

Methods of chromosomal examination

- **Karyotyping** (see question No. 33 - Human karyotype, methods of its examination)
- **Analysis of acquired chromosomal aberrations:**
 - conventional staining of chromosomes with *Giemsa dye solution only* - detection of chromosome breakage results (acentric fragments, double minutes, dicentric chromosomes, ring chromosomes, chromosome breaks, chromatid breaks,...)
 - **SCE** (sister chromatid exchange) – using of BrdU during cultivation, differential staining of sister chromatids
- **FISH** (fluorescent in situ hybridization) (see question No. 32 – Molecular cytogenetics)

Microarrays (see question No. 32 – Molecular cytogenetics)

- **NIPT** (non-invasive prenatal testing):
 - analysis of cell-free DNA (cfDNA) fragments of fetus in maternal circulation (test performed from maternal blood sampling), used as a screening method to search primarily for chromosomal aneuploidies, sex of the fetus or Rhesus blood type
 - different modifications include also detection of microdeletions or even whole karyotype scan
 - 10 weeks of pregnancy or later
 - methods of analysis are different (different companies) – based on PCR, CGH-arrays or next generation sequencing