

47, XXX syndrome

Chromosome X trisomy, also called the Triple X syndrome (and formerly the „Superfemale syndrome“), belongs in gonosomal numerical aberrations. As the name suggests, it is caused by the 47,XXX karyotype and mosaicism is also possible. Karyotypes 48,XXXX or 49,XXXXX may also occur very rarely and those cases have a different and more severe manifestation. The 47,XXX syndrome itself does not have a very prominent clinical picture, some women are treated for infertility^[1]. Minor psychosocial problems, such as difficulty learning, may occur.

Clinical pictures

The phenotype is **female**. The clinical picture varies from inconspicuous to normal. Puberty is without any problem, fertility is generally preserved, some women with this syndrome tend to have a worse reproductive history, based on which they are indicated for a genetic examination, during which the trisomy of the X chromosome is diagnosed. The syndrome is not connected with psychomotor retardation, some patients may have delayed speech development and emotional development^[2].

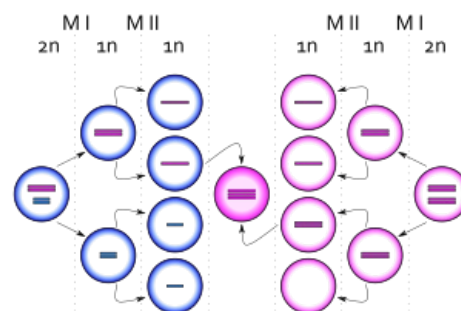


Schéma vzniku syndromu 47,XXX

ICD-10 classification

47,XXX syndrome according to the ICD-10 Template:MKN

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

References

Related articles

- Chromosomal abnormalities
- Numerical chromosomal abnormalities
- Syndromes due to gonosomal aneuploidy
 - Turner syndrome
 - Klinefelter syndrome
 - Syndrome 47,XYY

External links

- Triple-X females – AN ORIENTATION (<http://www.aaa.dk/TURNER/ENGELSK/TRIPLEX.HTM>)
- Triple X syndrome – Genetics Home Reference (<https://ghr.nlm.nih.gov/condition/triple-x-syndrome>)

References

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

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