Autosomal aneuploidy syndromes in man

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

Trisomy – three copies of particular chromosome

Monosomy – one copy of particular chromosome; no autosomal monosomy is viable in man

Clinically important (viable) syndromes with autosomal trisomy in man

Down syndrome

- trisomy of chromosome 21; karyotype 47,XX,+21 (girl) or 47,XY,+21 (boy)
- Frequency 1:600-800 (due to effective prenatal diagnostics the frequency is decreased)
- Phenotype:
  - constant symptoms - hypotonia in newborns, mental retardation
  - variable symptoms - upslanting palpebral fissures, flat face, neck webbing, dysplasia of ears, flat occiput, single palmar crease, epicanthus, macroglossia, short and broad hands, brachydactyly, male hypogenitalism
  - Life expectancy is limited variably by: congenital heart defects, defects of other organs, immune system defects, Alzheimer disease, leukemia

Patau syndrome

- trisomy of chromosome 13; karyotype 47,XX,+13 or 47,XY,+13
- Frequency 1:15 000-20 000
- Phenotype:
  - severe developmental retardation, congenital heart defects, urogenital defects, microcephaly, craniosynostosis, CNS malformations, microphtalmia (rarely cyclopia), cleft palate, cleft lip, malformed low-set ears, polydactyly
  - Life expectancy: approx. 1 month

Edwards syndrome

- trisomy of chromosome 18; karyotype 47,XX,+18 or 47,XY,+18
- Frequency 1:5 000-10 000
- Phenotype:
  - severe developmental retardation, failure to thrive, congenital heart defects, kidney defects, malformed low-set ears, hypoplastic nails, digits overlapping, micrognathia, prominent occiput, pedes equinovares (clubfoot), microcephaly
  - Life expectancy: approx. 1 year

Prenatal screening and diagnostics (see questions No. 127 - Prenatal screening of inborn errors of development; No. 129 - Prenatal diagnostics of chromosomal aberrations, possibilities of prevention; No. 130 - Prenatal diagnostics of inborn errors of development, possibilities of prevention)