B10 201 (INTRODUCTORY GENETICS)

LECTURE NOTE

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MENDEL'S LAWS OF INHERITANCE

- Mendelian inheritance (or Mendelian genetics or Mendelism) is a set of primary tenets relating to the transmission of hereditary characteristics from parent organisms to their children; it underlies much of genetics. The tenets were initially derived from the work of Gregor Mendel published in 1865 and 1866, which was "re-discovered" in 1900.

Terminology Used in Genetics Experiment

- **Genotype**: This is the genetic make up of an individual.
- **Phenotype**: This is an organism's appearance or observable traits.
- **Homozygous**: A diploid with identical alleles at a locus.
- **LOCUS**: The site of a gene on a chromosome.

Terminology Used in Genetics Experiment

- **Parental generation**: The plants which are used as parents in a cross are called parental generation(P).
- **F1 generation** (The first filial generation): The offspring of two pure breeding parents.
- **F2 generation** (The second filial generation): The offspring resulting from selfing the F1 generation.
- **F3 generation** (The third filial generation): The offspring resulting from selfing the F2 generation.

Terminology Used in Genetics Experiment

- **Hybridisation:** This is the process of combining two complementary single-stranded DNA or RNA molecules and allowing them to form a single double-stranded molecule through base pairing.
- **Hybrids**: The offspring obtained from the crossing. The parents differ in at least one characteristic.
- **Selfing**: Pollination of an organism with itself, or with others like itself.

Terminology Used in Genetic Experiment

- **Pure-breeding or True breeding**: An organism which, when crossed with itself or others like itself, always produces offing like itself.
- **Dominant and Recessive**: The character which appears in F1 generation is called **dominant.**
- The alternative character that fails to show itself in the presence of dominant gene called **recessive.**
- **Monohybrid Cross**: The cross which involves a single pair of contrasting characters is called monohybrid cross.

Terminology Used in Genetic Experiment

- **Dihybrid Cross**: The cross which involves two pairs of contrasting characters is called dihybrid cross.
- **Polyhybrid Cross**: The cross which involves more than two pairs of contrasting characters is called polyhybrid cross.
- **Reciporcal Cross**: A pair of crosses between a male of one strain and a female of another, and vice versa is called reciprocal cross.
- **Varieties**: A group of individuals that differ distinctly from one another but can interbreed with other varieties of the same species is called variety.
- **Punnett Square (checker board)**: It is a square board with vertical and horizontal columns to study all possible results of various crosses.

Mendel's Experiment

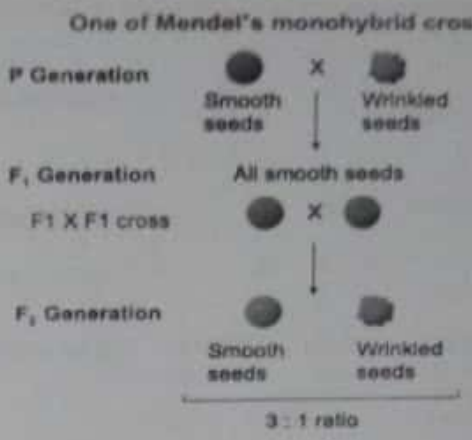
- **Gregor Mendel** spent his time crossing pea plants. As he did this over & over again, he noticed some patterns to the inheritance of traits from one set of pea plants to the next. By carefully analyzing his pea plant numbers, he discovered three laws of inheritance.
- Mendel's Laws are as follows:
- 1. The Law of Dominance
  2. The Law of Segregation
  3. The Law of Independent Assortment

Mendel's First Law

❑ LAW OF DOMINANCE:

- It states that when two pure plants with contrasting characters are crossed, only one form of the character appears in F1 generation, the other remains unexpressed. The character which appears itself in F1 generation is called **dominant** , the alternative factor that fails to show itself in F1 generation is called **recessive**.

Mendel's Experiment



Mendel's Experiment

❑ Mendel crossed a pea plant true breeding for smooth seeds with one true breeding for wrinkled seeds

❑ All the F_1 were smooth and not a blend of both parental phenotypes. When F_1 was allowed to self-fertilize to produce F_2, the wrinkled phenotype reappeared, along with smooth.

❑ When Mendel counted the F_2 progeny, he found a **3:1** ratio of smooth to wrinkled.

Mendel's Experiment

❑ How can a trait present in the P generation disappear in the F_1 and then reappear in the F_2?

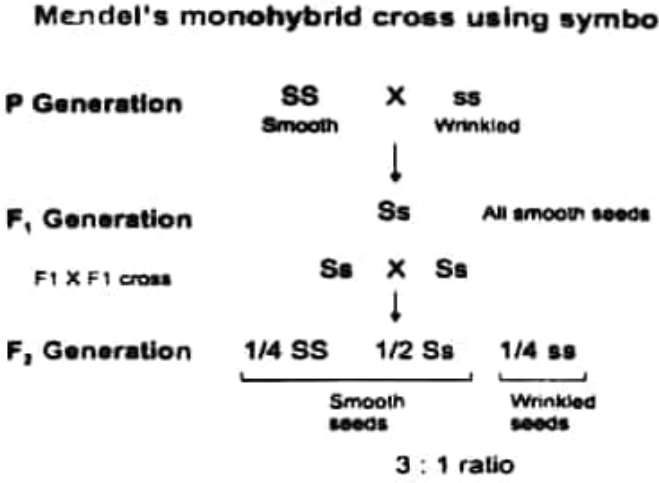
❑ Mendel reasoned that alternative traits are determined by **particulate factors** (i.e. genes) which are transmitted from parents to offspring through the gametes.

❑ Each factor was considered to exist in alternative forms (i.e. alleles), each of which specified one of the traits.

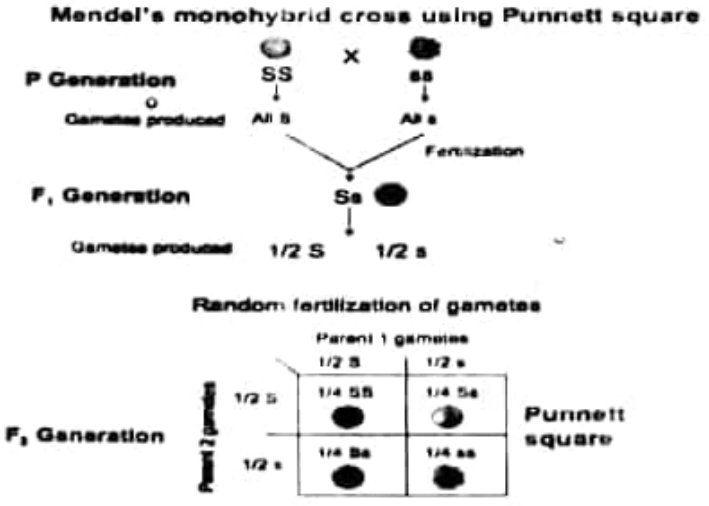
Mendel's Experiment

- Mendel also reasoned that a true-breeding strain of peas must contain a pair of identical factors.
- Since the F_2 exhibited both traits, the F_1 also contained both factors, one for each trait.
- The expression of the missing trait in the F_1 must have been masked by the visible trait. This is called **dominance** while he called the missing trait in F1 **recessive**.
  - Smooth trait (allele) is said to be **dominant**.
  - Wrinkled trait (allele) is said to be **recessive**.

Mendel's monohybrid cross using symbol



Mendel's monohybrid cross using Punett square



Mendel's Experiment

- Note that in F_2 three types of genotypes are produced in 1:2:1 ratio
- But because S is dominant, heterozygotes have smooth phenotype
- So phenotypic ratio is **3:1** smooth to wrinkled.

Mendel's Second Law

- **The Law of Segregation**

It states that during the formation of gametes (eggs or sperm), the two alleles responsible for a trait separate from each other. Alleles for a trait are then "recombined" at fertilization, producing the genotype for the traits of the offspring.

Mendel's Experiment

- Mendel analyzed the behaviour of six other pairs of traits. All gave the same results:
  - results of reciprocal crosses were always the same.
  - all F_1 progeny resembled one of the parents, indicating dominance of one allele over the other.
  - trait that disappeared in F_1, reappeared in F_2, and there was always a 3:1 phenotypic ratio in F_2.

Mendel's Experiment

- Note that segregation is due to the fact that genes occupy a specific location on a chromosome (**locus**),
- and that during meiosis homologous chromosomes separate from one another to produce haploid cells.

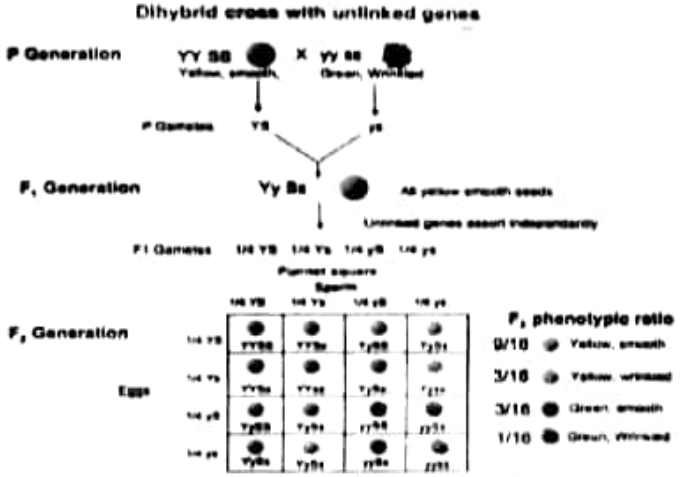
Mendel's Third Law

- **The Law of Independent Assortment**
  - It states that genes located on different chromosomes assort independently of one another.
  - Or Alleles for different traits are distributed to sex cells (& offspring) independently of one another.

Mendel's Experiment

- Mendel noticed during all his work that the height of the plant and the shape of the seeds and the colour of the pods had no impact on one another.
- In other words, being tall didn't automatically mean the plants had to have green pods, nor did green pods have to be filled only with wrinkled seeds, the *different* traits seem to be inherited INDEPENDENTLY.
- It involves what's known as a "dihybrid cross", meaning that the parents are hybrid for two *different* traits.

Mendel's Third Law



Mendel's Dihybrid Cross

- According to the rules of probability, if characters are inherited independently in a dihybrid cross, then the F_2 from an $F_1 \times F_1$ cross will always yield a **9:3:3:1** phenotypic ratio (assuming a simple dominant/recessive relationship between alleles).

MULTIPLE CHOICE QUESTIONS

1. Which of the following is a possible abbreviation for a genotype?

   A. 3C B. Pp C. Ty D. fg

2. What is the best way to determine the phenotype of the feathers on a bird?

   A. analyze the bird's DNA (genes) B. look at the bird's feathers

   C. look at the bird's beak D. examine the bird's droppings

3. Which of the following pairs is not correct?

   A. kk = hybrid B. hybrid = heterozygous

   C. heterozygous = Hh D. homozygous = RR

4. The genes present in an organism represent the organism's _____.

   A. genotype B. phenotype C. physical traits

MULTIPLE CHOICE QUESTIONS

5. Which choice represents a possible pair of alleles?

A. k & t B. K & T C. K & k D. K & t

6. How many alleles for one trait are normally found in the genotype of an organism?

A. 1 B. 2 C. 3 D. 4

7. Which statement is not true?

A. genotype determines phenotype
B. phenotype determines genotype
C. a phenotype is the physical appearance of a trait in an organism
D. alleles are different forms of the same gene

MULTIPLE CHOICE QUESTIONS

8. Which cross would best illustrate Mendel's Law of Segregation?

A. TT x tt B. Hh x hh C. Bb x Bb D. rr x rr

9. In the cross Yy x Yy, what percent of offspring would have the same phenotype as the parents?

A. 25% B. 50% C. 75% D. 100%

10. In a certain plant, purple flowers are dominant to red flowers. If the cross of two purple-flowered plants produces some some purple-flowered and some red-flowered plants, what is the genotype of the parent plants?

A. PP x Pp B. Pp x Pp C. pp x PP D. pp x pp

THEORY

1. In a pea plant that breeds true for tall, what possible gametes can be produced? Use the symbol D for tall, d for dwarf.

(b) In a pea plant that breeds true for dwarf, what possible gametes will be produced?

(c) What will be the genotype of F1 offspring from a cross between these two types?

(d) Assuming that the allele for tall is dominant, what will be the phenotype of F1 offspring from a cross between these two types?

(e) What will be the probable distribution of traits in the F2 generation? (Illustrate with a Punnett square).

THEORY

2. The ability to taste a bitter chemical, phenylthiocarbamide (PTC), is due to a dominant gene. Use T and t to symbolize the two alleles of this gene.

(a) What is the genotype of a nontaster? What are the possible genotypes of a taster?

(b) Could a person with two tasters as parents be a non-taster? How?

THEORY

3. Albinism, the total lack of pigment, is due to a recessive gene. A man and woman plan to marry and wish to know the probability of their having any albino children. What are the probabilities if: a. both are normally pigmentcd, but each has one albino parent.

b. the man is an albino, tbe woman is normal, but her father is an albino.

THEORY

4. A woman has a rare abnormality of the eyelids called ptosis, which prevents her from opening her eyes completely. This condition is caused by a dominant allele, *P*. The woman's father has ptosis, but her mother had normal eyelids. Her father's mother had normal eyelids.

(a) What are the genotypes of the woman, her father, and her mother?

(b) What proportion of the woman's children will have ptosis if she mates with a man with normal eyelids?

THEORY

5. In pigeons, a dominant allele *C* causes a checkered pattern in the feathers; its recessive allele *c* produces a plain pattern. Feather coloration is controlled by an independently assorting gene; the dominant allele *B* produces red feathers, and the recessive allele *b* produces brown feathers. Birds from a true-breeding checkered red variety are crossed to birds from true-breeding plain, brown variety.

(a) Predict the phenotype of their progeny.

(b) If these progeny are inter crossed, what phenotypes will appear in the F2, and in what proportions?

THE CHROMOSOMAL BASIS OF HEREDITY

- The chromosome theory of inheritance describes how the transmission of chromosomes account for the Mendelian patterns of inheritance.
- This theory was independently proposed in 1902-03 by Theodore Boveri, a German and Walter Sutton, an American

THE CHROMOSOMAL BASIS OF HEREDITY

- The chromosome theory of inheritance is based on a few fundamental principles:
- Chromosomes contain the genetic material
- Chromosomes are replicated and passed along from parent to offspring
- The nuclei of most eukaryotic cells contain chromosomes that are found in homologous pairs

THE CHROMOSOMAL BASIS OF HEREDITY

- During the formation of gametes, different types of (non homologous) chromosomes segregate independently.
- Each parent contributes one set of chromosomes to its Offspring. The sets are functionally equivalent. Each carries a full complement of genes

THE CHROMOSOMAL BASIS OF HEREDITY

- The chromosome theory of inheritance allows us to see the relationship between Mendel's laws and chromosome transmission
  - Mendel's law of segregation can be explained by the
    - homologous pairing and segregation of chromosomes during meiosis

THE CHROMOSOMAL BASIS OF HEREDITY

- Mendel's law of independent assortment can be explained by
- the relative behaviour of different (non homologous chromosomes) during meiosis

THE CHROMOSOMAL BASIS OF HEREDITY

CHROMOSOMAL THEORY OF INHERITANCE

The salient features of **chromosomal theory** (Sutton and Boveri) of inheritance are as follows

- Bridge between one generation and the next is through sperm and ovum. The two must carry all the hereditary characters
- Both the sperm and egg contribute equally in the heredity of the offspring.
- Nucleus contains chromosomes. Therefore, chromosomes must carry the hereditary traits
- Every chromosome or chromosome pair has a definite role in the development of an individual. Loss of a complete or part of the chromosome produces structural and functional deficiency in the organism.
- Like the hereditary traits the chromosomes retain their number, structure and individuality throughout the life of an organism and from generation to generation.
- Both chromosomes as well as genes occur in pairs in the somatic or diploid cells
- A gamete contains only one chromosomes of a type and only one of the two alleles of a trait
- The paired condition of both chromosomes as well as Mendelian factors is restored during fertilization
- Genetic homogeneity and heterogeneity, dominance and recessiveness can be suggested by chromosomal type and behaviour.
- Homologous chromosomes synapse during meiosis and then separate or segregate independently into different cells which establishes the quantitative basis for segregation and independent assortment of hereditary factors

CHROMOSOME STRUCTURE

- Eukaryotic **chromosome** contains a single DNA molecule of enormous length in a highly coiled-stable complexes of DNA and protein called **chromatin**
- The basic structural unit of chromatin is the **nucleosome**, a core particle of • histone proteins that the DNA wraps around in ~200bp segments

CHROMOSOME STRUCTURE

- The **centromere** is a specific region of the eukaryotic chromosome where the kinetochore (the complex of DNA and proteins to which the spindle fibers) attach and pull the chromosomes during both mitosis and meiosis
- The chromosome complement = the complete set of chromosomes of plants and animals

CHROMOSOME STRUCTURE

- Structurally, a chromosome is differentiated into the following parts:
- **Pellicle**: This is the outer thin covering or sheath of the chromosome.
- **Matrix** or **ground substance** of the chromosome is made up of protein, small quantities of RNA and lipid. It has one or two chromonemata depending upon the state of chromosome.
- **Chromonemata** (Sing. Chromonema) are coiled threads which forms the bulk of chromosomes.

CHROMOSOME STRUCTURE

- **Primary Constriction**: This is a narrow non stainable area where the two chromatids are attached in the prophase. The primary constriction is also called **centromere**.
- The surface of centromere bears a special trilaminar plate called **kinetochore.**
- **Secondary Constrictions** are narrow areas other than the primary constriction.

CHROMOSOME STRUCTURE

- **Secondary Constrictions** are of two types. One type are produced by breaking and subsequent fusion of chromosome segments. The other type are metabolically active and function as **nucleolar organisers.**
- In human beings, 5 chromosomes (13,14,15,21 and 22) have nucleolar organiser regions. The chromosomes having nucleolar organiser regions also possess satellites and are called **SAT chromosomes.**

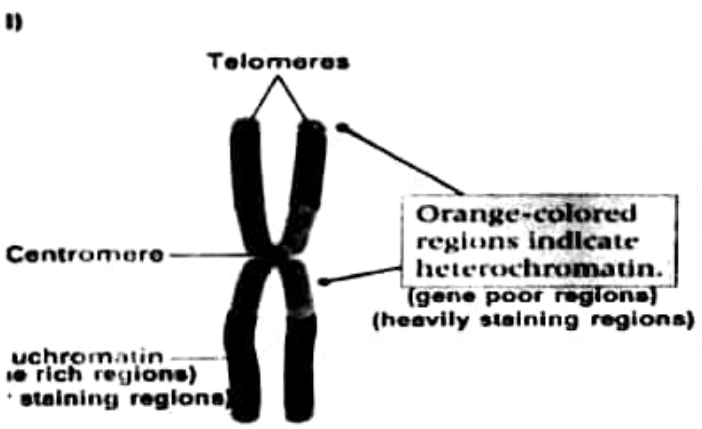
CHROMOSOME STRUCTURE

- Satellite: The area of a chromosome distal to a nucleolar organiser is called satellite or trabant.
- The chromosome bearing a satellite is known as **sat chromosome**.
- The word "sat" is not derived from satellite but from poor staining ability of the nucleolar organiser region as its DNA content is low.

CHROMOSOME STRUCTURE

- **Telomere**: The terminal ends of chromosome are referred to as telomeres. A telomere is a special area of the chromosome having moderately repetitive DNA.
- It allows chromosome to get attached to the nuclear envelope but not to any other chromosome.
- Even when a chromosome breaks, the separated segment fuses in the region other than telomere.

CHROMOSOME STRUCTURE



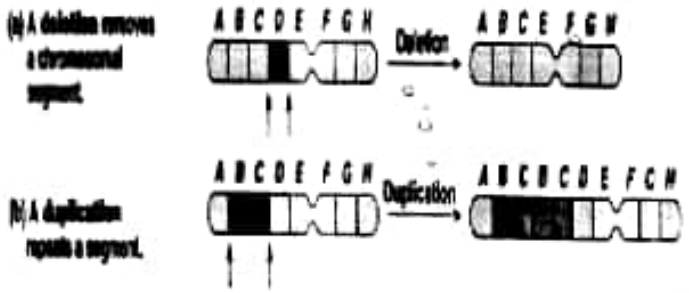
FUNCTIONS OF CHROMOSOMES

1. Chromosomes contain genes. All the hereditary information is located in the genes.
2. Chromosomes controls the synthesis of structural proteins and thus helps in cell division and cell growth.
3. Sat chromosomes produce nucleoli for synthesis of ribosomes.
4. Some chromosomes called sex chromosome (X and Y) determine the sex of an individual.

Errors and Exceptions in Chromosomal Inheritance

- Breakage of a chromosome can lead to four types of changes in chromosome structure.
- A **deletion** occurs when a chromosome fragment lacking a centromere is lost during cell division.
  - This chromosome will be missing certain genes.
- A **duplication** occurs when a fragment becomes attached as an extra segment to a sister chromatid.

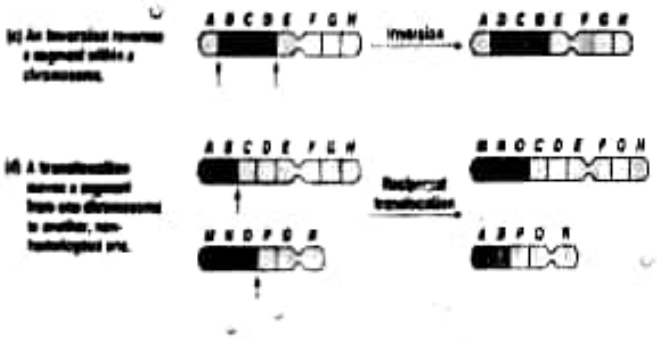
Errors and Exceptions in Chromosomal Inheritance



Errors and Exceptions in Chromosomal Inheritance

- **An inversion** occurs when a chromosomal fragment reattaches to the original chromosome but in the reverse orientation.
- **In translocation**, a chromosomal fragment joins a non homologous chromosome.
  - Some translocations are reciprocal, others are not

Errors and Exceptions in Chromosomal Inheritance



Errors and Exceptions in Chromosomal Inheritance

- Deletions and duplications are common in meiosis.
- Homologous chromatids may break and rejoin at incorrect places, such that one chromatid will lose more genes than it receives.

Errors and Exceptions in Chromosomal Inheritance

- A diploid embryo that is homozygous for a large deletion or male with a large deletion to its single X chromosome is usually missing many essential genes and this leads to a lethal outcome.
  - Duplications and translocations are typically harmful.
- Reciprocal translocation or inversion can alter phenotype because a gene expression is influenced by its location.

GENETIC INTERACTION

The condition where one pair of gene reverses or inhibits the effect of another pair of genes by causing the modification of the normal phenotype is called genetic interaction.

- It is also a condition where a single character is governed by two or more genes and every gene affects the expression of other genes involved.

Types of genetic interaction

- Gene interaction is of two types:
  - 1. Allelic or intragenic or non-epistatic gene interaction
  - 2. Non-allelic or intergenic or epistatic gene interactions

Allelic or intragenic or non-epistatic gene interaction

- This is when one allele affects the expression of another allele in the same gene locus. Examples include : complete dominance, incomplete dominance and co-dominance.

Non-allelic or intergenic or epistatic gene interactions

- These interactions occur between alleles of different genes present either on the same or different chromosome and alter the normal phenotype. Complementary gene interaction, supplementary gene interaction, duplicate factors and inhibitory factors are examples of intergenic interactions.

Epistatic gene

- When a gene or locus suppress or mask the phenotypic expression of another gene at another locus, such gene is known as **epistatic gene**.

Hypostatic gene

- The gene or locus which is suppressed by an epistatic gene is called **hypostatic gene.**

Allelic interaction: Complete Dominance

- This is a form of dominance in heterozygous condition wherein the allele that is regarded as dominant completely masks the effect of the allele that is recessive. For instance, an individual carrying two alleles that are both dominant (e.g. AA), the trait that they represent will be expressed.

complete dominance contd.

- But if the individual carries two alleles in a manner that one is dominant and the other one is recessive, (e.g. Aa), the dominant allele will be expressed while the recessive allele will be suppressed.
- Hence, the heterozygote (Aa) will have the same phenotype as that of the dominant homozygote (AA). This condition is called **complete dominance**.

Complete Dominance Contd.

- In complete dominance, the effect of one allele in a heterozygous genotype completely masks the effect of the other.
- The allele that masks the other is said to be **dominant** to the latter, and the allele that is masked is said to be **recessive** to the former.

Complete Dominance Contd.

- Complete dominance therefore means that the phenotype of the heterozygote is indistinguishable from that of the dominant homozygote.
- A classic example of dominance is the inheritance of seed shape (pea shape) in peas. Peas may be round (associated with allele *R*) or wrinkled (associated with allele *r*).

Complete Dominance Contd.

- In this case, three combinations of alleles (genotypes) are possible: *RR* and *rr* are homozygous and *Rr* is heterozygous. The *RR* individuals have round peas and the *rr* individuals have wrinkled peas.
- In *Rr* individuals the *R* allele masks the presence of the *r* allele, so these individuals also have round peas. Thus, allele *R* is dominant to allele *r*, and allele *r* is recessive to allele *R*.

Allelic interaction: Incomplete dominance

- A heterozygous organism carrying two alleles wherein one is dominant and the other one is recessive, (e.g. Aa), the dominant allele will only be partially expressed. Hence, the heterozygote (Aa) will have an intermediate phenotype. This is also refer to as **partial dominance**

Incomplete Dominance Contd.

- A typical example is the color of the flower in which *R* symbolizes the dominant allele for red pigment and *r* is the recessive allele for no pigment.
- In incomplete dominance, the heterozygous plant carrying both alleles, Rr, will not be able to produce enough red pigment (since the dominant allele is only partially expressed) and therefore will appear pink.

Allelic interaction: Codominance

- This is a form of dominance wherein the alleles of a gene pair in a heterozygote are fully expressed. This results in offspring with a phenotype that is neither dominant nor recessive.

Codominance Contd.

- A typical example showing codominance is the ABO blood group system. For instance, a person having A allele and B allele will have a blood type AB because both the A and B alleles are codominant with each other.

Codominance Contd.

- Codominance is different from incomplete dominance in a way that the former has both alleles manifesting the phenotypes whereas the latter produces an intermediate phenotype.

Codominance Contd.

Genotype	Phenotype
I^AI^A or I^Ai^O	A
I^BI^B or I^Bi^O	B
I^AI^B	AB
i^Oi^O	O

Codominance Contd.

- I^A and I^B are codominant, I^O is recessive to both I^A and I^B. If you inherit I^A from your father and I^B from your mother, you will be AB blood group.
- To be blood group O, both parents must have at least I^O alleles.

Codominance Contd.

Mother genotype \ Father genotype	I^A	I^O
I^B	I^AI^B	I^BI^O
I^O	I^AI^O	I^OI^O

Codominance Contd.

EXAMPLES

1. A man with type AB blood marries a woman with type B blood. Her mother has type O blood. List the expected phenotype and genotype frequencies of their children.

Solution

	A	B
B	AB	BB
O	AO	BO

Phenotype frequencies : ¼ AB, ½ B and ¼ A
Genotypes frequencies : ¼ AB, ¼ BB, ¼ BO and ¼ AO.

. Non-allelic or intergenic or epistatic gene interactions

- **Epistasis**
  - There are two pairs of independent non-allelic genes affecting a single trait.
  - The suppression of the gene on one locus of a chromosome by the gene present at some other locus is called **epistasis** meaning "standing over". The gene which is suppressed is called **hypostatic** and the other is the epistatic or inhibiting gene which is also called the **suppressing gene.**

Epistatic gene interactions contd.

- Epistasis can be of the following types.
  - Due to **recessive gene** : Recessive gene a masks the effect of dominant gene **B.**
  - Due to **dominant gene** : Dominant gene **A** masks the effect of the dominant gene **B.** Apart from this, the term epistasis refers to all non-allelic interactions involving a pair of genes.

Epistatic gene interactions contd.

- Therefore epistasis may be responsible for the production of several modified dihybrid ratios as follows:
  - Duplicate recessive epistasis (9:7)
  - Dominant epistasis (12:3:1)
  - Recessive epistasis (9:3:4)
  - Dominant recessive epistasis (13:3)
  - Duplicate dominant epistasis (15:1)

Duplicate Recessive Epistasis (9:7)

- This type of inheritance is also called **complementary gene interaction** observed in *Lathyrus odoratus*(Sweet pea) by Bateson and Punnett. Inheritance of flower colour was studied.

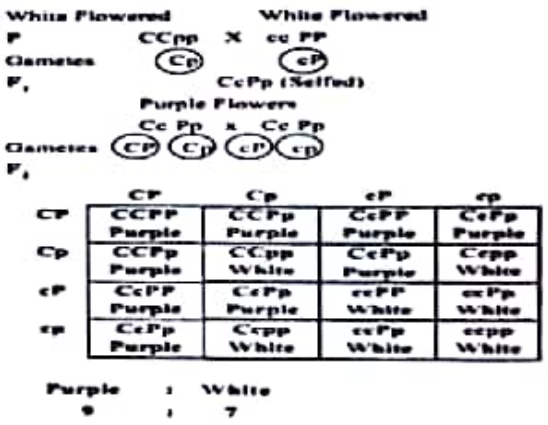
Duplicate Recessive Epistasis (9:7)

- When two pure breeding white flowered varieties of sweet pea where crossed, the F_1 hybrids were all purple flowered plants. When the F_1 hybrids were selfed, purple and white flowered varieties were produced respectively in the ratio of 9:7.

Duplicate Recessive Epistasis (9:7)

- Here two dominant genes C and P interact to produce purple colour. When any one of the genes is present in recessive condition, colour is not produced. Thus both the genes in the recessive state inhibit the formation of purple colour and so this has been referred to as Duplicate recessive epistasis.

Duplicate Recessive Epistasis (9:7)



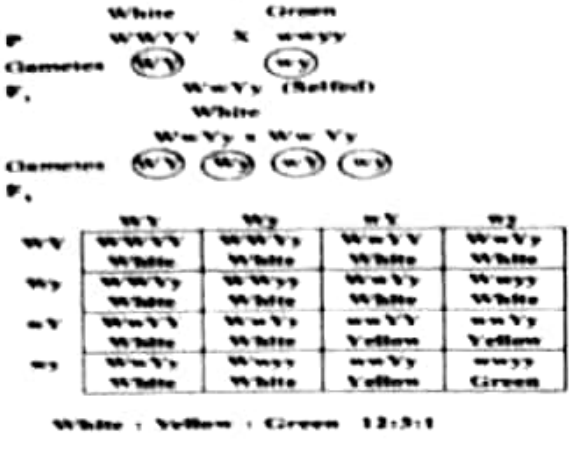
Dominant Epistasis - 12:3:1

- In *Cucurbita pepo* there are three common fruit colours white, yellow and green. White colour is produced due to the presence of dominant gene W. In the absence of W, the dominant gene Y produces yellow fruit colour and the double recessive is green. The effect of dominant gene `Y' is masked by dominant gene `W' which is the epistatic gene so this is called **dominant epistasis.**
-

Dominant Epistasis - 12:3:1

- When pure breeding white fruited variety is crossed with the double recessive green variety, the F1 hybrids are all white. When the hybrids are selfed, white, yellow and green fruited plants arise respectively in the ratio of 12:3:1

Dominant Epistasis - 12:3:1



Recessive epistasis - 9:3:4

- In *Sorghum* the dominant gene (P) is responsible for purple colour which is dominant over brown (q).
- When both the dominant genes (P and Q) are brought together either in homozygous or heterozygous condition, the purple colour is changed to red.

Recessive epistasis - 9:3:4

- A cross between purple (PPqq) and brown (ppQQ) results in plants with red colour in F_1 and when the F_1 heterozygotes are selfed, three kinds of phenotypic classes are produced in the ratio of 9:3:4 (9 Red, 3 Purple and 4 Brown).
- Thus in this example, the gene `p' is epistatic to the other colour genes.

Recessive epistasis - 9:3:4

- If the *Sorghum* is pp, it is brown inspite of other genotypes. The expression of the colour genes is masked if pp is present.
- The genes for recessive epistasis are also called **supplementary genes** because the gene P determines the formation of colour. The alleles of the other gene Q and q specify the colour.

Recessive epistasis - 9:3:4

Duplicate Recessive Epistasis (9:7)

- This type of inheritance is also called **complementary gene interaction** observed in *Lathyrus odoratus*(Sweet pea) by Bateson and Punnett. Inheritance of flower colour was studied.

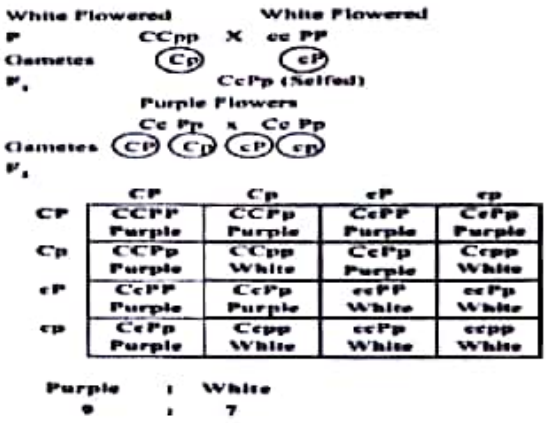
Duplicate Recessive Epistasis (9:7)

- When two pure breeding white flowered varieties of sweet pea where crossed, the F_1 hybrids were all purple flowered plants. When the F_1 hybrids were selfed, purple and white flowered varieties were produced respectively in the ratio of 9:7.

Duplicate Recessive Epistasis (9:7)

- Here two dominant genes C and P interact to produce purple colour. When any one of the genes is present in recessive condition, colour is not produced. Thus both the genes in the recessive state inhibit the formation of purple colour and so this has been referred to as Duplicate recessive epistasis.

Duplicate Recessive Epistasis (9:7)



Dominant Epistasis - 12:3:1

- In *Cucurbita pepo* there are three common fruit colours white, yellow and green. White colour is produced due to the presence of dominant gene W. In the absence of W, the dominant gene Y produces yellow fruit colour and the double recessive is green. The effect of dominant gene `Y' is masked by dominant gene `W' which is the epistatic gene so this is called **dominant epistasis.**
-

Dominant Epistasis - 12:3:1

- When pure breeding white fruited variety is crossed with the double recessive green variety, the F1 hybrids are all white. When the hybrids are selfed, white, yellow and green fruited plants arise respectively in the ratio of 12:3:1

Recessive epistasis - 9:3:4

P ppQQ x PPqq

Purple Brown

Gametes (pQ) (Pq)

F_1 PpQq(Selfed)

Red

Pp Qq x Pp Qq

Gametes (PQ) (Pq) (pQ) (pq)

F_2

	PQ	Pq	pQ	pq
PQ	PPQQ Red	PPQq Red	PpQQ Red	PpQq Red
Pq	PPQq Red	Ppqq Purple	PpQq Red	Ppqq Purple
pQ	PpQQ Red	PpQq Red	ppQq Brown	ppQq Brown
pq	PpQq Red	Ppqq Purple	ppQq Brown	ppqq Brown

Red: Purple :Brown

9 : 3 : 4

Table : 4.2. Differences between Epistasis and Dominance

Epistasis	Dominance
i. This type of gene interaction involves two non-allelic pairs of genes.	Only one pair of genes is involved, therefore there is no interaction.
ii. One pair of genes masks the effect of another pair of genes	An allele masks the effect of another allele of the same gene pair
iii. Expression of both the dominant and recessive alleles may be suppressed by the epistatic gene	Expression of a recessive allele is masked by the dominant allele
iv. Number of phenotypes in the F_2 generation are reduced	There is no reduction in the number of phenotypes of F_2 generation

LINKAGE AND GENETIC RECOMBINATION

- Linkage is the phenomenon of certain genes staying together during inheritance through generations without any change or separation due to their being present on the same chromosome

LINKAGE AND GENETIC RECOMBINATION

- It is also the tendency of parental combinations to remain together, which is expressed in terms of low frequency of recombinations.

LINKAGE AND GENETIC RECOMBINATION

- Linked genes occur on the same chromosome The strength of linkage between genes increases with the decrease in the distance between them.
- According to T.H. Morgan,

$$Linkage \propto \frac{1}{Distance\ between\ genes}$$

LINKAGE AND GENETIC RECOMBINATION

- The strength of linkage between two genes is inversely proportional to the distance between them.
- i.e Two linked genes show higher frequency of crossing over if the distance between them is higher and lower frequency if the distance between them is small.

LINKED GENES

- These genes are placed very closely on the chromosome and do not show independent assortment at the time of gamete formation.
- They show monohybrid ratio of 3:1. Genes that are located very close together on the same chromosome may show complete linkage.

LINKED GENES

- They may be so close to each other that they cannot be separated by recombination during meiosis.
- Genes located far apart on the same chromosome typically show incomplete (partial) linkage because they are easily separated by recombination.

UNLINKED GENES

- These are genes located distantly and are found on different chromosomes. Genes located on different chromosomes are not linked.
- This allows independent assortment – in a di-hybrid cross, the traits show the classic 9:3:3:1 inheritance pattern.

TYPES OF LINKAGE

- **Complete linkage:** This is the phenomenon in which parental combinations of characters appear together for two or more generations in a continuous and regular fashion.
- The genes located on the same chromosome do not separate and are inherited together over the generations due to absence of crossing over.

TYPES OF LINKAGE

- Complete linkage allows the combination of parental traits to be inherited, as such it is rare but has been reported in male drosophila and some other heterogametic individuals.

TYPES OF LINKAGE

- **Incomplete /partial linkage:** Genes present far apart in the same /other chromosomes have a tendency to separate due to crossing over and hence produce recombinant progeny besides the parental type.
- For example, incomplete or partial linkage has been reported in female Drosophila and various other organisms such as tomato, maize, pea, mice, chicken and human being.

CHROMOSOME THEORY OF LINKAGE

- Morgan and Castle formulated the chromosome theory of linkage which is as follows:
  - The genes which show the phenomenon of linkage are situated on the same chromosome and these linked genes usually remain bounded by the chromosomal material so that they cannot be separated during the process of inheritance.

CHROMOSOME THEORY OF LINKAGE

- The distance between the linked genes determines the strength of linkage. The closely located genes show strong linkage than the widely located genes which show weak linkage.
- The genes are arranged in linear fashion on the chromosomes.

LINKAGE GROUPS

- All the genes linked together on a single chromosome (which do not show independent assortment), constitute a linkage group.
- The number of linkage groups of a species is equal to the haploid chromosome number of that species.

Examples

- Drosophila has 4 pairs of chromosomes and 4 linkage groups.
- A human being has 23 pairs of chromosomes and 23 linkage groups.
- Corn(Zea mays) has 10 pairs of chromosomes and 10 linkage groups.

Examples

- However, in organism the female or male sex having dissimilar sex chromosomes (e.g., human beings, Drosophila, fowl, etc.), one more linkage group occurs than the haploid number. For example,

➢ Female human beings = 22 pairs of autosomes + 1 pair of homomorphic X chromosomes

= 22 autosomal linkage groups + 1 X chromosomal linkage group

= **23 linkage groups.**

Examples

- Male human beings = 22 pairs of autosomes + 2 heteromorphic sex chromosomes, i.e., 1 X chromosome + 1 Y chromosome

= 22 autosomal linkage group + 1 X chromosomal linkage group + 1 Y chromosomal linkage group = **24 linkage groups.**

SIGNIFICANCE OF LINKAGE

- The possibility of variations (variability) in gametes is reduced by linkage (unless crossing over occurs).
- It provides a strong proof in favour of linear arrangement of genes on the chromosomes.

DYAD AND TETRAD

- The familiar pattern of a two-chromatid chromosomes is called a **dyad**.
- When two homologous pairs are aligned (side by side), we call the pair a **tetrad**.
- Therefore, a tetrad is composed of two chromosomes- one maternal (M) and one paternal (P).

Dyad and Tetrad

- A tetrad will have a two centromeres and four chromatids (because it is made from two chromosomes).
- A dyad is a single (X-shaped) chromosome, so tetrad is composed of two dyads.

A TETRAD

A Tetrad

CROSSING OVER

- This is the random exchange of genetic materials between two non-sister chromatids of homologous chromosomes.
- It results to recombination of genetic material and prevalence of recombination is dependent on the distance between linked genes.

CROSSING OVER

- Crossing over occurs in prophase I of meiosis in a process called **synapsis**, where homologous chromosomes break at identical locations and rejoin with each other.
- Genes that are far from each other on a chromosome are more likely to be separated by crossing over than genes that are close to each other.

CHARACTERISTICS OF CROSSING OVER

- Crossing over or recombination occurs at two levels : at gross chromosomal level called **chromosomal crossing over** and at DNA level called **genetic recombination.**
- A reciprocal exchange of material between homologous chromosomes in heterozygotes is reflected in crossing over.

CHARACTERISTICS OF CROSSING OVER

- The crossing over results basically from an exchange of genetic material between non-sister chromatids by break and-exchange following replication.
- The frequency of crossing over appears to be closely related to physical distance between genes on chromosome and serves as a tool in constructing genetic maps of chromosomes.

TYPES OF CROSSING OVER

- According to its occurrence in the somatic or germ cells, the following two types of crossing over have been recognised:
- **Somatic or Mitotic Crossing Over**: When the process of crossing over occurs in the chromosomes of body or somatic cells of an organism during the mitotic cell division, it is known as somatic or mitotic crossing over.
- The somatic or mitotic crossing over is rare in its occurrence and it has no genetical significance. It has been reported in the body or somatic cells of *Drosophila* and in the fungus *Aspergillus nidulans*.

TYPES OF CROSSING OVER

- **Germinal or Meiotic Crossing Over:** Usually, the crossing over occurs in germinal cells during the gametogenesis in which the meiotic cell division takes place.
- This type of crossing over is known as **germinal or meiotic crossing over**. The meiotic crossing over is universal in its occurrence and is of great genetic significance.

KINDS OF CROSSING OVER

- According to the number of chiasma, the following three types of crossing over have been described.
- **Single crossing over**: This is when the chiasma occurs only at one point of the chromosome pair, then the crossing over is known as single crossing over.

KINDS OF CROSSING OVER

- **Double crossing over**: This is when the chiasmata occur at two points in the same chromosome.
- **Multiple crossing over**: This is when crossing over takes place at more than two places in the same chromosome pair. This occurs rarely.

MECHANISMS OF CROSSING OVER

- It comprises of four steps:
- Synapsis
- Duplication of chromosomes/Tetrad formation
- Exchange of chromatids and
- Disjunction/Terminalisation.

SYNAPSIS

- Synapsis or intimate pairing between two homologous chromosomes (one paternal and another maternal) is initiated during zygotene stage of prophase I of meiosis I.

SYNAPSIS

- Synapsis often starts when the homologous ends of the two chromosomes are brought together on the nuclear envelope
- And it continues inward in zipper-like manner from both ends, aligning the two homologous chromosomes side by side.

SYNAPSIS

- Synapsis is the phase of prolonged and close contact of homologous chromosomes due to attraction between two exactly identical chromosomes.
- The paired homologous chromosomes are called **bivalent**. Their chromatids are not visible at this stage.

Duplication of chromosomes or tetrad formation

- The synapsis is followed by duplication of chromosomes in pachytene.
- ❑ During this stage, each homologous chromosome of bivalent splits longitudinally and form two identical sister chromatids which remains held together by an unsplitted centromere.
- ❑ At this stage, each bivalent contains four chromatids, so it is known as **tetrad**.

Crossing over/exchange of chromatids

- ❑ Crossing over occurs in the pachytene sub-stage. Chromosomal crossing over occurs due to exchange of chromosomal material between non-sister chromatids of each tetrad.
- ❑ During the process of crossing over, two non-sister chromatids first break at the corresponding points due to the activity of a nuclear enzyme known as **endonuclease**.

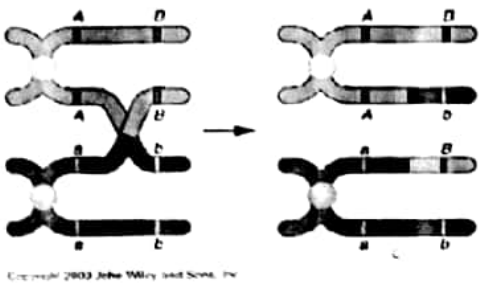
Crossing over/exchange of chromatids

- ❑ Then a segment on one side of each break connect with a segment on the opposite side of the break, so that the two non-sister chromatids cross each other.
- ❑ This takes place due to the action of an enzyme known as **ligase**. The crossing of two chromatids is known as **chiasma**.
- ❑ The crossing over thus include the breaking of chromatid segment, their transposition and fusion.

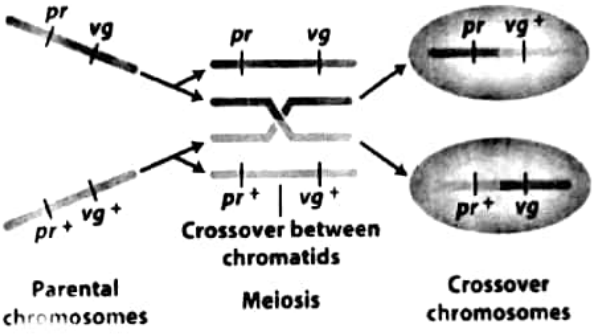
DISJUNCTION/TERMINALISATION

- ❑ After the completion of crossing over, the synaptic forces end and the homologous chromosomes move apart.
- ❑ The sites where crossing over occurs are called **chiasmata**. Therefore, the above explained mode of crossing over is called chiasma-type hypothesis.

Crossing over/exchange of chromatids



Crossing over/exchange of chromatids



FACTORS AFFECTING CROSSING OVER

Factors affecting crossing over

- **Temperature** : Frequency of crossing over is increased by **high and low temp.**
- **X-rays** : Frequency of crossing over is increased by X-rays.
- **Radium radiations also** increase crossing over.
- **Sex** : Crossing over **is more in females.**
- **Age** : Older females **have** more chances of crossing over.

HUMAN KARYOTYPE

- **Karyotype** is the organized profile of metaphase chromosomes of individual cell. Karyotype is specific to an individual or to related group [species]
- **Karyotyping** is the process by which doctors and geneticists take pictures of the chromosomes while the cell are undergoing mitosis.
- The picture of the chromosomes are then cut up so that each chromosome is removed. The chromosomes are matched up and attached to a paper according to size. The chromosomes pairs are numbered from largest to smallest.

KARYOTYPING

- This technique can be used to assess the "normalcy" of an individual's chromosomes and to assay for various genetic diseases such as Down's syndrome and Klinefelter's syndrome.
- It is estimated that one in 156 live births have some kind of chromosomal abnormality.

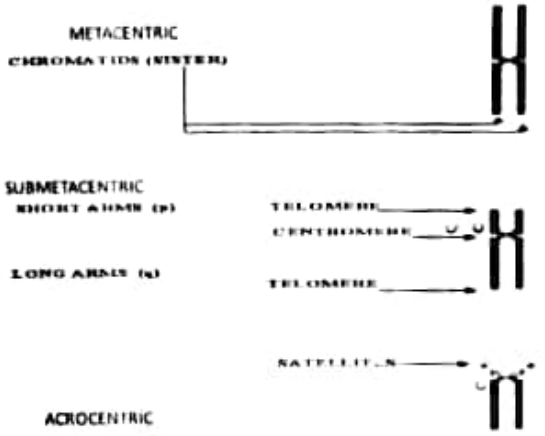
KARYOTYPE

- Karyotype include information about:
  - chromosome number
  - chromosome size
  - chromosome shape [morphology]
  - composition of the sex chromosomes
  - Some chromosomal abnormalities

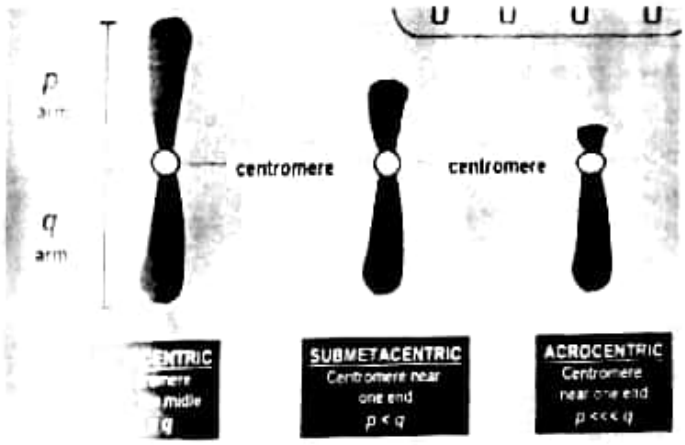
CHROMOSOME MORPHOLOGY

- A chromosome is divided by its centromere into short arm **(p) and long arm (q).**
- chromosomes can be classified by the position of their **centromere:**
- - **Metacentic: If its two arms are equal in length.**
- - **Submetacentric: If p arm is shorter than q arm.**
- - **Acrocentric: If the p arm is so short that is hard to observe, but still present**

CHROMOSOME MORPHOLOGY



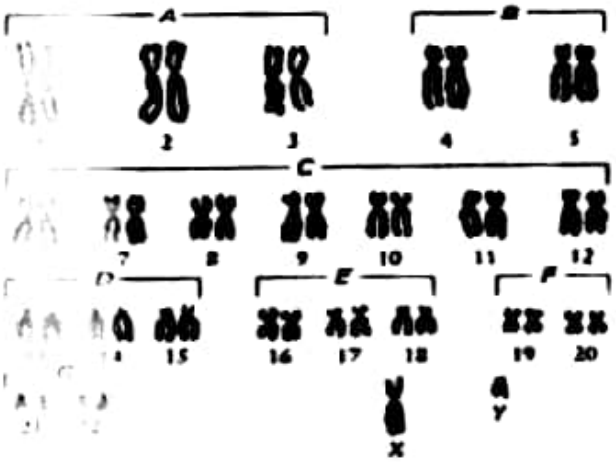
CHROMOSOME MORPHOLOGY



KARYOTYPE ARRANGEMENT

- In karyotype, chromosomes are arranged according to:
- **Size:** chromosomes are arranged and numbered from largest to smallest, with the short p-arm on the top and the long arm on the bottom.
- **Centromere location**
- **Banding patterns**
- Chromosomes can be arranged in 7 groups (A, B, C,D, E, F, G).

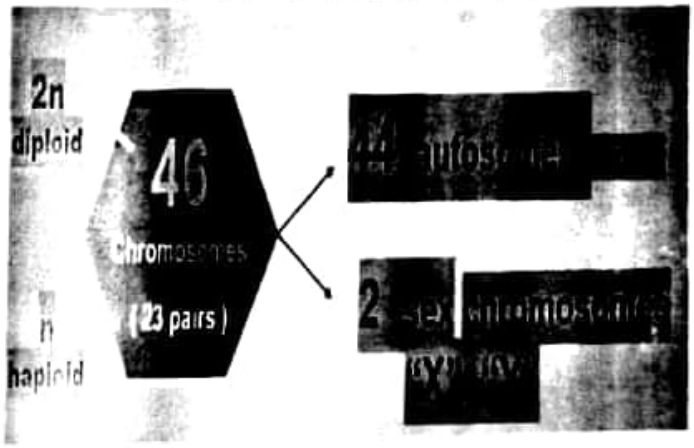
KARYOTYPE ARRANGEMENT



KARYOTYPE ARRANGEMENT

Group	Chromosomes	Size and Shape
C	6 - 12 and X	Medium submetacentric
D	13 - 15	Medium acrocentric
	16 - 18	Short [illegible]
F	19 and 20	Short metacentric
G	21 and 22 and Y	Short acrocentric

HUMAN KARYOTYPE



Human karyotype: Male & Female

ALTERATIONS IN CHROMOSOME NUMBER

- Chromosomal aberrations are abnormalities in the number or microscopically observable structure of chromosomes.
- The number of chromosomes in human cells is 46 with 22 autosomal pairs (one of each type contributed by the mother and one of each type from the father) and
- 2 sex chromosomes - 2X chromosomes for females (one from father and one from mother) or

ALTERATIONS IN CHROMOSOME NUMBER

- X and a Y chromosome for males (the X from the mother and the Y from the father).
- The chromosomes are visible only at the **metaphase stage of mitosis, 22** homologous pairs of **autosomes and two sex chromosomes.**
- **Each** chromosome has a characteristic size and shape in the "normal" cell.

CHROMOSOMAL ABERRATIONS

- **Nondisjunction** occurs when either homologues fail to separate during anaphase I of meiosis, or sister chromatids fail to separate during anaphase II.
- The result is that one gamete has 2 copies of one chromosome and the other has no copy of that chromosome.

CHROMOSOMAL ABERRATIONS

- If either of these gametes unites with another during fertilization, the result is
- **Aneuploidy** (abnormal chromosome number)
- **A trisomic cell has one extra chromosome (2n +1) example: trisomy21** (Down syndrome).
- **A monosomic cell has one missing chromosome (2n - 1) = usually lethal except for one known in humans: Turner's syndrome** (monosomy XO).

CHROMOSOMAL ABERRATIONS

- The frequency of non disjunction is quite high in humans, but the results are usually so devastating to the growing zygote that miscarriage occurs very early in the pregnancy.
- If the individual survives, he or she usually has a set of symptoms - a syndrome - caused by the abnormal dose of each gene product from that chromosome.

Examples of chromosomal aberrations:

- ❖ Human disorders due to chromosome alterations in autosomes (Chromosomes 1-22).
  - **Trisomy 13, XX (Patau Syndrome).** The karyotype here demonstrates trisomy 13 (47, XX, +13).
  - It is rare for babies to survive for very long if live born because of the multitude of anomalies that are usually present.

Examples of chromosomal aberrations:

- There is severely abnormal cerebral development and virtually always leads to death in early infancy.
- This baby has very pronounced clefts of the lip and palate, broad nose, small cranium, polydactyl (An extra finger), deafness, and non functional eyes.
- Heart defects and severe mental retardation are also part of the clinical picture

TRISOMY 18 (EDWARDS SYNDROME):

- ❖ There are severe mental retardation and highly characteristic pattern of malformations such as elongated skull, a very narrow pelvis, and a grasping of the two central fingers by the thumb and little finger.
- ❖ In addition, the ears are often low set and the mouth and teeth are small.
- ❖ Nearly all babies born with this condition die in early infancy

TRISOMY 21 (DOWN SYNDROME)

- This is an example of trisomy 21 (47, XY, +21) also known as Down syndrome. Note the extra chromosome 21.
- The non-dysjunctional event in meiosis that produces this anomaly increases in incidence with increasing maternal age.
- Trisomy 21, one of the most common causes of mental retardation. The child can have an IQ between 25-74. An average person has an IQ between 90-110.

TRISOMY 21 (DOWN SYNDROME)

- This results in a number of characteristic features, such as short stature, broad hands, stubby fingers and toes, a wide rounded face, a large protruding tongue that makes speech difficult.
- Individuals with this syndrome have a high incidence of respiratory infections, heart defects, and leukemia.
- The average risk of having a child with trisomy 21 is 1/750 live births.
- Mothers in their early twenties have a risk of 1/1,500 and women over 35 have a risk factor of 1/70, which jumps top 1/25 for women 45 and over.

TRISOMY 16 WITH MONOSOMY X

- This is the most common chromosomal abnormality, but the fetuses **NEVER survive past the first trimester.**
- Many first trimester fetuses are lost in this fashion (many are "silent" abortions).
- Note in this case that a sex chromosome is missing as well. Intrauterine demise is nature's way of eliminating abnormal karyotypes.

Nondisjunction of the sex chromosomes (X or Y chromosome)

- **Klinefelter syndrome (47, XXY males).** Male sex organs: unusually small testes, sterile. Breast enlargement and other feminine body characteristics. Normal intelligence.
- This is Klinefelter's syndrome with a 47, XXY karyotype. A non-dysjunctional event in meiosis left two X chromosomes in an ovum.
- This particular anomaly is relatively common (about 1 in 500 males), with affected persons being relatively normal.
- Characteristics associated with this condition are tall stature and sterility.

47, XYY MALES (JACOBS SYNDROME)

- A chromosome aberration which is caused by non disjunction of the Y chromosome during the second phase of meiosis giving a **47 XYY** karyotype.
- Occurence is 1/1000 live male births.
- Men with this karyotype are tall and have low mental ability/intelligence.

TRISOMY X: 47, XXX FEMALES

- 1:1000 live births
- Healthy and fertile - usually cannot be distinguished from normal female except by karyotype.

somy X (Turner's syndrome)

- This is nosomy X (Turner's syndrome, with karyotype 45, XO).
- This can ccur in about 1 per 2,700 births. It is not related to maternal age.
- Women with Turner's syndrome can live relatively normal lives, though they are unable to bear children.
- The phenotype of this female includes short stature, short broad neck, and a broad chest.
- Intelligence does not seem to be affected.
- 95% of these fetuses die before birth).

MULTIPLE CHOICE QUESTIONS

1. The number of chromosomes in human beings is
   (a) 36 (b) 46 (c) 26 (d) 16
2. One of the following is a mismatch
   (a) Down syndrome – Autosomal aneuploidy
   (b) Haemophila - Sex-linked
   (c) Klinefelter syndrome - XO complement
   (d) Turner syndrome – Female with retarded growth

IPLE CHOICE QUESTIONS

3. Turner's yndrome is depicted by
   XY XY (c) XYY (d) XO
4. A human male has enlarged breasts, sparse hair on t body and sex complement as XXY. He then ffers from
   Do wn syndrome (b) Edward syndrome
   Klinefelter's syndrome (d) Turner's syndrome
5. Number of chromosomes in human beings
   24 pairs (b) 28 pairs (c) 23 pairs (d) 20 pairs

MULTIPLE CHOICE QUESTIONS

6. Different types of chromosomes can be recognised by the position of the following separating the two arms

(a) centromere (b) genes (c) spindle (d) nucleus

7. Webbed neck is characteristic of

(a) XXY (b) XY male (c) XO female (d) XXX female

8. If various types of chromosomal abnormalities either in their number or morphology and these abnormalities may reside in autosomes or sex chromosomes and cause symptoms or a particular disease is known as

(a) gene expression (b) chromosomal aberrations

(c) syndromes (d) mutagens