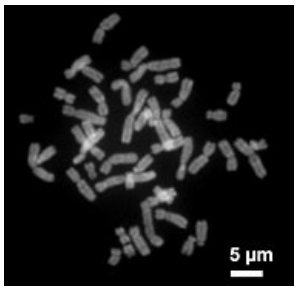
Chromosome

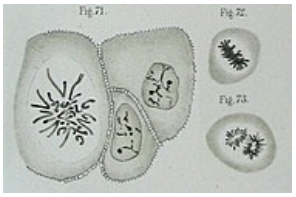
The nuclear chromosomes of human cells are linear structures composed of DNA and proteins; histones (basic proteins) and non-histone type proteins (acidic proteins). These proteins are essential in spiraling DNA and regulating its function. E.g. histones regulate DNA activity using epigenetic mechanisms (imprinting). In addition to the chromosomes located in the nucleus, semi-autonomous **genetic information** is stored in cells in **the circular chromosomes** of the mitochondria. Cytogenetics deals with the study of chromosomes. At this point, we will focus only on nuclear chromosomes in connection with the transmission of genetic information from parents to offspring.



Metaphase chromosomes of a human (female) lymphocyte

Each animal species has a characteristic number and structure of chromosomes. Diploid (**somatic**) human cells have 46 chromosomes (2n), which form 23 pairs. Of these, 22 pairs are homologous autosomes and one pair are sex chromosomes (synonyms - gonosomes/heterochromosomes). In each pair of chromosomes, one chromosome is of maternal origin, the other is of paternal origin.

Gametes have a haploid number of chromosomes (n), meaning they contain only one chromosome of a given pair; one autosome from each pair and one gonosome. During the first meiotic division, when the chromosomes separate to the poles of the cell, random combinations of chromosomes of maternal and paternal origin occur, which subsequently leads to new combinations of genetic equipment in the next generation. From the point of view of chromosomal makeup, women create the same type of gametes (homogametic sex) with chromosomal makeup 22,X; in males, two types of gametes with chromosomal makeup of either 22,X or 22,Y are produced with a 50% probability for each type.


Probably the earliest depiction of human chromosomes (Walther Flemming, 1882)

Each chromosome carries **linearly arranged** genes that form a binding unit; we are talking about the chromosome, or genetic map. The chromosome map indicates the position of individual loci (genes), which is unchangeable. The order of loci on each chromosome is determined by linkage analysis. Linkage analysis makes it possible to determine both the order of loci and their relative distance on the respective chromosome. In the next step, the genetic map is refined by physical mapping. Linkage groups on pairs of homologous chromosomes are identical. In the case of homologous chromosomes, either identical or different gene forms - alleles - can occur in paired loci.


Walther Flemming (1843-1905)

Links

Related Articles

- Autozomes
- Gonosomes (Chromosome X, Chromosome Y)
- Chromosomal aberrations
- Identification of chromosomes
- Human karyotype
- Structure of the metaphase chromosome
- Types of metaphase chromosomes