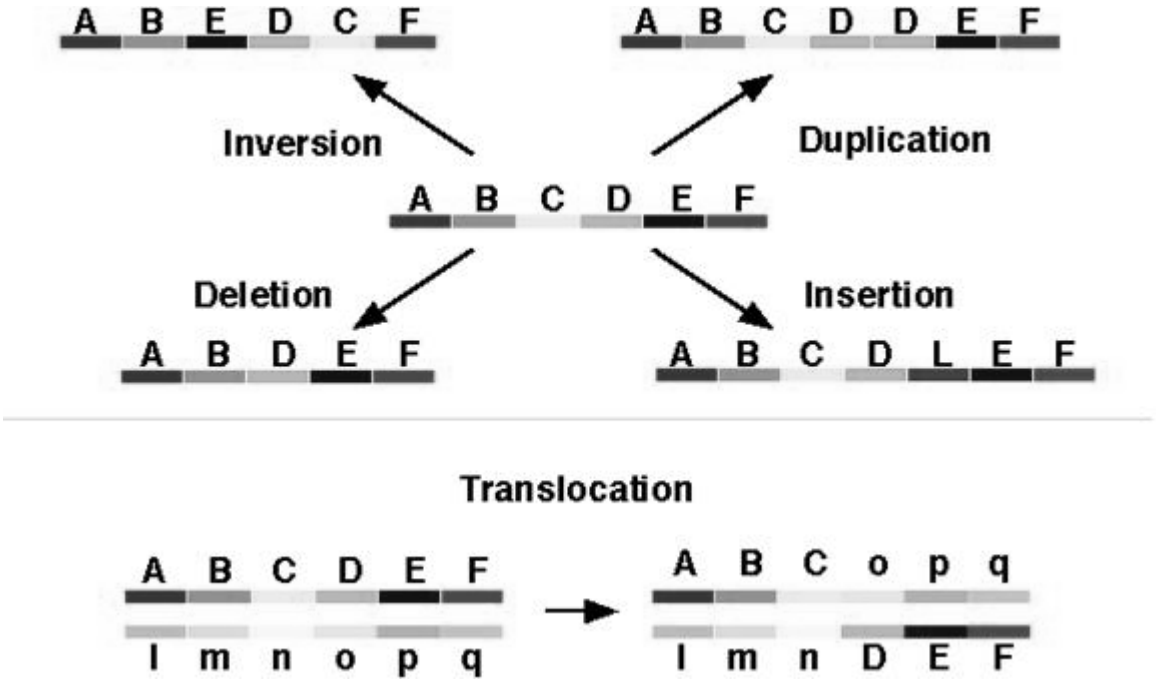


Fig. : Structural changes in chromosome.

B) Numerical Changes in Chromosomes:

Each organism, plant or animal has a fixed number of chromosomes in its cells. The set of chromosomes present in one cell is known as Genome. Each organism has a fixed or specific genome in the cell, which brings specific characters of that particular organism. However

sometimes this fixed number changes suddenly, which may occur during cell division, mitosis or meiosis or during fertilization causing a change in the fixed number.

Variation in chromosome numbers or numerical change is chromosome is called as Ploidy. A ploidy can occur either in the complete set (genome) of the chromosome or it may occur in a single chromosome.

Numerical changes in chromosomes are two types - Euploidy and Aneuploidy.

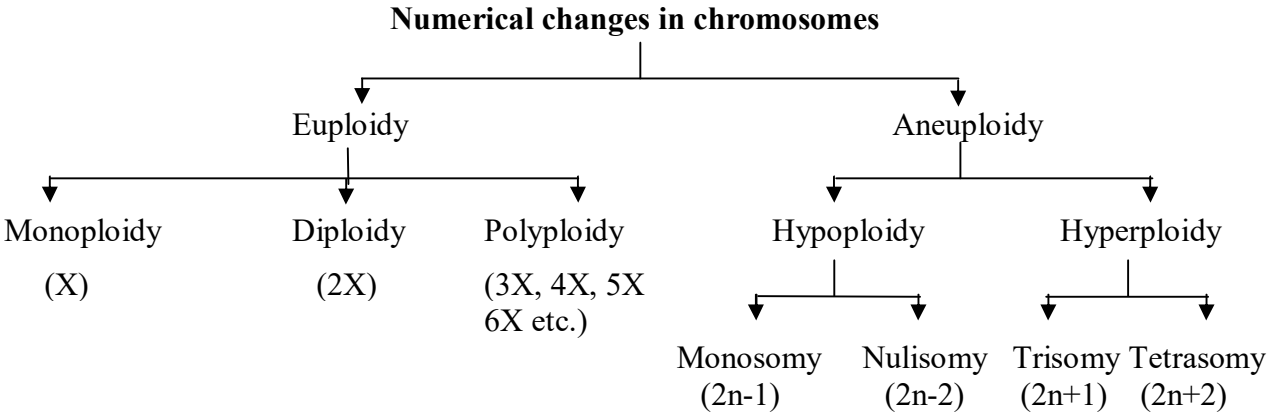


Fig. : Different kinds of numerical changes in chromosomes

1) Euploidy:

Living organisms have either have n or 2n number of chromosomes. Euploidy is the condition in which changes in the number of chromosomes in the living cells occur in multiples of odd numbers. The change in number could occur as a single set (monoploidy) or in many multiples of the basic set of chromosomes. It does not result in genetic unbalance.

The change in number (ploidy) takesplace in a set of chromosomes is called as Euploidy. The euploidy is divided into -

i) Monoploidy or Haploidy (n): The cells of an individual contain one genome or basic set in their nuclei, this condition is called as monoploidy or haploidy and the organism is called as monoploid. The number of chromosomes in haploid cells is denoted by 'n'. Thus the individual is hemizygous for containing single set.

Monoploidy is rare in animals. Exa. – Rotifers, drones of honey bee and wasps.

The haploidy may be normal or abnormal to the particular species. The haploidy can be produced in plants, but these plants are small, with small leaves usually remain sterile. Exa. – Algae, fungi, sorghum, Triticum, hordem, datura, nicotiana etc, Haploid amphibian embryos have been reported but they rarely reach the adult stage.

Haploids originate spontaneously due to parthenogenetic development of egg in plants and animals. Such rare haploids have actually been obtained in tomatoes and cotton under cultivation. Rarely haploids may originate from pollen tube rather than form egg, synergids or antipodals of embryo sac. These haploids will be called androgenic haploids.

Haploids can be artificially produced by X-rays treatment, delayed pollination, temperature shocks, colchicine treatment, distant (interspecific or intergeneric) hybridization and anther or pollen culture.

ii) Diploidy (2n): Diploidy is a condition in which due to certain reasons the chromosomes occur as twice the haploid number. Diploidy is possible in living organisms which are haploid like the lower group of plants. Higher living organisms cannot be considered diploid because normally they have twice the basic number of chromosomes (2n).

iii) Triploidy (3n): The triploid organisms contain 3 sets of chromosomes (3n) in the nuclei of their body cells. The triploidy may originate from the union of a haploid (n) and diploid gametes (2n) of different strains of the same species, such organisms are always sterile because of the difficulties in chromosome pairs or non-disjunction during meiosis.

Ohno (1963) and Pennock (1965) studied triploidy in reptiles, man and some cells of birds and other vertebrates.

However, triploid plants can be propogated by asexually shows much economic value. Exa. – Several varieties of apples that possess superior qualities are triploid and are propogated by grafting or budding technique.

iv) Tetraploidy (4n): The organisms with four sets or four genomes (4n) in the nuclei or their somatic cells are called as Tetraploids. This doubling in the chromosomal number may be due to either spontaneously or artificially by treating the cells of organisms with chemicals such as chochicine, acenaphthene and vevarine or by exposure to temperature and electric shocks. Exa. – Wheat, solanum. Chrysanthemum, salamander and man.

v) Polyploidy (Multiple Set of Chromosomes)**:** Polyploidy is the condition in which an organism contains more than the usual two sets or genomes of chromosomes. Such animals are called as polyploidy organisms. They may have three, four or more sets of chromosomes.

This phenomenon occurs very rarely among animals. Polyploidy has been induced in the tomatoes, apples, chickoos, grapes, strawberry plants, wheat and jowar. Among human beings polyploidy is rare and has been observed only in cancer cells.

There are three kinds of polyploids - autopolyploids, allopolyploids and segmental allopolyploids.

a) Autopolyploids: Autopolyploids are those polyploids, which have the same basic set of chromosomes multiplied. For example, if a diploid species has two similar sets of chromosomesorgenomes (AA), an autotriploid will have three similar genomes (AAA) andan autotetraploidwill have four such genomes (AAAA).

The autopolyploids may occur in nature or may be artificially produced. When they are found in nature, their autopolyploid nature is inferred mainly by their meiotic behaviour.

The very common examples of natural auto-polyploidy relevant to Northern India is ‘doob’ grass (*Cynodon dactylon*). In U.P. and Bihar, common ‘doob’ grass was found to be an autotriploid as

inferred from its meiotic behaviour. It is perhaps successful due to efficient vegetative reproduction, because, it is normally triploids and set no seeds.

Autotriploids are also known in watermelons, sugarbeet, tomato, grapes and banana although in several of these cases the polyploids have been artificially produced.

Similarly autotetraploids are known in rye (*Secale cereale*), corn (*Zea mays*), red clover (*Trifolium retense*), berseem (*Trifolium alexandrium*), marigolds (*Tagetes*), snapdragons (*Antirrhinum*), *Phlox,* grapes, apples, etc.

b) Allopolyploids: Polyploidy may also result from doubling of chromosome number in a F_1 hybrid which is derived from two distinctly different species. This will bring two different sets of chromosomes in F_1 hybrid. The number of chromosomes in each of these two sets may differ.

Let A represent a set of chromosomes (genome) in species X and let B represent another genome in a species Y. The F_1 will then have one A genome and another B genome. The doubling of chromosomes in this F_1 hybrid (AB) will give rise to a tetraploid with two A and two B genomes (Fig.). Such a polyploid is called an Allopolyploid or Amphidiploid.

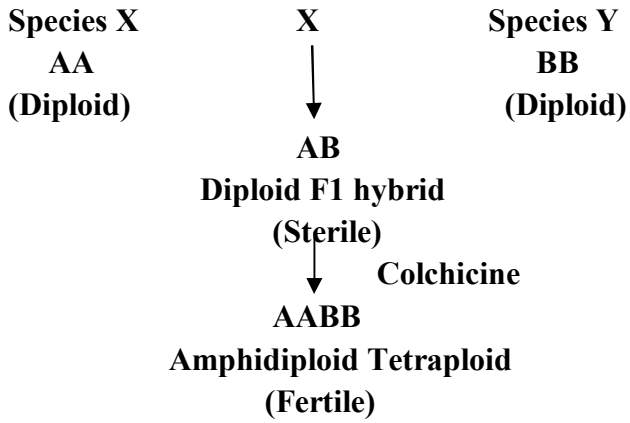


Fig. : Derivation of a Tetraploid amphidiploid from two Diploid species.

Raphanobrassica is a classical example of allopolyploidy. In 1927, G.D.Karpechenko a Russian geneticist reported a cross between *Raphanus sativus* ($2n = 18$) and *Brassica oleracea* ($2n = 8$) to produce F_1 hybrid which was completely sterile. This sterility was due to lack of chromosome pairing, since there is no homology between genomes from *Raphanus sativus* and *Brassica oleracea.* Among these sterile F_1 hybrids, Karpechenko found certain fertile plants. On cytological examination these fertile plants were found to have $2n = 36$ chromosomes, which showed normal pairing into 18 bivalents.

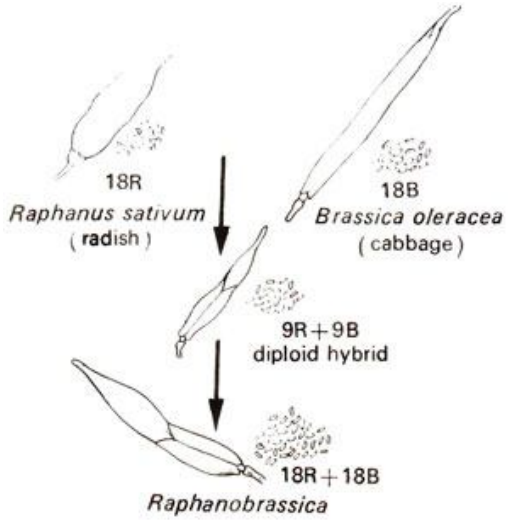


Fig. : Artificial synthesis of Raphanobrassica.

c) Segmental Allopolyploids: In some allopolyploids, the different genomes which are present are not quite different from one another. Consequently, in these polyploids chromosomes from different genomes do pair together to some extent and multivalents are formed. This means that segments of chromosomes and not the whole chromosomes are homologous. Therefore, such allopolyploids are called segmental allopolyploids. These segmental allopolyploids are intermediate between autopolyploids and allopolyploids and can be identified by their meiotic behaviour.

It is also believed that most of the naturally occurring polyploids are neither true autopolyploids nor true allopolyploids but are rather segmental allopolyploids. Our common hexaploid bread wheat is also regarded to be a segmental ailopolyploid, because the three diploid genomes (A, B and D) are related (homoeologous) to each other.

iii) Aneuploidy:

Aneuploidy is a change in the number of chromosomes in a set (i.e. basic number). The chromosome set either has one or more extra chromosome or one or more chromosomes less than the normal complement. Organisms with chromosome numbers that are not exact multiple of the monoploid set are called as Aneuploid.

Thus, the somatic chromosome number of Aneuploid differs from normal by an amount that is not an exact multiple of some basic haploid number. The number may be 2n + 1, 2n + 2 or 2n – 1, 2n – 2 and so on.

Aneuploidy was first discovered by Bridges in Drosophila in 1916. This condition is due to the non-disjunction, where homologous chromosomes fail to separate during meiotic division. As a result some gametes have both homologous chromosomes while others do not have them. Fertilization of such gametes will produce zygotes either with one additional chromosome or with

one less chromosome, resulting in Aneuploidy.Aneuploids are therefore, unbalanced individuals and show phenotypic changes.

Aneuploidy may be of two types Hypoploidy and Hyperploidy.

i) Hypoploidy:

One or more chromosomes are loss from a set of genome is called as Hypoploidy. It may be monosomic or nulliosomic.

a) Monosomic condition (2n – 1): In this case there is one chromosome less in one pair i.e. 2n – 1 condition. The monosomic parent forms two types of sex cells – (n) type (haploid) and (n–1) type (one less than normal haploid cell) during gametogenesis.

Normally (n – 1) type gametes will die, but if they survive the resulting offsprings will have a genetic imbalance resulting in reduced fertility or high mortality.

In plants, gametes with (n – 1) genome do not survive while in animals that cause genetic imbalance and leads either high mortality or reduced fertility.

Exa. – In Fruit fly, Drosophila, small chromosome (4^{th}) is missing leads to monosomic condition referred to Haplo-IV, such individuals develop slowly and shows reduced body size and have impaired viability.

Among human beings, individuals with a single chromosome (Y or the other X chromosome is missing) of sex chromosome resulting in Turner's syndrome. Such individual has the external appearance of a female but lacks ovaries or a degenerate ovary, is sterile.

b) Nullisomic (2n – 2): Here the organisms loss a pair of chromosomes. They have 2n – 2 genomic formula. Nullisomic type can arise from the union of two monosomic gametes or from non-disjunction of chromosomes. Nullisomic individuals usually do not survive but their polyploidy forms may survive but weak and sterile.

Exa. – Blackslee and his co – workers produced nullisomic condition in *Datura stramonium* and Sears (1904) established 17 hexaploid nullisomic in wheat (*Triticum vulgare*), but the plant has reduced vigour and vitality.

ii) Hyperploidy:

The hyperplpoids are individuals with increase in one or more chromosomes to a set i.e. trisomic (2n + 1), tetrasomic (2n + 2) and so on. Such condition usually called as Plolysomic. It may be trisomic, tetrasomic or pentasomic.

a) Trisomic condition (2n + 1):

In this case there is an extra chromosome i.e. 2n + 1condition. During gametogenesis, the resultant gametes will be two types – (n) type gamete, usual haploid sex cell and (n+1) type gamete, containing an extra chromosome.

The individuals produced due to trisomy will not have normal characters. In the human being trisomies can occur with any chromosome, but often result in miscarriage.

b) Tetrasomic condition (2n+2): In this case diploidcontains two extra chromosomes (2n + 2). The tetrasomic condition produces some useful hybrids like wheat.

Sometime instead of addition of chromosomes a pair of chromosomes are lost, it is called as Nullisomic condition, resulting in change in the genetic formula 2n – 2. Such a nullisomic individuals are rarely survives. In wheat a hexaploid nullisome is reported to survive, but the plant has reduced vigour and vitality.

Disorders related to Chromosomal Number:

Turner Syndrome:

In 1938, H. H. Turner first described this syndrome. These individuals are phenotypically females with 45 (44 + XO) number of chromosomes. This is the monosomic condition due to missing Y chromosome. The frequency of such individuals in population is one in about 5000 people.

The individuals of this syndrome are characterized by ovarian dysgenesis shows incomplete development of ovaries. The females show other physical abnormalities like short stature, a short neck with webbed skin, low set of ears, dystrophy of nails, ahigh arched palate with abnormal jaws. The chest is broad with widely spaced nipples, colorblindness. The uterus may be represented by hard string in the mid-line is more common than the normal women.

Inspite of these physical abnormalities of various organs, the nervous system is not affected and individuals with this syndrome have normal brain development that is they do not have tendencies towards mental retardation.

Klinefelter Syndrome:

In 1942 H. F. Klinefelter reported this abnormality in phenotypically male individuals. Later on in 1959 Jacob and Strong demonstrated that these individuals have 47 chromosomes i.e. trisomy (XXY) to chromosome number. They possess two X chromosomes and a Y chromosome. It is caused by non-disjunction of XX chromosomes.

The affected individuals appear normal in childhood but the abnormalities become visible only in adult males. This syndrome is characterized by enlargement of the breasts, small testes and absence of spermatogenesis, amount of male hormone is low, genitalia are poorly developed and sterile. They are tall and feminine fat deposits and female distribution of abdominal and facial hair.

In a population the persons of this syndrome can be detected by mentally retarded with infertility and develop a variety of psychiatric problems.

Down Syndrome:

It is a genetic condition that causes delays in physical and intellectual development. It was first described by Down in 1866. These individuals are phenotypically females. It is causes due to trisomy in 21st pair of chromosome. It is causes due to non-disjunction of chromosomes of pair 21 during meiosis. Hence both chromosomes of this pair enter the same egg. When this egg is fertilize by a normal sperm, Down syndrome results.

It occurs in one in every 691 live births. Individuals with Down syndrome have 47 chromosomes instead of the usual 46. It is the most frequently occurring chromosomal disorder.

Down syndrome is also known as Trisomy 21, because the person has three copies of chromosome 21 instead of two. There are three types of Down syndrome.

a) Trisomy 21: About 95 percent of the time, Down syndrome is caused by trisomy 21. The child has three copies of chromosome 21 (instead of the usual two copies) in all cells. This is caused by abnormal cell division (non-disjunction) during the development of the sperm cell or the egg cell.

b) Mosaic Down syndrome: This is rare form of Down syndrome. Children have some cells with an extra copy of chromosome 21. This mosaic of normal and abnormal cells is caused by abnormal cell division after fertilization.

c) Translocation Down syndrome: Down syndrome can also occur when part of chromosome 21 becomes attached (translocated) onto another chromosome, before or at conception. These children have the usual two copies of chromosome 21, but they also have additional material from chromosome 21 attached to the translocated chromosome.

The facial features of the victims resemble Mangolian race.Flattened face, mouth is constantly open and the tongue is protruded, teeth small. Outwardly slanted eyes, neck is short and broad, nose is oblique. The ears are flat, set low on the head. They are mentally retarded. She is dwarf, arms and legs are short.

Important Questions

Q.1 Multiple choice questions (1 marks each)

1) Point mutations are

a)Change in chromosome number b) Change in chromosome structure

c) Change at DNA level **d) the single base substitution**

2) A micromutation is

a) Polyploidy b) Additions of Chromosome

c) a small-scale or highly localized mutation d) Deletion of chromosome

3) Polyploidy refers to –

a) Extra copy of gene

b) Extra set of chromosomes

c) The chromosome which has replicated but not divided

d) Multiple ribosomes on the single mRNA

4) Klinefelter syndrome is the result of

a) **Sex chromosome abnormality** b) Autosomal abnormality

c) Morphological abnormality d) only (b) and (c) correct

5) Aneuploidy arising through loss of chromosomes

a)Hypoploidy b) Nanoploidy c) Lethoploidy d) Aploidy

6) Which of the following are capable of causing chromosomal mutations?

a) hydrogen peroxide b) high body temperature

c) X-rays d) **all of the above**

7) What type of mutation has occurred when a plant inherits a complete extra set ofchromosomes?

a) **Polyploidy** b) Euploidy c) Aneuploidy d) all of the above

8) Polyploidy refers to:

a) extra copies of a gene adjacent to each other on a chromosome

b) an individual with complete extra sets of chromosomes

c) a chromosome which has replicated but not divided

d) multiple ribosomes present on a single mRNA

9) Which of the following is correct with regard to aneuploidy?

a) Inversion **b) 2n + 1**

c) All aneuploid individuals die before birth d) 4n

10) Mongolian idiocy due to trisomy in 21st chromosome is called

a) **Down's syndrome** b) Turner's syndrome

c) Klinefelter's syndrome d) Triple X syndrome.

11) Those mutations that arise in the absence of known mutagen are known

a) Induced mutations b) Fused mutations

c) **Spontaneous mutations** d) None of the above

Q.2 Define /explain /comment (2 marks each)

1) Mutations 2) Pseudo-dominance

3) Deletion of chromosomal segment 4) Duplication

5) Translocation 6) Position effect

Q. 3 Question for (4mark each)

1) Write a brief note on Turner's syndrome. 2) Position effect

3) Write a brief note on Klinefelter's syndrome. 4) Causes of polyploidy

5) Write a brief note on Down's syndrome.
6) Aneuploidy
7) Phenotypic effect of autopolyploidy.
8) Euploidy
9) Autopolyploid
10) Allopolyploid
11) Haploids
12) Syndrome
13) Nullisomic
14) Trisomic
15) Tetrasomic

Q.4Question for (8mark each)

1) Give a brief account on origin of numerical chromosomal abnormalities.
2) Explain in brief the different types of structural changes causing chromosomalaberrations.
3) Describe in brief various kinds of numerical changes in chromosomes.
4) What are the genetic effects structural changes in chromosome.
5) Discuss the any two syndromes in human, known to result from numerical changes inchromosome.
6) Write a detail account on different chromosomal aberrations.
7) Write the chromosomal complement of Down's syndrome.
8) What is the chromosomal complement of Turner's syndrome?
9) Explain intrachromosomal changes in structure of chromosome with reference toduplication and inversion of segments.
10) What are the different kinds of polyploids? How will you distinguish betweenautopolyploids and allopolyploids?
11) What do you understand by spontaneous mutation and induced mutation? Discussvariation in mutation frequencies at different loci within an organism.
12) Why are salivary gland chromosome of *Drosophila* commonly used for study ofstructural changes in chromosomes?
13) Describe and illustrate how: 1) deletions, 2) inversion, and 3) reciprocal translocationarise nature.
14) Sketch and label diagrams to show various types of structural changes inchromosomes.

Q.5 Distinguish between (4 marks)

1) Double monosomic and nullisomic
2) Primary trisomy and secondary trisomy
3) Autopolyploids and allopolyploids
4) Haploidy and monoploidy

Unit - 10

Population Genetics

Basic Concepts of Population Genetics:

Population genetics is the study of the distributions and changes of allele frequency in a population, as the population is subject to the four main evolutionary processes - natural selection, genetic drift, mutation and gene flow.

Gene Pool:

The collection of all the alleles of all of the genes found within a freely interbreeding population is known as the **Gene pool of the population**. Each member of the population receives its alleles from other members of the gene pool (its parents) and passes them on to other members of the gene pool (its offspring). Population genetics is the study of the variation in alleles and genotypes within the gene pool and how this variation changes from one generation to the next.

Factors influencing the genetic diversity within a gene pool include population size, mutation, genetic drift, natural selection, environmental diversity, migration and non-random mating patterns.

Gene frequency and Genetic drift:

Genetic drift is a change in allele frequencies caused by random sampling. That is, the alleles in the offspring are a random sample of those in the parents. Genetic drift may cause gene variants to disappear completely, and thereby reduce genetic variability. In contrast tonatural selection, which makes gene variants more common or less common depending on their reproductive success, the changes due to genetic drift are not driven by environmental or adaptive pressures, and may be beneficial, neutral, or detrimental to reproductive success.

The effect of genetic drift is larger for alleles present in few copies than when an allele is present in many copies. Scientists wage vigorous debates over the relative importance of genetic drift compared with natural selection. Ronald Fisher held the view that genetic drift plays at the most a minor role in evolution, and this remained the dominant view for several decades. In 1968 Motoo Kimura rekindled the debate with his neutral theory of molecular evolution which claims that most of the changes in the genetic material are caused by neutral mutations and genetic drift. The role of genetic drift by means of sampling error in evolution has been criticized by John H Gillespie and Will Provine, who argue that selection on linked sites is a more important stochastic force.

The population genetics of genetic drift are described using either branching processes or a diffusion equation describing changes in allele frequency. These approaches are usually applied to the Wright-Fisher and Moran models of population genetics. Assuming genetic drift is the only

evolutionary force acting on an allele, after t generations in many replicated populations, starting with allele frequencies of p and q, the variance in allele frequency across those populations is

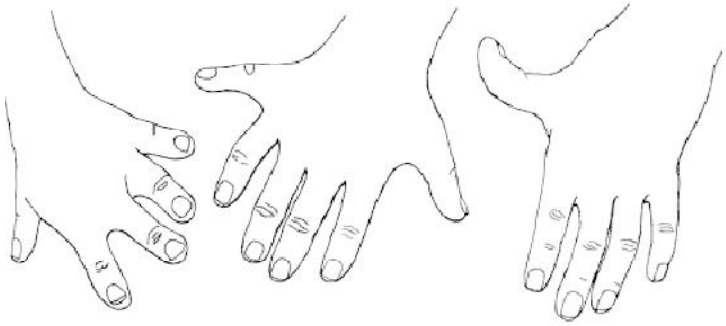
$$V_t \approx pq\left(1 - \exp\left\{-\frac{t}{2N_e}\right\}\right).$$

Hardy Weinberg equilibrium and its significance:

During the 20th century, scientists showed great interest in applying the theories of Mendel to human traits. They were able to identify a dominant trait namely brachydactyly that agreed with the Mendelian theories and showed Mendelian ratio in the human populations. Brachydactyly is characterized by abnormal shortening of the phalanges while the rest of the portion of the arms remains normal.

An objection to this example was raised. If the trait brachydactyly is a dominant character, three-fourth of the human populations should possess the abnormally shortened phalanges. But in reality brachydactyly is seen rarely in human population. This occurrence appeared to contradict Mendelian theories. However, until 1908 it remained unexplained among the scientists.

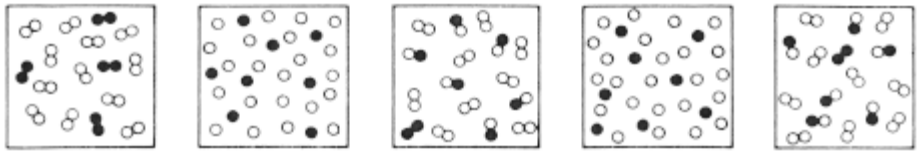
Two scientists G.E. Hardy and Wilhelm Weinberg (worked independently) put forth the theory Hardy-Weinberg Law. This theory explains why some characters though dominant (like brachydactyly) appear rarely in a population.



brachydactyly polydactyly hitchhiker's thumb

Fig. Human traits

This law states that under specified conditions the genotypic frequency in a population remains constant. It simply means, the common traits remain common and the rare traits remain rare.It further explains that the Mendelian terms dominantand recessive refer to the phenotypic appearance of the heterozygotes and not to the abundance of any trait in the population. The relative abundance of a genotypye in apopulation is referred to as its genotypic frequency.



parents (homozygotes) **gametes** **offspring** **gametes** **offspring**

A second component of the Hardy–Weinberg principle concerns the effects of a single generation of random mating. In this case, the genotype frequencies can be predicted from the allele frequencies. For example, in the simplest case of a single locus with two alleles: the dominant allele is denoted **A** and the recessive **a** and their frequencies are denoted by p and q; freq (**A**) = p; freq (**a**) = q; $p + q = 1$. If the genotype frequencies are in Hardy–Weinberg proportions resulting from random mating, then we will have freq (**AA**) = p^2 for the **AA** homozygotes in the population, freq (**aa**) = q^2 for the **aa** homozygotes, and freq(**Aa**) = $2pq$ for the heterozygotes.

It is clear that this law is applicable only to populations with followingfeatures

a) Infinitely large populations, b) No mutation

c) No selection d) Random mating

e) Isolated from any other population of the same species.

This law could be mathematically proved not only to a new populationbut also to the populations already existing with equilibrium. Such populationsthat has reached equilibrium remain unchanged from generation aftergeneration. This means that they remain constant and have no chance forevolution.

1. If a population of fruit flies has a gene frequency of 20% for the recessiveallele causing vestigial wings, what proportion shows this trait phenotypically?What proportion of the population is heterozygous?
2. Assume that brown eyes are dominant to blue eyes. On a certain islandabout 9% of the population is blue eyed. What proportion of the population isheterozygous.

Important Questions

Q.1 Multiple choice questions (1 marks each)

1) Imagine a species where there is a locus with two alleles, *A* and *B*. Further imaginethat *A* and *B* are codominant. How many different *phenotypes* are possible in thisspecies?

a) 1 b) 2 c) **3** d) 4

2) Members of the same species which are capable of interbreeding is best described as a(n)

a) Community **b) population** c) ecosystem d) biosphere

3) Which of the following formulas lets you predict the genotypic frequencies of thenext generation?

a) p + q = 1 b) X2 = sum [(o - e)2 / e]

c) p2 + 2pq + q2 = 1 d) d = 0.5 lambda / n * sin (theta)

4) The total aggregate of alleles in a population is referred as:

a) The gene pool b) The allelic frequency

c) The genotypic frequency d) The genetic structure

5) The *ABO* blood group has three alleles (*A*, *B*, *O*). If a woman with genotype *AO* mateswith a man with genotype *BB*, the expected proportion of *heterozygotes* among theiroffspring is

a) 25 percent. b) 50 percent. c) 75 percent. **d) 100 percent.**

6) The ABO blood group has three alleles (A, B, O) where A and B are codominant andO is recessive. If a man with type O blood mates with a woman with type A blood, theexpected proportion of heterozygotes among their offspring is _____ dependingon the genotype of the woman.

a) 0 or 25 percent. b) 25 or 50 percent. **c) 50 or 100 percent.** d) 75 or 100 percent.

Q.2 Define /explain /comment (2 marks each)

1) Genome 2) Progeny 3) Pure line 4) Population genetics

Q.3 Attempt/ Question for (4mark each)

1) Explain the significance of pedigree analysis.
2) Gene frequency
3) Genetic drift.
4) Natural populations do not usually fit the Hardy-Weinberg Equilibrium model. Whatdoes this tell us about most natural populations?
5) Describe Hardy Weinberg equilibrium and its significance.

Q.4 Problem solving (4mark each)

1) The compound phenylthiocarbamide (PTC) tastes very bitter to most persons. Theinability to taste PTC is controlled by a single recessive gene. In the American whitepopulation, about 70% can taste PTC while 30% cannot (are non-tasters). Estimate thefrequencies of the Taster (T) and nontaster (t) alleles in this population as well as thefrequencies of the diploid genotypes.
2) p + q = 1 for any population, so if p = 0.8, then q must = 0.2. Hardy-Weinberggenotypic frequencies are calculated by plugging p and q into the Hardy-Weinbergequation.
3) The allele for the ability to roll your tongue is dominant over non-rollers. In apopulation of 500 people, 100 people are non-rollers. How many people would youexpect to be homozygous dominant or Heterozygous?
4) In a certain population, the dominant phenotype of a certain trait occurs 91% of thetime. What is the frequency of the dominant allele?
5) The ability to test PTC is a dominant character to inability to test. So In SNDT College,among the graduate student out of 252 students, 189 are found taster, 63are found non-tester Calculate the, a) Frequency of recessive allele, b) Frequencyof dominant allele, c) Frequency of heterozygote and Verify the Hardy Weinbergequilibrium

Unit – 11

Genetic Disorders in Human beings

Introduction, purpose, hereditary diseases and disorders:

A genetic disorder is an illness caused by one or more abnormalities in the genome, especially a condition that is present from birth (congenital). Most genetic disorders are quite rare and affect one person in every several thousands or millions.

Genetic disorders may or may not be heritable, i.e. passed down from the parents' genes. In non-heritable genetic disorders, defects may be caused by new mutations or changes to the DNA. In such cases, the defect will only be heritable if it occurs in the germ line. The same disease, such as some forms of cancer, may be caused by an inherited genetic condition in some people, by new mutations in other people and mainly by environmental causes in still other people. Whether, when and to what extent a person with the genetic defect or abnormality will actually suffer from the disease is almost always affected by environmental factors and events in the person's development.

Genetic disorders are conditions that have some origin in an individual's genetic make-up. Many of these disorders are inherited and are governed by the same genetic rules that determine dimples and red hair. However, some genetic disorders, such as Down syndrome, characterized by heart malformation, poor muscle tone and a flattened face, result from a spontaneous mutation (gene change) that takes place during embryonic (earliest life) development.

Genetic disorders can be classified according to the way in which they develop. If the disorder is transmitted by genes inherited from only one parent, it is said to be an **autosomal dominant disorder**. The term autosome applies to any of the 22 chromosomes that are identical in human males and females. (Chromosomes are structures that organize genetic information in the nucleus of cells). By contrast, disorders that can be inherited only by the transmission of genes from both parents are called an **autosomal recessive disorder**.

Other genetic disorders are associated with the X (female) or Y (male) chromosome and are called **sex-linked disorders** because the X and Y chromosomes are related to sexual characteristics in humans. Finally, the development of some genetic disorders involves environmental factors, factors present outside the organism itself. Such disorders are known as **multifactorial genetic disorders**.

Haemoglobin disorders:

Decreased levels of haemoglobin, with or without the concomitant decrease in red blood cells, can cause anaemia. Iron deficiency is one cause of anaemia, as it directly affects the ability to produce haem molecules, but there are several other causes of anaemia. There can also be other

disease profiles associated with abnormalities in haemoglobin, known generally as haemoglobinopathies, as well as abnormalities affecting the production of haem molecules, known as porphyrias.

Thalassemia:

Thalassaemia is caused when the production of haemoglobin chains is impaired, the most common forms affecting the alpha globin chain (alpha Thalassaemia) or the beta globin chain (beta Thalassaemia). The chains themselves can be normal, but the amounts produced are not; sometimes the genes can even be missing. There are four genes needed to make the alpha globin chain, with moderate to severe anaemia resulting when more than two genes are affected. With the beta globin chain there are two genes required, the most severe form of the disease affecting both genes. An equal number of alpha and beta globin proteins are required to make functional adult haemoglobin, and a deficiency in either chain will cause an imbalance that damages and destroys red blood cells, thereby producing anaemia. The deficiency in globin chains can cause the an abnormal association of globin chains: in the case of alpha Thalassaemia, beta globin chains combine to produce abnormal beta tetramers that cannot bind oxygen, whereas with beta Thalassaemia no such alpha tetramers exist – instead the alpha globin chains become degraded in the absence of beta globin chains.

Sickle Cell Anaemia:

Sickle Cell Anaemia affects the shape of red blood cells, changing them from a flattened disc to a sickle or crescent shape. Whereas normal red blood cells are smooth and move easily through blood vessels, sickle blood cells are hard, inflexible and tend to clump together, causing them to get stuck in blood vessels as blood clots, thereby blocking the flow of blood. This can cause pain, blood vessel damage and a low red blood cell count (anaemia) due to the more fragile nature of sickle blood cells. The abnormal sickle shape is due to the presence of abnormal haemoglobin (haemoglobin S), which contains abnormal beta polypeptide with a single amino acid substitution at position 6 along the polypeptide chain (the alpha chain is normal). The abnormal b chain reduces the amount of oxygen inside the red blood cell, altering its shape.

Heterozygotes, where one beta chain gene is affected and the other is normal, usually display normal red blood cells, and it is only when both beta chain genes are affected (homozygote) that the sickle cell disease is seen. However, heterozygote carriers of the disease are better protected against malaria than people with two normal beta chain genes. This malarial protection has caused the sickle cell gene to reach high levels in indigenous populations in Africa and India.

Inborn Errors of Metabolism:

The biosynthetic pathway involves a number of enzymes and reactions each controlled by a gene. A mutation or defect in a single gene can block a metabolic pathway and produce a heritable genetic disease.

The defective metabolic reactions lead to certain metabolic diseases which appear at the time of birth. So these diseases are called as Inborn errors of metabolism. 1909, A. E. Garrod first studied inborn errors of metabolism in human being. There are three types of inborn error of metabolism – Albinism, Phenylketonuria and Alkaptonuria.

Albinism:

It is the condition where the skin and the hair of the whole body appear colourless due to total or nearly total absence of pigmentation (melanin). It is an inborn error in metabolism. A person with this defect is referred as Albino. In albinos, usually the iris is also devoid of pigment and appears pinkish due to the blood vessels flowing through it. This is why albinos cannot stand in bright light and usually resort to the use of sun glasses.

The production of melanin pigment is the result of a chain of reactions involving the amino acids phenylalanine and tyrosine. Phenylalanine is an essential amino acid supplied in the human diet. Phenylalanine from the digestive tract, diffused into body cells where it is converted into tyrosine with the help of a dominant gene (A). This tyrosine is oxidized to dihydroxyphenylalanine by enzyme tyrosine. Further oxidation results in the production of melanin.

An absence of enzyme tyrosine blocks the chain of reaction in one or more places causing albinism. Thus, albinism is due to absence of tyrosinase enzyme.

When the recessive gene (a) is present in homozygous condition (aa), the enzyme tyrosinase is not produced and it blocks the conversion of tyrosine into dihydroxyphenylalanine results in the non-production of melanin. This condition is known as Albinism. The person suffering from this deficiency is called Albinos.

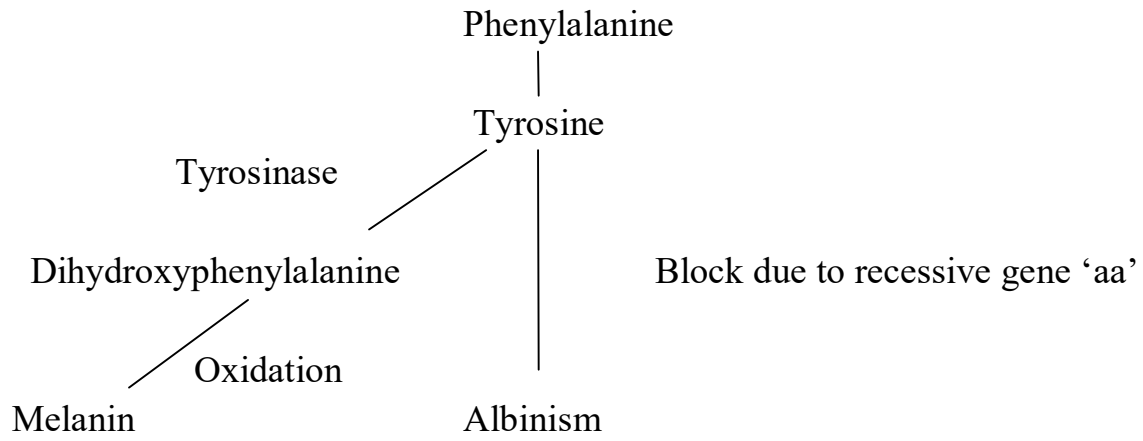


Fig.: Blocking of Tyrosine metabolism in human body.

Phenylketonuria:

This is a rare human abnormality due to a hereditary block in the metabolism of phenylalanine. The affected person excrete large amount of phenylpyruvic acid and phenylalanine in the urine.

An enzyme phenylalanine hydroxylase converts phenylalanine into tyrosine. When this enzyme is absent there is high level of phenylalanine in the body fluids like blood, cerebro-spinal fluid (CSF) and sweat. Besides tyrosine there are few more breakdown products of phenylalanine such as phenylpyruvic acid also formed.

A dominant gene (P) in homozygous or heterozygous condition brings about the conversion of phenylalanine into tyrosine. Its recessive mutant in homozygous state causes failure in the production or functioning of this enzyme. Thus, non-conversion of phenylalanine into tyrosine.

This brings about the accumulation of phenylalanine in large amount in the body fluid. More of phenylalanine is now available to be converted into phenylpyruvic acid. This result in excess of phenylpyruvic acid and phenylalanine in the blood and some amount of these substances is excreted in the urine.

This abnormality is known as Phenylketonuria (PKU). In patients 15-63 mg of phenylalanine/100 ml of plasma present as compared to 1-2 mg/100 ml in normal persons.

People suffering from Phenylketonuria are mentally retarded, having an I.Q. below 20, the idiot level. The high level of phenylalanine in the body tissues interferes with normal development of brain and often fair skin and light hair.

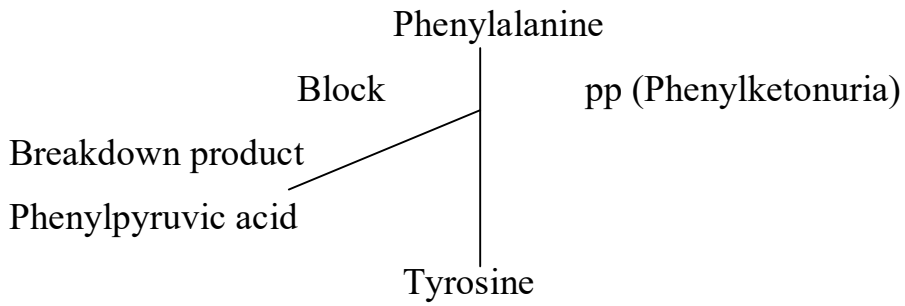


Fig.: Blocking of Phenylalanine metabolism in human body.

Alkaptonuria:

This is the first metabolic disease described by A. E. Garrod in 1909. According to him this is the hereditary block in metabolism of phenylalanine and tyrosine result in the disease Alkaptonuria and characterized by darkening of the cartilage. In the presence of this abnormality certain areas lie close to surface in ear, wrist and elbow showed discolouration.

Another symptom of this disease is that urine of affected person turns black when exposed to air or brought in contact with alkaline solution. This is due to the presence of unusual substance, Alkapton or Homogentisic acid.

Tyrosine is broken into CO_2 and water through a gradual series of reactions with the help of enzymes. This degradation releases energy. This process is normal part of cellular metabolism.

In this series of reaction, tyrosine is converted into parahydroxyphenylpyruvic acid which is then converted into dihydroxyphenylpyruvic acid under the direction of dominant gene (T). The recessive gene (t) in its homozygous condition inhibits the conversion. Which is a result in increase in amount of tyrosine in the system and affected person is said to Tyrosinosis. There is no serious symptom of tyrosinosis except that it is excreted in the urine.

In the next step of reaction, under normal circumstances, the dihydroxyphenylpyruvic acid is converted into homogentisic acid. This is in turn converted into maley-laceto acetic acid under the direction of a dominant gene (H). But its recessive allele (h) in the homozygous condition blocks the conversion of homogentisic acid into maley-laceto acetic acid. This result in excess accumulation of homogentisic acid called Alkapton. This is excreted in urine. It is oxidized on exposure to air and turns black.

A person suffering from this disease does not have ill-effects in early life but in old age it leads to degenerative arthritis (inflammation or swelling of joints) due to slow deposition of homogentisic acid pigments in the body cartilage and joints with advancing age.

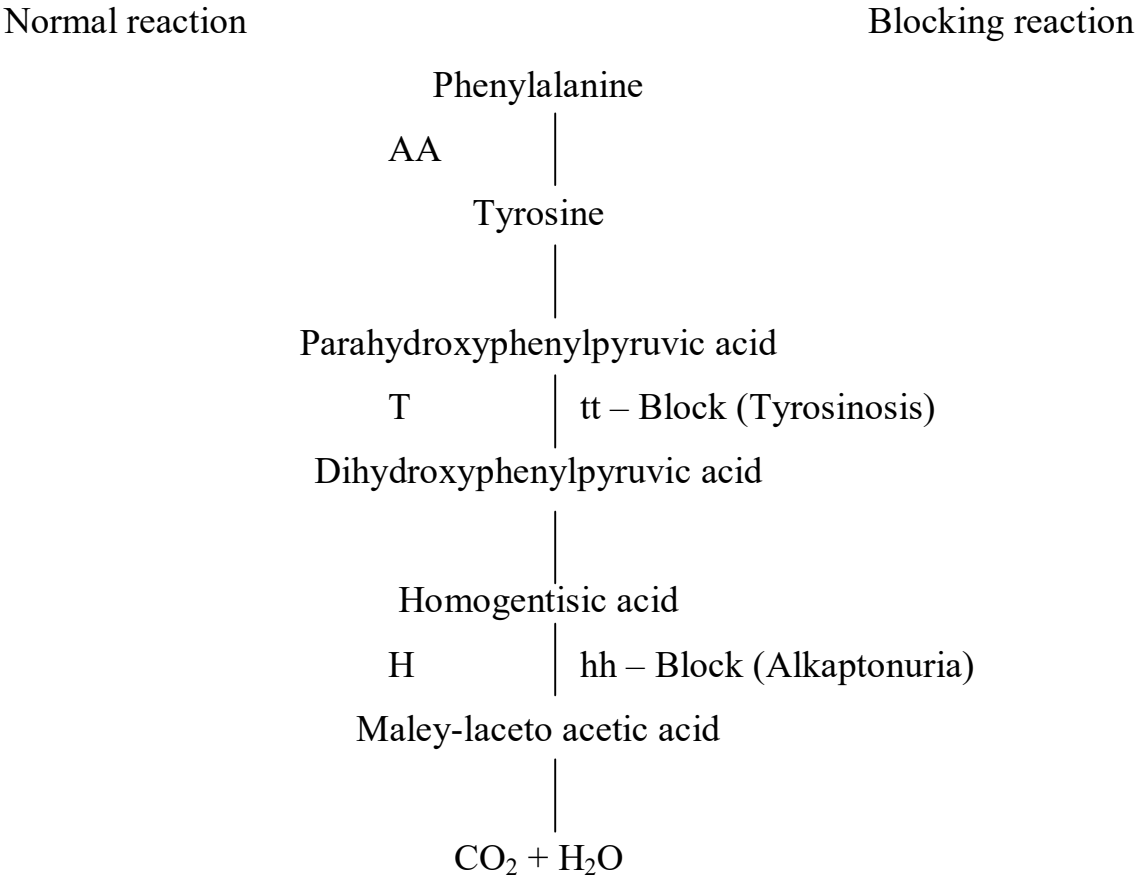


Fig. ; Blocking of Tyrosine metabolism in human body.

Eugenics:

In 1983, Sir Francis Galton derived the term eugenics from Greek word, well born. According to him eugenics is the science which deals with all influences that improve the inborn qualities of a race or the study of eugenics under social control that may improve the racial qualities of the future generation, either physically or mentally. In short, eugenics means to improving the race by ensuring improve progeny.

The Mendelian principles are equally applicable to man and several traits are known which concerns in certain families. Although, is not possible to perform experiments on man. This mode of inheritance of a number of diseases and traits are studied by pedigree analysis. Number of diseases and defects like syphilis, colour blindness, hysteria, haemophilia, polydactylism and syndactylism etc. are all inherited from the parents.

Not only defects and diseases but good qualities like memory, intelligence, ability of speech, mechanical skill, muscle ability and various bodily characters are transferred from one generation to the next.

The work of eugenist is to improve the human race by guiding young people. It is possible to improve the overall qualities of the race by carefully applying the principles of eugenetics.

The eugenic measures are two types – Positive and Negative eugenics.

1) Positive Eugenics:

It includes effective increasing the frequency of desirable traits in the population. There are many ways to bring the positive eugenics. It can done by better education, social hindrance, prevention of germinal waste, subsidizing the fit, immigration, improvement of environmental condition and promotion of genetic research.

2) Negative Eugenics:

It includes the eugenic measures which help in preventing the multiplication of defective genes. It can be done by restriction on marriage, control of immigration, sterilization, segregation and genetic advice.

Genetic Counseling:

It is also called as genetic advice. Eugenist can help in the improvement of the human race by giving guidance to defective couple from having children. To produce healthy progeny should be endeavor (effect) of man as this alone can ensure a better future for humanity.

The families with history of genetic disease, genetic counseling can provide the much needed relief. Genetic counseling can make a significant contribution in this direction.

By careful examination the genetic counselor can detect genetic abnormalities like Down's syndrome and even recessive genes for disease like sickle cell anaemia. Knowledge of the possibilities of transmitting these diseases to offsprings can help a person choosing a marriage partner.

Genetic counseling can do great benefit to human society. The role of genetic counselor is to inform concerned individuals, nature of the mutant condition that concerns them.

Genetic Engineering:

Genetic engineering refers to artificial transfer of genes and their manipulation, addition, deletion, alteration and amplification. Genetic engineering or recombinant technology is the manipulation of DNA from different sources to recombine desired DNA portions for repair, improvement, perfection and matching of a genotype.

Genetic engineering is a set of technologies used to change the genetic makeup of cells, including the transfer of genes within and across species boundaries to produce improved or novel organisms. The techniques involve sophisticated manipulations of genetic material and other biologically important chemicals.

Genetic engineering has applications in medicine, research, industry and agriculture and can be used on a wide range of plants, animals and microorganisms.

In medicine, genetic engineering has been used to mass-produce insulin, human growth hormones, human albumin, monoclonal antibodies, vaccines and many other drugs. In research, organisms are genetically engineered to discover the functions of certain genes.

Industrial applications include transforming microorganisms such as bacteria or yeast or insect mammalian cells with a gene coding for a useful protein. Mass quantities of the protein can be produced by growing the transformed organism in bioreactors using fermentation, then purifying the protein.

Genetic engineering is also used in agriculture to create genetically modified crops or genetically modified organisms.

Applied Genetics:

DNA Fingerprinting:

DNA fingerprinting is a test to identify and evaluate the genetic information called DNA in a person's cells. It is called a fingerprint because it is very unlikely that any two people would have exactly the same DNA information, in the same way that it is very unlikely that any two people would have exactly the same physical fingerprint. The test is used to determine whether a family relationship exists between two people, to identify organisms causing a disease, and to solve crimes.

Only a small sample of cells is needed for DNA fingerprinting. A drop of blood or the root of a hair contains enough DNA for testing. Semen, hair or skin scrapings are often used in criminal investigations.

A person who has DNA fingerprinting done voluntarily usually provides a sample of blood taken from a vein. DNA testing also can be done on cells obtained by a simple mouthwash or a swab of the cheeks inside themouth, but these methods are not recommended.

DNA fingerprinting is done to find out who a person's parents or siblings are. This test also may be used to identify the parents of babies who were switched at birth.

Solve crimes (forensic science) - Blood, semen, skin, or other tissue left at the scene of a crime can be analyzed to help prove whether the suspect was or was not present at the crime scene.

Identify a body - This is useful if the body is badly decomposed or if only body parts are available, such as following a natural disaster or a battle.

Amniocentesis:

During pregnancy, the fetus is surrounded by amniotic fluid, a substance much like water. Amniotic fluid contains live fetal skin cells and other substances, such as alpha-fetoprotein (AFP). These substances provide important information about baby's health before birth.

Amniocentesis is a prenatal test in which a small amount of amniotic fluid is removed from the sac surrounding the fetus for testing. The sample of amniotic fluid (less than one ounce) is removed through a fine needle inserted into the uterus through the abdomen, under ultrasound guidance. The fluid is then sent to a laboratory for analysis. Different tests can be performed on a sample of amniotic fluid, depending on the genetic risk and indication for the test.

Amniocentesis is performed to look for certain types of birth defects, such as Down syndrome, a chromosomal abnormality.

Because amniocentesis presents a small risk for both the mother and her baby, the prenatal test is generally offered to women who have a significant risk for genetic diseases like, family history of certain birth defects, previously had a child or pregnancy with a birth defect, will be 35 or older at the time of delivery.

Amniocentesis does not detect all birth defects, but it can be used to detect the Down syndrome, Sickle cell disease, Cystic fibrosis, Muscular dystrophy, Tay-Sachs and similar diseases conditions if the parents have a significant genetic risk.

Amniocentesis can also detect certain neural tube defects (diseases where thebrain and spinal column don't develop properly), such as spinal bifida andanencephaly.

An amniocentesis can also be done during the third trimester of the pregnancy to determine if the baby's lungs are mature enough for delivery or to evaluate the amniotic fluid for infection.

Antenatal Diagnostic Tests:

Diagnostic tests are used to provide fetal diagnoses antenatally and are only offered to those women who are at risk of having a fetus with a congenital abnormality.

Screening tests differ: they are offered to all pregnant women and include:

- Blood tests to look for infections (Hepatitis B, HIV or syphilis), abnormalities (Sickle Cell Disease, Thalassaemia) and Rhesus status

- Down's screening (serum screening, nuchal translucency or a combination, depending on gestation)
- An anomaly scan between 18 and 20 weeks gestation.

Screening tests do not carry any risk to the pregnancy; they are performed for conditions which have a reasonably high prevalence and are implemented in order to identify the 'high risk' pregnancies. Any pregnancy identified as 'high risk' is then offered further, 'diagnostic' testing. Diagnostic tests are usually invasive and thus carry some risks, including an increased risk of miscarriage. The antenatal diagnostic tests most commonly used are amniocentesis and chorionic villus sampling.

Table to compare screening and diagnostic tests:

	Screening Tests	Diagnostic Tests
Who	All pregnant women	Only 'High Risk' woman
Why	Identify 'high risk' group	Diagnose abnormality
What	Blood tests, ultrasound, maternal history including age.	Amniocentesis, Chorionic villus sampling
What are the Risks	No risk to pregnancy. Anxiety if identified as 'high risk'.	Miscarriage (1%)

Sperm Banks:

A sperm bank, semen bank or cryobank is a facility or enterprise that collects and stores human sperm from sperm donors for use by women who need donor-provided sperm to achieve pregnancy. Sperm donated by the sperm donor is known as donor sperm and the process for introducing the sperm into the woman is called artificial insemination, which is a form of third party reproduction.

From a medical perspective, a pregnancy achieved using donor sperm is no different from a pregnancy achieved using partner sperm and it is also no different from a pregnancy achieved by sexual intercourse.

Sperm banks serve our patients in many ways - Need donor sperm due to problems with own sperm, an LGBT couple, single woman, want to store frozen sperm to preserve for future fertility after medical treatments for cancer or other illnesses.

Position Effect:

Position effect is the effect on the expression of a gene when its location in a chromosome is changed, often by translocation. This has been well described in Drosophila with respect to eye color and is known as position effect variegation (PEV).

The phenotype is well characterized by unstable expression of a gene that results in the red eye coloration. In the mutant flies the eyes typically have a mottled appearance of white and red sectors. These phenotypes are often due to a chromosomal translocation such that the color gene is now close to a region of heterochromatin. The heterochromatin can spread stochastically and switch off the color gene resulting in the white eye sectors.

Position effect is also used to describe the variation of expression exhibited by identical transgenes that insert into different regions of a genome. In this case the difference in

expression is often due to enhancers that regulate neighboring genes. These local enhancers can also affect the expression pattern of the transgene. Since each transgenic organism has the transgene in a different location each transgenic organism has the potential for a unique expression pattern.

Important Questions

Q.1 Multiple choice questions (1 marks each)

1) Branch of science related with improvement of mankind genetics

a) Human genetics b) General genetics **c) Eugenics** d) Genetics

2) Improvement of mankind by genetic engineering is studied under

a) Eugenics b) Euthenics c) Euphenics d) Pathology

3) Intelligence in man is

a) Polygenic trait b) Mendelian trait c) Congenital trait d) Morphological trait

4) Which of the following refers to the science concerned with the application of geneticprinciples to the improvement of the human species?

a) assortive mating b) **eugenics** c) holandricism d) none of these

5) Of the following, which is not an autosomal dominant disorder?

a) Huntington's disease b) Neurofibromatosis **c) Phenylketonuria** d) Achondroplasia

6) Which one is a hereditary disease

a) cataract b) leprosy c) blindness **d) phenylketonuria.**

6) Sickle cell anaemia induce to

a) change of amino acid in a- chain of haemoglobin

b) **change of amino acid in b- chain of haemoglobin**

c) change of amino acid in both a and b chains of haemoglobin

d) change of amino acid either a or b chains of haemoglobin.

7) Which one of the following conditionsthough harmful in itself, is also potentialsaviour from a mosquito borne infection disease?

a) Thalassaemia **b) Sickle cell anaemia**c) Perniciofls anaemia d) Leukaemia

8) Sickle cell anaemia has not been eliminated from the African population because

a) it is controlled by dominant genes

b) it is controlled by recessive genes

c) it is not a fatal disease

d) it provides immunity against malaria.

9) Sickle cell anaemia caused by the presence of mutant allele in ..... chromosome

a) Seventh b) Eighth c) Tenth **d) Eleventh**

10) HbA2 raised in

a) Thalassaemia b) Sickle cell anaemia

c) Myelofibrosis d) Sideroblastic anaemia

9) In thalassaemia, treatment used as,

a) Iron b) Deferrioamine c) Folic acid **d) Blood transfusion**

10) A classical example of epistasis is

a) ABO blood groups in humans. b) height in garden peas.

c) coat color in rabbits. d) **albinism in humans and animals**.

11) What causes the effects of albinism?

a) the lack of pigment production b) inability to produce normal connective tissue

c) an extra chromosome 21 d) the presence of two different codominant alleles

12) Hereditary Alkaptonuria is due to

a) Catalase deficiency **b) Homogentisic acid oxidase deficiency**

c) Protease deficiency d) Tryptase deficency

Q.2 Define /explain /comment (2 marks each)

1) Albinism, 2) Eugenics 3) Gene therapy

Q. 3 Question for (4mark each)

1) Effect of eugenics. 2) DNA fingerprinting, 3) amniocentesis,

4) sperm banks, 5) karyotyping 6) Phenylketouria

7) Qualities of a good genetic counsellor. 8) Alkaptouria

9) Give an overview of the ways and means of prevention of Genetic Diseases

Q. 4 Question for (6 mark each)

1) Discuss the purpose and limitation of genetic counselling

2) Describe any three hereditary diseases and disorders in human.

3) Discuss the role of human genetics in medical science with reference to

a. Genetic counselling, b. Antenatal diagnosis, c. Gene therapy

Q.5 Distinguish between (4 marks)

1) Positive eugenics and negative eugenics

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