Chromosome Number and Structure

Chromosome Number

Sexually reproducing species have:

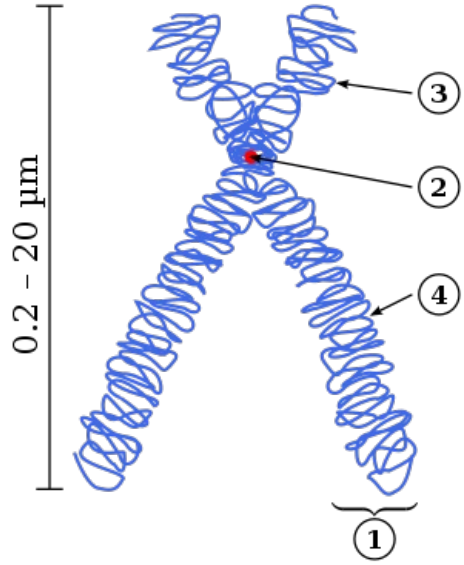
1. Somatic or body cells, which are DIPLOID [2n] having two sets of chromosomes, one from the mother and one from the father.
2. Gametes or reproductive cells, are HAPLOID [n]. They are produced by the meiosis of a diploid germ cell, where the matching chromosomes of the father and mother go through crossover i.e. exchange small parts, and thus create new chromosomes. When a male and a female gamete merge (fertilization), a new diploid organism is formed.

Examples:

1. In humans, a normal cell nucleus contains 46 chromosomes (22 pairs of autosomes, 1 pair of sex chromosomes);
2. In gorillas/chimpanzees, there are 48 chromosomes;
3. In dogs, there are 78 chromosomes.

Chromosome Structure

Chromosomes are packaged and condensed into a complicated structure with proteins called chromatin. Chromosomes are best seen during cell division when they are most condensed, especially during metaphase. They are seen by light microscope or by special stains. The centromere (number 2 on picture) is the point where the two chromatids touch, and where the microtubules attach therefore helping in the movement of chromosomes at cell division. It divides it into short (p; number 3 on picture) and long arms (q; number 4 on picture). The telomere is the tip of each end.



Chromosome

- Size & shape classification: this is based on the length of chromosomes, i.e. large, medium, or short chromosomes.
- Morphological classification: this is based on the position of the centromere. Chromosomes can be:

1. Metacentric: when the two chromosome arms are equal in length.
2. Submetacentric: when the two chromosome arms are unequal in length, e.g. shorter p arms and longer q arms.
3. Acrocentric: when the p (short) arm is too short to observe but is still present. There are 5 acrocentric chromosomes in the human genome: 13, 14, 15, 21, and 22.
4. Telocentric: when the centromere is located at the end of the chromosome. There are no telocentric chromosomes in the human genome.
5. Holocentric: when the centromere makes up the entire length of the chromosome. There are no holocentric chromosomes in the human genome.

Techniques of Examination

Staining methods originally available for human cytogenetic analysis did not allow all 24 types of chromosome (22 autosomes, X and Y) to be individually identified. With techniques now in common all chromosomes can be identified:

Staining Methods for Routine Analysis

These are banding methods for chromosomal identification and analysis of chromosomal structure. Banding can be used to identify chromosomal abnormalities, such as translocations because of the unique pattern of bands.

1. G-Banding (GIEMSA): chromosomes in metaphase are treated with trypsin (to partially digest the protein) and stained with GIEMSA (specific stain for phosphate groups of DNA). This gives each chromosome a characteristic pattern of light & dark bands. Dark regions are Adenine-Thymine rich. This method will normally produce 300-400 bands in a normal human karyotype.
2. Q-Banding (Quinacrine): involves staining with quinacrine mustard or related compounds and examination with a fluorescent microscope. The chromosomes stain in a specific pattern of bright & dim bands. The bright

Q-bands correspond almost exactly to the dark G-bands.
3. R-Banding (Reverse): the reverse of G-banding. The dark regions are euchromatic (guanine-cytosine rich regions) and the bright regions are heterochromatic (thymine-adenine). It is a result of the chromosomes receiving heat pretreatment before Giemsa staining.

Special Procedures

1. C-Banding (Centromeric Heterochromatin): chromosomes pretreated with acid followed by alkali prior to G-banding; centromeres and other heterochromatic regions containing highly repetitive DNA sequences are preferentially stained. i.e. sections of chromosomes 1q, 9q, 16q and Yq
2. High Resolution Banding: achieved through G-banding or R-banding techniques to stain chromosomes that have been prepared at an early stage of mitosis (prophase or prometaphase) when they are still in a relatively uncondensed state. Prometaphase banding is routinely used. Metaphase banding may also be used when a structural abnormality is suspected.
3. Fragile Sites: non-staining gaps, occasionally observed in characteristic sites on several chroms. Many such sites are known to be heritable variants. It is seen near the end of Xq. It is clinically significant in males with a specific and quite common form of X-linked mental retardation, and female carriers of the same defect. In the fragile X-syndrome the fragile site is visualized only in cells that have been cultured under conditions of thymidine deprivation. This is achieved by a culture medium low in thymidine and folic acid, and an addition of an inhibitor of enzyme thymidine synthetase to the medium.
4. Molecular cytogenetics: use of cloned DNA probe labeled with different colored fluorescent tags to visualize one or more specific regions. [FISH – Fluorescent In Situ Hybridization].

Links

Related articles

- Karyotype
- The Sex Chromosomes and Sex Determination
- Disorders of the Autosomes
- Disorders of the Sex Chromosomes
- Abnormalities of Chromosome Structure

External links

References

Bibliography