KARYOTYPE

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Karyotyping is the process by which photographs of chromosomes are taken in order to determine the chromosome complement of an individual, including the number of chromosomes and any abnormalities. The term is also used for the complete set of chromosomes in a species or in an individual organism and for a test that detects this complement or measures the number.

Karyotypes describe the chromosome count of an organism and what these chromosomes look like under a light microscope. Attention is paid to their length, the position of the centromeres, banding pattern, any differences between the sex chromosomes, and any other physical characteristics The preparation and study of karyotypes is part of cytogenetics.

Karyogram of human male using Giemsa staining

The study of whole sets of chromosomes is sometimes known as *karyology*. The chromosomes are depicted (by rearranging a photomicrograph) in a standard format known as a *karyogram* or *idiogram*: in pairs, ordered by size and position of centromere for chromosomes of the same size.

The basic number of chromosomes in the somatic cells of an individual or a species is called the *somatic number* and is designated *2n*. In the germ-line (the sex cells) the chromosome number is *n* (humans: n = 23). Thus, in humans 2n = 46.

So, in normal diploid organisms, autosomal chromosomes are present in two copies. There may, or may not, be sex chromosomes. Polyploid cells have multiple copies of chromosomes and haploid cells have single copies.

Karyotypes can be used for many purposes; such as to study chromosomal aberrations, cellular function, taxonomic relationships, medicine and to gather information about past evolutionary events.(*karyosystematics*).

Six different characteristics of karyotypes are usually observed and compared:

1. Differences in absolute sizes of chromosomes. Chromosomes can vary in absolute size by as much as twenty-fold between genera of the same family. For example, the legumes *Lotus tenuis* and *Vicia faba* each have six pairs of chromosomes, yet *V. faba* chromosomes are many times larger. These differences probably reflect different amounts of DNA duplication.
2. Differences in the position of centromeres. These differences probably came about through translocations.
3. Differences in relative size of chromosomes. These differences probably arose from segmental interchange of unequal lengths.
4. Differences in basic number of chromosomes. These differences could have resulted from successive unequal translocations which removed all the essential genetic material from a chromosome, permitting its loss without penalty to the organism (the dislocation hypothesis) or through fusion. Humans have one pair fewer chromosomes than the great apes. Human chromosome 2 appears to have resulted from the fusion of two ancestral chromosomes, and many of the genes of those two original chromosomes have been translocated to other chromosomes.
5. Differences in number and position of satellites. Satellites are small bodies attached to a chromosome by a thin thread.
6. Differences in degree and distribution of heterochromatic regions. Heterochromatin stains darker than euchromatin. Heterochromatin is packed tighter. Heterochromatin consists mainly of genetically inactive and repetitive DNA sequences as well as containing a larger amount of Adenine-Thymine pairs. Euchromatin is usually under active transcription and stains much lighter as it has less affinity for the giemsa stain. Euchromatin regions contain larger amounts of Guanine-Cytosine pairs. The staining technique using giemsa staining is called G banding and therefore produces the typical "G-Bands"

Human karyotype

The normal human karyotypes contain 22 pairs of autosomal chromosomes and one pair of sex chromosomes (allosomes). Normal karyotypes for females contain two X chromosomes and are denoted 46,XX; males have both an X and a Y chromosome denoted 46,XY. Any variation from the standard karyotype may lead to developmental abnormalities.