Gonosomal aneuploidy syndromes in man

Aneuploidy – change in chromosome number by less than a complete set, usually ± 1 chromosome

Trisomy – three copies of sex chromosomes

Monosomy – one copy of chromosome; monosomy of chromosome X is only monosomy viable in man

Clinically important syndromes with gonosomal aneuploidy in man

Turner syndrome

- monosomy of chromosome X; karyotype 45,X
- Frequency 1:2 000-2 500
- Phenotype (female):
  - short stature, broad/shield chest, underdeveloped breast, hygroma colli cysticum (usually detected by ultrasound prenatally), short webbed neck (pterygium colli), palms and feet oedemas (newborns), low posterior hairline

gonadal dysgenesis (rudimentary fibrous ovaries), primary amenorrhoea, average intelligence (not retarded), but frequently learning difficulties

- Therapy: hormonal therapy – growth hormone (height) and sex hormones (sexual features); in vitro fertlilization with oocyte donor
- Life expectancy: usually not limited (more than 90 % of fetuses do not survive, monosomy X is most frequent finding in spontaneous abortions

Klinefelter syndrome

- karyotype 47,XXY
- Frequency 1:500-1 000
- Phenotype (male):
  - tall stature, sterility – azoospermia, testicular atrophy, cryptorchism, female pubic hair pattern, poor beard growth, female type of fat distribution, gynaecomastia, average intelligence (not retarded), male psychosexual orientation

XXX syndrome („superfemale")

- trisomy X; karyotype 47,XXX
- Frequency 1:1 000
- Phenotype:
  - no specific phenotype, average intelligence, normal sexual development, decreased fertility (spontaneous abortions), without higher risk of chromosomal aberrations in offspring, no increased occurrence of congenital disorders

XYY syndrome („supermale")

- karyotype 47,XYY
- Frequency 1:1 000
- Phenotype:
  - „robust" growth (proportional), especially height, average intelligence, normal sexual development, normal fertility, without higher risk of chromosomal aberrations in offspring, controversial hypothesis - affected psychosocial development