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Lecture – 8

Blood group, Linkage and its estimation

by

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ABO Blood Group

Discovered by Karl Landsteiner in early 1900s

- An excellent example of multiple allele inheritance is human blood type. Blood type exists as four possible phenotypes: A, B, AB, & O.
- The human ABO gene is on chromosome 9.
- Everyone has two copies of chromosome 9 so you have two *ABO* genes.



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ABO Blood Group

One copy is inherited from our mother, the other from our father.

There are 3 alleles (A, B and O) for the gene that determines blood type.

A person's blood type is determined by which allele he/she inherits from each parent.



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- The “A” allele is dominant and so is the “B” allele.
- Together though, the “A” and “B” alleles are co-dominant.
- The “O” allele is recessive.
- One combination of alleles in the ABO system exhibits a codominant mode of inheritance which is blood type AB



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Determining the Genotype

- Human blood type is controlled by three alleles : I^A , I^B and i .
- Alleles I^A and I^B are dominant over i
- I^A and I^B are codominant

Phenotype (blood type)	Genotypes
A	$I^A I^A$ or $I^A i$
B	$I^B I^B$ or $I^B i$
AB	$I^A I^B$
O	ii



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The ABO System

Genotype	Antigen	Phenotype
$I^A I^A$ $I^A I^O$	A A	A
$I^B I^B$ $I^B I^O$	B B	B
$I^A I^B$	A and B	AB
$I^O I^O$	Neither	O

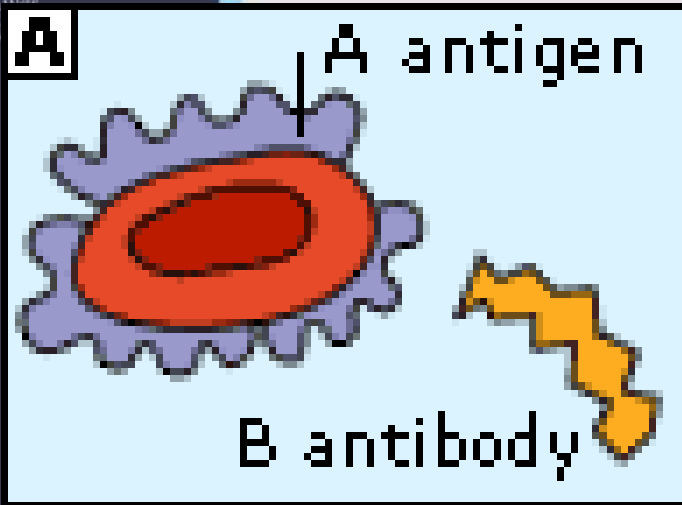
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Allele	Codes
I^A	Type A
I^O	Type O
I^B	Type B

ABO Blood Grouping System

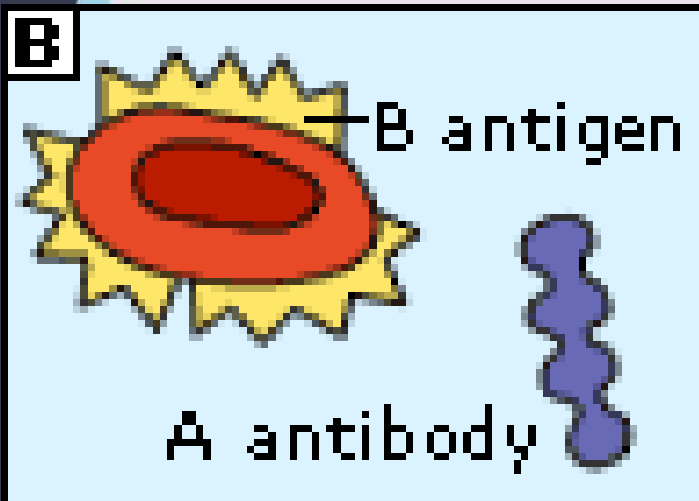


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Blood group A

If you belong to the blood group A, you have A antigens on the surface of your RBCs and B antibodies in your blood plasma.



Blood group B

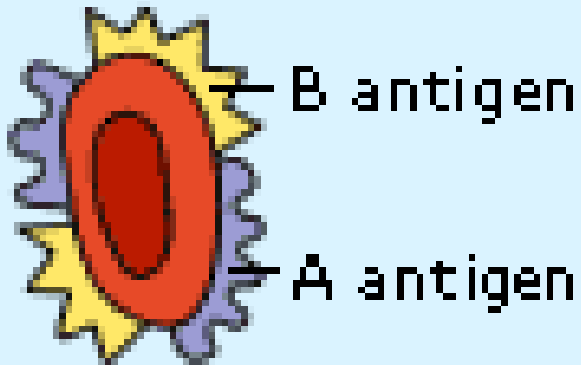
If you belong to the blood group B, you have B antigens on the surface of your RBCs and A antibodies in your blood plasma.



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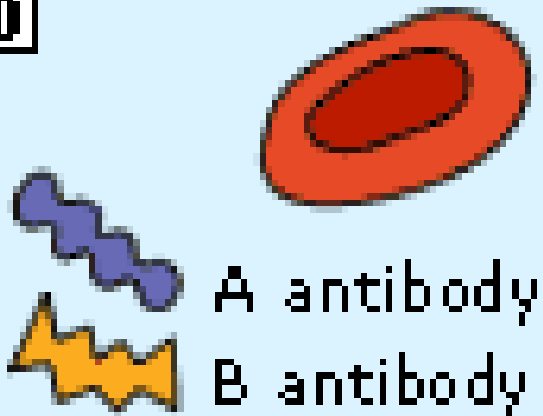
AB



Blood group AB

If you belong to the blood group AB, you have both A and B antigens on the surface of your RBCs and no A or B antibodies at all in your blood plasma.

O



Blood group O

If you belong to the blood group O (null), you have neither A or B antigens on the surface of your RBCs but you have both A and B antibodies in your blood plasma.



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Blood Transfusions

- It is important to carefully match the donor and recipient blood types.
- If the donor's blood cells have antigen that are different from those of the recipient, antibodies in the recipient's blood recognize the donor blood as foreign.
- This triggers an immune response resulting in blood clotting or **agglutination**.



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Donor

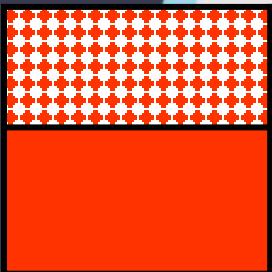
Donor-recipient compatibility

Recipient

Type	A	B	AB	O
A	Safe transfusion	Agglutination	Safe transfusion	Agglutination
B	Agglutination	Safe transfusion	Safe transfusion	Agglutination
AB	Agglutination	Agglutination	Safe transfusion	Agglutination
O	Safe transfusion	Safe transfusion	Safe transfusion	Safe transfusion

Note:

- **Type O blood** may be transfused into all the other types = the **universal donor**.
- **Type AB blood** can receive blood from all the other blood types = the **universal receivers**.



= Agglutination

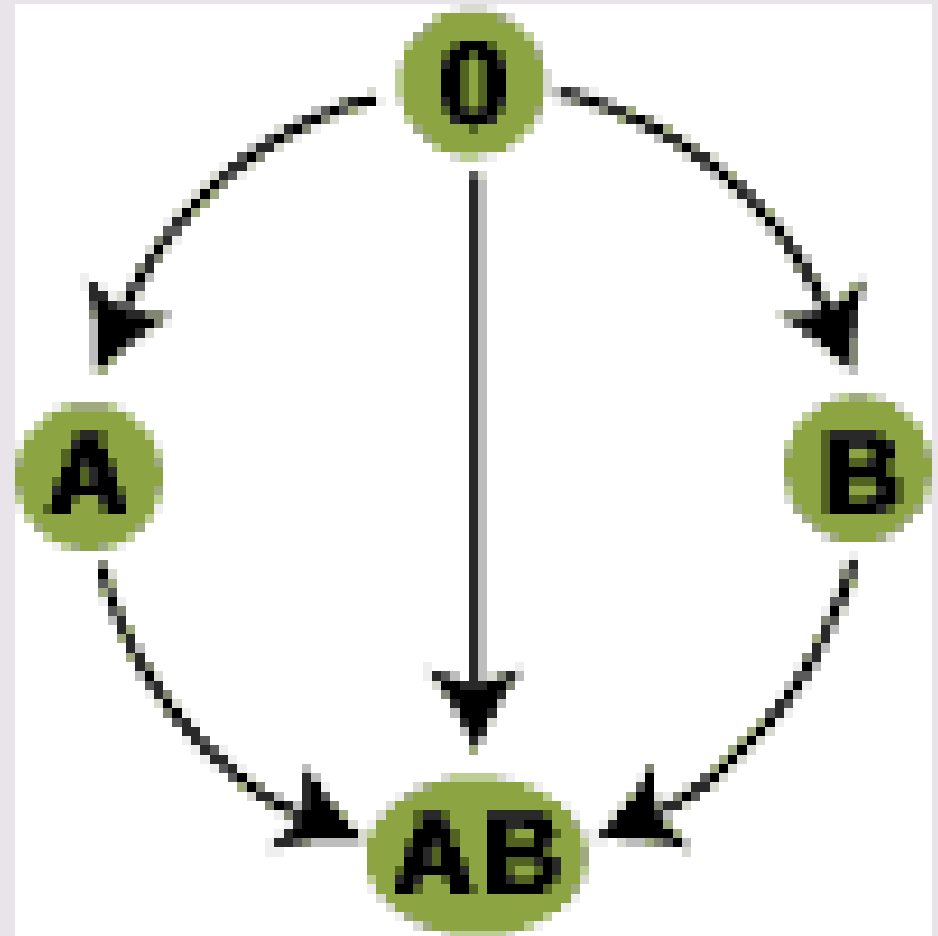
= Safe transfusion



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Donor-recipient compatibility

- Blood group O is called "**universal donor**" because it has no antigens on RBC.
- Blood group AB are called "**universal receivers**" because it has no antibodies in the plasma.












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Relative Abundance of Blood Types

<u>A</u>	<u>B</u>	<u>AB</u>	<u>O</u>
40-42%	10-12%	3-5%	43-45%



The ABO Blood System

Blood Type (genotype)	Type A (AA, AO)	Type B (BB, BO)	Type AB (AB)	Type O (OO)
Red Blood Cell Surface Proteins (phenotype)	 <p>A agglutinogens only</p>	 <p>B agglutinogens only</p>	 <p>A and B agglutinogens</p>	 <p>No agglutinogens</p>
Plasma Antibodies (phenotype)	 <p>b agglutinin only</p>	 <p>a agglutinin only</p>	<p>NONE.</p> <p>No agglutinin</p>	 <p>a and b agglutinin</p>



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Linkage and its estimation

- ❖ Sutton and Boveri - **chromosomal theory of inheritance.**
- ❖ Many genes are located in each chromosome in a linear fashion.
- ❖ It may therefore be expected that all genes located in same chromosome would move to same pole during cell division.
- ❖ As a consequence, such genes will fail to show independent segregation and would tend to be inherited together.



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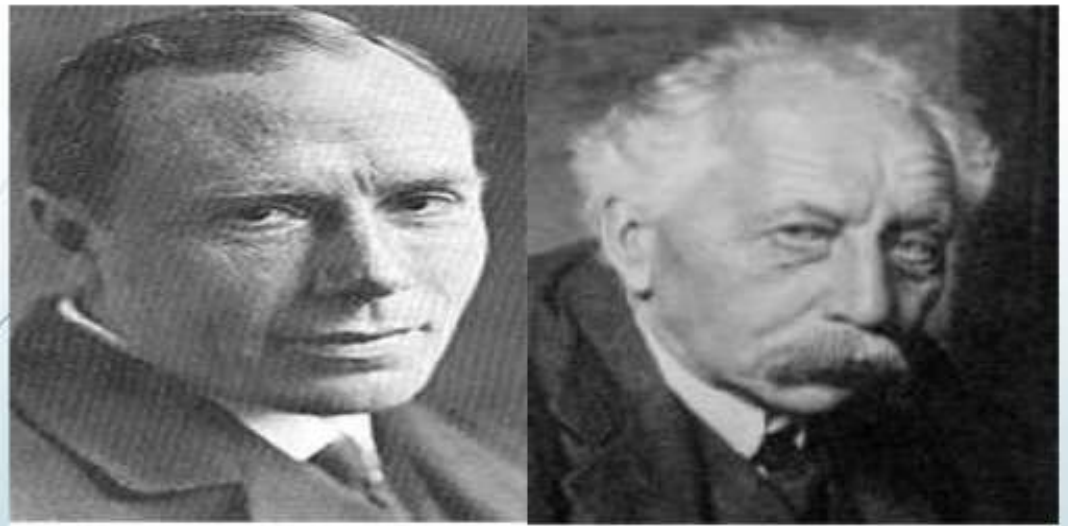
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- ❖ This tendency of genes to remain together in their original combination during inheritance is called **linkage**
- ❖ The phenomenon of linkage was first reported by **Bateson and Punnet** in 1906



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Discovery of Linkage



Reginald Crundall Punnett

William Bateson, 1922.

The phenomenon of linkage was first reported by Bateson and Punnett in 1906. They studied purple (P) vs. red (p) flowers, and a for long pollen grains (L) vs. round pollen grains (l) involving two varieties / races.

The character purple (P) is a simple monogenic dominant to red (p); while long (L) pollen is dominant to round (l) pollen, when these two plants were crossed the F_1 (PL/pl) was purple flowered with long pollen. But in F_2 , the ratio of four types of plants deviated from the normal dihybrid ratio of 9:3:3:1 expected on the principle of independent assortment of flower colour and pollen shape.

Bateson and Punnett also studied peas:

Flower Colour: P = purple p = red

Pollen seed shape: L = long l = round

True Breeding lines: PPLL x pp ll P

Pp Ll F1

F2 Phenotype	Actual Number	Exp Ratio
Purple long	284	9
Purple round	21	3
Red long	21	3
Red round	55	1

Crosses produced a deviation from the predicted Mendelian independent assortment ratio

What is going on????





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Test cross F1 to double recessive:

Parents $PpLl \times ppll$

Gametes $PL \quad pl$

Pl

pL

pl

Expect 1:1:1:1 ratio of phenotypes

Bateson and Punnett observed 7:1:1:7

Some gamete types more common than others...but why???

PARENTAL VERSES RECOMBINANTS



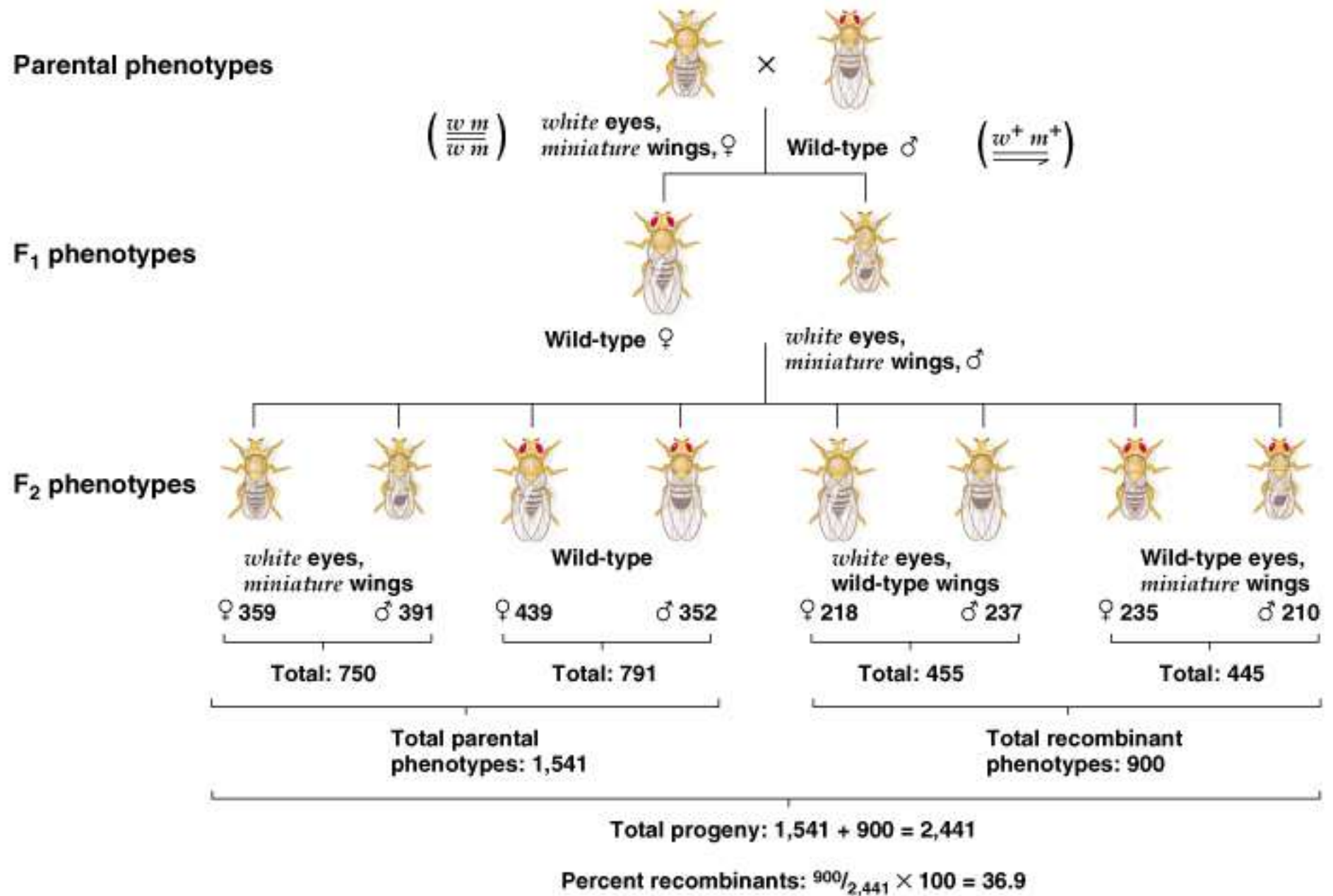
- ❖ The plants with the character combination as in the original parents are known as **parental forms or parental combinations**, i.e., (i) Plants with purple flowers and long pollen and (ii) Plants with red flowers and round pollen.
- ❖ Plants possessing one character from one parent and another character from the second parent are known as **recombined types or recombinants or new combinations or cross-overs**, i.e. (i) plants with red flowers and long pollen and (ii) plants with purple flowers and round pollen.



Conclusion of the experiment

- In F_2 , there are both parental forms and recombinants.
- The chief peculiarity of the results in the above table was that **parental forms are in far excess of the expected number while the recombinants were fewer.**
- the researchers hypothesized that there was a coupling, or connection, between the parental alleles for flower color and pollen grain shape
- This coupling resulted in the observed deviation from independent assortment.
- But why are certain alleles linked? Bateson and Punnett weren't sure.
- In fact, it was Thomas Hunt Morgan that first relate linkage to the segregation of homologous chromosomes and occurrence of crossing over during meiosis in *Drosophila*

Fig. 13.1 Morgan's experimental crosses of white-eye and miniature-wing variants of





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Types of Linkage:

Linkage is generally classified on the basis of three criteria viz., (i) Crossing over, (ii) Genes involved and (iii) Chromosomes involved

Based on crossing over:

Linkage may be classified into (a) complete and incomplete / partial depending up on absence or presence of recombinant phenotypes in test cross progeny.

Complete linkage: It is known in case of males of *Drosophila* and females of silkworms, where there is complete absence of recombinant types due to absence of crossing over.

Incomplete / partial linkage: If some frequency of crossing over also occurs between the linked genes, it is known as incomplete / partial linkage. Recombinant types are also observed besides parental combinations in the test cross progeny. Incomplete linkage has been observed in maize, pea, *Drosophila* female and several other organisms.



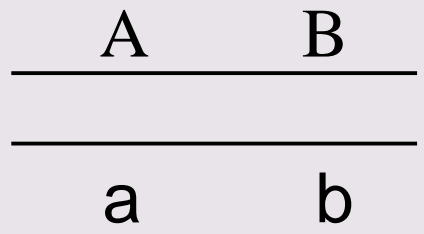
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(ii) Based on genes involved : Depending on whether all dominant or some dominant and some recessive alleles are linked together, linkage can be categorized into (a) Coupling phase and (b) Repulsion phase

• **Coupling phase/ Cis-configuration:** All dominant alleles are present on the same chromosome or all recessive alleles are present on same chromosome.



Coupling phase



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• **Repulsion phase:** Dominant alleles of some genes are linked with recessive alleles of other genes on same chromosome.

$$\begin{array}{c} A \quad b \\ \hline \\ \hline a \quad B \end{array}$$

Repulsion phase

(iii) Based on chromosomes involved: Based on the location of genes on the chromosomes, linkage can be categorized into (a) autosomal linkage and (b) X-chromosomal linkage / allosomal linkage / sex linkage

Autosomal linkage: It refers to linkage of those genes which are located in autosomes (other than sex chromosomes).

X-chromosomal linkage / allosomal linkage / sex linkage: It refers to linkage of genes which are located in sex chromosomes i.e. either 'X' or 'Y' (generally 'X')



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Characteristic features of Linkage:

1. Linkage involves two or more genes which are located in same chromosome in a linear fashion.
2. Linkage reduces variability.
3. Linkage may involve either dominant or recessive alleles (coupling phase) or some dominant and some recessive alleles (repulsion phase).
4. It may involve either all desirable traits or all undesirable traits or some desirable and some undesirable traits.
5. It is observed for oligo-genic traits as well as polygenic traits.
6. Linkage usually involves those genes which are located close to each other.
7. The strength of linkage depends on the distance between the linked genes. Lesser the distance, higher the strength and vice versa.



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8. Presence of linkage leads to higher frequency of parental types than recombinants in test cross. When two genes are linked the segregation ratio of dihybrid test cross progeny deviates significantly from 1:1:1:1 ratio.
9. Linkage can be determined from test cross progeny data.
10. If crossing over does not occur, all genes located on one chromosome are expected to be inherited together and form a **LINKAGE GROUP**. Thus the maximum number of linkage groups possible in an organism is equal to the haploid chromosome number.
Eg. Onion $2n = 16$; $n = 8$ maximum linkage groups possible = 8
Maize $2n = 20$; $n = 10$ maximum linkage groups possible = 10
11. Besides pleiotropy, linkage is an important cause of genetic correlation between various plant characters.
12. Linkage can be broken by repeated intermating of randomly selected plants in segregating population for several generations or by mutagenic treatment.



Significance of Linkage in Plant Breeding:

1. Linkage limits the variability among the individuals.
2. Linkage between two or more loci controlling different desirable characters is advantageous for a plant breeder. A linkage between genes controlling two different desirable characters will help in simultaneous improvement of both the characters.
3. Linkage is undesirable when desirable and undesirable genes are linked together.
4. The estimates of genetic variances for quantitative characters are greatly influenced by the presence of linkage



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