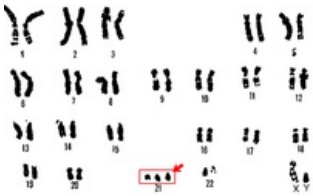
Syndromes due to aneuploidy of autosomes

Aneuploidy refers to the loss (monosomy) or presence of (one or more) chromosomes in the genome of a cell. **The cause** may be a division disorder (nondisjunction) of homologous chromosomes in the I. maturation division or chromatids in the II. I see division.


Down syndrome karyotype

Down's syndrome

We find the karyotype **47,XX or XY + 21** (trisomy of chromosome 21). Clinically described for the first time by Dr. by Langdon Down (1866), but molecularly by Lejeun (1959)

- **incidence** : 1:600 – 1:800 births
- **significant dependence on the age of the mother** at the time of delivery
- 65-70% of cases are detected prenatally in the Czech Republic and the number of unterminated pregnancies is increasing

Clinical picture

- pronounced muscle **hypotonia** in newborns
- brachycephaly
- broad, flat face, slanted eye slits, **epicanths** (skin folds, bridging the inner corner of the eye), broad nasal bridge, large tongue, and dental anomalies
- 50% of individuals **palmar** (monkey, four-fingered) **grooves** on the palms (in the normal population 2-3%)
- **fingers** are short and wide, little fingers are roll-shaped (clinodactyly) with a short middle link
- a wide **gap** between the first and second toes
- smaller figure, around **150 cm**
- psychomotor retardation of varying degrees
- IQ is usually in the range of **25-75** , on average 40-45
- socially **adaptable** and thanks to better health care, they live to be 50-60 years old (ability to treat infections, cancer, congenital heart defects and developmental defects of other organs - they are **prone to them** and can be operated on today)
- in most adults later in life **Alzheimer's disease** - third dose effect of gene on chromosome 21 - gene for **amyloid precursor protein**
- **fertility** greatly reduced, especially in men
- **cases of pregnancy** more frequent in women , almost zero fertility in men
- the risk of **the offspring** of parents with Down syndrome being affected is difficult to determine due to the number, but there are also cases of healthy children

Types of Down syndrome

- **simple trisomy** of chromosome 21 (95%)

- **translocation form** of trisomy 21 (4-5%)

- trisomic and normal line **mosaicism** (1%) – individuals generally have milder manifestations of Down syndrome

🔎 *For more information see Down Syndrome.*

Edwards syndrome

Karyotype: **47,XX or XY + 18** (trisomy of chromosome 18). The prognosis of individuals with **trisomy 18** is poor, they rarely live for a few months to a year. 80% of affected individuals are **female** , boys generally have a lower chance of survival

- **incidence**: approximately 1:5000 live births

- directly **related to the age of the mother**
- most fetuses conceived in this way are spontaneously **aborted**

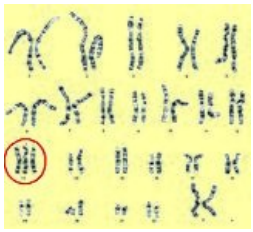
Clinical picture

- severe **psychomotor retardation** and failure to thrive
- congenital developmental defects **of the kidneys and heart**
- microcephaly and prominant headache
- receding chin, low-set malformed acorns

- the characteristic **holding of the fingers** in closed fists with the second finger crossed over the third and the fifth over the fourth
- nails tend to be **hypoplastic**

🔎 *For more information see Edwards syndrome.*

Patau syndrome



Karyotype in Patau syndrome

The karyotype is **47,XX or XY + 13** (trisomy of chromosome 13). It has the worst prognosis of the **three syndromes** – survival 1 month after birth at most.

- rarer occurrence – 1:10,000

Clinical picture

- severe mental and growth retardation
- severe CNS malformations, **holoprosencephaly**
- malformations of the eyes, which can merge into one or be completely absent
- craniofacial dysmorphia with severe cleft palate and lip
- earlobes malformed and low-set
- **congenital heart and urogenita**l defects are present
- postaxial polydactyly of the upper and lower limbs

🔎 *For more information see Patau Syndrome.*

Links

Related articles

- Syndromes due to aneuploidy of gonosomes
- 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>.