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)
- patological 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 dysmorphy (flat occiput, epicantus, 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:
   - amenorhoea
   - 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