

# 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.<sup>[1]</sup> 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.<sup>[2]</sup>

## 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

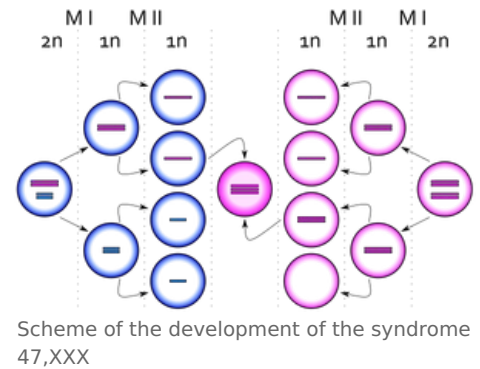
- Chromosomal abnormalities
- Numerical chromosomal abnormalities
- Syndromes due to aneuploidy of gonosomes
  - Turner syndrome
  - Klinefelter syndrome
  - 47,XYY syndrome

### 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. *Pediatric*. 2. edition. Praha : Grada, 2014. ISBN 978-80-247-4588-6.



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