BIO 201 LECTURE NOTE

BY MRS R.J. KOMOLAFE

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 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
- 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.

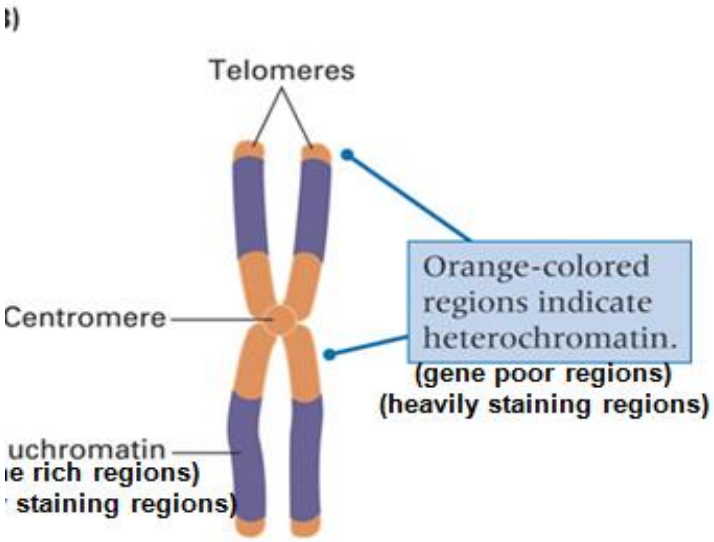
The chromosome theory of inheritance allows us to see the relationship between Mendel's laws and chromosome transmission

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

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
- 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
- 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.
- **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.
- **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.**
- 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.
- **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.



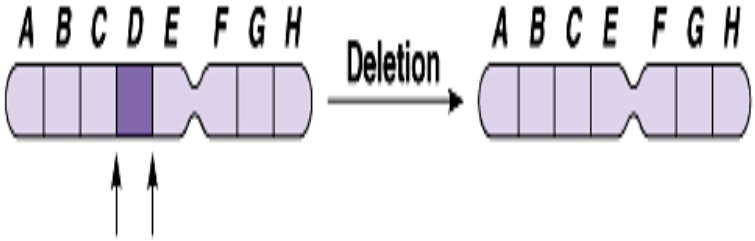
FUNCTIONS OF CHROMOSOMES

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.

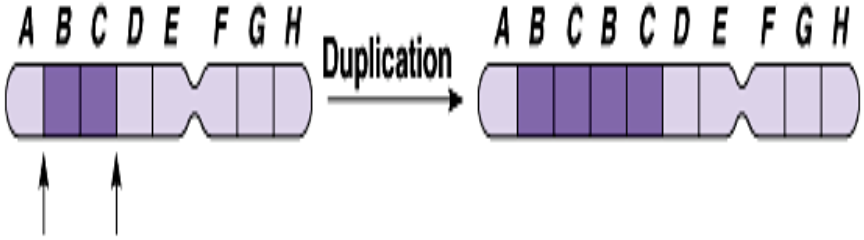
Errors and Exceptions in Chromosomal Inheritance

- 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.

(a) A **deletion** removes a chromosomal segment.

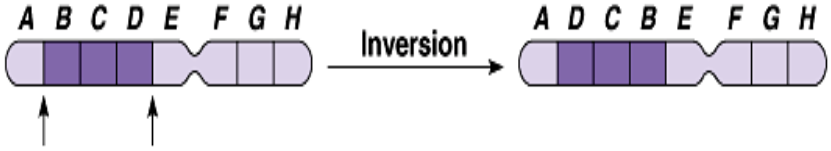


(b) A **duplication** repeats a segment.

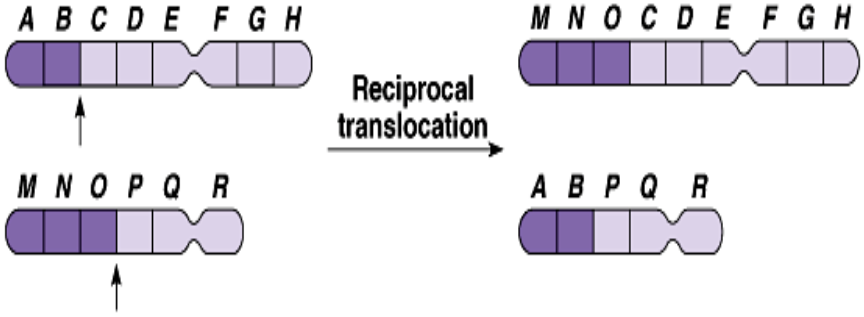


- An **inversion** occurs when a chromosomal fragment reattaches to the original chromosome but in the reverse orientation.
- In **translocation**, a chromosomal fragment joins a non homologous chromosome.
  - Some translocations are reciprocal, others are not.

(c) An **inversion** reverses a segment within a chromosome.



(d) A **translocation** moves a segment from one chromosome to another, non-homologous one.



- 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.
- 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.**

Examples of allelic interaction

1.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. 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**.

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 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*). 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*.

2. 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 referred to as partial dominance

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.

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

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.

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
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

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.

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.**

- Epistasis can be of the following types.
  - Due to **recessive gene**: Recessive gene **a** mask 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.

- 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)

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

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,

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

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. 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.

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.

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

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**.

- 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).
- ❑ 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

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 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.
- 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.*
- **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 point at which two chromatids join during the fusion and exchange of genetic material/crossing over in cell division), 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.

- **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

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

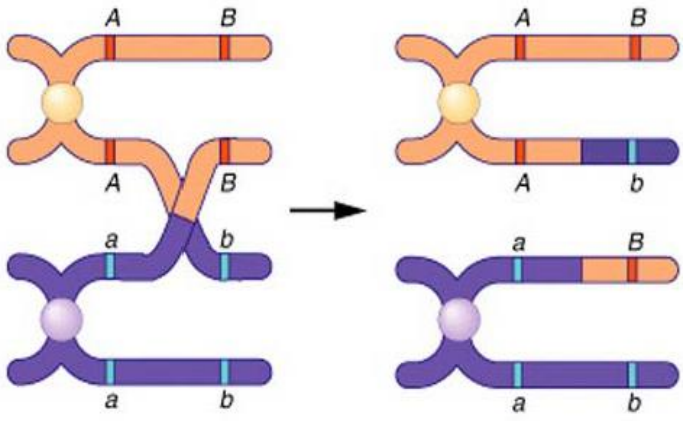
- At this stage, each bivalent contains four chromatids, so it is known as **tetrad**.

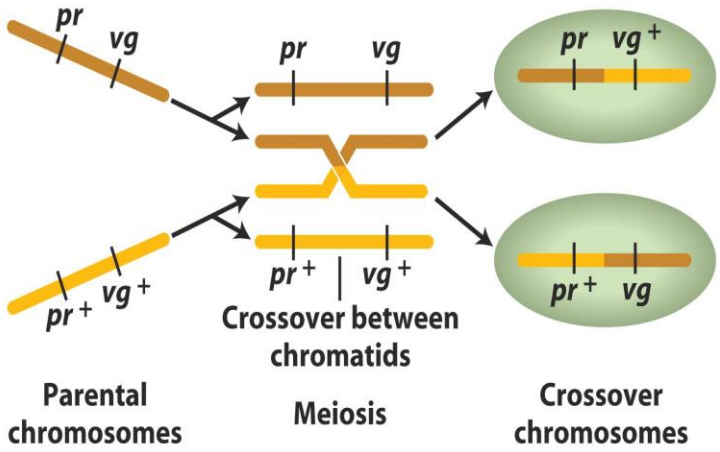
Crossing over/exchange of chromatids

- 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**.
- 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.





HUMAN KARYOTYPE

- Karyotype is the organized profile of metaphase chromosomes of individual cell. It is a photographic image that shows the sum of all the chromosome information in an individual cell.
- 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

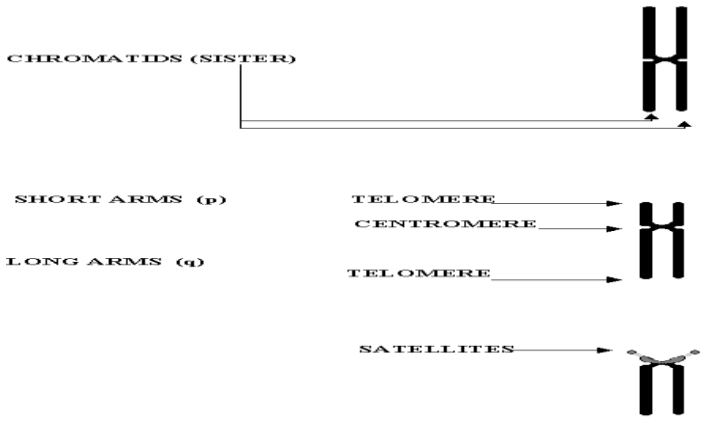
- A complete karyotype helps doctors determine if a person has extra chromosomes or missing chromosomes, or chromosomes that have attached to one another in unusual ways.
- It is estimated that one in 156 live births have some kind of chromosomal abnormality.
- Karyotype include information about:
  - chromosome number
  - chromosome size
  - chromosome shape [morphology]
  - composition of the sex chromosomes
  - Some chromosomal abnormalities

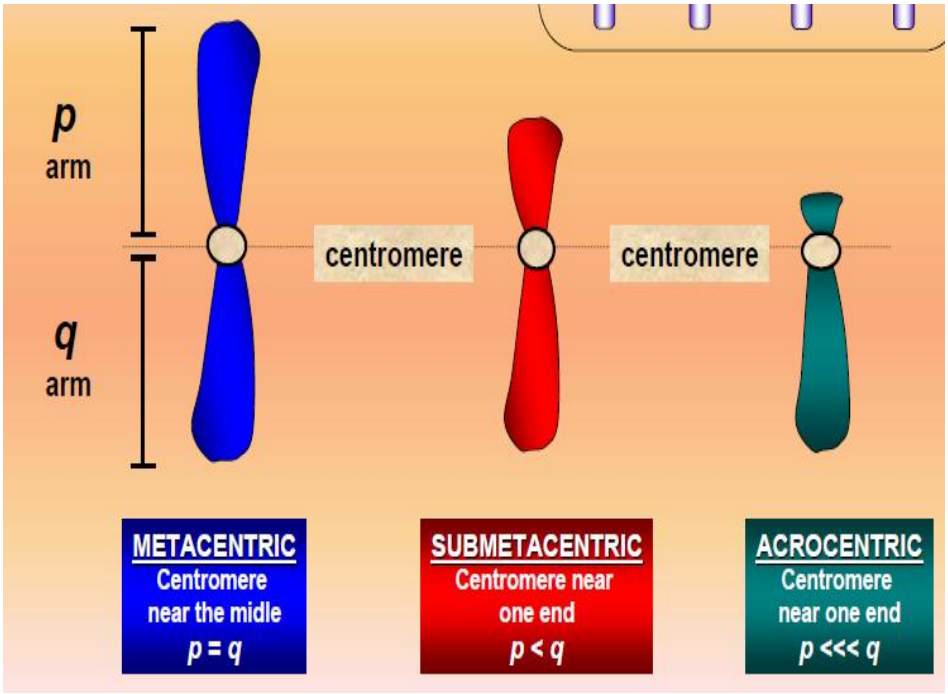
CHROMOSOME MORPHOLOGY

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:

- - Metacentric: 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

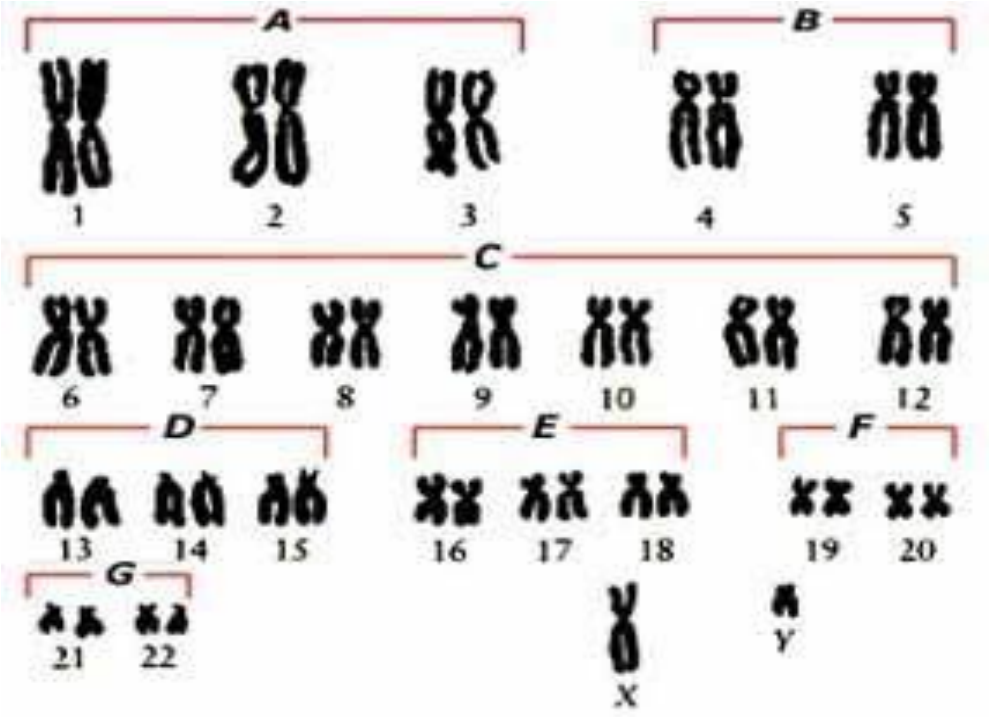




KARYOTYPE ARRANGEMENT

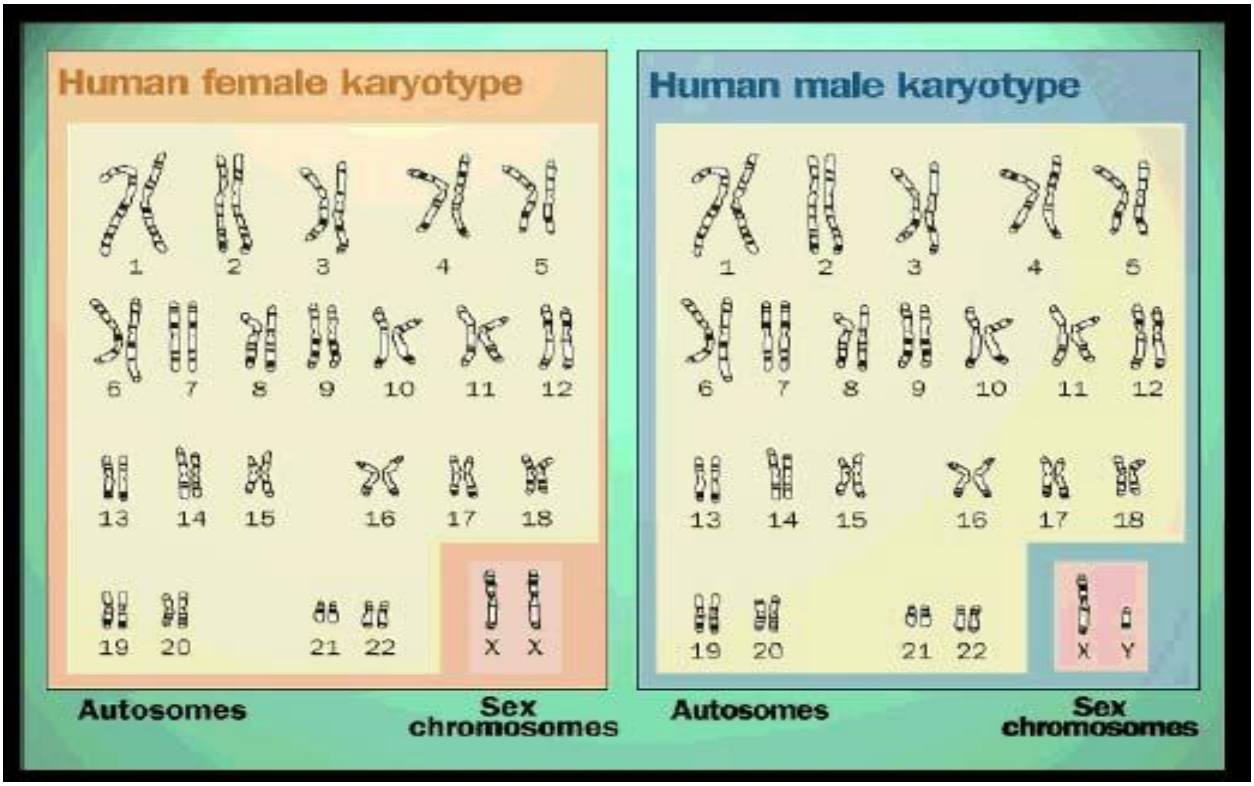
- In karyotype, chromosomes are arranged according to:

- **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).



Group	Chromosomes	Size and Shape
A	1 - 3	Large metacentric
B	4 and 5	Large submetacentric
C	6 - 12 and X	Medium submetacentric
D	13 - 15	Medium acrocentric
E	16 - 18	Short submetacentric
F	19 and 20	Short metacentric
G	21 and 22 and Y	Short acrocentric

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 - XX chromosomes for females (one from father and one from mother) or an 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.
- 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: trisomy**21 (Down syndrome).
- **A monosomic cell has one missing chromosome (2n - 1) = usually** lethal except for one known in humans: **Turner's syndrome** (monosomy XO).
- 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.
  - There is severely abnormal cerebral functions 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)

- These 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.
- 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.

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.

MONOSOMY X (TURNER'S SYNDROME)

- This is monosomy X (Turner's syndrome, with karyotype 45, XO).
- This can occur in about 1 per 2,700 births. It is not linked 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.
- (98% 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

3. Turner’s syndrome is depicted by

(a) XY (b) XXY (c) XYY (d) XO

4. A human male has enlarged breasts, sparse hair on the body and sex complement as XXY. He then suffers from

(a) Down syndrome (b) Edward syndrome

(c) Klinefelter’s syndrome (d) Turner’s syndrome

5. Number of chromosomes in human beings

(a) 27 pairs (b) 28 pairs (c) 23 pairs (d) 20 pairs

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

GENE MAPPING

Gene mapping is the map/location of genes that are present inside the chromosome.

- Gene mapping is divided into two types:
- Linkage mapping
- Physical mapping
- Sturtevant proposed that recombination frequencies

reflect the distances between genes on a chromosome.

- It is assumed that the chances of crossing over is equal at all points on a chromosome
- If so, then the farther apart two genes are:
- (a) the higher the probability that a cross-over will occur between them and therefore
- (b) the higher the recombination frequency
- Because the greater the distance between two genes the more points there are between them where crossing over can occur
- The knowledge of gene linkage can be used to:
- Determine the order of genes on the chromosome
- Determine how far apart genes are on the chromosome
- Construct a map of the chromosome.

APPLICATION OF GENE MAPPING

- Gene mapping is used to locate diseased genotypes
- To locate a particular gene in total genome.
- To screen different types of plants for a desired trait and breed them to get superior products.

LINKAGE MAPPING

- A diagrammatic, graphical representation of relative distances between linked genes of a chromosome is called linkage or genetic map.
- It is based on the use of genetic techniques to construct maps showing the positions of genes and other sequence features on a genome
- Genetic techniques include cross-breeding experiments or in case of humans, the examination of family histories (pedigrees).
- Linkage mapping is the type of mapping which is able to tell us the location of one gene to the other. It tells us whether G1 and G2 are closely related or distantly related. Utilizing linkage map, we can get the distance between two genes.
- Linkage mapping is not satisfactory enough because it can only give us the relative location and relative placement of a gene but it cannot give us the relative presence of a gene inside the chromosome.

Physical Mapping

Physical map is the type of map which shows the presence of gene inside the chromosome.

- For example,we can tell that G1 is present in chromosome x and G2 is present in chromosome y.
- Physical map can tell us the idea of the exact location of a gene inside the chromosome.
- Physical map is much more specific by giving us the idea of the presence of gene in the chromosome or not.