Syndromes due to aneuploidy of gonosomes

Aneuploidy = loss (monosomy) or presence of 1 or more chromosomes in the genome of a cell.

- Causes: division disorder (**nondisjunction**) of homologous chromosomes in the I. maturation division or chromatids in the II. I see division.

Overview of syndromes

Turner syndrome

🔎 *For more information see Turner syndrome.*

- Karyotype **45,X** ;
- incidence of about 1/10,000 girls born;
- diagnosis: already in the prenatal period ultrasound diagnosis is possible - the fetus is affected by extensive **lymphedema** mainly in the neck area, after their resorption the skin folds remain permanently visible - **pterygia coli** ;
- signs: **small height** (100%) – up to 150 cm, wide chest (53%), low hairline on the neck, hypoplasia of nails (60%), pigmented nevi (63%), delayed sexual development, ovaries normally established, gradually however, it turns into **fibrous bands** , mostly **sterility** , absence of secondary sexual characteristics (genital dysgenesis – 100%), puberty disorder;
- exceptionally congenital defects of internal organs;
- frequent mosaics (46,XX/45,X; 45,X/46,XX/47,XXX).

Syndrome XXX (so-called Superfemale)

More detailed information can be found on the XXX Syndrome page .

- Karyotype 47,XXX ;
- incidence of about 1/1,000 girls born;
- signs: **no typical phenotypic manifestations** , non-constant psychomotor retardation, irregular puberty, more frequent spontaneous abortions, limited fertility;
- there is no increased incidence of congenital malformations above the population risk.

Klinefelter syndrome

🔎 *For more information see Klinefelter syndrome.*

- Karyotype **47,XXY** (other variants are also possible, e.g. 48,XXXY);
- incidence of about 1/1,000 male births;
- signs: affected boys usually without problems until adolescence, infertility (100%), eunuchoid features, small rigid testes, azoospermia , in all forms **hypogenitalism** (reduced genitals, 100%), **gynecomastia** (50%), male psychosexual orientation;
- average intelligence, tall stature;
- patients with three or more X chromosomes have more often associated congenital developmental defects of internal organs and more severe mental retardation.

XYY syndrome (so-called Supermale)

🔎 *For more information see XYY syndrome.*

- Karyotype **47,XYY** ;
- incidence of about 1/1,000 male births;
- signs: body height **over 180 cm** , previously considered increased aggressiveness was not confirmed, sometimes mild mental retardation, slightly limited fertility.

Links

Related articles

- Syndromes due to aneuploidy of autosomes
- Chromosomal aberrations
- Numerical chromosomal aberrations
- Indications for karyotype examination
- Clinical genetics

Source

- ŠTEFÁNEK, Jiří. *Medicína, nemoci, studium na 1. LF UK* [online]. [cit. 11. 2. 2010]. <http://www.stefajir.cz>.