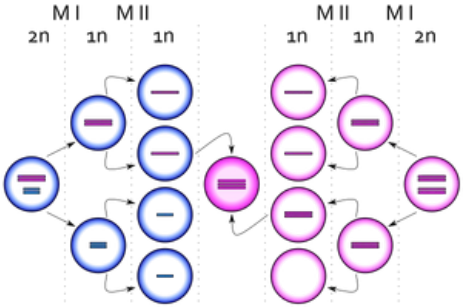
Syndrome 47,XXX



Scheme of the development of the syndrome 47,XXX

Chromosome X trisomy , also called Triple X syndrome (and formerly "Superfemale syndrome"), is a gonosomal numerical aberration . As the name already suggests, it is caused by 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.[1] There may be minor psychosocial problems, such as learning problems.

Clinical picture

The phenotype is **female** . The clinical picture is inconspicuous to normal. The course of puberty is without problems, fertility is generally preserved, but some women with this syndrome tend to have a worse reproductive history, on the basis of which they are indicated for a genetic examination, in which the trisomy of the X chromosome is subsequently diagnosed. The syndrome is not associated with psychomotor retardation, some patients may have delayed speech development and emotional maturation.[2].

ICD-10 classification

Syndrome 47,XXX according to ICD-10 Q97 (https://mkn10.uzis.cz/prohlizec/Q97)

- Q97.0 Karyotype 47,XXX
- Q97.1 Female with more than three X chromosomes

Links

related articles

- Chromosomal abnormalities
- Numerical chromosomal abnormalities
- Syndromes due to aneuploidy of gonosomes
  - Turner syndrome
  - Klinefelter syndrome
  - 47,XYY syndrome

External links

- Triple-X females – AN ORIENTATION (http://www.aaa.dk/TURNER/ENGELSK/TRIPLEX.HTM)
- Triple X Syndrome - Genetics Home Reference

References

1. THOMPSON, James Scott – THOMPSON, Margaret Wilson – NUSSBAUM, Robert L, et al. *Klinická genetika: Thompson & Thompson.* 6. edition. Praha. 2004. 426 pp. ISBN 80-7254-475-6.
2. MUNTAU, Ania. *Pediatrie.* 2. edition. Praha : Grada, 2014. ISBN 978-80-247-4588-6.