RESEARCH JOURNAL OF TRAUMA AND DISABILITY STUDIES

Volume: 01 Issue: 11 | Nov – 2022 **ISSN: 2720-6866**
http://journals.academiczone.net/index.php/rjtds

Ultrasound Prenatal Diagnosis of Chromosomal Anomalies in the First and Second Trimester of Pregnancy

Ibotova Mahfuza Oybekovna

Assistant of the Department of General Hygiene and Ecology

Abstract

On ultrasound in the second trimester of pregnancy, as well as in the first trimester, each chromosomal disorder has specific syndromic features. In this regard, if any anomaly/marker is detected in the fetus during a routine ultrasound examination, a detailed search for other manifestations characteristic of this chromosomal pathology should be carried out, since if other markers or malformations are detected, the individual risk of this pathology increases significantly. When an isolated anomaly is detected in the fetus, the decision to perform an invasive intervention depends on the nature of this anomaly.

Keywords: ultrasound prenatal diagnosis, chromosomal abnormalities, prenatal diagnosis, second trimester.

Ultrasound examination in the second trimester of pregnancy: phenotypic signs of chromosomal diseases Trisomy 21 is characterized by: hypoplasia of the nasal bones, an increase in the thickness of the cervical fold, heart defects, hyperechoic focus in one of the ventricles of the heart, duodenal atresia, hyperechoic intestine, hydronephrosis, shortening of the length of the femoral and brachial bones, sandal gap of the foot, clinodactyly or hypoplasia of the middle phalanx of the little toe. Trisomy 18 is characterized by: strawberry-shaped head, CCC, agenesis of the corpus callosum, enlargement of the large cisterna, cleft lip and palate, micrognathia, increased thickness of the cervical fold, heart defects, diaphragmatic hernia, esophageal atresia, omphalocele (usually containing only intestinal loops) , CAP, renal anomalies, hyperechoic bowel, meningomyelocele, IUGR and limb shortening, aplasia of the radius, crossed toes, clubfoot or rocking foot. Trisomy 13 is characterized by: holoprosencephaly, microcephaly, malformations of the facial skull, heart defects, enlarged and hyperechoic kidneys, omphalocele, and postaxial polydactyly.

Triploidy is characterized by: with an additional set of chromosomes of paternal origin, the presence of an altered placenta by the type of hydatidiform mole, while pregnancy, as a rule, is interrupted before 20 weeks. With an additional set of chromosomes of maternal origin, pregnancy can progress into the third trimester. The placenta is thin, although it has a normal structure, which is combined

Volume: 01 Issue: 11 | Nov – 2022 ISSN: 2720-6866
http://journals.academiczone.net/index.php/rjtds

with a pronounced asymmetric slowdown in fetal development. Typical findings are mild ventriculomegaly, micrognathia, heart defects, meningomyelocele, syndactyly, and abducted thumb. Turner's syndrome is characterized by: severe cystic hygroma of the neck, generalized edema, pleural effusion, ascites, heart defects, horseshoe kidney, which can be suspected when bilateral moderate hydronephrosis is detected.

VENtriculomegaly The incidence of ventriculomegaly in newborns is 1 in 1000. The causes of ventriculomegaly are fetal chromosomal and gene diseases, hemorrhages and infectious brain lesions, but in most cases the etiology of ventriculomegaly remains unknown. The average incidence of chromosomal abnormalities in ventriculomegaly is 10%, with trisomies 21, 18, 13 and triploidy being the most common. As a rule, chromosomal disorders are more often observed in mild and moderate than in severe ventriculomegaly.

HOLOPROSENCEPHALY The incidence of holoprosencephaly in newborns is approximately 1 in 10,000. Although chromosomal or gene disorders are common causes of holoprosencephaly, in most cases the etiology of holoprosencephaly remains unclear. The frequency of occurrence of chromosomal disorders in holoprosencephaly is 30%, and the most common of these are trisomies 13 and 18. Holoprosencephaly is often associated with anomalies in the development of the midline structures of the face, however, the frequency of chromosomal anomalies increases only in those fetuses in which holoprosencephaly is combined with anomalies in the development of other structures fetus, and not in those in which holoprosencephaly is isolated or combined only with facial anomalies.

Vascular plexus cysts Choroid plexus cysts are detected in 2% of fetuses at 16–24 weeks of gestation, but in more than 95% of cases they spontaneously disappear by the 28th week of pregnancy, without any clinically significant consequences. There is a relationship between the detection of choroid plexus cysts and the presence of chromosomal disorders in the fetus, mainly trisomy 18. However, in most cases in the presence of trisomy 18, the fetus has other multiple developmental anomalies, so the detection of choroid plexus cysts should lead the researcher to exclude other possible manifestations of this trisomy . If choroid plexus cysts are the only finding, then the risk of trisomy 18 in the fetus increases slightly.

DANDY-WALKER SYNDROME Dandy-Walker syndrome includes a wide range of anomalies in the development of the cerebellar vermis, enlargement of the fourth ventricle, and enlargement of the large cisternae. There are Dandy-Walker anomaly (partial or complete agenesis of the cerebellum in combination with a posterior fossa cyst), Dandy-Walker variant (partial agenesis of the cerebellar vermis without enlargement of the large cisterna) and enlargement of the large cisterna (in the absence of agenesis of the cerebellar vermis or expansion of the fourth ventricle). The incidence of Dandy-Walker syndrome in newborns is 1 in 30,000. The causes of its occurrence include

Volume: 01 Issue: 11 | Nov – 2022 ISSN: 2720-6866
http://journals.academiczone.net/index.php/rjtds

chromosomal abnormalities, more than 50 genetic syndromes, congenital infections of the fetus, exposure to teratogens such as warfarin, but in some cases its etiology remains unknown. The overall incidence of chromosomal abnormalities in the presence of Dandy-Walker syndrome is 40%, and the most common are trisomies 13 and 18, as well as triploidy.

Diaphragmatic hernia The incidence of diaphragmatic hernia in newborns is 1 in 4000, and in most cases this malformation occurs sporadically. The incidence of fetal chromosomal diseases (mainly trisomy 18) in the presence of diaphragmatic hernia in the fetus is 20%.

ANOMALIES OF DEVELOPMENT OF THE HEART Anomalies of the development of the heart and great vessels occur in 4-7 per 1000 live births and in 30 per 1000 stillbirths. The etiology of cardiac anomalies is very diverse and is most likely the result of a combination of genetic factors and the impact on the fetus of adverse external factors. Cardiac anomalies occur in 90% of fetuses with trisomy 18 and 13 and in 40% of fetuses with trisomy 21 or Turner syndrome. Studies have shown that with prenatal detection of heart defects in 25% of fetuses, various chromosomal diseases are detected. OMPHALOCELE The incidence of omphalocele in newborns is 1 in 4000. As a rule, this developmental feature is sporadic, but in some cases it can be combined with genetic syndromes. Chromosomal abnormalities, mainly trisomy 18 and 13, are detected in omphalocele in the second trimester of pregnancy in 30% of fetuses, while among newborns only in 15%. If only intestinal loops are part of the omphalocele, then the probability of a chromosomal disease in the fetus is four times higher than if there is an intestine and liver in the hernial sac.

Esophageal atresia The incidence of esophageal atresia in newborns is 1 in 3000. This developmental anomaly is sporadic and in 90% of newborns is combined with the presence of a tracheoesophageal fistula. Chromosomal diseases in the presence of esophageal atresia are detected in 3–4% of newborns. In the case of prenatal detection of esophageal atresia, chromosomal diseases, mainly trisomy 18, are detected in 20% of fetuses.

DUODENAL ATRESIA The incidence of duodenal atresia or stenosis in newborns is 1 in 5000. As a rule, this malformation occurs sporadically, but in some cases it can have an autosomal recessive pattern of inheritance. Trisomy 21 in the presence of duodenal atresia is detected in 40% of fetuses.

ANOMALIES OF THE URINARY SYSTEM Studies have shown that anomalies of the urinary system are often combined with various chromosomal diseases. The risk of chromosomal abnormalities does not change depending on whether the anomaly is unilateral or bilateral, the nature of the kidney pathology, the presence or absence of ureteral or urethral obstruction, and the amount of amniotic fluid. However, in female fetuses in the presence of anomalies of the urinary system, the incidence of chromosomal pathology is twice as common as in male fetuses. For various chromosomal diseases, various types of pathology of the urinary system are characteristic. In moderate hydronephrosis, as a rule, trisomy 21 is detected, while in severe hydronephrosis, in

Volume: 01 Issue: 11 | Nov – 2022 ISSN: 2720-6866
http://journals.academiczone.net/index.php/rjtds

multicystic or kidney agenesis, trisomy 18 and 13 are the most common chromosomal diseases.

With trisomy 21, 18, triploidy and Turner syndrome, shortening of long tubular bones is observed. If syndactyly is detected, the most common chromosomal pathology is triploidy, in the presence of clinodactyly and a sandal gap of the foot, trisomy 21 is detected, the presence of polydactyly is characteristic of trisomy 13, and the identification of crossed fingers, rocking foot or clubfoot is characteristic of trisomy 18.

CLEFT LIP AND PALATE The frequency of cleft lip/palate is 1 in 800 live births, and the causes of this pathology are both genetic factors and adverse external influences. Postnatal karyotyping for cleft lip/palate reveals chromosomal disorders in less than 1% of newborns. However, with prenatal karyotyping with these features of fetal development, the frequency of chromosomal diseases reaches 20%, and the most common of them are trisomies 13 and 18. Such differences in the frequency of prenatal and postnatal occurrence of cleft lip/palate of the fetus are explained by the fact that when they are combined with other multiple developmental anomalies, there is a high probability of intrauterine death of the fetus.

MICROGNATIA The incidence of micrognathia in newborns is 1 in 1000. This finding is a non-specific feature of many genetic syndromes and chromosomal disorders, usually trisomy 18 and triploidy. Data from two studies showed that if the fetus had micrognathia, the probability of chromosomal diseases was 60%, but all fetuses had malformations of other organs or intrauterine growth retardation.

NOSE HYPOPLASIA Studies from 15 to 24 weeks of gestation have shown that about 65% of fetuses with trisomy 21 have nasal bone hypoplasia, which is defined as the absence of visualization of the nasal bones, or their length is less than 2.5 mm. With a normal fetal karyotype, the incidence of nasal bone hypoplasia depends on the ethnic origin of the mother and is less than 1% in women of European descent and 10% in women of Afro-Caribbean descent. It is still premature to talk about the specific significance of the frequency of detection of chromosomal abnormalities during screening in the second trimester of pregnancy based on the age of the mother, biochemical screening data and assessment of the length of the nasal bones and other ultrasound markers of fetal chromosomal pathology. However, based on currently available data, it can be concluded that fetal nasal bone hypoplasia is probably the most sensitive and specific marker of trisomy 21 in the second trimester of pregnancy.

References

1. Monteagudo A., Timor-Tritsch I. E. Second- and third-trimester ultrasound evaluation of chorionicity and amnionicity in twin pregnancy. A simple algorithm. J Reprod Med 2000; 45:476–80.

Volume: 01 Issue: 11 | Nov – 2022 **ISSN: 2720-6866**
http://journals.academiczone.net/index.php/rjtds

2. Sawidou M. D., Karanastasi E., Skentou C., Geerts L., Nicolaides K. H. Twin chorionicity and pre-eclampsia. Ultrasound Obstet Gynecol 2001; 18:228–31.
3. Ibodova M.O. SOIL ECOLOGY AND FUNCTIONS. // Journal of Advanced Research and Stability -2022. - Б.177-181
4. Ibotova M.O. Condition of Endemic Goiter. // Vital Annex: International Journal of Novel Research in Advanced Sciences (IJNRAS) – 2022.- P. 63-66
5. Ibodova M.O. Ecological Problems of the Environment Under Modern Conditions. // AMALIY VA TIBBIYOT FANLARI ILMIY JURNALI – 2022. Б. 82-84
6. Ибадова М.О. Шумовой Фактор Окружающей Среды: Оценка Влияния На Самочувствие Студентов. // AMALIY VA TIBBIYOT FANLARI ILMIY JURNALI – 2022. Б. 92-94
7. Олимова Д. В. USE OF MODERN METHODS IN THE TREATMENT OF GLOSSALGIA // Analytical Journal of Education and Development, https://sciencebox.uz/index.php/jars/issue/view/45 P. - 197-200
8. Олимова Д. В. ДИАГНОСТИКИ И ЛЕЧЕНИЯ ГЛОССАЛГИИ И ГЛОССОДИНИИ // Journal of Advanced Research and Stability ISSN: 2181-2608. - Special Issue | 2022. P – 147-152
9. Олимова Д. В. СТОМАЛГИЯ, МЕТОДЫ ЛЕЧЕНИЯ // Analytical Journal of Education and Development, https://sciencebox.uz/index.php/ajed/issue/view/43 P. - 237-244