ISBN:978-81-951982-5-2

Genetics

(Powerpoint Presentation e-Book for Students)

K. J. Adate
V. V. Ajagekar

Published By:
Bhumi Publishing, India

Genetics

ISBN: *978-81-951982-5-2*

Mr. Kishor J. Adate

Head,

Department of Zoology,

Shivraj College, Gadhinglaj,

Dist – Kolhapur, M.S., India

Dr. Vinayak V. Ajagekar

Head,

Department of Zoology,

Ajara College, Ajara,

Dist – Kolhapur, M.S., India

Bhumi Publishing

2021

First Edition: 2021

ISBN: 978-81-951982-5-2

Published by:

Bhumi Publishing,

Nigave Khalasa, Kolhapur 416207, Maharashtra, India

Website: www.bhumipublishing.com

E-mail: bhumipublishing@gmail.com

Book Available online at: https://www.bhumipublishing.com/books/

Preface

We are very happy to handover this E-Book based on power point presentation of Genetics. During the Covid-19 pandemic teachers and students are doing work from home. This is the right time to provide teaching material to the students and teachers. Taking this in to consideration we prepared this E-Book which is based on syllabus of Shivaji University, Kolhapur. Various reference books have been referred while preparing this book. Information from the internet has also been collected. For the easy understanding of the student's simple language, large, accurate, and neat labeled diagrams are used.

We hope sincerely that this book will meet the needs and demands of B.Sc. first year Zoology students. We also request the readers to point out any mistakes, typographical errors, and also make suggestions if any. The mistakes will be corrected and worthy suggestions will be incorporated in the next edition.

- ***Authors***

Acknowledgement

We are really grateful because we managed to complete the book for students. The assignment cannot be completed without the encouragement and cooperation of our colleagues and board members of our education society.

We are specifically thankful to President Prof. K. V. Kurade, Prof. Anil Kurade (Secretary of Karmveer Vittal Ramaji Shinde Shikshan Sanstha, Gadhinglaj) and Ashokanna Charati (President, Janata Education Society, Ajara) and our management for their continuous and favourable encouragement.

We are also very much thankful to Dr. S. M. Kadam, Principal, Shivraj College, Gadhinglaj and Dr. A. N. Sadale, Principal, Ajara Mahavidyalaya, Ajara for their guidance and trust.

We would like to express our gratitude to our great source of inspiration i.e. students, who motivated us to write and publish this work.

Finally we thank all our friends and well wishers for their support and wishes.

Prof. K. J. Adate

Dr. V. V. Ajagekar

Unit : 1

1) INTRODUCTION TO GENETICS

a) Mendel's work on transmission of traits

b) Genetic Variation

a) Molecular basis of Genetic Information

2) MENDELIAN AND POST MENDELIAN GENETICS

a) Principles of Inheritance.

b) Incomplete dominance and co-dominance.

c) Gene interaction.

d) Multiple alleles w.r.t. ABO, Rh blood groups and coat colour in rabbit.

e) Sex linked inheritance.

3) LINKAGE, CROSSING OVER

a) Linkage and process of crossing over.

b) Coupling and repulsion theory.

c) Cytological evidence of crossing over.

UNIT: 2

4) MUTATIONS

a) Chromosomal Mutations: Deletion.

   Duplication, Inversion, Translocation.

b) Aneuploidy and Polyploidy.

c) Induced gene mutation.

5) SEX DETERMINATION

a) Sex Chromosomal theory of sex determination,

b) Genic balance theory,

c) Haploidy, Diploidy mechanism,

d) Environmental sex determination,

e) Dosage compensation.

GENETICS

Unit : 1

1) INTRODUCTION TO GENETICS

a) Mendel's work on transmission of traits.

b) Genetic Variation.

a) Molecular basis of Genetic Information .

INTRODUCTION TO GENETICS

A. MENDEL'S WORK ON TRANSMISSION OF TRAITS

1) The branch of biology which deals with the study of heredity and variation is called as 'Genetics'.
2) The contribution of Mendel to Genetics is called as "Mendelism"
3) Gregor Johann Mendel is called the "Father of Genetics"
4) Mendel was born in a peasant family in Austria (22nd July 1822 to 6th January 1884).

INTRODUCTION TO GENETICS

5) Mendel selected pea plants and did his experiments of hybridization.

6) He selected pea plants because they shows clear cut contrasting characters, self fertilized, hybrids are perfectly fertile, cross pollination is not very much difficult, cross pollination is always successful, having short growth period and short life cycle.

7) The most success of the Mendel's work is that the seven characters selected by him were located on seven separate homologous chromosomes and he studied only one character at a time and maintained statistical record of it.

INTRODUCTION TO GENETICS

FOLLOWINGS ARE THE SEVEN CHARACTERS AND THEIR CONTRASTING ALTERNATIVES

SR. NO.	CHARACTERS	DOMINANT	RECESSIVE
1	The length of the stem	Tall (T)	Dwarf (t)
2	The position of the flower	Axial (A)	Terminal (a)
3	The colour of the pod	Green (G)	Yellow (g)
4	The shape of the pod	Inflated (I)	Constricted (i)
5	The shape of the seed	Round (R)	Wrinkle (r)
6	The colour of the seed coat	Green (G)	White (g)
7	The colour of the cotyledon	Yellow (Y)	White (y)

INTRODUCTION TO GENETICS

B) GENETIC VARIATION

- In genetic variation, the genes of organisms within a population change.
- Gene variation is important to the process of natural selection.

CAUSES OF GENETIC VARIATION

i) DNA Mutation

ii) Gene Flow

iii) Sexual Reproduction

GENETIC VARIATION

EXAMPLES OF GENETIC VARIATION

A person's skin colour, hair colour, multi-coloured eyes, dimples, and freckles are all examples of genetic variations that can occur in a population. Examples of genetic variation in plants include the modified leaves of carnivorous plants and the development of flowers that resemble insects to lure plant pollinators. Gene variation in plants often occurs as the result of gene flow. Pollen is dispersed from one area to another by the wind or by pollinators over great distances. Examples of genetic variation in animals include cheetahs with stripes, snakes that fly, animals that play dead, and animals that mimic leaves. These variations enable the animals to better adapt to conditions in their environments.

GENETIC VARIATION

C) MOLECULAR BASIS OF GENETIC INFORMATION

1. Structure of DNA
2. Central dogma of molecular biology
3. DNA replication
4. Transcription
5. Genetic Code
6. Translation
7. Regulation of Gene Expression

GENETICS

2) MENDELIAN AND POST MENDELIAN GENETICS

- Principles of Inheritance.
- Incomplete dominance and co-dominance,
- Gene interaction.
- Multiple alleles w.r.t. ABO and Rh- blood groups and coat colour in rabbit.
- Sex linked inheritance.

MENDELIAN AND POST MENDELIAN GENETICS

PRINCIPLES OF INHERITANCE

A. Principle of unit character

B. Principle of Dominance (Monohybrid cross)

C. Principle of Segregation (Monohybrid cross)

D. Principle of Independent Assortment (Dihybrid cross)

A. PRINCIPLE OF UNIT CHARACTER

Mendel carried out several experiment on pea plants and stated that there is a something which carries the characters from one generation to the other. This something also controls the transmission of characters. This transmission of characters takes place from parents to offspring through the gametes. Letter on this something is called “factor” or “unit” by Mendel. He also stated that this factor or unit is present in the sex cells (gametes). They form the connecting bridge between the parents and their offspring. These factors occur in a pair. Each parent passes only one factor of a pair to the f1 offspring.

Based on this, Mendel stated the principle of unit of characters which states that ***“every trait is controlled by a factor that occurs in a pair.” The factors in a pair are always contrasting. These factors are called alleles or allelomorphs. For ex. Tallness is a factor which is dominant over dwarfness in a pea plant or tallness and dwarfness are two contrasting alleles of a trait.***

B) PRINCIPLE OF DOMINANCE (MONOHYBRID CROSS)

Mendel's law of dominance states that ***'In crossing between pure (homozygous) organisms for contrasting characters of a pair, only one character of the pair appears in the filial generation'.***

He called the variety that appeared in the F1 generation of his monohybrid cross as dominant and those which did not appear in the F1 generation as recessive. A recessive factor freely expresses itself in the absence of its dominant allele. This law is based on monohybrid experiment**.**

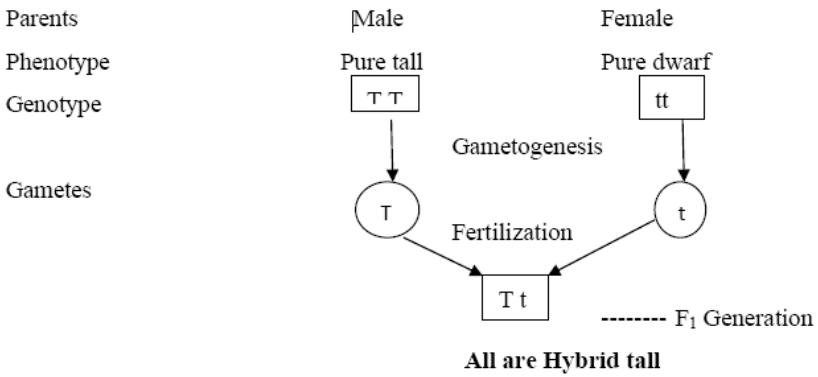
Example: For its monohybrid cross, Mendel selected two varieties of pea plants, pure tall (TT) and pure dwarf (tt). When a cross has between taken from these two plants he observed following results.

F1 Generation – All are tall pea plants

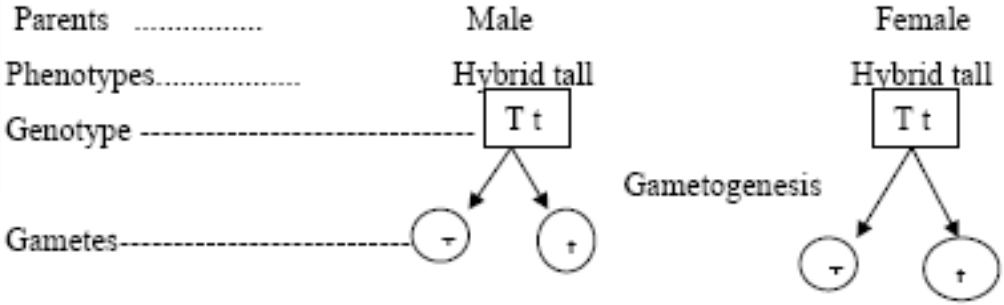
F2 Generation --1:2:1 Genotypic monohybrid ratio and

3:1 Phenotypic monohybrid ratio

PRINCIPLE OF DOMINANCE



All are Hybrid tall



CHECKER BOARD:

	T	T
T	T T	T t
T	T t	t t

OBSERVATIONS:

OBS. NOS.	GENOTYPES	PHENOTYPES.
1	T T	Pure Tall
2 & 3	Tt	Hybrid Tall
4	t t	Pure Dwarf

Phenotypic ratio – 3:1 and Genotypic ratio – 1: 2: 1

C) PRINCIPLE OF SEGREGATION (MONOHYBRID CROSS)

This law is also called law of purity of gametes. This law of segregation states that ***'The hybrids or heterozygotes of F1 generation have two contrasting characters or allelomorphs of dominant and recessive nature. These alleles though remain together for longer time but do not contaminate or mix with each other and separate or segregate at the time of gametogenesis so that each gamete receives only one allele of a character either dominant or recessive.'*** This law is based on monohybrid experiment.

EX.: During gamete formation in the F1 hybrid tall plant, the paired factor Tt present in the plant, segregate independently and enter different gametes. So each gamete receives either T or t from the paired factors Tt, which is responsible for the expression of a single character.

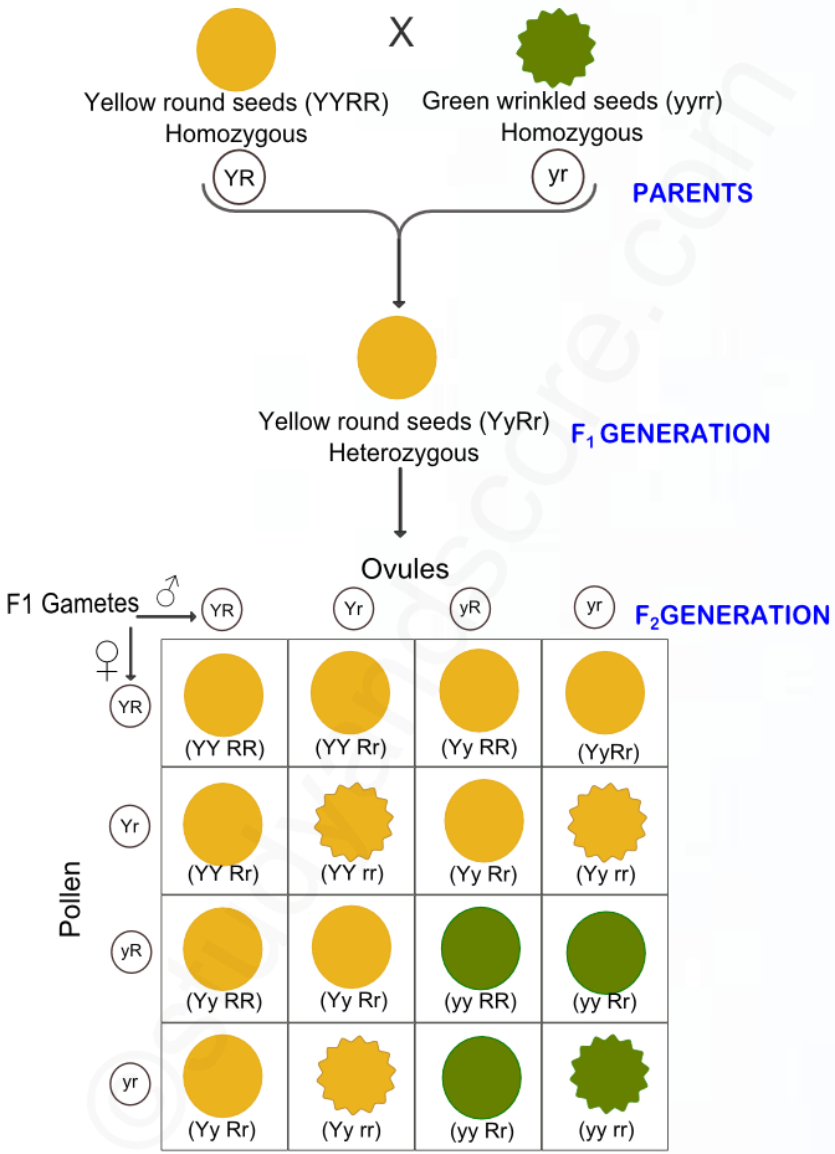
D) PRINCIPLE OF INDEPENDENT ASSORTMENT (DIHYBRID CROSS)

This law is based on Dihybrid experiment. The crossing of two plants differing in two characters are considered at a time is called Dihybrid experiment. According to this law, *'when the parents differ from each other in two or more pairs of contrasting characters or factors then the inheritance of one pair of factors is independent to that of the other pair of factors.'*

MECHANISM OF INDEPENDENT ASSORTMENT

It can be understood very easily by assuming that the homozygous pea plants with yellow round seed has the alleles YY and RR for yellow colour and roundness of the seed, respectively. Similarly the homozygous pea plant with green wrinkled seeds contains the allele's yy and rr. The gametes produced by YYRR and yyrr plants are YR and yr types respectively. When both parents are crossed the union of both types of gametes takes place to give the F1 hybrid (YyRr). According to the law of dominance it is heterozygous yellow round plant. Now the F1 hybrids have four types of alleles, viz., Y for yellow colour, y for green colour, R for round shape and r for wrinkledness of seed. During Gametogenesis these four alleles are assorted independently to produce four types of gametes i.e. YR, Yr, yR and yr. Thus each gamete of Dihybrid cross consist alleles of both the characteristics. These four types of gametes of F1 hybrid unite at random in the process of fertilization and produce sixteen types of individuals in F2 generation.

MECHANISM OF INDEPENDENT ASSORTMENT



CHECKER BOARD (F_2 GENERATION)

Female / Male	YR	Yr	yR	yr
YR	YYRR	YYRr	YyRR	YyRr
Yr	YYRr	Yyrr	YyRr	Yyrr
yR	YyRR	YyRr	YYRR	Yyrr
yr	YrRr	Yyrr	YYRr	Yyrr

OBSERVATIONS:

OBS. No.	Phenotype	Total
1, 2, 3, 4, 5, 7, 9, 10, 13	Yellow round (Y&R)	9
6, 8, 14	Yellow wrinkled (Y&rr)	3
11, 12, 15	green round (yy&R)	3
16	green wrinkled (yyrr)	1

Phenotypic Dihybrid ratio = 9:3:3:1

1) CO-DOMINANCE AND INCOMPLETE DOMINANCE

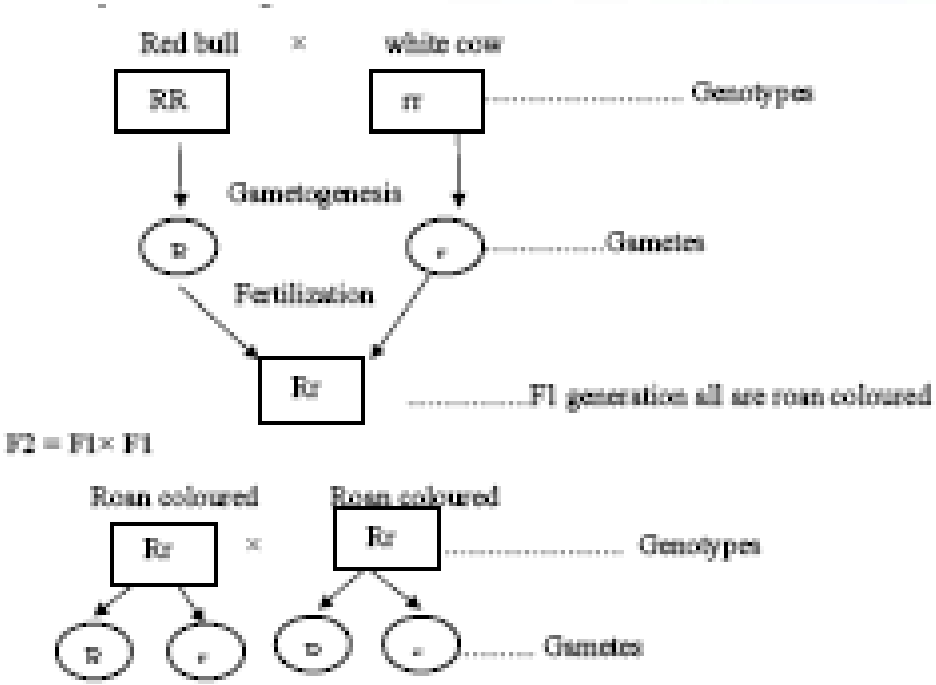
i) CO-DOMINANCE

In co-dominance both dominant and recessive alleles lack their dominant and recessive relationship and both the genes express their expression independently. In this case the dominant character is not mixed with the recessive character. Thus in co-dominance both the dominant and recessive alleles expresses their character in F1 generation, none is masked. Co-dominance is an allelic interaction. Examples are ---

1) The AB blood group is due to co-dominance. AB group is controlled by the genes IA and IB. The IA and IB are equally dominant. IA produces antigen A and IB produces antigen B.

2) Another example of co-dominance is coat colour in short horn cattle. In short horn cattle there are two colours of hair, red and white. Red colour is controlled by R and white by r. When red and white are crossed the F1 has roan colour having both red and white coloured hairs. This is because r is also expressed in F1 generation.

CO-DOMINANCE



	R	r
R	RR	Rr
r	Rr	r r

OBSERVATIONS

SR NO.	GENOTYPES	PHENOTYPES
1	RR	Red colour
2	Rr	Roan colour
3	r r	White colour

Genotypic ratio 1:2:1 & Phenotypic ratio 1:2:1

ii) INCOMPLETE DOMINANCE

In incomplete dominance both alleles of a character has partial expression in F1 generation. So the F1 individual has a mixture of characters of both the parents. Ex. *Mirabilis Jalapa (Four o' clock plant)*

When a homozygous red flowered (R_1R_1) four o' clock plant is crossed with a homozygous white flowered plant (R_2R_2) a pink coloured variety is produced (R_1R_2). This is due to the incomplete dominance of the gene R over the r. The expression of the two genes(R & r) in the same individual leads to the production of an individual with mixed character.

INCOMPLETE DOMINANCE

P ɔotype of Pare ɔts → Red flower × White flower

Genotype → RR × rr

Gametes → R × r

F_1 generation → Rr

Pink flower

Selfing of F_1 generation → Rr × Rr

Gametes → R r × R r

F_2 generation →

♀ \ ♂	**R**	**r**
R	RR Red	Rr Pink
r	Rr Pink	rr White

GENE INTERACTION

Experimentally it has been proved that most of the characters of living organisms are controlled/ influenced /governed by a collaboration of several different genes. This is the condition where a single character is governed by more genes and every gene affect the expression of the other genes involved; is called interaction of genes.ie expression of one gene depends on the expression of other gene. Followings are the types of gene interactions.

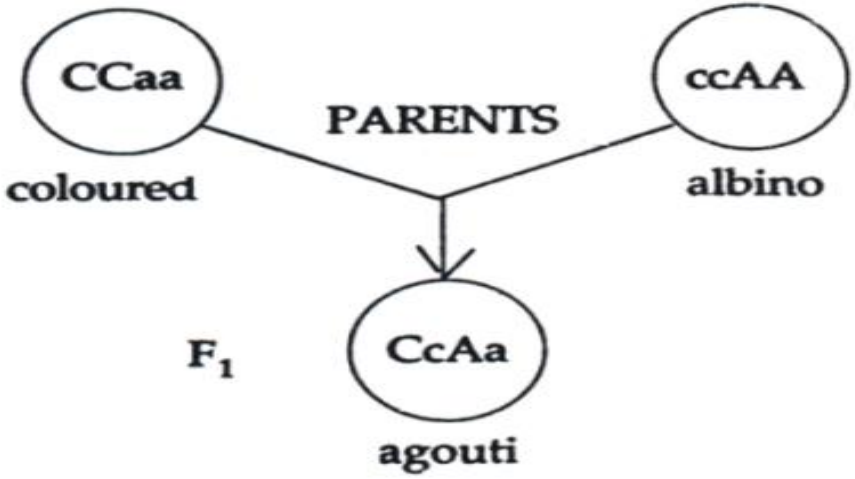
1. Supplementary gene interaction
2. Complementary gene interaction

1. SUPPLEMENTARY GENE INTERACTION

These are two independent pairs of genes interacting in such a manner that one dominant produces its effect whether the other is present or not while the second one produces its effect only in the presence of the first is called supplementary gene interaction. This has been demonstrated in the inheritance of coat colour of rabbits and other rodents by **Castle.**

Example: In case of Guinea pigs it has been found that the black colour of the coat C is dominant over albino c. Apart forms this there is a wild variety the Agouti in which the colour of the coat is more or less greyish. Here the hairs are black at the base and tip, with a yellow band in between. This produces a kind of neutral gray colour, a protective colour pattern characteristic of the wild variety and it is due to presence of a dominant gene A. This gene when present either in a single or double dose turns black fur into Agouti. It naturally follows that a black guinea pig is therefore always homozygous for the recessive allele in an addition to possessing at least one dominant gene C. In the absence of C the dominant gene A or its recessive ‘a’ has absolutely no effect. Therefore the albino varieties may or may not possess the gene A. The genetic constitution of the 3 different kinds of Guinea pigs may be represented as follow.

SUPPLEMENTARY GENE INTERACTION



F_2 GENERATION

MALE GAMETES \ FEMALE GAMETES	CA	Ca	cA	ca
CA	CCAA agouti	CCAa agouti	CcAA agouti	CcAa agouti
Ca	CCAa agouti	CCaa coloured	CcAa agouti	Ccaa coloured
cA	CcAA agouti	CcAa agouti	ccAA albino	ccAa albino
ca	CcAa agouti	Ccaa coloured	ccAa albino	ccaa albino

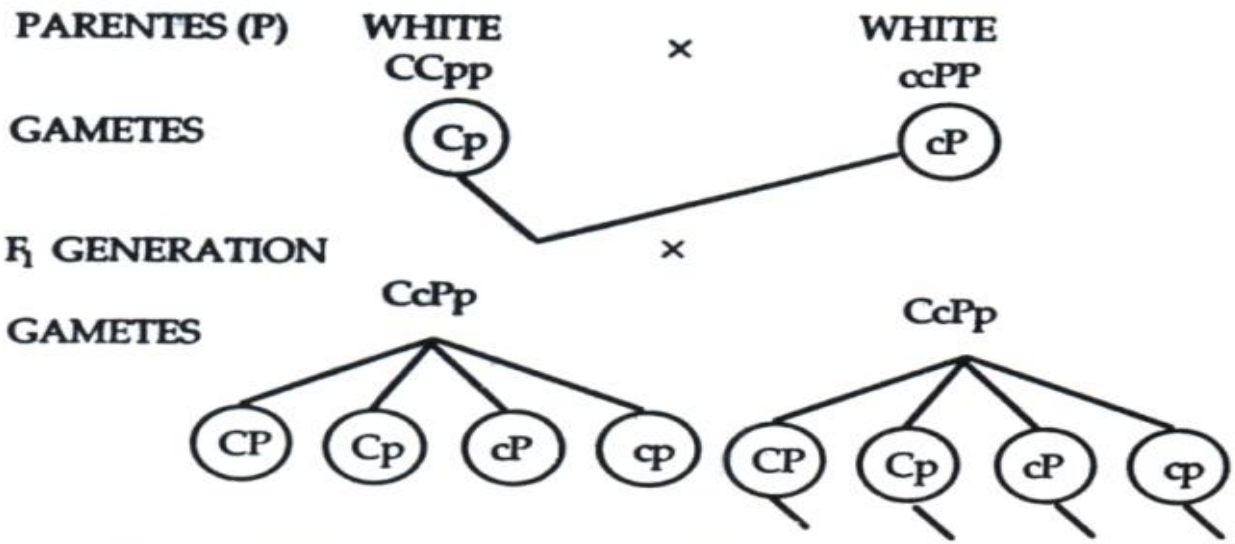
Inheritance of supplementary genes showing 9:3:4 ratio in the F_2 generation

2) COMPLEMENTARY GENE INTERACTION

Certain characters are expressed as a result of the interaction between the two dominant non allelic genes coming from different parents. These genes, if alone, remain unexpressed and become effective only when they come together. Such genes are called complementary genes, because their action is complementary to each other for a particular trait.

Example: W. Batson and R. C. Punnet observed that, when two white flowered varieties of sweet pea, Lathyrus odoratus were crossed, F1 progeny had coloured flowers. When F2 progeny obtained from F1 was classified plants with 9 coloured flowers and 7 white flowers i.e. 9:7 ratios this is again a modification of 9:3:3:1 ratio.

COMPLEMENTARY GENE INTERACTION



F_2 GENERATION — MALE GAMETES

♀ \ ♂	CP	Cp	cP	cp
CP	CCPP Purple	CCPp Purple	CcPP Purple	CcPp Purple
Cp	CCPp Purple	CCpp White	CcPp Purple	Ccpp White
cP	CcPP Purple	CcPp Purple	ccPP White	ccPp White
cp	CcPp Purple	Ccpp White	ccPp White	ccpp White

FEMALE GAMETES

PURPLE : WHITE = 9 : 7

Inheritance of complimentary genes showing 9 : 7 ratio in the F_2 generation

2) MULTIPLE ALLELES

According to Mendel a character is controlled by a single pair of genes. The two genes of a character are located in the same locus of the homologous chromosomes. The two genes are called alleles. These alleles undergo mutation to gives rise to three or more alleles located in the same locus of homologous chromosome. These mutant alleles express different alternatives of the same character. Such genes are called multiple alleles. It may be defined as 'a series of three or more genes which control the same character and occupy the same locus in the homologous chromosomes".

i) COAT COLOUR IN RABBITS

In Rabbits coat colour is controlled by multiple alleles. There are four varieties of rabbits. A) Agouti B) Chinchilla B) Himalayan and D) Albino

A) Agouti : This is the wild type rabbit and its body is brownish gray in colour. This has banded hairs; the portion near the skin is gray, followed by yellow band, and finally a black or brown tip. The dominant gene C is responsible for the brown coat colour. The dominant gene undergoes mutation to give rise to three mutant alleles C^{ch}, Ch, and 'c' located in the same locus. These mutant alleles express different shades of coat colour and are recessive to dominant allele C.

B) Chinchilla - In some rabbits the coat colour lacks the yellow pigment and due to the optical effect of black and gray hairs body appears silvery gray in colour. The mutant allele C^{ch} is responsible for the production of the silver gray coat colour and is dominant to another mutant alleles C^{h} and c.

C) Himalayan - In these individuals the extremities such as ear nose and tips of the limbs are coloured while rest of the body is white. This type of pigmentation is known as acromelanism. The mutant allele for Himalayan is C^{h} and is dominant to the mutant allele c.

D) Albino - From these individuals the pigments are completely absent. The allele for albino coat is represented as 'c'

PHENOTYPES	GENOTYPES
Agouti	$C C / C C^{ch} / C C^{h} / C c$
Chinchilla	$C^{ch} C^{ch} / C^{ch} C^{h} / C^{ch} c$
Himalayan	$C^{h} C^{h} / C^{h} c$
Albino	cc

ii) ABO BLOOD GROUP SYSTEM

Karl Landsteiner (1900) discovers the presence of two types of antigens on the extraneous coat of human RBCs. These two antigens are antigen A and antigen B. Based on this he divided human blood groups into three types namely blood group A, B and O. A blood group person contain antigen A on the RBC, B blood group person contain antigen B on the RBC and O blood group person do not have the antigens on their RBC coat. Latter on in 1902 A. Von Decastello and A.Sturil recognized the presence of both antigens together i.e. antigen A and antigen B. Based on this they discovered fourth blood group called blood group AB.

With the antigens there are certain naturally occurring antibodies are present in the serum of the blood. A blood group person has antigen A and antibody b. B blood group person has antigen B and antibody a, AB group person has antigen A and B and no antibody. O blood group person has both antibody ‘a’ and antibody ‘b’. Thus it is clear that antigen A cannot coexist with antibody ‘a’ in any man. Similarly antigen B cannot coexist with antibody ‘b’.

BLOOD GROUP	ANTIGEN	ANTIBODY
A blood group	A	b
B blood group	B	a
AB blood group	AB	-
O blood group	-	a & b

The synthesis of antigen -A is controlled by a dominant allele I^A (I-Isoaglutination), antigen- B synthesis is controlled by another dominant allele represented by I^B. The absence of antigen is due to the presence of recessive allele represented by ‘i’. These three alleles are responsible for the inheritance of ABO blood group. They are I^A, I^B, and i. ‘i’ is recessive allele and is recessive to both I^A, & I^B. I^A & I^B are dominant alleles and are co-dominant. In co-dominance both genes express their character. None is masked. When I^A, & I^B is present in the same man I^A produces antigen - A and I^B produces antigen - B.

As I^A, I^B, and i occurs in the same locus on the homologous chromosomes they are called multiple alleles. Though there are three alleles each person may contain only two alleles. For example ‘A’ blood group person contains I^AI^A or I^Ai, B blood group person contains I^B I^B or I^B i and O blood group person contains ii. The inheritance of ABO blood group follows simple Mendelian inheritance.

GENOTYPES	PHENOTYPE
$I^A I^A$ / I^A i	A - blood group
$I^B I^B$ / I^B i	B - blood group
$I^A I^B$	AB - blood group
Ii	O - blood group

APPLICATIONS OF ABO BLOOD GROUP SYSTEM

1) Blood transfusion

2) Disputed parentage

3) Identification of culprits

iii) Rh- BLOOD GROUPS

Rh- blood group was discovered by *Landsteiner and Wiener in 1940*. *It is controlled by a set of multiple alleles located in the same locus of the homologous chromosomes. According to this theory there are two types of human beings namely Rh positive (Rh+) and Rh negative (Rh-). Rh+ positive persons contain an antigen called Rh antigen present on the surface of the RBC. Rh- antigen is Rhesus antigen as it was first discovered by Rhesus monkey. Rhesus antigen is also called Rh factor. The Rh- person do not contain Rh antigen.*

The Rh- antigen has no natural antibody. However Rh antibody can be produced artificially. An Rh- person develops Rh antibody when he receives blood from Rh+ person. Even a small amount of Rh+ blood (as small as 0.5ml) can evoke the production of Rh antibody in the Rh- person. The antibody once formed remains throughout the life.

There are several varieties of Rh antigen and of antibody. The commonest Rh antigen is called antigen D and its antibody is called anti D. the production of antigen is controlled by a dominant gene represented by R. when this gene is recessive it cannot produce the antigen. Hence the Rh+ persons may be homozygous dominant (RR) or heterozygous (Rr). The Rh- persons are always homozygous recessive (rr).

SEX LINKED INHERITANCE

In most of the sexually reproducing organisms there are two types of chromosomes are present. These are called autosomes and allosomes or sex chromosomes. Sex chromosomes are mostly responsible for the determination of the sex of the individuals. These sex chromosomes also transmit other characters along with the sex. Such characters are called sex linked characters and transmission of these characters is called sex linked inheritance. It is also called sex linkage. It is defined as the transmission of body characters from parents to offspring along with sex is called sex linked inheritance. The common examples of sex linked inheritance are Colour blindness, Haemophilia, Eye colour in Drosophila, Hypertrichosis (hair on ear pinna), and Ichthyosis hystrix (scales on the body).

SEX LINKED INHERITANCE

The sex linked genes are located on the X chromosomes or Y chromosomes or both X and Y chromosomes. The genes located on X chromosomes are called X- linked genes and the inheritance of X- linked genes is called X- linked inheritance. For example - Colour blindness, Haemophilia, Eye colour in Drosophila.

Similarly the genes located on Y chromosome are called Y- linked genes and the inheritance of these genes is called Y- linked inheritance. For example - Hypertrichosis (hair on ear pinna), and Ichthyosis hystrix (scales on the body).

Sometimes genes located on both X and Y chromosomes controls the body characters such genes are called XY- linked genes and the inheritance is called XY- linked inheritance. Xeroderma pigmentosa, Retinitis pigmentosa and nephritis are the examples of the XY- linked inheritance.

GENETICS

3) LINKAGE, CROSSING OVER

a) Linkage and process of crossing over.

b) Coupling and repulsion theory.

c) Cytological evidence of crossing over.

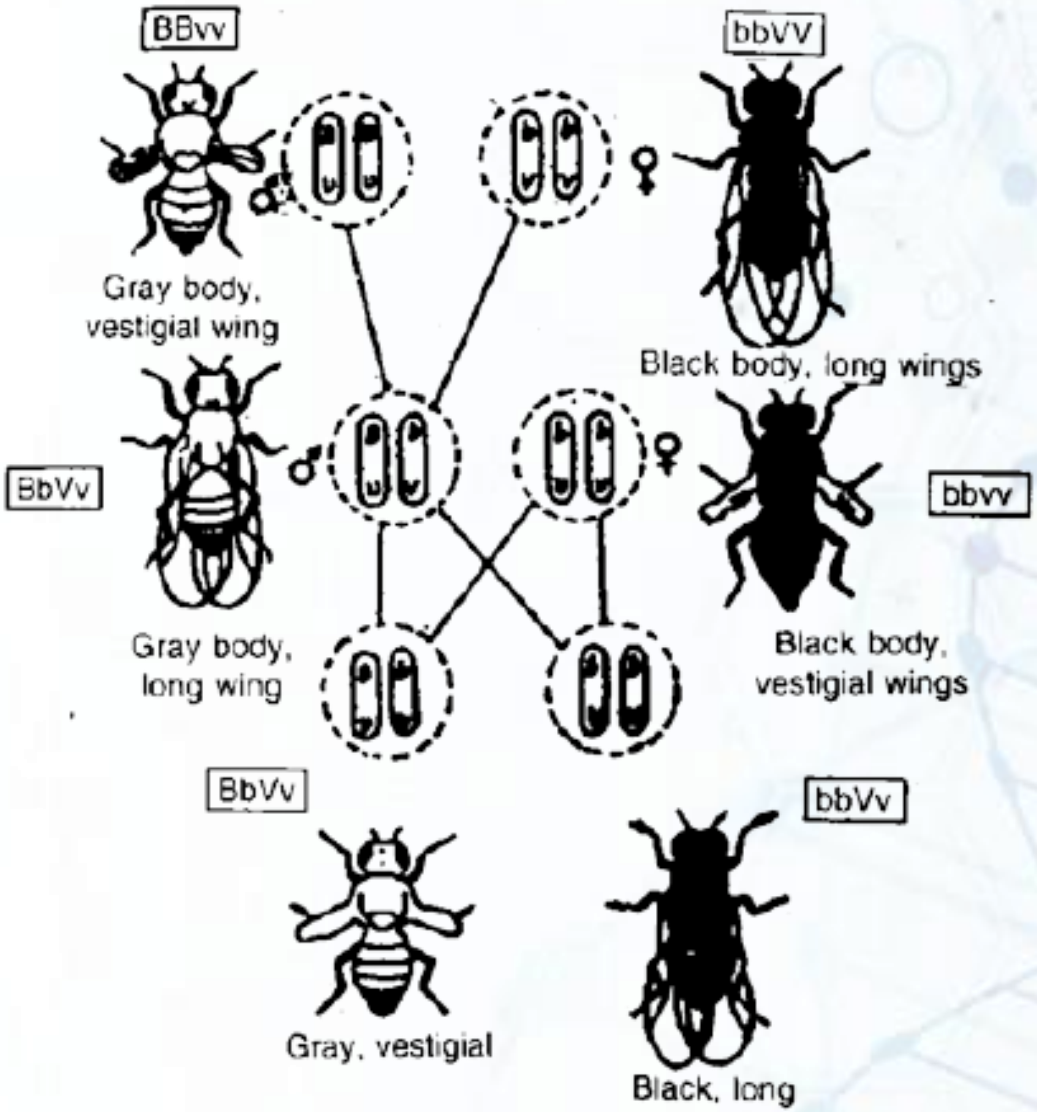
LINKAGE, CROSSING OVER

LINKAGE: Linkage may be defined as the tendency of are two more genes to remain together in the same chromosome during the process of inheritance.

1) COMPLETE LINKAGE:

When the linked genes are so closely located in chromosomes that they inherit in same linkage groups for two or more generations in a continuous and regular fashion then, they are called completely linked genes and the phenomenon of inheritance of completely linked genes is called complete linkage.

Ex. - In Drosophila, Gray body colour (B) is dominant over black (b) where as long winged condition (V) is dominant over vestigial wing (v). If gray bodies vestigeal winged male fly (BBvv) is crossed with black long winged (bbVV). The F1 individuals obtained are Gray long. If F1 male hybrids are crossed with double recessive female (Test cross) only two kinds of files i.e. gray long and black vestigial as those of the parents. I.e. all the genes of male Drosophila remain completely linked.

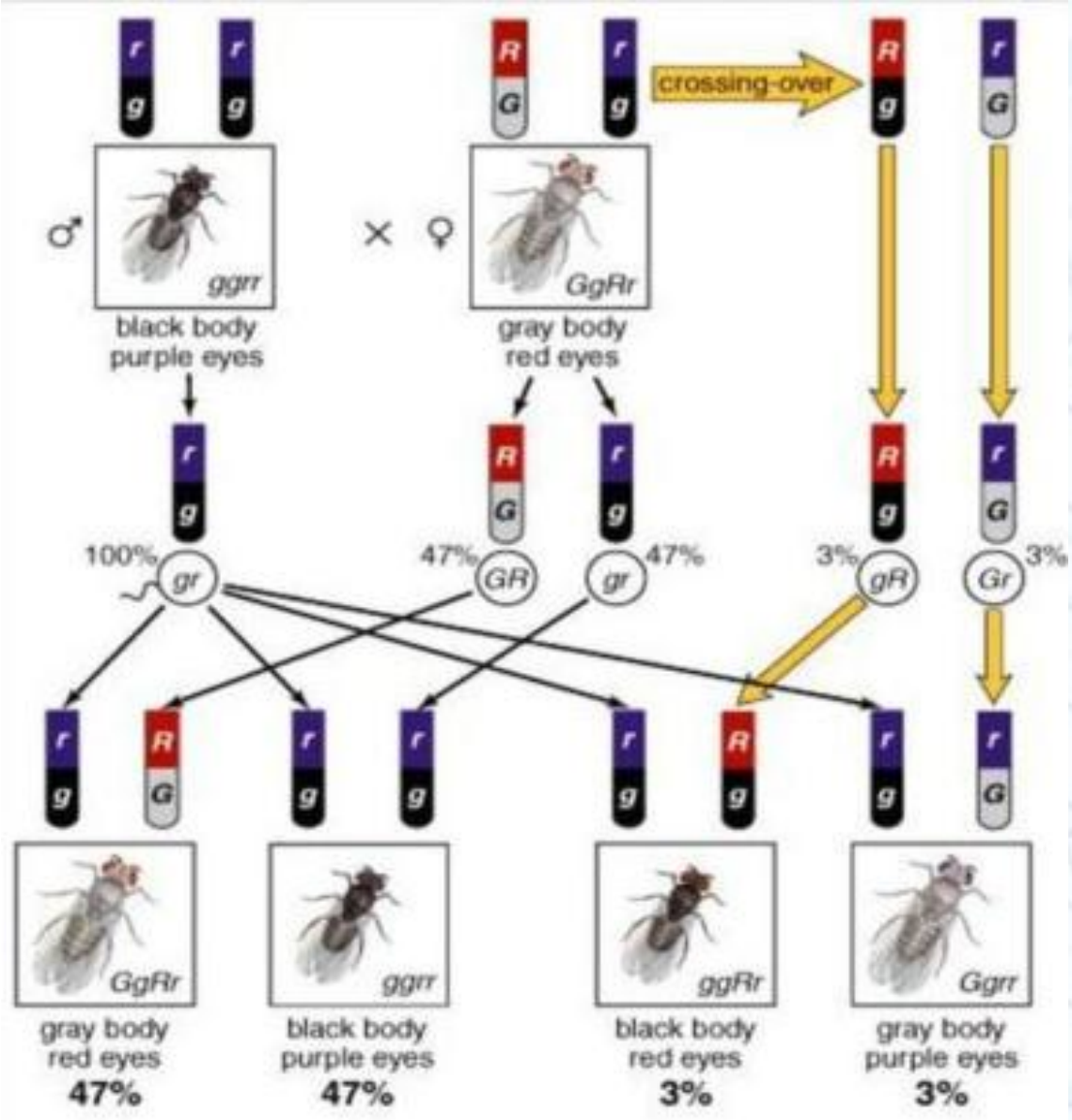


COMPLETE LINKAGE IN MALE DROSOPHILA

2) INCOMPLETE LINKAGE

The linked genes do not always stay together because homologous non sister chromatids may exchange segments of varying lengths (Which bearing many linked genes) with one another during meiotic prophase, by the process of crossing over. The linked genes which are widely located in chromosome and have chances of separation by crossing over are called incompletely genes and the phenomenon of their inheritance is called incomplete linkage.

INCOMPLETE LINKAGE



CROSSING OVER

"The reciprocal exchange of segments between homologous chromosomes (Generally occurring during meiosis) bringing the recombination of the linked genes particularly of those which are not very closely situated."

OR

It may be defined as the mutual exchange of genetic material between the non sister chromatids of a homologous chromosomes during the pachytene stage of the prophase first of the meiosis first.

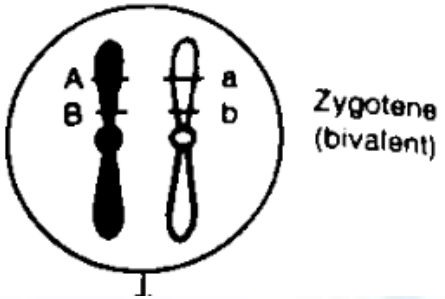
MECHANISM OF CROSSING OVER

The mechanism of crossing over takes place during early prophase stage (Pachytene of IST meiotic division). The entire process takes place in four stages.

1. Synapsis / Pairing / Bivalent stage
2. Duplication (Tetrad stage)
3. Proper crossing over
4. Terminalization.

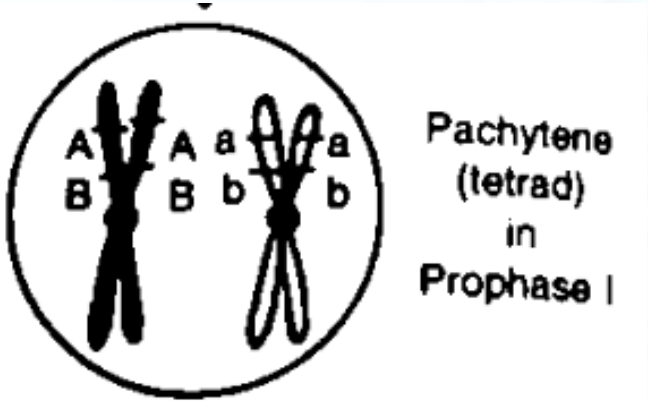
1) Synapsis / Pairing / Bivalent stage

The homologues chromosomes present in the germinal cells come closer and pair longitudinally. This process called pairing or synapsis takes place during the zygotene stage of the prophase I. The paired chromosomes are called bivalent. Synapsis, which begins during the zygotene, continues up to pachytene. The force of attraction is synaptic force.



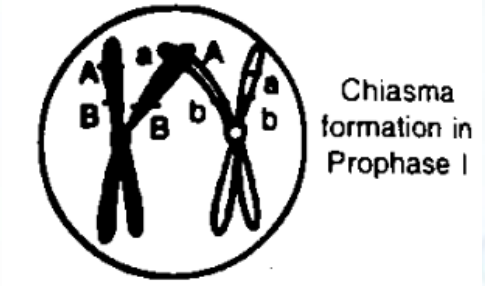
2) Duplication / Tetrad stage

The bivalent undergoes duplication during pachytene. Each chromosome now consists of two chromatids. (But the centromere will not divide) Thus there will be four chromatids (tetrad)



3) Crossing over

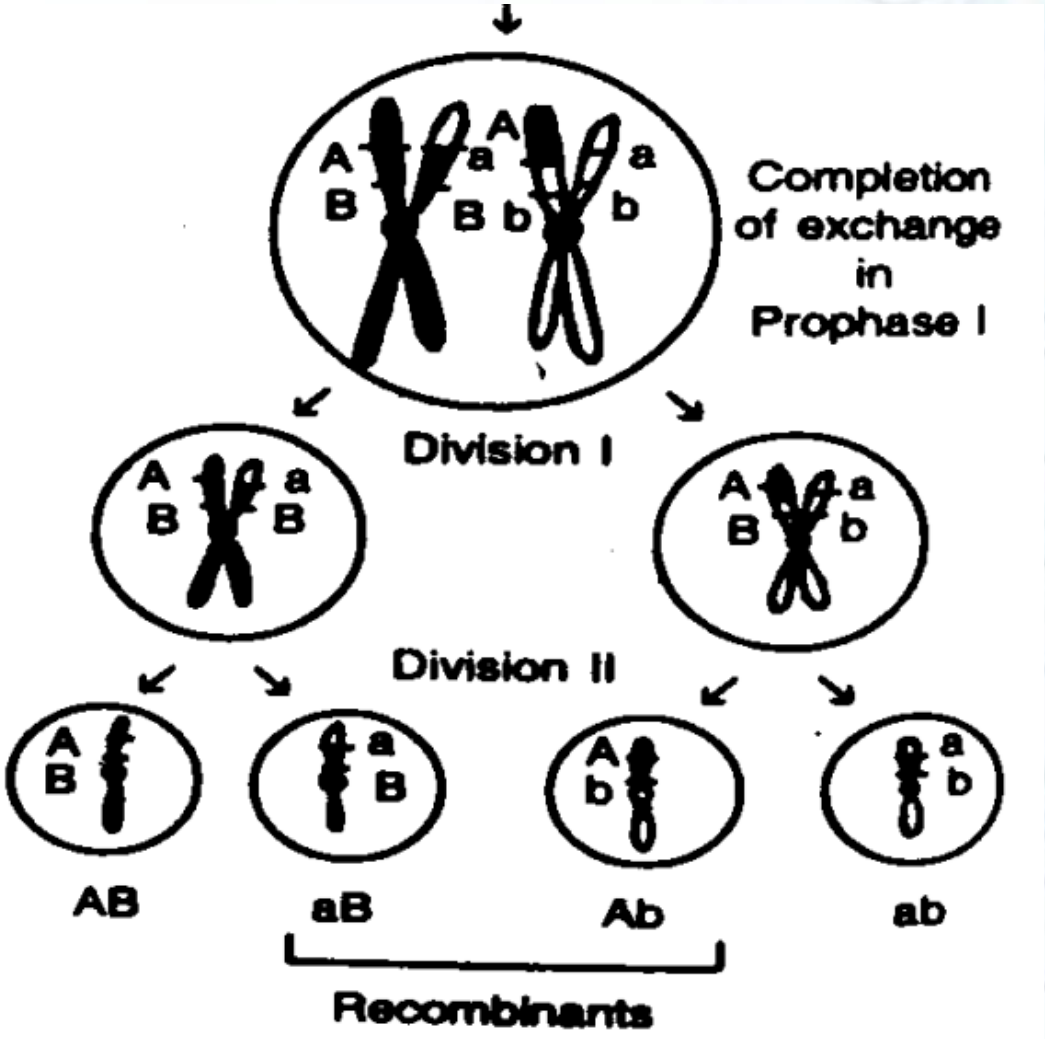
This takes places at the tetrad or the four strand stage. At the time of cross over two opposing non sister chromatids (chromatids belonging to two different chromosomes) have a brake at identical points. This is brought about by the action of to enzyme endoneculease. The two chromatids exchange an identical length of the genome. After exchange the segments fuse with the chromatids due to the action of the enzyme called Ligases. This crossing over is physically demonstrated in the form of chiasma.



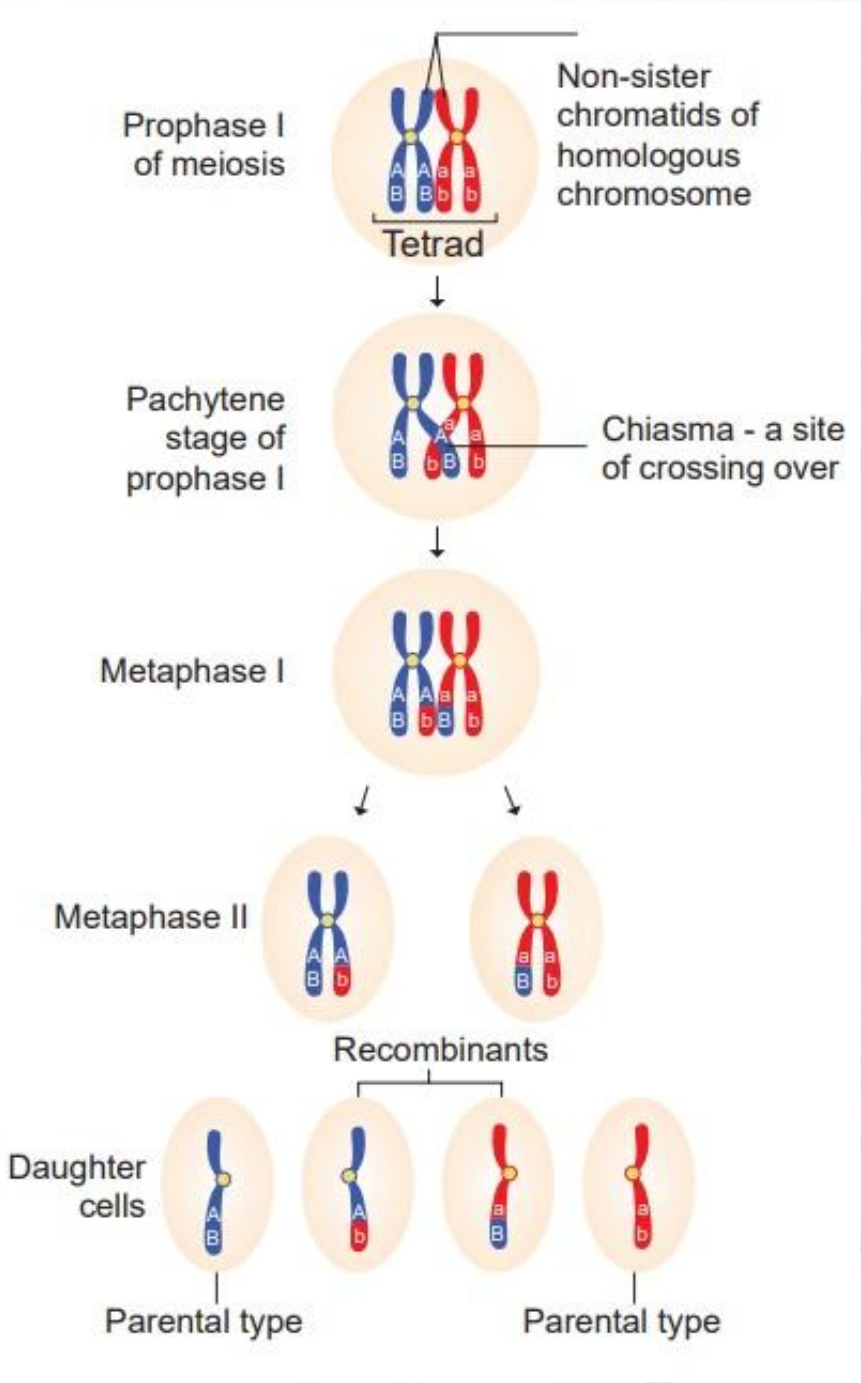
4) Terminalization

After exchange of segments, the two chromosomes start moving away from each other as the synaptic force lapses. The separation begins from the centromere and moves towards the ends of the chromosome. It is the uncoupling of chiasma that is called terminalization. During dikinesis the homologous chromosomes get separate except at their ends.

4) Terminalization



MECHANISM OF CROSSING OVER



COUPLING AND REPULSION THEORY

The theory of coupling and repulsion was formulated by Batson and Punnett (1910). According to this theory linkage may be either ‘Cis’ or ‘Trans’ type. When two dominant alleles are linked on the same chromosome and the recessive alleles on the homologue, the genes are said to be in the cis arrangement, sometimes called coupling phase.

In trans arrangement or repulsion phase there is a combination of dominant and recessive genes on the same chromosome. On the basis of this coupling and repulsion phenomenon can be explained as follows.

Coupling phenomenon: when two dominant alleles are transmitted from one and the same parent to its offspring they try to remain together is called coupling phenomenon and the arrangement of the genes is called ‘Cis’ arrangement.

Repulsion phenomenon: when two dominant alleles are transmitted from two different parents to its offspring they try to remain separate is called repulsion phenomenon and the arrangement of the genes is called ‘Trans’ arrangement

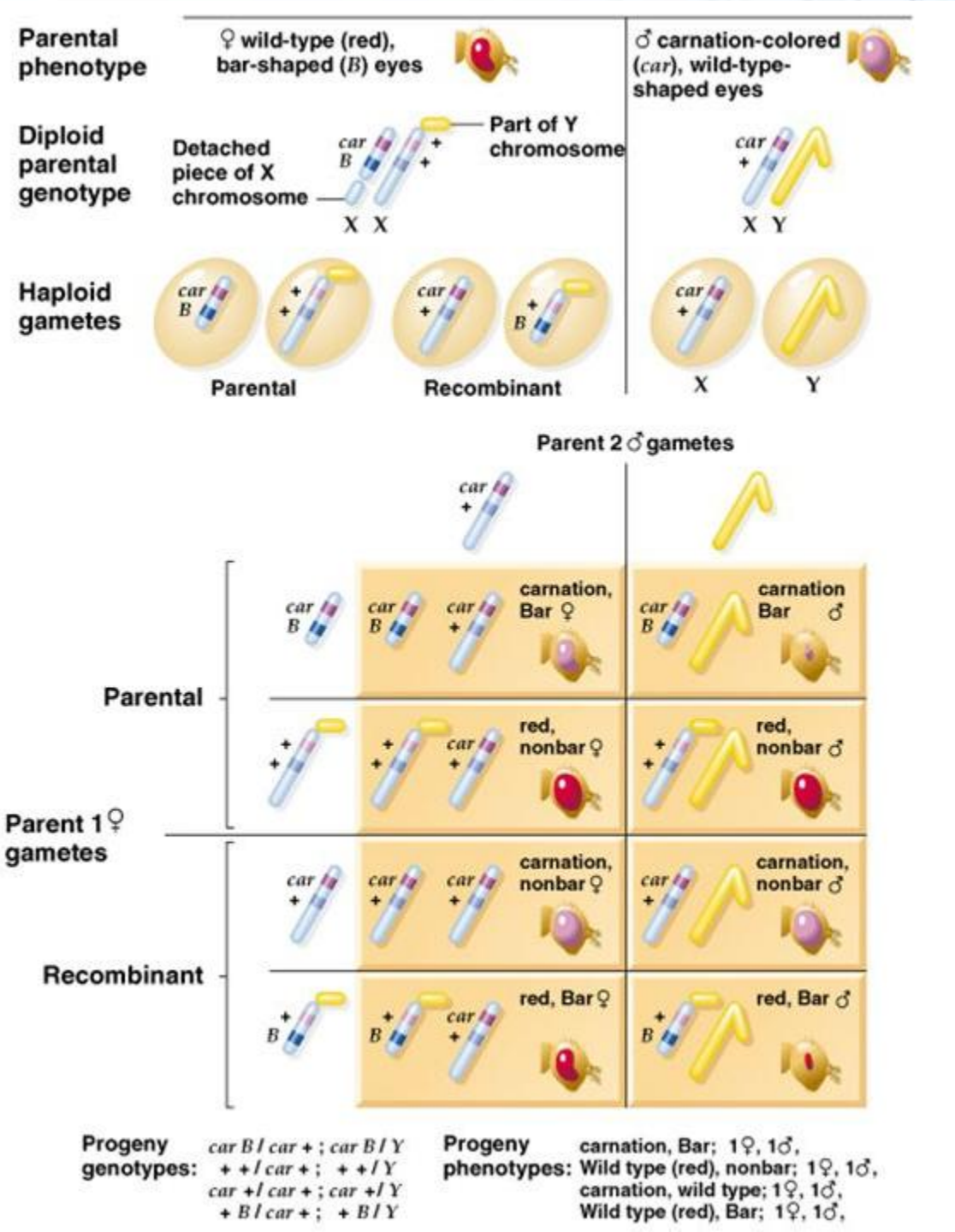
CYTOLOGICAL EVIDENCE OF CROSSING OVER

Crossing over between homologous chromosomes can be physically and visually demonstrated when the chromosomes have special physical parameters. Stern (1931) has demonstrated this very clearly in Drosophila where the two members of the homologous pair can be identified as a result of translocation. A translocated chromosome has a modified configuration and can be identified visually.

In one of the strain of Drosophila, a portion of the Y chromosome has been attached to the X chromosome resulting in “L” shaped chromosome, while the normal “X” chromosome is rod shaped. In another strain, the “X” chromosome is broken into two parts. On the broken X chromosome he placed a recessive eye colour gene carnation (car) and the dominant eye shaped gene bar (B). The unbroken X with the translocated Y piece had wild alleles (++) of the two genes. He crossed this female with such two abnormal X-chromosomes, with a male with X and Y chromosomes having recessive gene car and wild allele of B.

The progeny of the above cross showed four types of female individuals of which 50% were non-cross-over and remaining 50% were cross-over types. Another four kinds of male flies will be produced due to fertilization by Y carrying male gametes. Thus, of the four types of individuals the first two belongs to non-cross over category while the later two are of the cross over category. Such cytological observations suggested that an actual exchange of chromosome segment.

CYTOLOGICAL EVIDENCE OF CROSSING OVER



SIGNIFICANCE OF LINKAGE

Linked genes particularly of the desired trait play a great role in hybridization programme. The following are some of the significant features of the linkage.

1. As linkage help to hold the parental characters together, selection of an individual for breeding programme is based on the combination of characters which the breeder wants to remain together.
2. Another significance of linkage among genes in terms of evolution is that they tend to retain the identity and individuality of species by the clustering of characters.

SIGNIFICANCE OF CROSSING OVER

The following are some of the significant features of cross over which make it a unique phenomenon in providing variation within a parameter among the individuals of a species.

1. Intraspecific variation among individuals is due to crossing over. Crossing over produces new grouping of genes and thus produces new phenotypic traits. Hence it is of great significance in breeding programme.
2. Crossing over provides physical proof for the linear arrangement of genes on the chromosome.
3. Gene mapping on a chromosome is made possible by assessing the cross over value. The percentage of cross over is a function of distance between two gene loci.
4. Crossing over provides a new definition for the gene. According to this “Gene is the smallest section of the chromosome within no cross over can takes place.

MUTATION

4) MUTATION

1. Chromosomal Mutations
2. Deletion
3. Duplication
4. Inversion
5. Translocation
6. Aneuploidy and Polyploidy
7. Induced gene mutation.

MUTATION :

A mutation is defined as a sudden change of a gene or chromosome from one form to another is called mutation. According to Dobzhansky it is a mistake or misprint in cell division. The term mutation was introduced by De Vries. Chromosomal Mutations are also called chromosomal aberrations and is due to of following two types.

A. Change in structure of chromosome.

B. Change in number of chromosomes.

A) Change in structure of chromosome

The chromosome contains genes. The change in the structure of chromosome brings about the changes in the number and arrangement of genes. These are of four types:

a) Deletion

b) Duplication

c) Inversion

d) Translocation

a) Deletion:

It is the chromosomal aberration where segment of the chromosome is lost. It occurs during meiosis. Due to this some genes are also lost. Deletion is of two type's namely terminal deletion and intercalary deletion. In terminal deletion a terminal segment of chromosome is lost and in intercalary deletion an intermediate segment of the chromosome is lost.

When deletion occurs in one member of a homologous chromosome a deletion loop is produced in the normal homologous chromosome at the opposite to the deleted segment. In humans cridu chat syndrome is due to deletion of segment of 5th chromosome. In this syndrome baby cries like cat and is mentally retarded with small head.

b) Duplication:

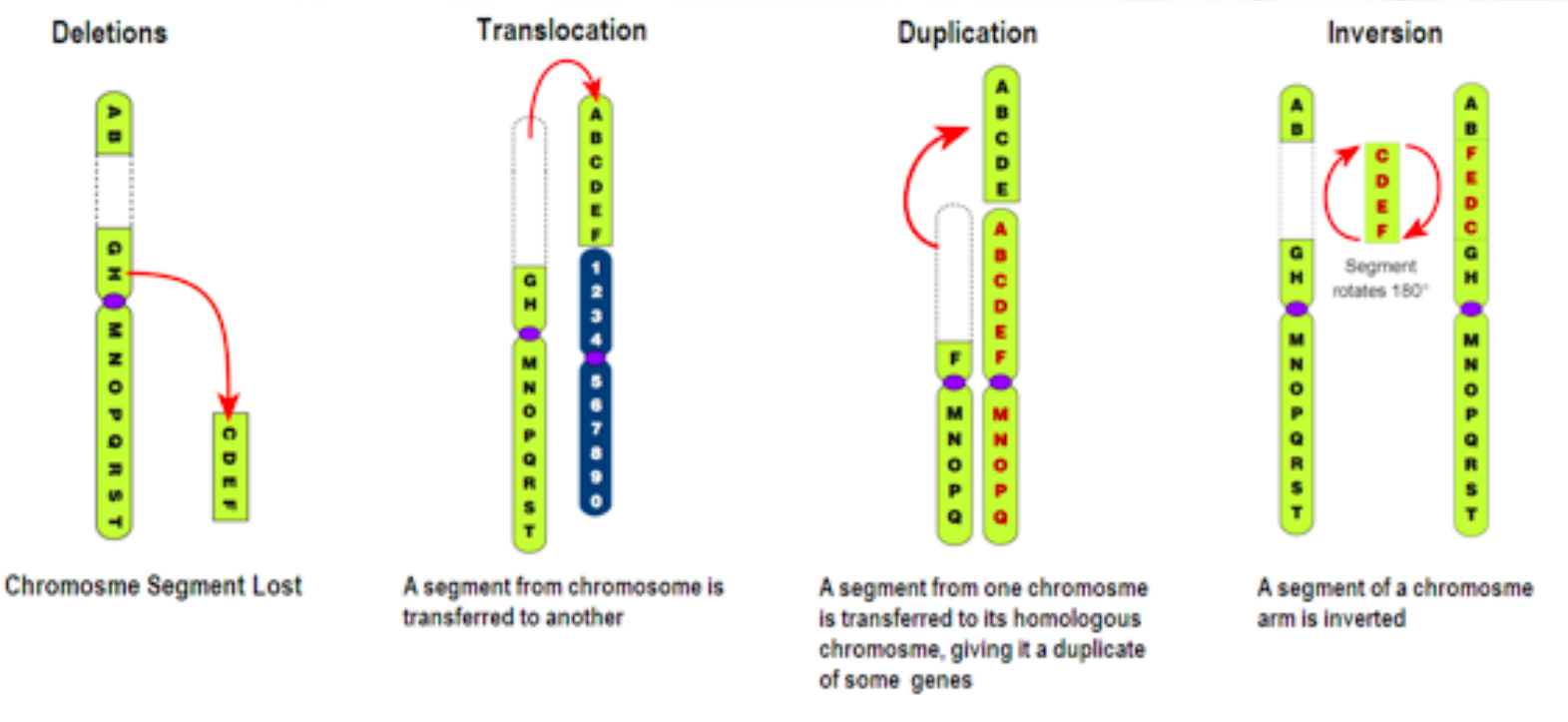
It is the chromosomal aberration where the segment of the chromosome repeated. Hence a set of genes is present in double doses. The duplicated segment forms a loop which shows position effect. Bar eye in Drosophila is the best example of duplication.

c) Inversion:

It is the chromosomal aberration where a segment of chromosome breaks and reunites in the reverse order. In inversion there is no loss or gain of genes but the genes are arranged in reverse order. It may be of pericentric or paracentric type. In pericentric inversion centromere is included in the inverted segment and in paracentric inversion centromere is not included in the inverted segment. The chromosome with the inverted segment produces inversion loop. Inversion prevents crossing over. It brings position effect and produces variation and speciation.

d) Translocation:

In translocation non homologous chromosomes exchange segments. It produces a cross shaped structure during pairing. It has position effect and it alters the linkage groups.



B) Change in number of chromosomes:

Change in number of chromosomes is called ploidy. Ploidy may be due to a change in one chromosome or a set of chromosomes. Based on this there are two kinds of ploidy called

1. **Aneuploidy**
2. **Euploidy**

1) Aneuploidy:

It is a chromosomal aberration where there is gain or loss of one or more chromosomes in a set. It is caused by non-disjunction of chromosomes and is of three types.

a) Monosomy:

In monosomy one chromosome is lost from a pair. Turner syndrome is due to monosomy.

b) Nullisomy:

In Nullisomy both chromosomes of a pair lost. Nullisomic individuals cannot survive.

c) Trisomy:

In trisomy one chromosome is added to a pair. It is of two types i.e. trisomy of autosomes(Down syndrome- Trisomy 21) and trisomy of sex chromosomes (Klinefelter's syndrome, 22AA + XXY)

2) Euploidy:

It is a chromosomal aberration involving the change in the number of chromosome set. It is of two types.

a) Haploidy:

Sometimes in the life of some animals a set of chromosomes will be lost and this leads to haploidy. So some characters which are present I any parent, will be lost from the resulting individual.

b) Polyploidy:

In polyploidy an organism contains more than the usual two sets of chromosomes such animals are called polyploid. Polyploid organisms may have three, four or more number of sets and are called Triploids (3N), Tetraploids (4N), and Pentaploids (5N) and so on.

INDUCED GENE MUTATION:

Artificial induction of mutation in the living organisms by exposing them to abnormal environment such as radiation, certain physical conditions (i.e. temperature) and chemicals is called induced gene mutation. The substances or agents which induce artificial mutations are called mutagens or mutagenic agents. These are of followings three types-

a) Radiations:

Ionizing radiations such as X-rays, gamma rays, alpha and beta rays, electrons, protons, neutrons and other fast moving particles. Non-ionizing radiation includes ultra-violet (UV) light.

b) Temperature as mutagen:

It is reported that rate of mutation is increased by increase in temperature. For example an increase of 100^0C temperature increases the mutation rate by two or three fold. Temperature probably affects the thermal stability of DNA and the rate of reaction of other substances with DNA.

c) Chemical mutagen:

Chemical substances which are responsible to increase the mutability of genes are called chemical mutagens. It was first of all demonstrated by Auerbach and Robson in 1947 using mustard gas and related compounds as the nitrogen and sulphur mustards, mustard oil and chloroacetone in experiments with Drosophila melanogaster.

SEX DETERMINATION

5) SEX DETERMINATION

1) Sex Chromosomal theory of sex determination
2) Genic balance theory
3) Haploidy, Diploidy mechanism
4) Environmental sex determination
5) Dosage compensation

1) CHROMOSOMAL THEORY OF SEX – DETERMINATION :

In dioecious, diploidic organisms following two systems of chromosomal determination of sex have been recognized. a) Heterogametic males b) Heterogametic females

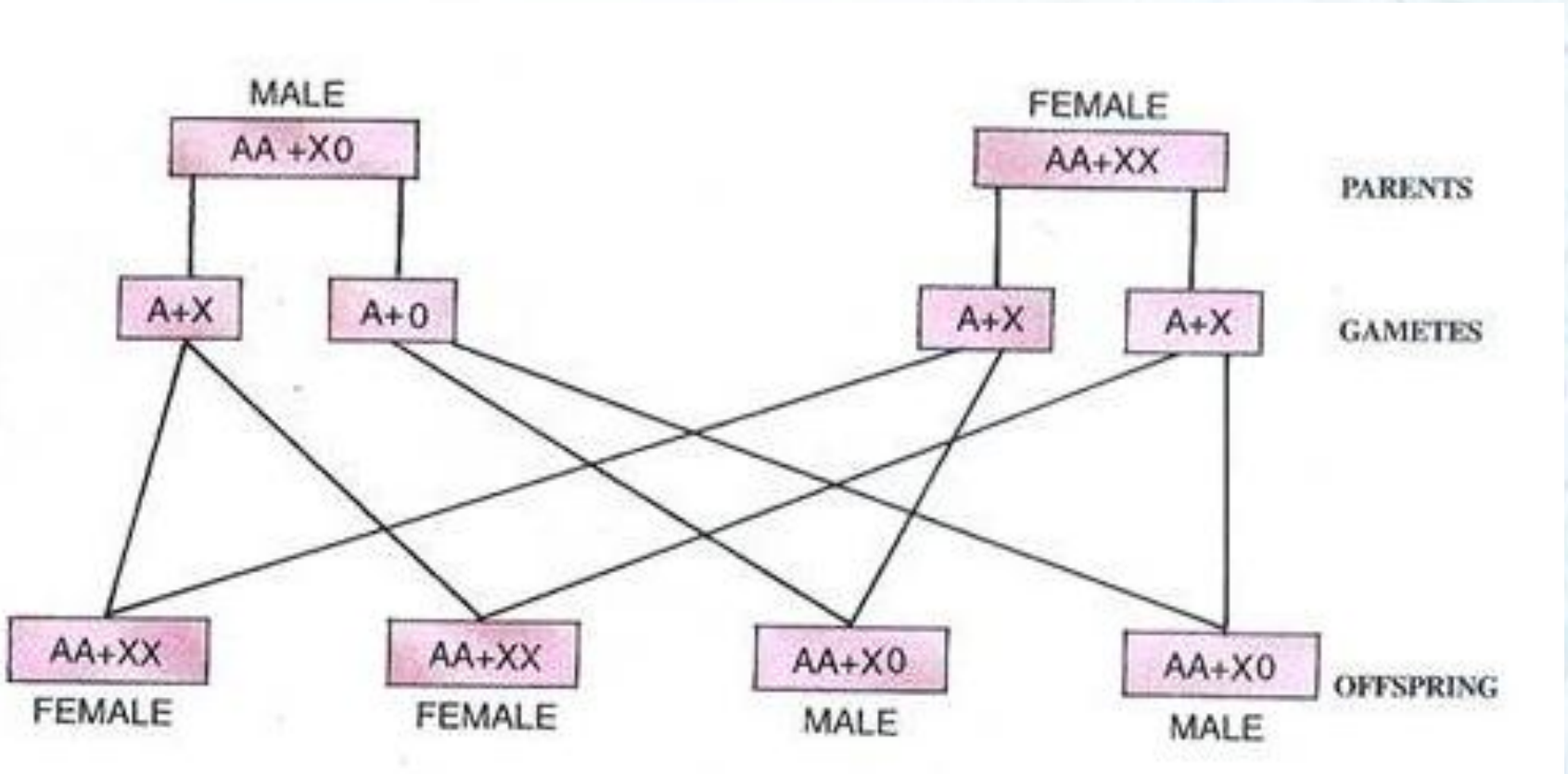
a) Heterogametic Males :

In this type of chromosomal determination of sex, the female sex has two X chromosomes, while the male sex has only one X – chromosome. Because male lacks a X chromosome therefore during Gametogenesis produces two types of gametes 50% gametes carry the X chromosomes while the rest 50% gametes lack in X chromosome. Such a sex which produces two different types of gametes in terms of sex chromosomes called heterogametic sex. The female sex therefore produces similar types of gametes is called homogametic sex. The heterogametic males may be of following two types.

i) XX-XO System:

The somatic cells of a female grasshopper contain 24 chromosomes, where as those of the male contain only 23 chromosomes. Thus in Grasshopper (and in many other insects) there is a chromosomal difference between the sexes, females referred to as XX (having two X chromosomes) and males as XO ("X-oh", having only one X chromosome). The X chromosome is called a sex chromosomes, the remaining chromosome are called autosomes. Thus in XX- XO system, all the eggs have one X chromosome where as the sperms are of two types X and O that is half the sperms have one X chromosome and the other half have none.

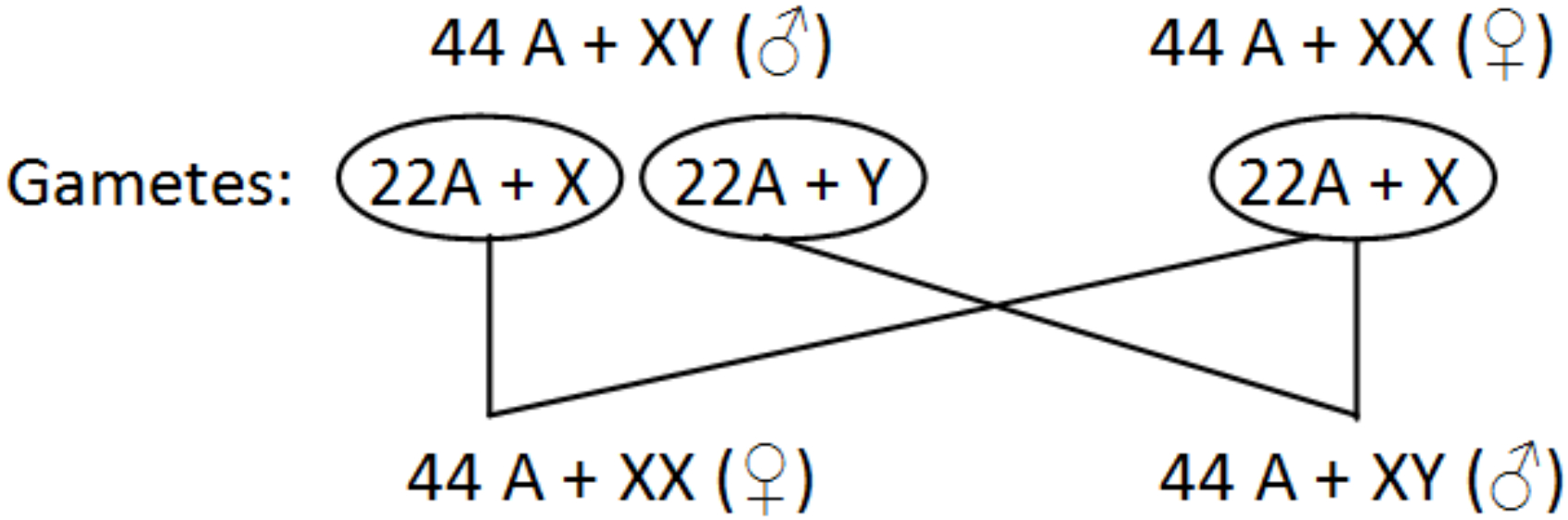
XX-XO System


MALE
AA +X0
FEMALE
AA+XX
PARENTS
A+X
A+0
A+X
A+X
GAMETES
AA+XX
FEMALE
AA+XX
FEMALE
AA+X0
MALE
AA+X0
MALE
OFFSPRING

ii) XX – XY System:

In mammals, Drosophila and some plants (eg. The angiosperms genus lichens) etc. the females are generally referred to as XX (having two X chromosomes) and male as XY (having only one X chromosomes and another one called Y chromosome) . In Drosophila there are four pairs of chromosomes, three pairs of these chromosomes (that is two pairs of V- shaped chromosomes and a pair of small dot like chromosomes) in both sexes are called autosomes. The fourth pair of chromosomes is different in the two sexes, these are sex chromosomes, In female Drosophila both the sex chromosomes are identical and each is called X chromosome in male one of the sex chromosome is straight (X- chromosome) but the other is bent having two unequal arms Y-chromosome) In man, the female have 44 autosomes and one X- chromosome and a Y chromosomes (44+XY). In the XX-XY system, all the ova have one X- chromosomes whereas the sperms are of two kinds, X and Y. in both the XX-XO and XX-XY types, the male is the heterogametic sex (Producing two types of sperms) Where as the female is the homogametic sex (Producing only one type of ovum)

XX–XY System



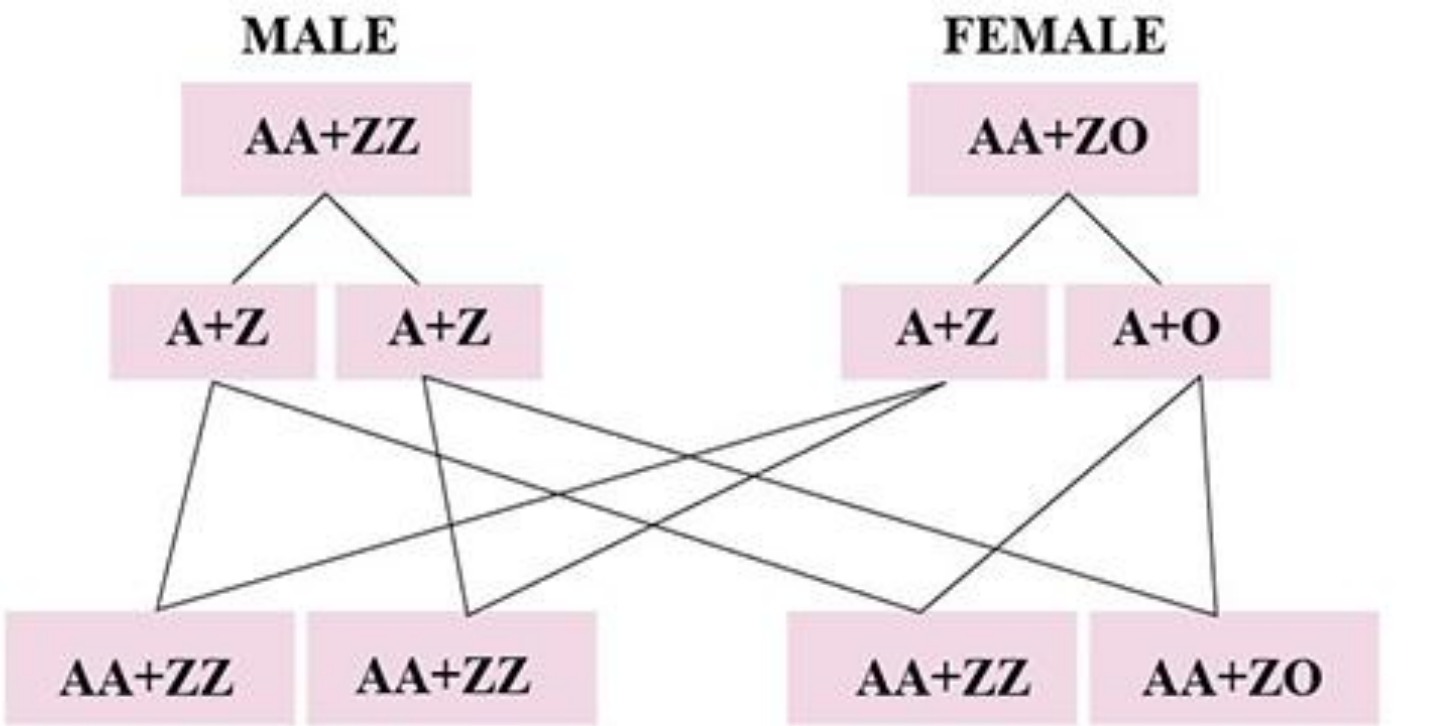
B) Heterogametic Females:

In this type of chromosomal sex determination, the male sexes possesses two homomorphic X chromosomes, therefore, is homogametic and produces single type of gametes, each carries a single X chromosome. The female sex either consists of single X chromosome or one X chromosome and one Y chromosome. The female sex is, thus, heterogametic and produces two types of eggs, Half with a X chromosome and half without a X chromosome (With re without a Y chromosome) To avoid confusion with that of XX-XO and XX-XY type of sex determining mechanisms, instead of the X and Y alphabets Z and W alphabets are generally used respectively. This kind of sex determination mechanism is called Abraxus mechanism of sex determination. (Kuspira and Walker 1973) The heterogametic females may be of following two types.

I) ZZ - ZO System:

This system of sex determination is found in certain moths, Butterflies and domestic chickens. In this case, the female possesses single Z chromosome in its body cells (Hence is referred to as ZO and is heterogametic, producing two kinds of eggs half with a Z chromosome and half without any Z chromosome. The male possesses to Z chromosome (hence referred to as ZZ) and is homogametic, producing single type of sperms each of which carries a single Z chromosome

ZZ-ZO System

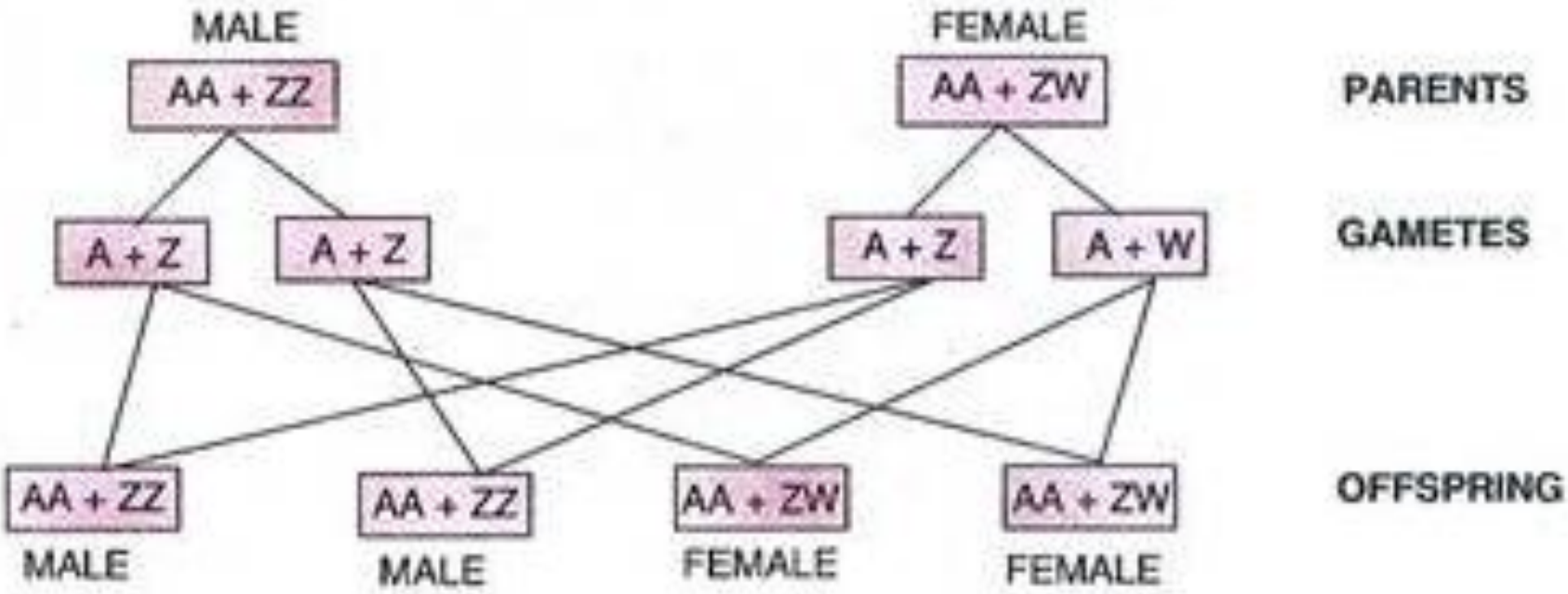


ZO-ZZ DETERMINATION OF SEX IN BUTTERFLY

II) ZZ - ZW System:

In birds (Including the domestic fowl), butterflies, moths and some fishes the female is heterogametic but the male is homogametic. To avoid confusion, the sex chromosome in this case are often designated as Z (instead of X) and W (instead of Y) Thus in these cases females are ZW (or XY) and males are ZZ (or XX).

ZZ-ZW System


MALE
AA + ZZ
FEMALE
AA + ZW
PARENTS
A + Z
A + Z
A + Z
A + W
GAMETES
AA + ZZ
MALE
AA + ZZ
MALE
AA + ZW
FEMALE
AA + ZW
FEMALE
OFFSPRING

2) GENIC BALANCE THEORY:

This theory was put forward by Bridges, 1922 who opined that in Drosophila and other organisms the Y chromosome is practically inert as far as the determination of sex was concerned. He opined that the sex of the individuals actually depends on a delicate balance between the sex chromosomes and autosomes. According to him the X chromosome consists of genes for femaleness, while the autosomes carry the genes for maleness, while. Any increase in the dose of the X chromosome (XX) will result in femaleness. While any reduction will produces maleness. The development of an individual either into male or into female depends on the ratio or balance between X chromosome and autosomes.

- Male X: 3 pairs of autosomes.
- Female XX: 3 pairs of autosomes

If we give a value of 1 to autosomes and 1 to X chromosome 1:1 will be male and 1:2 will be female. Genic balance may be defined as the ratio between the sex chromosomes and autosomes having a prominent role in deciding the sex of the offspring. A few examples from Drosophila will clearly illustrate this phenomenon that is not nearly the quantity of the X chromosome that decides the sex of the individual, but the relative production of X chromosome to autosomes that is important in sex determination.

GENIC BALANCE THEORY

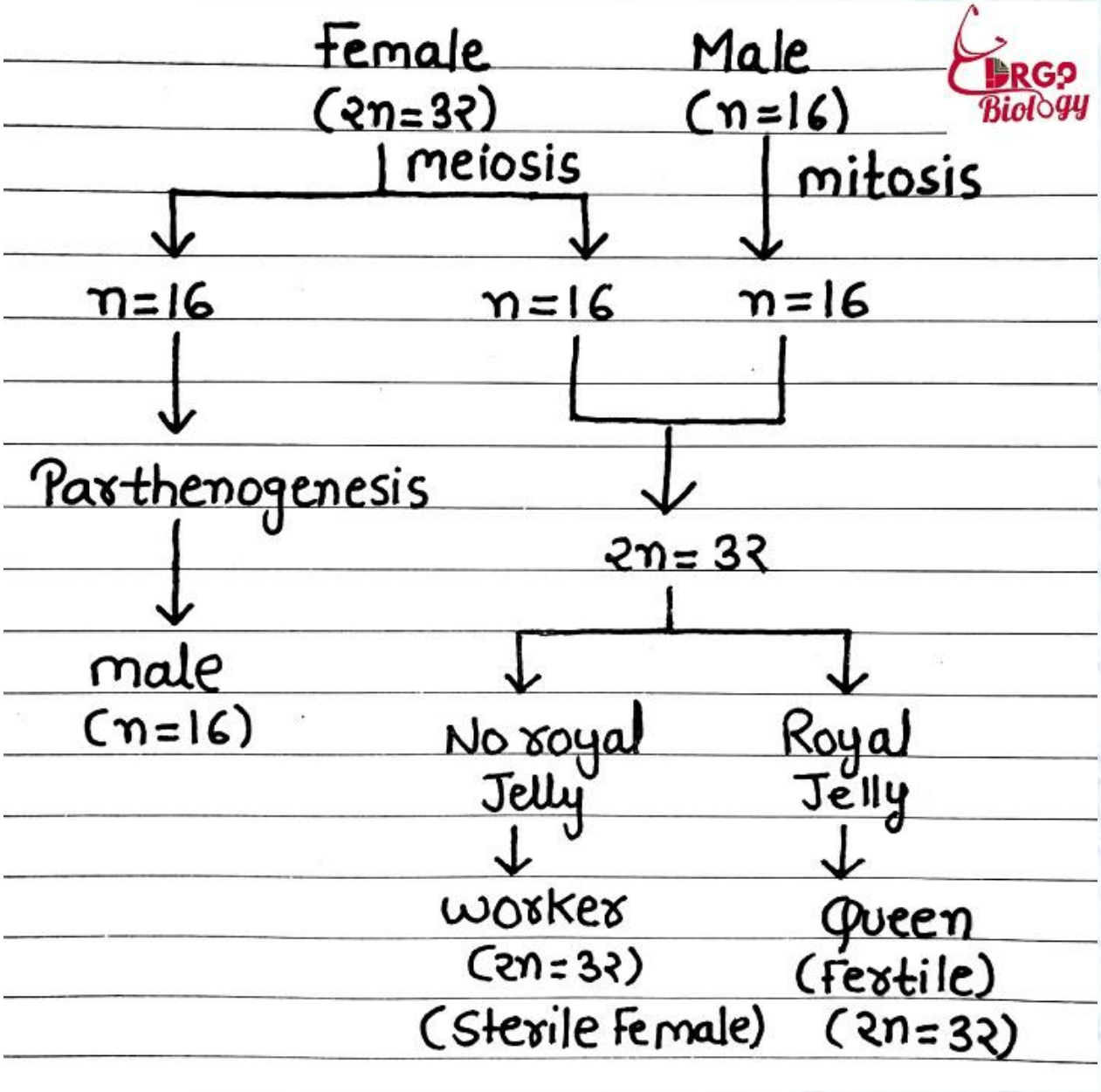
Chromosome Complement	X / A Ratio	Sexual Morphology
X X X + 2A	3/2 or 1.5	Metafemale
X X X + 3A	3/3 or 1.0	Female
XX + 2A	2/2 or 1.0	Female
X X + 3A	2/3 or 0.67	Inter sex
X X X + 4A	3/4 or 0.75	Inter sex
XO + 2A	1/2 or 0.5	Male
XY + 2A	1/2 or 0.5	Male
XY + 3A	1/3 or 0.33	Metamale

3) HAPLOIDY DIPLOIDY MECHANISM:

It is also called haplodiploidy or arrhenotokous parthenogenesis and is found in most of the hymenopterous insects such as ants, bees, sawflies and wasps. In these insects since fertilized eggs develop into diploid females and unfertilized ones into haploid males.

For example in case of honey bee, the drones (males) are entirely derived from the queen, their mother. The diploid queen has 32 chromosomes and the haploid drones have 16 chromosomes. Drones produce sperm cells that contain their entire genome, so the sperm are all genetically identical except for mutations. The male bees' genetic makeup is therefore entirely derived from the mother, while the genetic makeup of the female worker bees is half derived from the mother, and half from the father.

HAPLOIDY DIPLOIDY MECHANISM

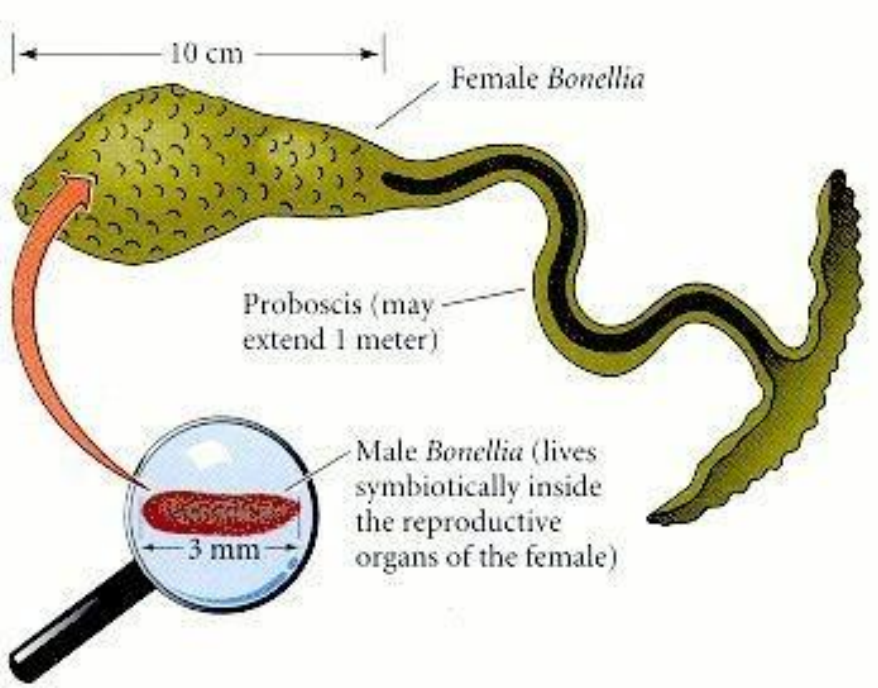


4) ENVIRONMENTALLY CONTROLLED SEX DETERMINATION:

In certain cases Environmental factors rather than chromosomes are known to influence sex as is seen in marine worms Bonelia, Molluscs, Daphnids, Rotifers, and Aphids etc. The mechanism of sex determination in Bonelia has been studied by Baltzer. The male and female individuals are strikingly dimorphic. The female worm is leaf like and several centimetres long. There is a long proboscis extending from the anterior part of the body. The ovaries are distended. The male individual is an extremely minute creature which lives in the reproductive tract of the female. The only function of the male is that to fertilize the eggs produced in the ovaries of the female.

Baltzer discovered that, if a single worm is reared separately from others, it invariably develops in to female, while the worms patched together and released into water containing mature females tend to develop into both male and female worms.

Some of the young ones attach themselves to the proboscis of the female and obtain the nourishment. These become male worms and eventually migrate down to the oviducts. It is assumed that the some hormonal secretions at the proboscis region in the female transfer the attached young ones in to males.



GYNANDROMORPHS

In Greek (Gyne: woman, aner: man and morphe: form) gynandromorph refers to the appearance of both male and female characters at different halves of the body in the same organism. In insect the secondary sexual characters are not influenced by the hormones secreted by the gonads and the differentiation occur independently depending upon the genetic constitution of the cells in various parts of the body. When the genotype of the cells in different parts of the body differs with respect to the sex linked characters a sex mosaic results and is known as gynandromorph. Gynandromorphs differ from the intersexes. The later are genetically sterile but Gynandromorphs consist of two genetically different tissues, some of the cells of gynandromorph are genetically male and others are female.

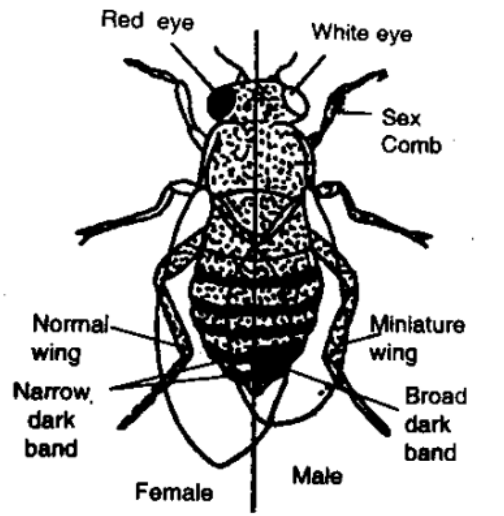
Gynandromorph cases have been studied in several species of insects like Drosophila, bees and in silkworms. In a gynandromorph of Drosophila obtained by Morgan the right half of the fly showed male characters viz. shorter wings, black tipped abdomen and a sex comb of the first leg. The left side showed contrasting female characters. The right eye was white and the left one red.

Kinds of Gynandromorphs:

Gynandromorphs can be classified into the following types on the basis of the position of sex tissues the gynandromorph have

1) Bilateral gynandromorph:

If the line of division between male and female tissues of a gynander passes through the middle of the body the stage is called bilateral gynandromorph.



BILATERAL GYNANDROMORPHS IN DROSOPHILA

2) Antero- posterior gynandromorph:

It is less common than bilateral gynandromorph. In such case the anterior region of the body has the characters of one sex and posterior region has the characters of other sex.

3) Sex piebalds:

This kind of gynandromorph is of very rare occurrence. In this the body consist of female tissue having spots of male tissue scattered irregularly or the vice versa.

Causes of formation of gynanaders:

1) Elimination of X-chromosome

2) Fertilization of binucleated eggs.

5. DOSAGE COMPENSATION:

Dosage compensation is the process by which organisms equalize the expression of genes between members of different biological sexes.

For example, in humans, females (XX) silence the transcription of one X chromosome of each pair, and transcribe all information from the other, by expressed X chromosome. Thus, human females have the same number of expressed X-linked genes as do human males (XY), both sexes having essentially one X chromosome per cell, from which to transcribe and express genes.

DETAILS ABOUT DOSAGE COMPENSATION OR LYON'S HYPOTHESIS

1. The inactive X hypothesis or the Lyon's hypothesis or the Dosage Compensation is widely known from 1961 which states that only one of the two X chromosomes in the homogametic sex is functional while the other condenses and is inactivated. The X inacti-vated in some cells would be that from the father, in other cells it would be that from the mother.
2. Hence any tissue in the body of a woman would be a mosaic of cells which would show dominance of all genes having diffusible products but would remain a fine--grained mosaic for other intracellular differen-ces.
3. Such a mosaic of cells might be difficult to demonstrate, particularly among rigid tissues, although cells which can be separated and cloned might show antigenic differences. This hypothesis has stimulated many new investiga-tions, some of which are currently being completed.

OBJECTIVES BEHIND THE PROPOSITION OF LYON'S HYPOTHESIS

Lyon was impressed by three observations relating to X chromosome

1. In normal mammalian females, one of the two X's is genetically inactive in the soma-tic cells (single active X-hypothesis).
2. Inactivation is random i.e., irrespec-tive of paternal and maternal origin (random inactivation).
3. (a) The inactivation occurs during early ontogeny (early ontogenic differentiation) and (b) The particular X which has thus become inactivated, remains inactive in all the succee-ding cell generation (fixed differentiation).

It has been observed that Dosage Compensation takes place by means of following three ways:

1. Random inactivation of one of the female's X chromosome.
2. Two-fold increased transcription of a single male's X chromosome
3. Decreased transcription of both hermaphroditic X chromosomes by half.

“Thank you”

Genetics (Powerpoint Presentation e-Book for Students)

About Authors

Prof. K. J. Adate, is working as a Head, Department of Zoology, Shivraj College Gadhinglaj, Dist-Kolhapur since last twenty years. He has published many research papers in reputed journals. Till today Published seven books for B.Sc.I and B.Sc.II, Zoology on Theory and practical syllabus of Shivaji University, Kolhapur.
Mob. 9271692747

Dr. V. V. Ajagekar is working as a Head, Department of Zoology, Ajara, Mahavidyalaya, Ajara, Dist-Kolhapur since last twenty six years. He is Research guide for M.Phil. and Ph.D. of Shivaji University Kolhapur. He has completed one Minor Research Project and published sixteen research papers in reputed Journals. Till today Published two books for B.Sc.I and two books for B.Sc.II, on syllabus of Shivaji University Kolhapur.
Mob. 9890087915

About Book

During the Covid-19 pandemic teachers and students are doing work from home. This is the right time to provide teaching material to the students and teachers. Taking this in to consideration this E-Book is prepared which is based on syllabus of Shivaji University, Kolhapur. Various reference books have been referred while preparing this book. Information from the internet has also been collected. For the easy understanding of the student's simple language, large, accurate, and neat labeled diagrams are used.

Bhumi Publishing

ISBN 978-81-951982-5-2