

Karyotype and Idiogram

What is a Karyotype?

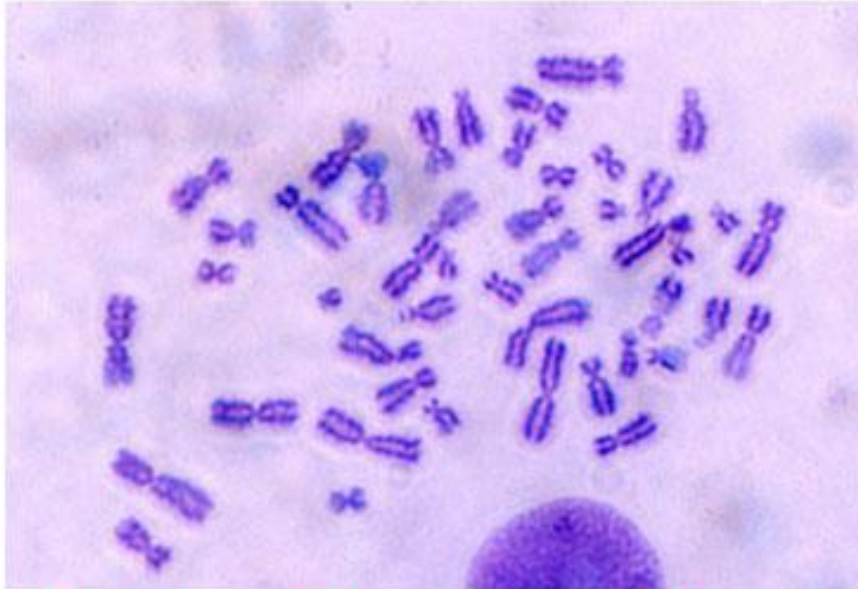
- The word 'karyotype' is derived from Greek word "kernel" which means content of nucleus.
- The term Karyotype is referred to a group of characteristics that allow the identification of particular chromosome pair on the basis of general morphology of a set of chromosome at metaphase of a somatic cell of an individual.
- Karyotype concept was derived by S. Navashin based on the observation that the number of chromosomes and morphology of each chromosome pair is normally constant and characteristics for a species.
- Karyotype concept is a phenotypic appearance of a chromosome and its homologous pair which helps to differentiate types of chromosome based on their morphology in an individual species.
- Karyotyping is a method of arrangement of pair of homologous chromosome of a cell in decreasing series of their size.
- Karyotype study is very important to identify abnormalities in chromosomal structure and number.
- Karyotype study also helps to determine origin and evolutionary relationship among different taxa.
- All species are characterized by a set of chromosomes to carry their genetic information.
- The chromosomal composition of each species has a number of characteristics.
- *The Karyotype is a set of characteristics that identifies and describes a particular set of chromosome.*

These characteristics which are described by a karyotype are: -

- (1). The chromosome numbers
- (2). Relative size of different chromosomes
- (3). Position of centromere and length of chromosomal arms
- (4). Presence of secondary constrictions and satellites
- (5). Banding pattern of the chromosome
- (6). Features of sex chromosomes

What is Karyotyping?

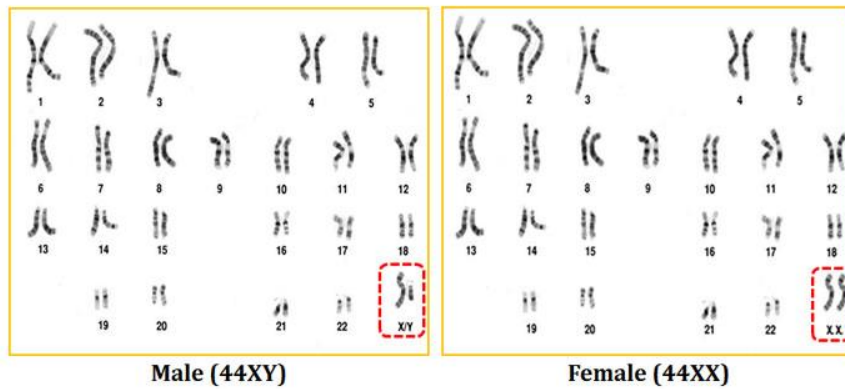
- *The process of preparation of the karyotype of a species is called Karyotyping.*
- Karyotyping is now most commonly used in **clinical diagnosis** and **clinical genetics**.
- Karyotype is prepared from the microphotographs of **metaphase** chromosomes.



Metaphase Chromosomes (Human)

- Ø The metaphase chromosome is selected because at this stage the chromosome will have **maximum condensation** (maximum thickness).
- Ø At metaphase stage, the chromosomes will be visible through an ordinary laboratory microscope.
- Ø For the clinical karyotyping, the sample materials used may be cells from biopsies, bone marrow cells, blood cells or cells from amniotic fluid or chorionic villus.
- Ø The sample cells were first cultured on artificial medium with suitable growth regulators.
- Ø The then the cells are arrested at their mitotic metaphase phase by treating with **Colchicine**.
- Ø Colchicine will arrest the cells at metaphase stage since it prevents the formation of **spindle fibres**.
- Ø In the absence of spindle fibres, the metaphase stage cannot proceed to anaphase.
- Ø Then the cells were fixed with suitable fixative and treated with specific stains to produce characteristic banding patterns in the chromosomes.
- Ø Specific staining or **banding** techniques are used to identify the homologous pairs of chromosomes within the cells.
- Ø Cells are then observed through the microscope and the photographs of the chromosomes were taken.
- Ø The individual chromosomes are cut out from the microphotographs and then they are lined up by size with their respective partners to form the karyogram
- Ø A uniformly accepted pattern is used for the arrangement of chromosomes in the preparation of karyogram.
- Ø In a karyotype, the chromosomes of the organism are ordered in a series of its decreasing size (largest chromosome at first and smallest at last).
- Ø In the case of human, the autosomes are numbered from 1 to 22 and arranged in the order of decreasing size.
- Ø Sex chromosomes are arranged after the autosomes.
- Ø Chromosomes in the karyogram are aligned along a horizontal axis shared by their centromeres.

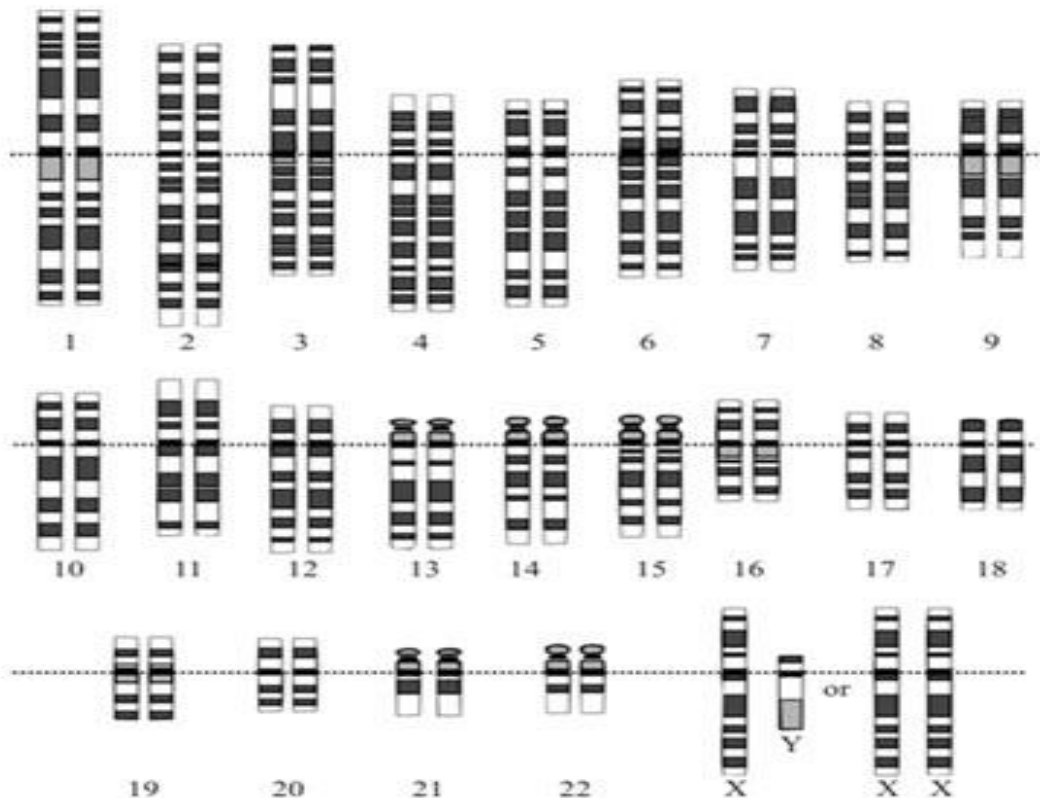
HUMAN KARYOTYPE (NORMAL)



Ø Individual chromosomes are always depicted with their short 'p' arms at the top, and their long 'q' arms at the bottom.

Ø The centromeric index is also noted in karyotype analysis.

Ø Centromeric index is the ratio of the length of long and short arms of the chromosome.



Idiogram (Human)

What is an Idiogram?

Ø The **diagrammatic** representation of a **karyotype** of a species is called **Idiogram**.

What is the Significance / Importance of Karyotype and Karyotyping?

Ø Karyotypes of different species can be easily **compared**.

Ø Similarities in the karyotypes represent the **evolutionary relationship**.

- Ø Karyotypes can be used to **solve taxonomic disputes**.
- Ø The karyotype can indicate **primitive** and **advanced** features of an organism.
- Ø The karyotype of an organism may be **symmetric** or **asymmetric**.
- Ø A symmetric karyotype possesses more or less similar sized and shaped chromosomes.
- Ø An asymmetric karyotype will have huge differences in small and large chromosomes and contain less metacentric chromosomes.
- Ø A symmetric karyotype is considered as primitive whereas, an asymmetric karyotype is considered as advanced.
- Ø The **zygomorphic flowers** in plants are associated with **asymmetric** karyotype.
- Ø Some species may have special characteristics in their karyotypes such as mouse has acrocentric chromosomes and many amphibians have only metacentric chromosomes.