

X chromosome

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Chromosome X' is one of a pair of human sex chromosomes. As a medium-sized submetacentric chromosome, it corresponds in size and shape to group C. In the latest assembly of the human genome, 2327 genes are annotated on the X chromosome ^[1]. Normally, each person receives one X chromosome from their mother. The second heterochromosome – either the X chromosome or the Y chromosome is then received from the father. In individuals with more than one X chromosome in their karyotype (that is, females and those with numerical aberrations of the X chromosome), these additional copies are inactivated in a process known as X inactivation.

Genes and Heredity

The inheritance of genes located on the X chromosome has its own characteristics, as a woman has two X chromosomes (lyonization is used here) and a man has only one (a man is a **hemizygote**) . We refer to this type of inheritance as gonosomal recessive or gonosomal dominant.

Pseudoautosomal regions

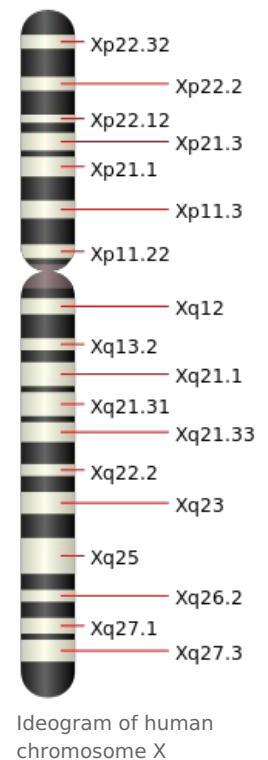
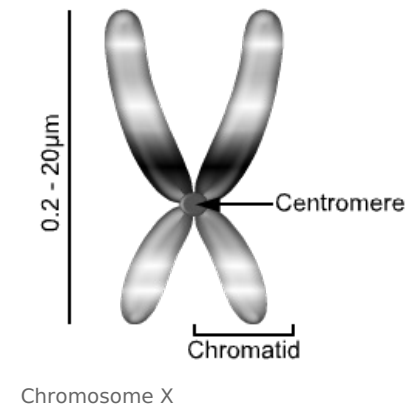
 For more information see *Pseudoautosomal region*.

Certain genes are located in the so-called ``pseudoautosomal sections of the X chromosome. These are two sections - ``PAR 1 (the larger section) at the end of the short arm of the X chromosome and ``PAR 2 ' (smaller section) at the end of the long arm of the X chromosome. These genes have their homologous copies in the same order on the Y chromosome. These regions allow the X and Y chromosomes to form a "homologous" pair during meiosis; crossing-over can occur between genes in these regions. An example is the gene **SHOX** (Short Stature Homeobox; Xp22.32; OMIM: *312865 (<https://www.omim.org/entry/312865>)) and its homologue the gene SHOXY ' (Yp11.2; OMIM: *400020 (<http://www.omim.org/entry/400020>)).

Selected genes

- **STS** (Steroid sulfatase; Xp22.32; OMIM: *300747 (<https://www.omim.org/entry/300747>)) - Encodes for **steroid sulfatase**; defect or deletion of this gene causes **X-linked ichthyosis**'.
- **KAL1** (Kallman syndrome 1; Xp22.3; OMIM: +308700 (<https://www.omim.org/entry/308700>)) - Encodes the protein **anosmin-1**; the mutation causes **Kallman syndrome** (central hypogonadism associated with anosmia).
- **PIGA** (Phosphatidylinositol glycan A; Xp22.1; OMIM: +311770 (<https://www.omim.org/entry/311770>)) - Acquired mutation of this gene (at the hematopoietic stem cell level) causes **Paroxysmal nocturnal hemoglobinuria**.
- **DMD** (Dystrophin; Xp21.2; OMIM: *300377 (<https://www.omim.org/entry/300377>)) - The gene encodes the muscle protein **dystrophin**; mutation of this gene causes muscular dystrophy of the Becker or Duchenne type.
- **WAS' (Wiskott-Aldrich syndrome; Xp11.33-p11.22; OMIM: *300392 (<https://www.omim.org/entry/300392>)) - A mutation in this gene causes a 'Wiskott-Aldrich syndrome** (congenital immunodeficiency, thrombocytopenia, eczema).
- **BTK** (Bruton agammaglobulinemia tyrosine kinase; Xq21.3-q22; OMIM: *300300 (<https://www.omim.org/entry/300300>)) - Mutations in this gene cause [**Bruton's agammaglobulinemia**|**Bruton's agammaglobulinemia**] (congenital immunodeficiency).
- **COL4A5** (Collagen, type IV, alpha-5; Xq22.3; OMIM: *303630 (<https://www.omim.org/entry/303630>)) - Encodes the alpha chain [collagen] in type IV, which is involved in the construction of basement membranes; mutation causes **Alport syndrome** (nephropathy associated with deafness).
- **F9** (Factor IX; Xq27.1-q27.2; OMIM: *300746 (<https://www.omim.org/entry/300746>)) - This gene encodes **coagulation factor IX** ; its mutation causes **Hemophilia B**.
- **FMR1** (Fragile X mental retardation protein; Xq27.3; OMIM: *309550 (<https://www.omim.org/entry/309550>)) - Mutations in this gene cause **Fragile X Chromosome Syndrome**.
- **F8** (Factor VIII; Xq28; OMIM: +306700 (<https://www.omim.org/entry/306700>)) - The gene encodes **coagulation factor VIII** ; its mutation causes **Hemophilia A**.
- **MECP2** (Methyl-CpG-binding protein 2; Xq28; OMIM: *300005 (<https://www.omim.org/entry/300005>)) - Mutations in this gene cause **Rett syndrome** (autism, stereotypical hand movements, affected almost exclusively by girls).

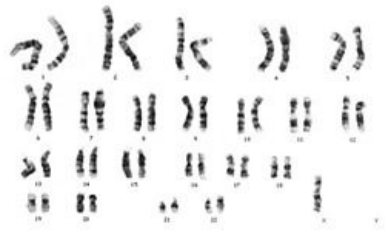
Chromosomal Abnormalities



Turner Syndrome

 For more information see [Turner syndrome](#).

It is most often caused by monosomy of the X chromosome (karyotype **45,X'**), **or by various loss structural aberrations (for example, deletion on the second X chromosome, etc.)**. **Numerical and structural changes can also occur in a mosaic. Classic manifestations are:** short stature, ovarian dysgenesis with all the consequences, typical skin fold (*pterygium colli*). Intellect is not significantly impaired. Treatment with growth hormone and hormone replacement therapy is possible.



Krk in Turner syndrome

Klinefelter syndrome

 For more information see [Klinefelter syndrome](#).

It is caused by the presence of an extra X chromosome in a man. It is most often caused by the karyotype **47,XXY**, **variants with more X chromosomes (48,XXXY or 49,XXXXY) are also possible, which have a more pronounced manifestation. There are also mosaic forms. The main symptoms are:** infertility' (*aspermia*), *hypogonadism*, average to tall height, long limbs, sparse hair, gynecomastia.



47,XXY

Syndrome 47,XXX

 For more information see [Syndrome 47,XXX](#).

Chromosome X trisomy, also called **'Triple X syndrome'** (and formerly "Superfemale syndrome"). As the name already suggests – it is caused by the karyotype **47,XXX**, it can also occur in mosaic. Karyotype 48,XXXX or 49,XXXXX can also occur very rarely, these cases have a different and more serious manifestation. The 47,XXX syndrome itself does not have a distinct clinical picture, some women are examined for "infertility". There may be minor psychosocial problems, such as learning problems.

Links

ws:Chromozom X

Related Articles

- Chromosome
- Gonosomes
- Y chromosome
- Human Karyotype
- Chromosomal aberrations
- Sex-linked inheritance
- X-linked inheritance

References

1. National Center for Biotechnology Information. *Homo sapiens Genome: Build 38 patch release 14 (GRCh38.p14)* [online]. ©2022. [cit. 7/11/2022]. <<https://www.ncbi.nlm.nih.gov/genome/?term=Human>>.