Indications for chromosomal examination

Chromosomal examination is one of the basic methods of clinical genetics. The main goal of this examination is to rule out numerical or structural chromosomal aberrations in the patient. If necessary, it can be a karyotype examination using standard banding methods, or one of the methods of molecular cytogenetics (FISH, etc.). Examination is indicated by a clinical geneticist (often in the framework of interdisciplinary cooperation - for example with paediatricians, gynecologists or obstetricians), for many different reasons and within the framework of different diagnostic programs.

List of indications

A karyotype examination may be indicated in the context of:

1. **Prenatal diagnostics**: The karyotype of the unborn fetus is examined. Material for cytogenetic examination needs to be obtained by some invasive method of prenatal diagnosis (amniocentesis, CVS, cordocentesis). The main indications are:
   - Results indicating an **increased risk of congenital chromosomal aberration** (CCA) - positive screening (biochemical or ultrasound) I. or II. trimester, or a positive result of the integrated screening (I. + II. trimester evaluated together).
   - Pregnancy in a woman **over the age of 35** (no longer an absolute indication, however, pregnant women over the age of 35 have the right to perform invasive prenatal diagnostics even without the presence of other risk factors).
   - Pregnancy with an a priori **higher risk of congenital chromosomal aberration** (chromosomal aberration or developmental defects in the family history; one of the parents is a carrier of a balanced chromosomal aberration; one of the parents after oncological treatment – cytostatics, radiation, etc.).
   - Certain cases of high-risk pregnancy - for example, pregnancy after **assisted reproduction**.
   - Pregnancies, the course of which could have been disturbed by serious **factors of the external environment** with a **clastogenic effect** (for example, conditions after exposure to ionizing radiation).
   - Today, a more historical indication is fetal karyotyping in order to determine the **gender of the fetus** in the case of a family occurrence of a serious genetic disease linked to sex (currently, DNA diagnosis is possible in most cases).
2. **Postnatal diagnostics:** The karyotype of a born individual (whether a child or an adult) is examined. The material for examination is most often peripheral blood (leukocytes) or (rarely) skin fibroblasts. The main indications include:
   - Examination of a newborn or child in case of **suspected chromosomal aberration** (phenotype corresponding to one of the typical syndromes, multiple congenital developmental defects, psychomotor or mental retardation, etc.). Short stature/failure to thrive in girls (to rule out Turner syndrome).
   - Examination of the karyotype of the parents, if the prenatal diagnosis of the fetus or the postnatal diagnosis of the child indicated a **congenital chromosomal aberration**. Furthermore, in the case of **repeated spontaneous abortions**, or in the event of frequent abortions or chromosomal aberrations in the family history.
   - Persons with **disorders of sexual development** (primary amenorrhea, etc.), persons (children) of clinically indeterminate or mixed sex.
   - Complex examination of **infertility** in an infertile couple; disorders of spermatogenesis in men.
   - Examination of the karyotype in **oocyte donors and sperm donors**.
   - Examination of the karyotype of cells of **solid tumors** or blood elements in **hemato-oncological diseases**. Cytogenetic examination in oncology has diagnostic and prognostic significance (and can also decide on appropriate therapy).
   - Examination of **acquired chromosomal aberrations** (ZCA) in persons exposed to clastogens in (for example, work) environment (various chemicals, ionizing radiation).
3. **Pre-implantation diagnostics**: Pre-implantation genetic diagnostics (performed within the In Vitro Fertilization program) is rarely used in the case of a *high risk of congenital chromosomal aberrations* (especially in persons with a balanced chromosomal aberration, who are at real risk of developing an unbalanced aberration in the offspring). The material for examination is most often the blastomeres of the developing embryo.

Links

Related articles

- Chromosomal aberrations
- Clinical genetics
- Prenatal diagnosis
- Family history

Source

- ŠÍPEK, Antonín, et al. *Vrozené vývojové vady - Důvody k vyšetření karyotypu* [online]. ©2008-2010. [cit. 2009-10-09]. <http://www.vrozene-vady.cz/vrozene-vady/index.php?co=indikace_k_vysetreni_karyotypu>.