Detection and prevention of congenital chromosomal abnormalities

Congenital chromosomal abnormalities are diseases caused by a change in the number of chromosomes (genomic abnormalities) or a change in the structure of a chromosome (chromosome abnormalities). Innate means that they are present in all cells of the organism and can be transmitted between generations. They are also called gametic. Incidence estimates are estimated at 5–8 cases per 1000 births.

Typical chromosomal aberrations

1. Numerical aberrations of autosomes: Down syndrome (trisomy 21), Edwards syndrome (trisomy 18), Patau syndrome (trisomy 13);
2. numerical aberrations of gonosomess: Turner syndrome (45,X), Klinefelter syndrome (47,XXY), 47,XXX syndrome, 47,XYY syndrome.

Capture

A very high number of chromosomal abnormalities affect the psychological and intellectual development of an individual, which is why prenatal detection plays an important role. Seizures began to a greater extent already in the 1960s. The main means was amniocentesis. 10 years later chorionic villus sampling became more widely used. In the future, less invasive examinations will be used - determination of maternal alpha-fetoprotein (AFP), hCG or estradiol levels, more recently also PAPP-A. A detailed ultrasound examination focused on specific markers of chromosome aberrations also plays an increasingly important role. cytogenetic examination plays a key role in confirming the diagnosis of chromosomal aberration of the fetus.

Comprehensive counseling and proposals for the investigation process are dealt with within genetic consultation. The means of examination are genetic tests.

Risk factors and warning signs

- Abnormal screening results – USG, biochemistry;
- pregnancy at an older age (over 35);
- infectious and non-infectious diseases of pregnant women;
- use of drugs or exposure to chemicals;
- parental couples with a positive family history;
- repeated spontaneous abortions;
- fertility disorders.

Prevention

Prevention of congenital chromosomal abnormalities is somewhat tricky. It is often a genetic disease where the chromosomes are inherited from the parents. External influences tend not to play such a significant role. Nevertheless, for example, an increased intake of folic acid is recommended for pregnant women. It mainly works against disorders of the development of the neural tube.

However, the screening of parents is more important. The examination is carried out from blood samples using a cytogenetic examination (*secondary prevention*). Elderly women are especially at increased risk. A mother's age over 35 is already considered risky. *Primary prevention* is the limitation of contact with potentially pathogenic factors – biological, chemical, physical.

Links

Related Articles

- Chromosomal Abnormalities
- Cytogenetic examination
- Amniocentesis
- Chorio villus collection
- AFP
- hCG

Source

- AYME, S. Prevention of chromosomal anomalies. *European Journal of Epidemiology* [online]. 1991, vol. 7, no. 6, p. 715-736, Available from <https://link.springer.com/article/10.1007%2FBF00218692>. ISSN 1573-7284.

External links

- Vrozene-vady.cz (http://www.vrozene-vady.cz)