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Prenatal Diagnosis

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Abstract: Prenatal diagnosis of congenital anomalies and hereditary human diseases is a new direction in clinical and medical genetics. The purpose of prenatal diagnosis is the diagnosis, prevention and intrauterine treatment of human hereditary diseases and congenital anomalies. Modern prenatal diagnostics has a complex of human genetic methods that make it possible to detect congenital pathology in the fetus at any stage of pregnancy.

Keywords: Prenatal diagnosis, congenital anomalies, genetic methods, chromosomal diseases, gene diseases, pregnancy, prevention.

Prenatal diagnosis of congenital anomalies and hereditary human diseases is a new direction in clinical and medical genetics, which arose at the intersection of clinical disciplines (obstetrics, gynecology, perinatology, neonatology) and fundamental sciences (biology, genetics, biochemistry, pathological physiology, teratology, embryology, molecular biology and also molecular genetics). The purpose of prenatal diagnosis is the diagnosis, prevention and intrauterine treatment of human hereditary diseases and congenital anomalies. Modern prenatal diagnostics has a complex of human genetic methods that make it possible to detect congenital pathology in the fetus at any stage of pregnancy. The choice of prenatal diagnostic method depends on the purpose, duration and characteristics of the course of pregnancy, the condition of the pregnant woman, and the technical capabilities of the medical genetic laboratory of the prenatal center. Each prenatal diagnostic method used has its own indications, contraindications, resolution options, and complications after the procedure.

Since 2019, by decree of the President of the Republic of Uzbekistan, district, city, regional and republican prenatal centers have been organized in all districts, regional centers and the capital.

In the programs being developed by prenatal centers to reduce perinatal morbidity and mortality, and prevent congenital pathologies, the leading place belongs to prenatal diagnosis, along with medical genetic counseling and mass screening for inborn errors of metabolism. To make an antenatal diagnosis of chromosomal diseases, gross anatomical malformations, metabolic disorders in the fetus, ultrasound scanning of the fetus, fetoscopy, chorion biopsy with subsequent examination of native or cultured chorionic villous cells, amniocentesis with further cytological, cytogenetic, biochemical, molecular genetic studies of amniotic tissue are used. cells or biochemical, immunochemical studies of amniotic fluid.

Prenatal diagnosis includes two stages:

Stage I - identification of families with a high risk of having children with congenital pathologies during medical genetic counseling or examination of a pregnant woman in a antenatal clinic.

Stage II is the actual prenatal diagnosis.

Main indications for prenatal diagnosis:

- mother's age is over 35 years, father's age is over 45 years;
- structural chromosomal abnormalities or congenital malformations in parents;
- consanguineous marriage between spouses;
- pathology in the previous child (chromosomal abnormality, congenital malformations, mental retardation); - the presence in the family of a clearly established hereditary disease (recessive, X-linked diseases, heterozygous carriage of a chromosomal abnormality);
- the presence of metabolic disorders in the family (lipidosis, mucopolysaccharidosis, galactosemia, abnormalities in the metabolism of amino acids - phenylketonuria, alkoptonuria, abnormalities in the metabolism of purine and pyrimidine bases);
- hematological disorders (thalassemia, chronic granulomatosis, hemophilia, incompatibility of the Rh factor and ABO systems, hemolytic anemia of the newborn);
- determination of lung maturity in the fetus; congenital malformations of the fetus identified by ultrasound examination;
- changes in indicators of screening factors (alpha-fetoprotein, choriogonic gonadotropin, estriol, 17-hydroxyprogesterone, PAPP-A) in high-risk pregnant women.

Relative indications for prenatal diagnosis:

- habitual miscarriage of unknown origin;
- complicated course of this pregnancy (long-term threat of miscarriage, high and low water levels, fetal growth retardation syndrome);
- exposure to adverse factors on the pregnant woman's body (carrying out diagnostic or therapeutic R-procedures, taking medications, infectious diseases).

Prenatal diagnostic methods

A. Direct (fetal) methods:

- ✓ R - graph of the skeleton or soft tissues of the fetus (amniography, fetography).
- ✓ Ultrasound examination (ultrasound).
- ✓ Electrocardiography (ECG), phonocardiography (PCG), cardiotocography (CTG).
- ✓ Electrocephalography (EEG).
- ✓ Fetoscopy.
- ✓ Amnioscopy.
- ✓ Amniocentesis.
- ✓ Biopsy of fetal tissue (amnion, chorion, skin and fetal blood).

B. Indirect or indirect (maternal) methods:

- ✓ Obstetric and gynecological examination.
- ✓ Medical genetics (genealogical, cytogenetic, molecular biological).
- ✓ Bacteriological, immunological, serological examination.
- ✓ Biochemical examination (screening tests for blood levels of alpha-fetoprotein, estradiol, betachoriogonic gonadotropin, PAPP-A, etc.).

B. Additional methods:

- ✓ Electron microscopy of fetal tissue.

- ✓ Fluorescent quantitative in situ PCR (QF-PCR, PRINS).
- ✓ Fluorescent in situ hybridization (FISH).

Indirect methods are aimed at examining pregnant women in order to select high-risk patients who require additional examinations. With direct PD methods, the object of study is the fetus itself, amniotic fluid, chorionic villus biopsies, placenta, and umbilical cord blood. Direct methods are divided into non-invasive (ultrasound, Doppler, CTG, EEG, PCG) and invasive (amniocentesis, cordocentesis, chorionic villus biopsy). A set of non-invasive methods makes it possible to further determine the group of pregnant women for whom invasive PD is indicated.

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