THE SIGNIFICANCE OF AMNIOCENTSIS IN THE PREVENTION OF CHROMOSOME-RELATED GENE DISEASES

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Annotation
The purpose of the study: to determine the most effective type of tests used to detect a fetus with chromosomal disorders in the womb. Research materials and methods: the test results of 45 patients who submitted the amniocentesis test to the "Genotechnology" laboratory, 42 Kokcha Darvoza Street, Shaikhontohur District, were studied. We collected information about other methods of amniocentesis method (FISH) in Tashkent Medical Academy Multidisciplinary Clinic 14 - educational building. Analysis and discussion of the results: in 45 patients, the amniocentesis method made a very accurate diagnosis. In the analysis of the results, 10 out of 45 mothers (16-21 weeks pregnant, average age 35.3) gave birth to a child with Edwards syndrome, although they passed the test through maternal blood and received a healthy result. 20 mothers who donated blood 3 times did not provide complete information each time. The remaining 15 mothers had a false abortion, that is, the fetus was aborted even though it was healthy, or it was given information about its health, but it was born with Down's syndrome. Later, these expectant mothers resorted to the method of amniocentesis. 3 of these 45 pregnant mothers had Down's syndrome, 2 had Edward's syndrome, and it was left to the discretion of the family to have a medical abortion or save the child's life. The rest of the patients had healthy fetuses.

Key words: Amniocentesis method, chromosomal diseases, FISH diagnostic method, perinatal biochemical screening test

Relevance: About 2,000 hereditary diseases caused by changes in the number and structure of chromosomes are currently known. Hereditary chromosomal diseases can occur due to changes in the number and structure of autosomes and sex chromosomes, and there is no cure for chromosomal diseases. Chromosomal diseases are a large group of congenital hereditary diseases, which are clinically characterized by the presence of several defects, and as an etiological basis, they have numerical or structural abnormalities of chromosomes.

In 1866, the English pediatrician Down described a specific type of mental retardation in children, which was later called Down syndrome. Shereshevsky-Turner and Klinefelter syndromes were identified in the first half of the 20th century. Study of chromosomal disorders In 1959, the presence of an additional 21+1 chromosome in the karyotype of patients with Down syndrome began to be actively identified. To date, about 2,000 different chromosomal abnormalities have been described in humans. There are about 100 syndromes for clinical diagnosis. The prevalence of chromosomal disorders is the same in all national and ethnic groups, with an average of 7-8 patients per 1000 newborns.

Chromosomal syndromes are usually sporadic, i.e. only one such patient was registered in the family. This is because most chromosomal changes occur as a result of random mutation. Such mutations occur more often during oogenesis than during spermatogenesis. It depends on the age of the woman, especially if she is over 35 years old, the risk of genetic mutations in germ cells increases significantly.

Chromosomal disorders include Klinefelter syndrome (XXY), Shereshevsky-Turner syndrome (XO), Down syndrome (21+1), Patau syndrome (13+1), Edwards syndrome (18+1), and others.

Chromosomal disorders can be detected using the amniocentesis method. In particular, the US study included all patients who underwent amniocentesis between January 2012 and June 2017. Maternal age, amniocentesis performance, gestational age at amniocentesis, karyotyping results, complications during and after the procedure, and fetal outcomes were reviewed and analyzed. Results The medical records of 114 patients were reviewed, and the majority of patients (50.9%) were between 30 and 39 years old, with an older maternal age (9.6%), with a mean age of 34.29 years. In the older age of the mother, polyhydramnios (7.9%), risk of Down syndrome (31.6%), risk of Patau syndrome (6.1%), increased risk of Edward's syndrome (4.4%). No complications were observed in patients with normal maternal age

(86.0%). Conclusion This 5-year retrospective study of the amniocentesis procedure showed that the majority of amniocentesis was safe, as 86.0% of patients were free of any complications. It is important to be aware of its complication in advance, because even though it is a generally safe procedure, there is always a risk. If the amniocentesis method is widely used in clinics, the birth of disabled children will decrease dramatically. According to statistics, the number of disabled people in the world, their share in the world population is more than 1 billion, which is 15%. The number of disabled people in Uzbekistan is about 760,000. About 120,000 of them are disabled children aged 18 and under.

Test material and methods: Amniocentesis is a (safe) process of taking approximately 20 ml of amniotic fluid from the amniotic fluid using a special method between 16-20 weeks. The procedure was performed under local anesthesia under ultrasound control. After the examination, the set of chromosomes of cells belonging to the fetus from the amniocyte cells in the amniotic fluid was checked in terms of quantity and quality. If a chromosomal abnormality is detected, the situation will be explained in detail to the family. The decision to give birth or abort a child is at the discretion of the family.
Amniocentesis was checked with the most modern cytogenetic testing method, FISH, with an accuracy of 99% or higher.

Indication for amniocentesis (when used): When the risk of homosomic diseases (Down syndrome, Pattau Shershevsky terner Kleifelter syndrome) is high in the results of perinatal biochemical screening. In pregnant women aged 35 and older. When one of their previous children had chromosomal disorders. In repeated abortions. In cases where there is or is suspected a hereditary or congenital disease in the offspring. In the ultrasound examination, the nasal bone is smaller than the norm or the neck fold is higher than the norm and other anomalies. Excess of amniotic fluid.

Contraindications to amniocentesis (when not used): Presence of an acute inflammatory process. Exacerbation of chronic diseases. Strong hypertonus of the uterus. Bleeding from the cervix. Rhesus imbalance (in some cases).

How long does it take to report results?
The results of the test can be known with high accuracy within 4-5 days using the rapid FISH (fluorescence in situ hybridization) testing method. It is a modern cytogenetic examination method that allows to identify qualitative and quantitative changes in chromosomes (including translocations and microdeletions) and is used for the diagnosis of chromosomal diseases.

How reliable are the amniocentesis results?

Amniocentesis is 99% accurate and if the chromosomal test results are "normal", the chances of the test being incorrect are very low.

Analysis and discussion of the results: In the analysis of the results, 10 of 45 mothers (16-21 weeks pregnant, average age 35.3) gave birth to a child with Edwards syndrome, although they passed a blood test and received a healthy result. 20 mothers who donated blood 3 times did not provide complete information each time. The remaining 15 mothers had a false abortion, that is, the fetus was aborted even though it was healthy, or it was given information about its health, but it was born with Down's syndrome. Later, these expectant mothers resorted to the method of amniocentesis. The method of amniocentesis, unlike other tests, made a diagnosis with great accuracy. Among these 45 expectant mothers, 3 had Down's syndrome, 2 had Edward's syndrome, and it was left to the discretion of the family to perform a medical abortion or save the child's life. The rest of the patients had healthy fetuses. Conclusion: Increasing the number of genetic counseling centers and perinatal screening centers. Reducing the cost of amniocentesis (current price 3.5 million) and gradually introducing it free of charge in family polyclinics. To study all types of amniocentesis method based on foreign models and to use it in our medicine. Explaining complete and accurate information about all types of amniocentesis to pregnant women and directing them to amniocentesis. As a result, children born with chromosomal disease can be prevented. It is necessary to save healthy children who are subjected to incorrect medical abortion as a result of tests with a low level of accuracy.

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