

Karyotype Encyclopedia Article

Karyotype

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Karyotype

The full complement of chromosomes, arranged in a logical order, is known as a karyotype. Chromosomes of a cell are visible with a microscope only during mitosis (nuclear division). When a cell's nuclei are in interphase chromosomes are not ordinarily visible. DNA replication occurs during interphase. Nuclear division begins with mitotic prophase. Chromosomes first appear as long threads in prophase. However, they become condensed as mitosis proceeds. The chromosomes are double because of prior DNA replication. The two parts of the chromosome are known as chromatids, which are held together at the centromere. Metaphase follows prophase and here the chromosomes are maximally shortened and readily stain (hence the name chromosome which means colored body). The nuclear membrane disappears at mitotic metaphase and the chromosomes can be prepared for karyotyping. First, a good metaphase spread must be prepared. Historically, this was done by literally squashing a fixed and stained cell in metaphase. With luck, the squashing separated individual chromosomes such that they could be photographed. Then the chromosomes were arranged in a logical order which was generally by size, from long chromosomes to short chromosomes.

Contemporary karyotyping involves first obtaining a culture of cells in a liquid medium. Many cells in mitosis can be obtained with culture. The cells are treated and stained which results in unambiguous identification of each chromosome pair. Formerly, short chromosomes were lumped together as were long chromosomes. Now, with the introduction of a diversity of stains which include some that fluoresce differentially in the ultraviolet, each chromosome pair may be clearly distinguished.

Some birth abnormalities (e.g., Down syndrome) and certain cancers are characterized by specific chromosomal aberrations. Further, exposure to teratogens, mutagens, and carcinogens may result in a non-specific karyotypic change from the normal.