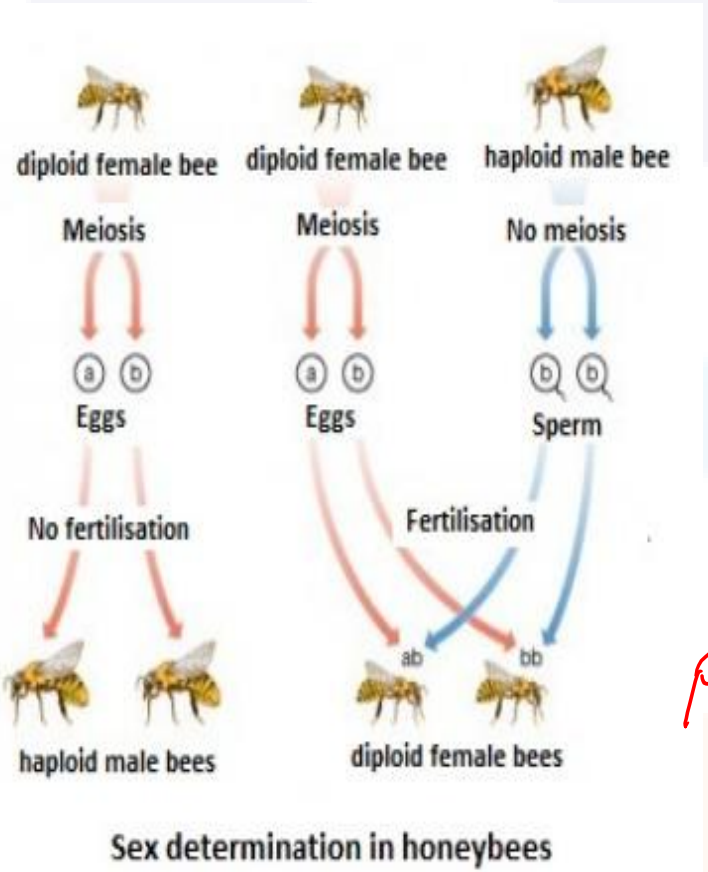
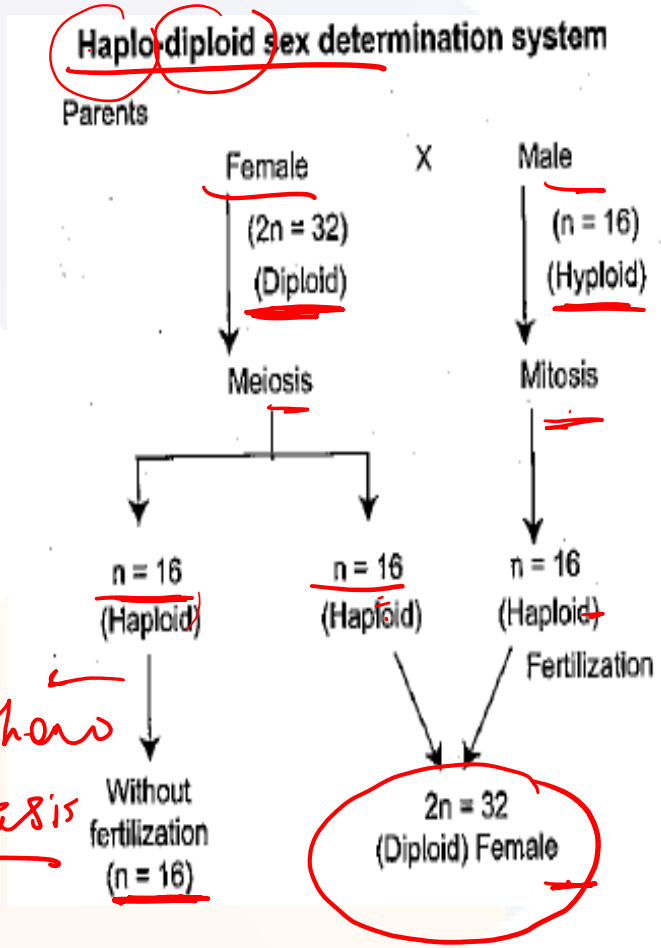
SEX DTERMINATION IN HONEY BEE



Sex determination in honeybees



Parthenogenesis

Mutation and Genetic Disorder

Mutation is a phenomenon which results in alternation of DNA sequence and consequently results in the change in the genotype and phenotype of an organism.

Point mutation

Mutations arise due to change in single base pair of DNA are called point mutation e.g. Sickle cell anaemia.

Frame shift Mutation

Frameshift mutation caused by a deletion or insertion in a DNA sequence that shifts the way the sequence is read.

Genetic Disorder

Mendelian disorder

Chromosomal disorder

Mendelian Disorders	Chromosomal disorders
These are due to alteration in a single gene.	These are caused due to absence or excess of one or more chromosomes or abnormal arrangement of one/more chromosomes.
They are transmitted into generations through Mendelian principles of inheritance.	They may be recessive or dominant in nature.
Examples: Colour blindness Pheffykenonia.	Examples: Downs syndrome, Turner's syndrome

Mendelian disorder

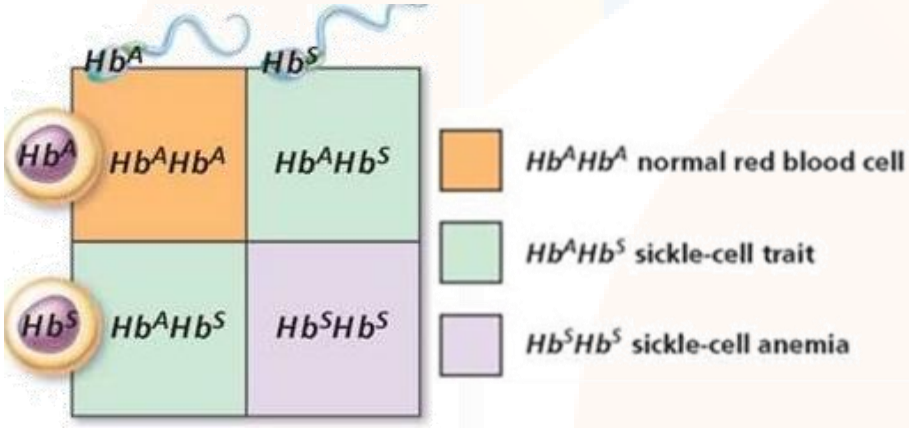
Haemophilia- sex linked recessive disease in which, in an infected individual, a minor cut leads to non-stop bleeding.

It is due to mutation in gene responsible for synthesis of one of the clotting factor.

Heterozygous female (carrier) can transmit the disease to their son. The possibility of a female becoming a haemophilic is extremely rare.

Sickle cell anaemia- an autosome linked recessive trait in which mutant haemoglobin molecules undergo polymerization under low oxygen tension causing change in shape of the RBC from biconvex disc to elongated sickle like structure.

The defect is caused by the substitution of Glutamic acid (Glu) by Valine (Val) at the sixth position of the beta globin chain of the haemoglobin molecule, due to the single base substitution at the sixth codon of the beta globin gene from GAG to GUG



Phenylketonuria- inborn error of metabolism inherited as autosomal recessive trait. The affected individual lacks an enzyme that converts the amino acids phenylalanine to tyrosine, as a result of this phenylalanine is accumulated and converted into phenyl pyruvic acid and other derivatives that results into mental retardation.

Chromosomal Disorder

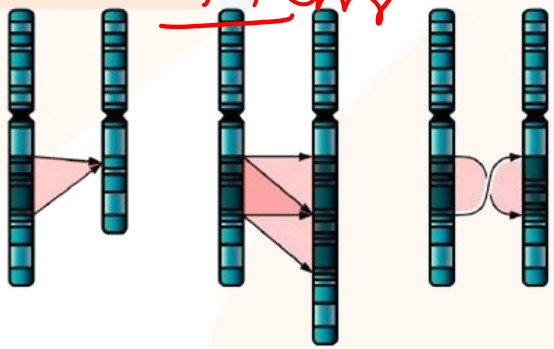
Failure of segregation of chromatids during cell division results in loss or gain of chromosome called **aneuploidy**.

The failure of cytokinesis leads to two sets of chromosome called **polyploidy**.

Down's Syndrome – is due to presence of additional copy of the chromosome number 21. The affected individual is short statured with small rounded head, furrowed tongue and partially opened mouth. Mental development is retarded.

Turner's Syndrome– caused due to the absence of one of the X chromosome. 45 with XO, such females are sterile as ovaries are rudimentary. They lack secondary sexual characters.

Klinefelter's Syndrome– due to presence of an additional copy of X-chromosome (XXY). Such persons have overall masculine development however, the feminine development (development of breast, i.e., Gynaecomastia) is also expressed. They are sterile.



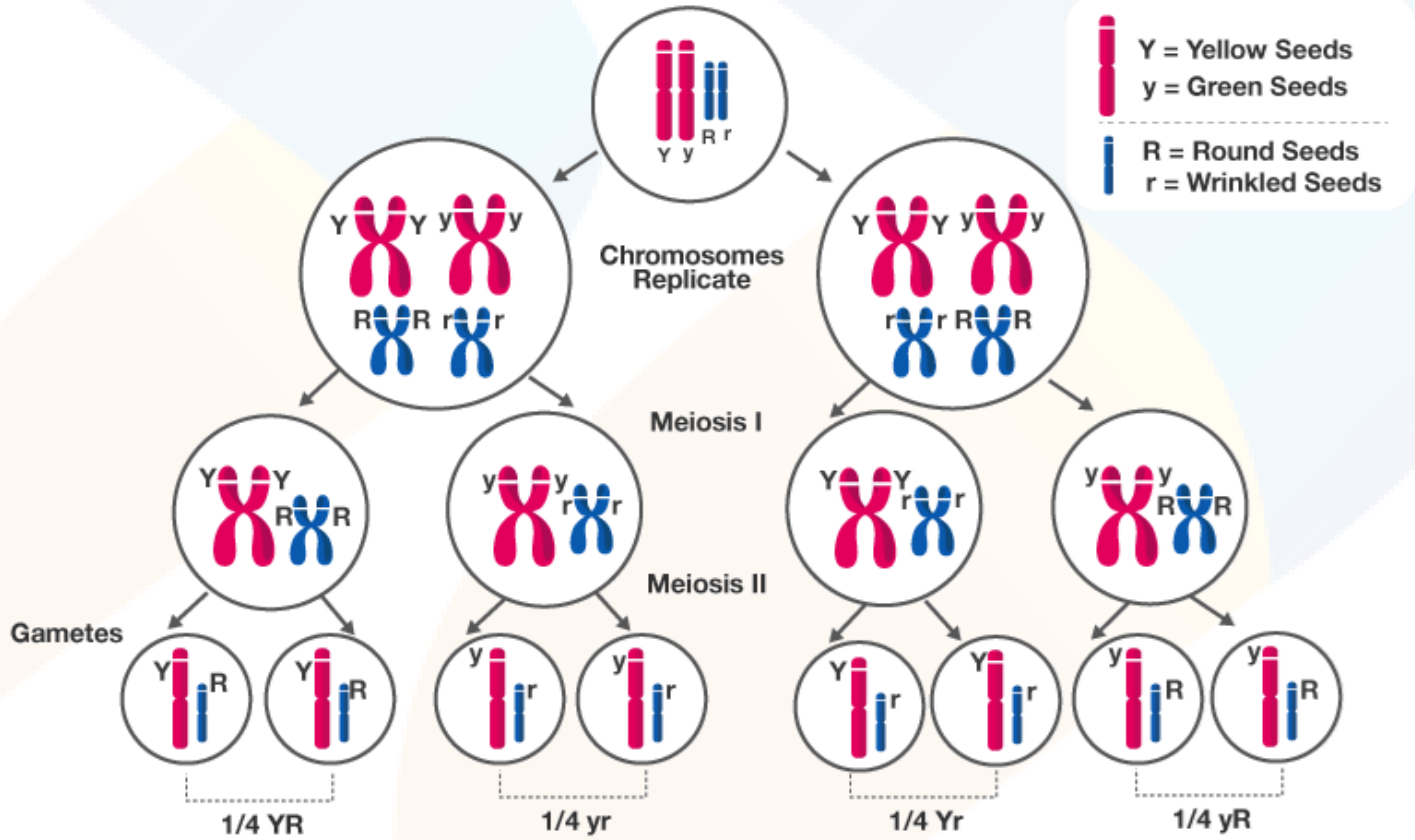
1. Deletion
   - Genetic material is missing
2. Duplication
   - Genetic material is present twice
3. Inversion
   - Genetic material is "flipped"

Chromosomal Theory of Inheritance

Chromosome as well as gene both occurs in pair. The two alleles of a gene pair are located on the same locus on homologous chromosomes.

Sutton and Boveri argued that the pairing and separation of a pair of chromosomes would lead to segregation of a pair of factors (gene) they carried.

Sutton united the knowledge of chromosomal segregation with mendelian principles and called it the chromosomal theory of inheritance.



Linkage and Recombination

When two genes in a Dihybrid cross were situated on same chromosome, the proportion of parental gene combination was much higher than the non-parental type. Morgan coined this as **linkage** to describe the physical association of genes on same chromosome.

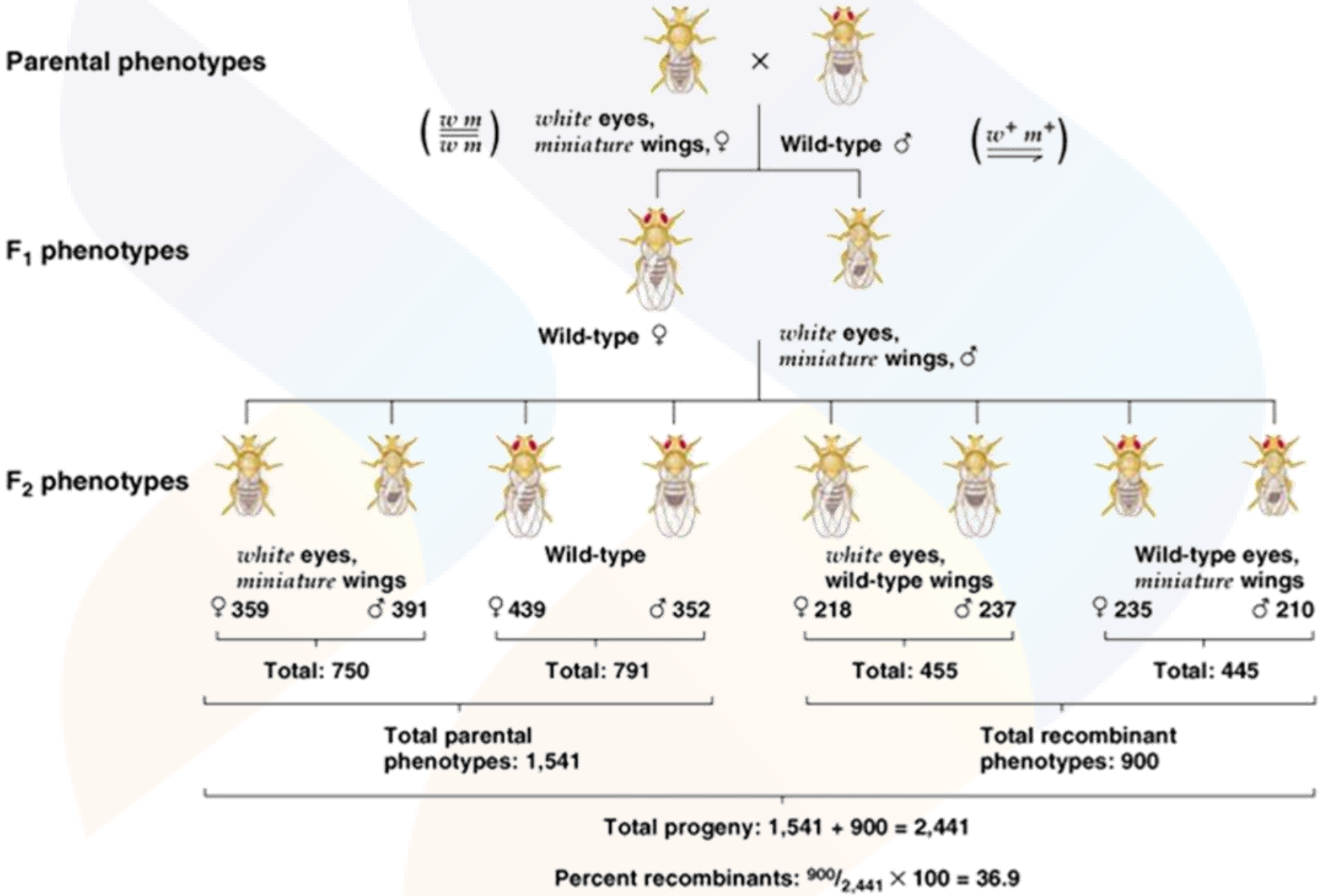
The generation of non-parental gene combination during Dihybrid cross is called recombination. When genes are located on same chromosome, they are tightly linked and show very low recombination.

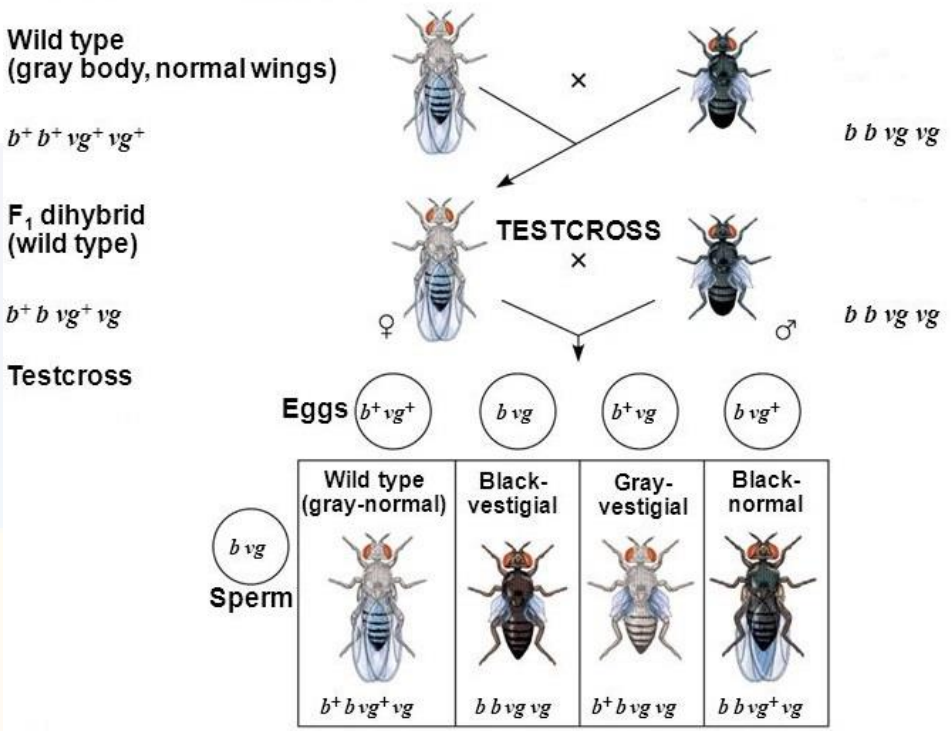
More linkage less recombination, less linkage more recombination

In Drosophila eye color and body color genes are linked show less recombination's, and eye color and size of wings are quite apart showing more recombination's.

Crossing over	Linkage
1. It leads to separation of linked genes	1. keeps the genes together
2. It involves exchange of segments between non-sister chromatics of homologous chromosomes.	2. It involves individual chromosomes.
3. The frequency of crossing over can never exceed 50%.	3. The number of linkage group can never be more than haploid Chromosome number.
4. It increases variability by forming new gene combinations.	4. It reduces variability.

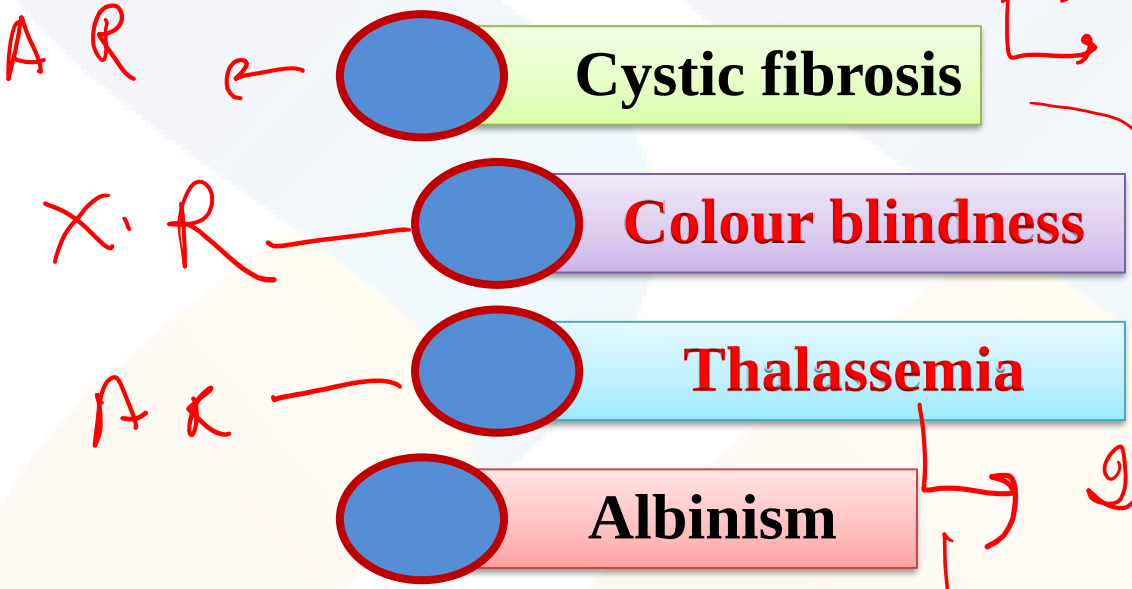
Morgan's Experiment on White eyes and Miniature Wings





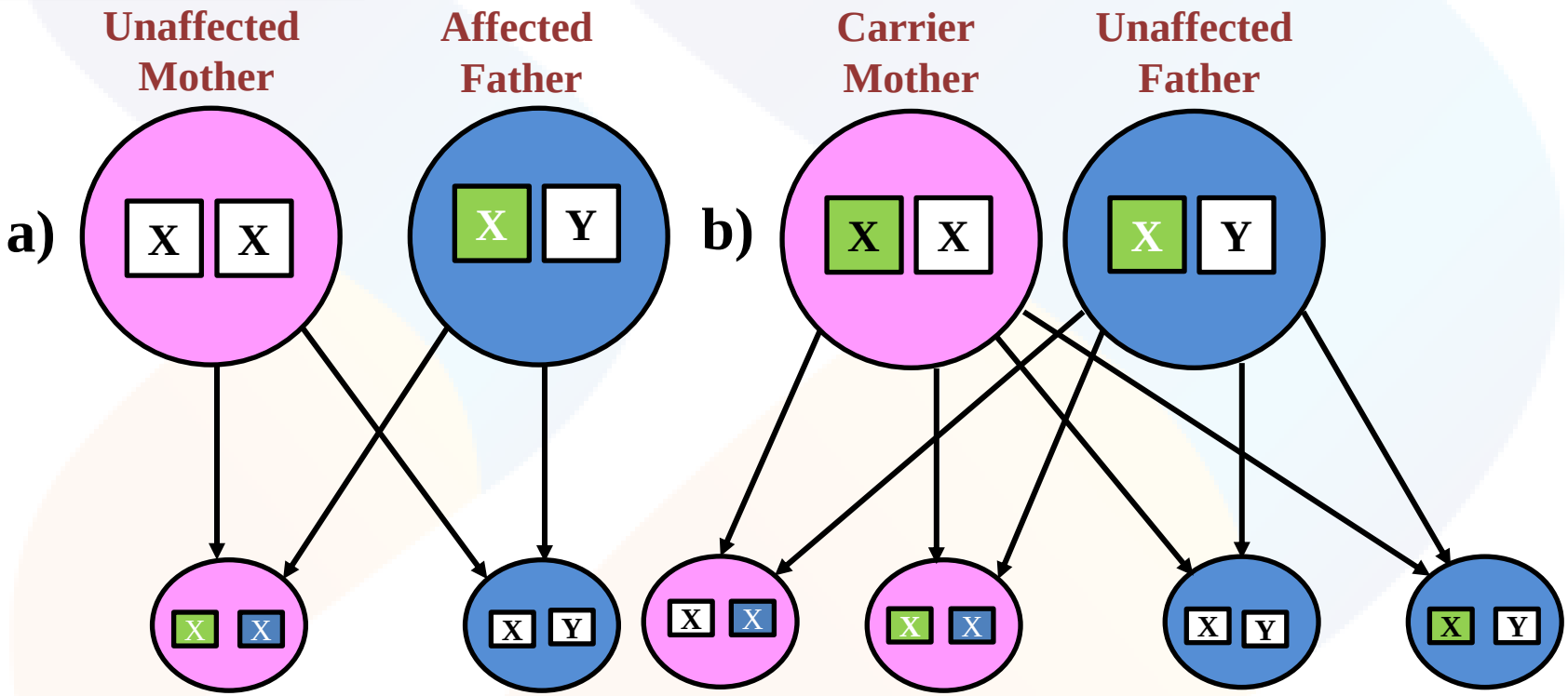
genes on different chromosomes:	1	:	1	:	1	:	1
genes on same chromosome & there is no recombination:	1	:	1	:	0	:	0
genes on same chromosome & there is recombination:	965	:	944	:	206	:	185

MENDELIAN DISORDERS



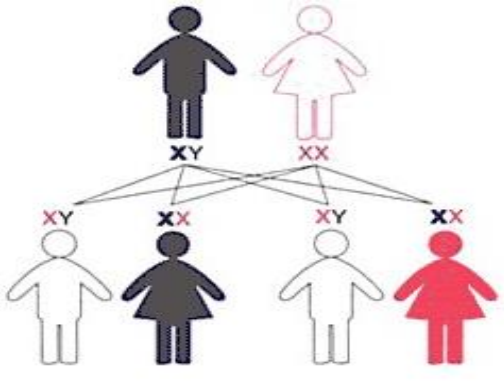
Queen Victoria passed Haemophilia on to some of her descendants

Haemophilia



When the father has haemophilia and the mother is unaffected

Father **Mother**

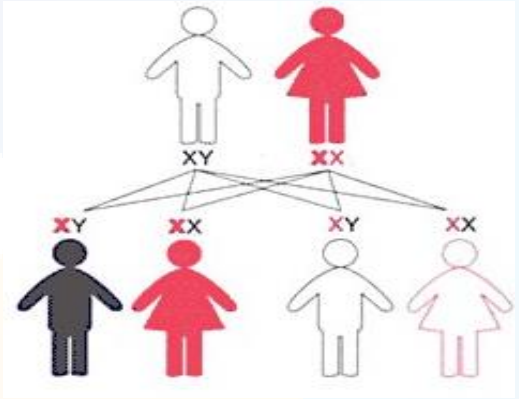


None of the sons will have haemophilia.

All of the daughters will carry the haemophilia gene.

When the mother carries the haemophilia and the gene is unaffected

Father **Mother**



There is a 50% chance at each birth that a son will have haemophilia.

There is a 50% chance at each birth that a daughter will carry the haemophilia gene.

Haemophilia

Carrier female, normal male = half sons affected, half daughter carrier

	X^H	Y
X^H	$X^H X^H$	$X^H Y$
X^h	$X^H X^h$	$X^h Y$

	X^h	Y
X^H	$X^H X^h$	$X^H Y$
X^h	$X^h X^h$	$X^h Y$

Carrier female, affected male = equal chance of affected male/female progeny

Normal female, affected male = no sons affected, all daughter carrier

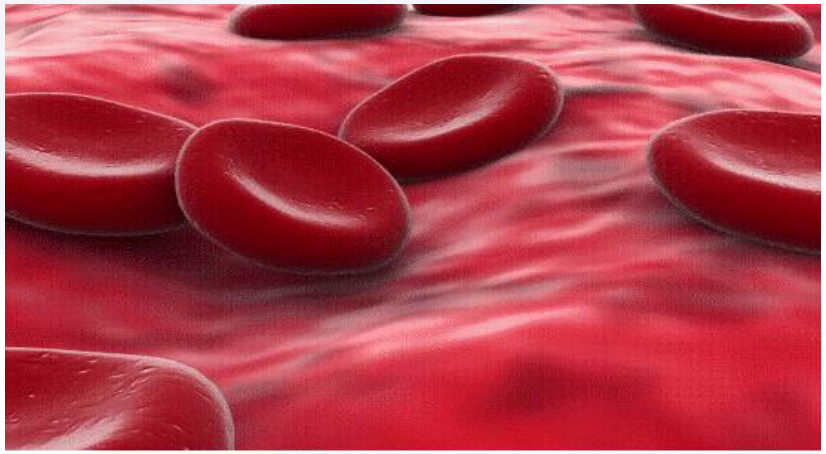
	X^h	Y
X^H	$X^H X^h$	$X^H Y$
X^H	$X^H X^h$	$X^H Y$

	X^h	Y
X^h	$X^h X^h$	$X^h Y$
X^h	$X^h X^h$	$X^h Y$

Affected female, affected male= all progeny affected

SICKLE-CELL ANAEMIA

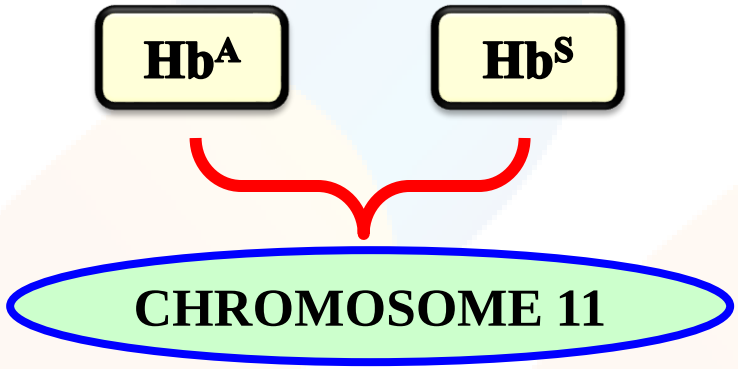
Autosomal recessive genetic blood disorder.

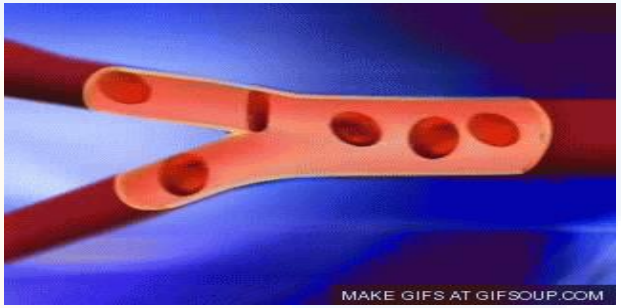


Red blood cells are characterized by an abnormal, rigid, sickle shape in hypoxia conditions.

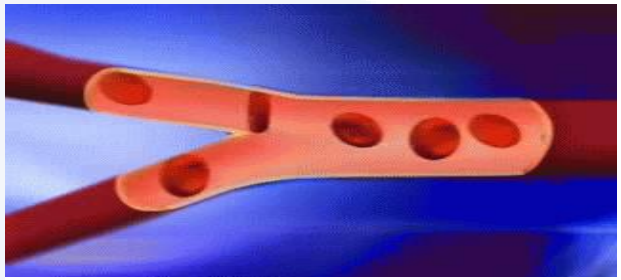
SICKLE-CELL ANAEMIA

Sickled-cell anemia is controlled by a single pair of alleles.





SICKLE-CELL ANAEMIA



Homozygous individuals for sickle-cell anaemia (Hb^S Hb^S) express the diseased phenotype.

Heterozygous individuals (Hb^A Hb^S) appear 'unaffected' but they are still, carriers of the disease.

Points to remember

Even though two sickle cell alleles are necessary to cause sickle cell anaemia, one dose can affect the phenotype.

Persons ‘heterozygous’ to sickle cell trait can usually lead a healthy life.

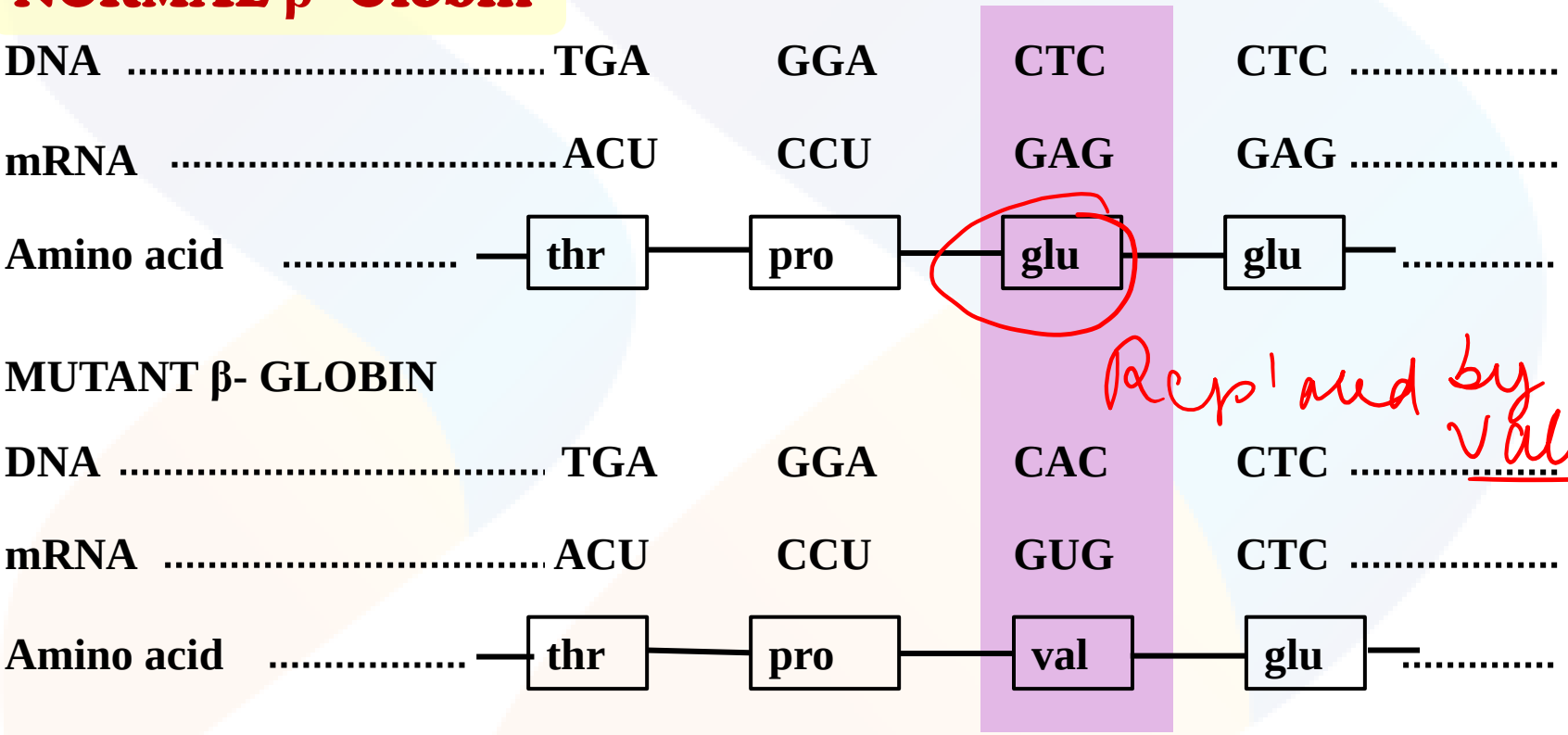
Sickle cell anaemia is caused by a point mutation in the DNA that codes for the beta globin polypeptide chains of the haemoglobin molecule.

Points to remember

It causes the replacement of the glutamic acid in the sixth position by valine.

In prolonged periods of reduced oxygen content in the blood the heterozygous persons may suffer from symptoms of SCD as both normal and sickle cell haemoglobin are formed in them.

NORMAL β- Globin



AUTOSOMAL DISORDERS

- Down syndrome (Trisomy 21)
- Patau syndrome (Trisomy 13)
- Edward syndrome (Trisomy 18)
- cri-du-Chat syndrome (5p minus syndrome)
- Chronic Myelogenous (Myeloid) Leukemia (CML)

DOWN SYNDROME-SYMPTOMS

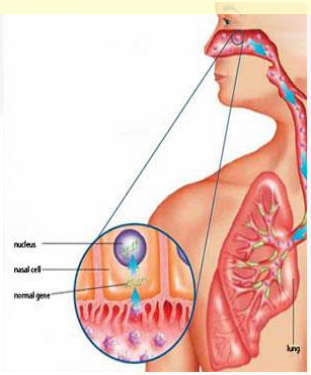
The affected individual is short statured with small round head, furrowed tongue and partially open mouth.

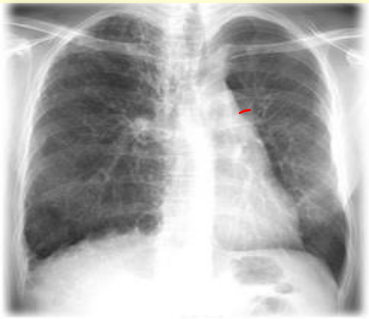
Physical, psychomotor and mental development is retarded.

This syndrome appears much more frequently in children born to ‘aged women’.

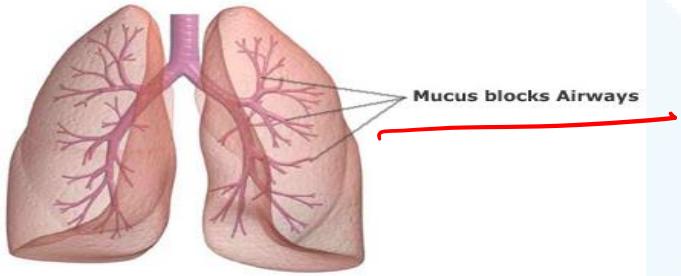
conceive babies rather late in their reproductive phase

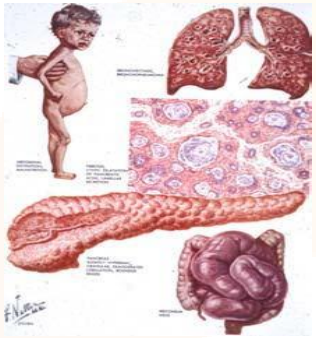
DOWN SYNDROME-SYMPTOMS

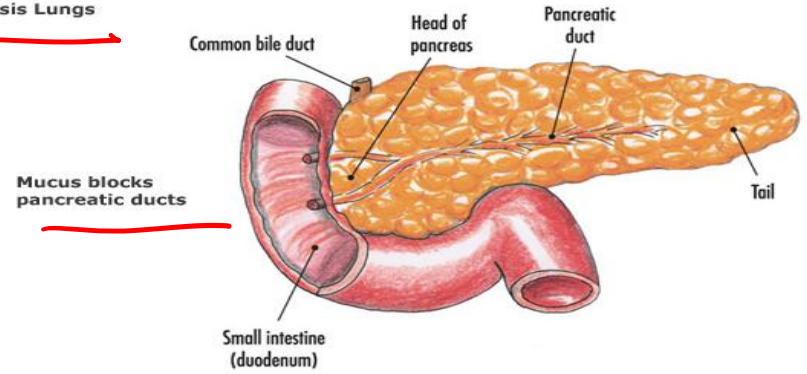




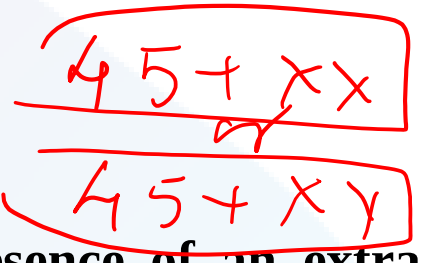
Cystic Fibrosis Lungs







EDWARD'S SYNDROME (TRISOMY 18)



- **Edward's syndrome is characterized by the presence of an extra copy of the genetic material on the 18th chromosome, either in whole (trisomy 18) or in part (such as due to translocations).**
- **The additional chromosome usually occurs before conception.**
- **The karyotype is designated as 47, XX, +18**

PATAU SYNDROME (TRISOMY 13)

45+XX or 45+XY

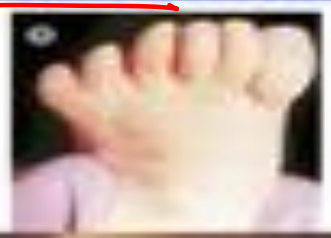
- **Patau syndrome, is a chromosomal condition associated with severe intellectual disability and physical abnormalities in many parts of the body.**
- **Most cases of trisomy 13 result from having three copies of chromosome 13 in each cell in the body instead of the usual two copies.**
- **The karyotype is designated as 47, XX, +13**

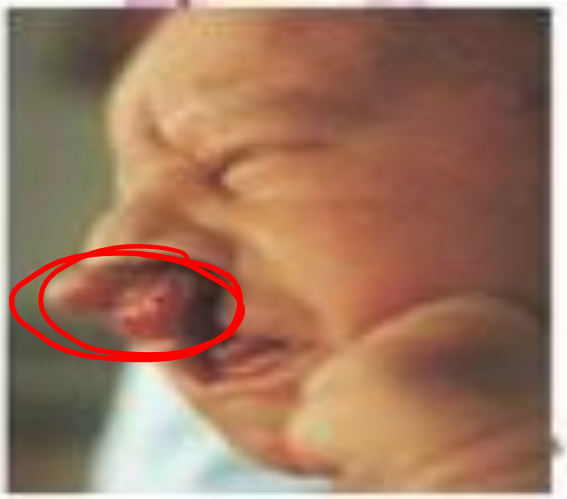
Trisomy 13 (Patau syndrome)

Genetic mechanism is nondisjunction during oogenesis

Features:

- Polydactyly
- Cleft lip and palate
- Microphthalmia
- Microcephaly
- Intellectual disability
- Cardiac anomalies





PATAU SYNDROME - SYMPTOMS

- **Individuals with trisomy 13 often have heart defects, brain or spinal cord abnormalities, very small or poorly developed eyes (microphthalmia), cleft palate etc.**

- **Due to the presence of several life-threatening medical problems, many infants with trisomy 13 die within their first days or weeks of life.**

CRI-DU-CHAT SYNDROME (5P minus syndrome)

- **Cri-du-chat syndrome (cat-cry) is due to a partial deletion of the short arm of chromosome 5**
- **It might be considered a case of *partial monosomy***
- **But since the region that is missing is so small, it is better referred to as *5p segmental deletion*.**
- **The karyotype is 46, XX, $5p^-$.**

CRI-DU-CHAT SYNDROME - SYMPTOMS

- **Cri-du-Chat is a French term referring to the characteristic *cat-like cry* of the affected children due to problems with the larynx and nervous system.**
- **Such infants are mentally retarded, have a small head with unusual facial features.**
- **They die in infancy or early childhood.**

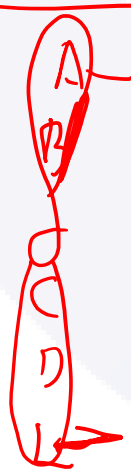
Linkage

Recomfreq = $\frac{\text{No. of Recom}}{\text{Total Progeny}} \times 100$

Crossing over % = Distance b/t genes

Lesser the dist more Linkage

& lesser is the Recombin



1% C.O = 1 map unit

eg Complete linkage - = 1cM

- Grey body & N dumal wings
- Black body & Vestigeal

} Drosophila

No. of Linkag groups = no. of chromosomes in a set

LINKAGE IN DROSOPHILA

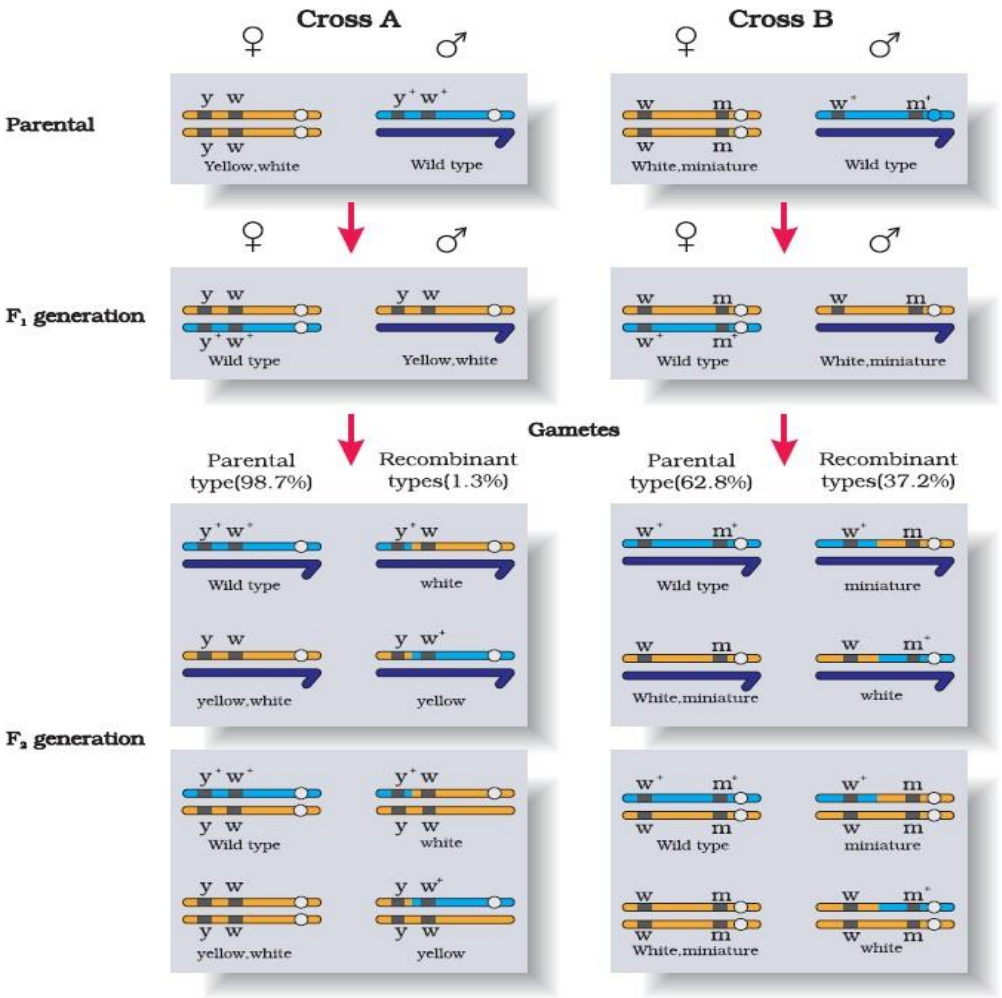


Figure 5.11 Linkage: Results of two dihybrid crosses conducted by Morgan. Cross A shows crossing between gene y and w; Cross B shows crossing between genes w and m. Here dominant wild type alleles are represented with (+) sign in superscript
Note: The strength of linkage between y and w is higher than w and m.