

# PRENATAL METHODS OF DIAGNOSING DOWN SYNDROME IN THE REPUBLIC OF BELARUS

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Down syndrome is the most common genetic pathology that causes characteristic mental and physical abnormalities in children with the condition.

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Down syndrome is a chromosomal disorder caused by abnormal cell division resulting in an extra full or partial copy of the 21 chromosomes

The disease occurs due to chromosome misalignment in the first meiotic division (66% of cases in the mother, 14% in the father), as well as due to chromosome misalignment in the second meiotic division. The main complex of pathology is associated with segment 22 of the long arm of the 21st chromosome [2].

According to the World Health Organisation recommendations, the following diagnostic methods are used:

Non-invasive prenatal testing - detects the presence of chromosome number abnormalities (aneuploidy) in the foetus. The study is possible from 9 weeks of singleton pregnancy. The technique is based on the quantitative analysis of DNA, carried out by mass parallel full-genome sequencing, which gives the most complete information about the genetic changes encountered.

Screening of maternal serum factors: Alpha-fetoprotein, an increased amount of which in the blood may indicate congenital malformations; chorionic gonadotropin (deviation from the norm indicates chromosomal abnormalities, fetal abnormalities in development); unconjugated estriol, an increased level of which indicates chromosomal abnormalities of the fetus. It is performed at 15-20 weeks of gestation.

Amniocentesis with subsequent examination of amniotic fluid - puncture of the amniotic membrane of the amniotic bladder for the purpose of taking amniotic water and subsequent analysis of fetal cells. The term is 16-19 weeks of gestation.

Chorion and placenta biopsy is a method of taking chorion villi and then counting the number of chromosomes in its cells. Timing is 9-11 weeks of gestation.

Cordocentesis is an examination of umbilical cord blood from the 18th week of gestation. The most accurate method of diagnosis.

The use of modern methods of prenatal diagnostics makes it possible to diagnose pathology at early stages of gestation, which makes it possible to make a timely decision on foetal elimination.

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