

# Indications for chromosome analysis in clinical genetics

## Chromosomal analysis

- prenatal
- postnatal

## Indications for prenatal analysis of chromosomes

- advanced maternal age
- higher than 35 ys in the date of delivery (increased risk of chromosomal aneuploidies, namely M. Down)
- positive screening of congenital anomalies (1st or 2nd trimester maternal serum screening test + increased NT - higher risk of chromosomal abnormalities)
- positive family history
- affected child/fetus in previous pregnancy (with chromosomal abnormality)
- parent - carrier of balanced chromosomal aberration (e.g. translocation)
- pathological or atypical ultrasound finding (IUGR - intrauterine growth retardation, microcephaly, hyperechogenic bowel, hydronephrosis, Fallot tetralogy, club foot, polyhydramnios/oligohydramnios, hygroma colli cysticum,...)
- important for differential diagnosis (ultrasound finding could be solitary - usually without chromosomal abnormality or syndromologic - in many cases caused by chromosomal abnormality)
- others (e.g. mother after chemotherapy, in vitro fertilisation pregnancy, ...)

## Indications for postnatal analysis of chromosomes

1. children:
  - craniofacial dysmorphism (flat occiput, epicanthus, hypertelorism, cleft lip, cleft palate, malformed ears, craniosynostosis, macroglossia,...)
  - congenital anomalies (heart defects, cryptorchism, NTD - neural tube defects, urogenital defects, agenesis corpus callosum,...)
  - psychomotoric retardation
  - developmental delay, failure to thrive
  - growth retardation, short stature
  - hypotonia
2. puberty:
  - amenorrhoea
  - gynecomastia
  - developmental defects of secondary sexual features
3. adults:
  - infertility/sterility
  - recurrent spontaneous abortions
  - abnormal spermiogram in men
  - positive family history (reproduction loss, affected child, chromosomal aberration in relatives,...)
  - gamete donors