Heterochromosome

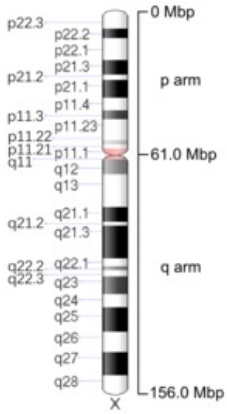
Position in karyotype

- The human karyotype consists of 22 pairs of **autosomes** and one pair of **gonosomes** (**heterochromosomes** , sex chromosomes)
- The X and Y chromosomes differ in their morphology, but especially in their genetic map, where the X chromosome has a rich and different genetic makeup compared to the Y chromosome in the heteronymous region.
- The X and Y chromosome have identical loci only in a short pseudoautosomal region (homonymic region) .

Role in sex determination

- The combination of X and Y chromosomes is involved in the **genetic determination of sex** in the mammalian type of sex determination, i.e. even in the human population.
- **Female sex** is determined by a pair of submetacentric XX chromosomes , which belong to **group C in the** karyotype.
- **Male gender** is determined by the **XY chromosomes** (the presence of the Y chromosome is key - see below). The Y chromosome belongs to **group G** chromosomes , but it is the only one from this group that **does not carry satellites**. [1][2]
- The SRY (sex determining region of the Y chromosome) gene, which is located in the heteronymous region of the Y chromosome, is responsible for the development of sexual dimorphism . It is a key gene on which the cascade of events that lead to the development of male genital organs depends.
- Until approximately the sixth week of embryonic development, sexual differentiation is preceded by the genetically conditioned possibility of the development of both sexes, regardless of the presence of XX or XY heterochromosomes. This means that the rudiments of both gonad types (Wolffian ducts and Müllerian ducts) are present in all embryos. This fact results in a situation where the chromosomally determined gender does not apply.

X-chromosome inactivation



- Since women, unlike men, have genes located on the X chromosome in two copies, they have a random inactivation of one of the two X chromosomes, thereby compensating for the dose of genes in men.
- Inactivation of the X chromosome occurs around the twelfth day of embryogenesis .
- In cells, the chromosome of paternal origin is inactivated with approximately 50% probability, and with the same probability of maternal origin.
- In further cell divisions, the "memory" for the type of inactivation is then preserved; at the cellular level, clones with an inactivated X chromosome of either maternal or paternal origin are thus created.
- In the case of a heterozygous combination of alleles in genes located on the X chromosome, mosaicism arises at the cellular level in the expression of one or the other allele (e.g. testicular feminization , anhidrotic ectodermal dysplasia and others).
- One inactivated X chromosome is demonstrable under a light microscope in the interphase nucleus of a woman as a heterochromatin **body** (synonyms - Barr body , X chromatin).
- In syndromes that are conditioned by the presence of an extra X chromosome, only one of the X chromosomes is always active in the cells, the rest are inactivated. E.g. in Klinefelter syndrome with karyotype **47,XXY** , there is one Barr body in the interphase nucleus.

Genes and heredity

Chromosome Y

- The difference in the heterochromosomal make-up of men and women is also reflected in the genetic determination of traits located on the X chromosome.
- Females can be dominant homozygotes , heterozygotes , or recessive homozygotes.
- Men, due to the presence of only one X chromosome, have a trait determined by only one of two possible alleles – they are hemizygotes .
- For genes located on the X chromosome, the difference between homogametic females and heterogametic males must be taken into account.
- **The father** passes the X chromosome **to all his daughters** , the Y chromosome to his sons (so-called Dutch inheritance).
- **A mother** passes on an X chromosome **to both daughters and sons** with a 50% probability for each of her two X chromosomes.

Links

Related articles

- Chromosome
- Autosome
- Human karyotype
- X Chromosome
- Y Chromosome
- Sex-linked inheritance

References

1. International Standing Committee on Human Cytogenetic Nomenclature. . *ISCN 2009: an international system for human cytogenetic nomenclature.* 1. edition. Basel : Karger, 2009. 138 pp. ISBN 978-3-8055-8985-7.
2. MARK, H. F.. *Medical cytogenetics.* 1. edition. New York : Marcel Dekker, 2000. 680 pp. ISBN 978-0824719999.