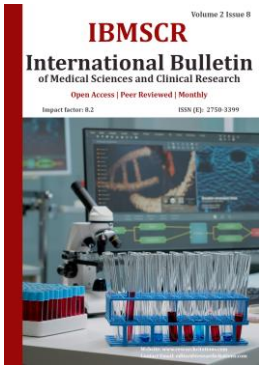

EARLY PRENATAL DIAGNOSIS AND PREVENTION OF CONGENITAL MALFORMATIONS IN CHILDREN. SPINA BIFIDA IS A DEFECT IN THE DEVELOPMENT OF THE NEURAL TUBE.

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Annotation: Spina bifida is a defect in the development of the neural tube, which is a cleavage of the spinal canal and is one of the most common congenital malformations of the fetus. Worldwide, the incidence of fetal neural tube defects ranges from 0.17 to 6.39 per 1000 newborns. This article describes the methods of early diagnosis of fetal neural tube development disorders in early pregnancy and the results of studies of pregnant women of different pregnancy periods. According to the static data, the conclusions of the research work are made.

Keywords: Spinal hernia, neural tube defect, ultrasound screening, biochemical screening, RARP-A test, alpha-fetoprotein, chorionic gonadotropin+ β, estriol, folic acid.

Relevance: On December 25, 2017, the President of Uzbekistan adopted the State Program "Early detection of congenital and hereditary diseases in children". This program is designed for 2018-2022 to prevent the birth of children with these diseases.The most common congenital malformations are "Spina bifida" – a defect in the development of the neural tube, which is a splitting of the spinal canal (spinal dysraphism), often with the formation of spinal cord hernias. Relevance of prenatal care
diagnostics of spina bifida is associated with the severity of pathology and the complexity of its correction. Spina bifida is one of the most difficult to diagnose congenital malformations of the neural tube with a multifactorial type of inheritance. The frequency of this defect depends on many factors, including geographical, ethnic and seasonal.

Goals and objectives of the work: The goal is to prevent the birth of children with various developmental abnormalities and genetically determined diseases. The upbringing of physically and mentally healthy children is considered the task of the family before society. However, in some cases, the fetus in the womb may develop incorrectly, and the child, whom the parents are waiting for with great joy and excitement, is born disabled or with hereditary diseases. To prevent such cases, it is reliable and convenient to use the screening service. As doctors, we have been convinced of this many times.

Within the framework of the State Program "Early detection of congenital and hereditary diseases in children", prenatal screening rooms were opened in the central multidisciplinary city and district polyclinics of the Fergana region and equipped with modern ultrasound scanners with high resolution. The Republican Center "Screening of mother and child" of the Ministry of Health of the Republic of Uzbekistan is equipped with a high-tech analyzer - tandem mass spectrometer for the diagnosis of a wide range of hereditary diseases in newborns and young children. As a result of the measures carried out, 1.1 million pregnant women were examined, which made it possible to prevent the birth of more than 21 thousand

children with congenital malformations. More than 1.7 million newborns were examined for the presence of hereditary and congenital diseases, 2.7 thousand children received qualified medical care to eliminate congenital malformations.

Research methods: Ultrasound screening provides for at least 3-fold examination of the fetus: 10-14 weeks; 20-24 weeks; 30-32 weeks. When examined at 10-14 weeks, gross developmental defects can be detected – anencephaly, omphalocele, acrania, exencephaly, cervical hygroma and some others. At 20-24 weeks, ultrasound can reveal the majority of gross anatomical anomalies of development – defects of the brain and spinal cord, kidneys, facial clefts, gross heart defects, defects in the development of limbs, gross anomalies of the gastrointestinal tract. Most of the developmental abnormalities detected in the middle of pregnancy are not subject to surgical treatment and are a medical indication for termination of pregnancy. Also at this stage, the presence of markers of fetal chromosomal pathology is assessed, which include: high and low water, fetal growth retardation, enlargement of the renal pelvis (pyelectasia), expansion of the ventricles of the brain (ventriculomegaly), reduction in the size of the fetal nasal bone, shortening of the length of tubular bones, hyperechogenic intestines, hyperechogenic inclusions in the fetal heart, cysts of the vascular plexus of the brain and a number of others.At the stage of 30-32 weeks, it is possible to detect developmental anomalies with low anatomical severity and late manifestation – heart defects, hydrocephalus, obstruction (narrowing) of the urinary tract. Many such developmental anomalies are subject to surgical correction after the birth of a child. Properly organized mass ultrasound screening allows you to identify most gross anatomical defects before the fetus reaches the age of viability. But ultrasound has its limits of informativeness. Ultrasound cannot detect a number of common genetic disorders of the fetus that do not have significant anatomical manifestations, in particular, chromosomal diseases (including Down syndrome).Biochemical screening based on the analysis of pregnant women's blood for serum markers serves as a means of forming a risk group for fetal chromosomal pathology. The fetoplacental complex, consisting of the fetus and fetal membranes (chorion, which transforms into the placenta, the aqueous membrane - the amnion) produces specific proteins that penetrate into the blood of a pregnant woman. Changes in the state of the fetoplacental complex, occurring for various, including genetic reasons, are reflected at the level of specific proteins (serum markers).

Currently, biochemical screening is carried out in two stages – screening of the first trimester (10-13 weeks) and screening of the second trimester (16-20 weeks).

In the first trimester (from 10-13 weeks), the PAPP-A test is performed. PAPP-A is a high–molecular glycoprotein. A number of serious clinical studies indicate the diagnostic significance of PAPP-A as a screening marker for the risk of fetal chromosomal abnormalities. The level of PAPP-A is significantly reduced if the fetus has trisomy 21 (Down syndrome) or trisomy 18 (Edwards syndrome). In addition, this test is also informative when assessing the threat of miscarriage and stopping pregnancy in short periods.

In the second trimester of pregnancy, an analysis is performed for AFP / HCG. Alpha-fetoprotein (AFP) is one of the indicators of the general condition of the fetus and the likelihood of congenital pathology. Elevated concentrations of AFP in maternal serum or amniotic fluid during pregnancy may indicate congenital cleavage of the spinous processes of the vertebrae, anencephaly, closure of the esophagus or multiple pregnancy.

In the Fergana regional screening center "Mothers and Children", pregnant women's blood serums are examined simultaneously for AFP, HCG and estriol. Currently, screening for the following types of congenital malformations in the fetus is recommended:Down syndrome (trisomy of 21 pairs of chromosomes); Edwards syndrome (trisomy of 18 pairs of chromosomes); Neural tube defects (spinal cord herniation, anencephaly).The most common congenital malformations are "Spina bifida" – a defect in the development of the neural tube, which is a splitting of the spinal canal (spinal dysraphism), often with the formation of spinal cord hernias.Typical for all types and forms of neural tube development defect is their posterior location with a defect of the posterior semicircle of the spinal canal. Extremely rarely, less than 1% of cases, non-infection is formed on the anterolateral surface of the canal, and anterior spinal hernias occur.The anterior and posterior clefts of the vertebra can pass along the median line, and also be located asymmetrically. In some cases, the gap is located obliquely. If the splitting of the vertebrae occurs along the median line, then the deformation of the spine may be insignificant or not at all pronounced. However, with an oblique and asymmetric location of the gap in combination with other anomalies of vertebral development (for example, unilateral microspondylia of half of the vertebra, anomaly of articular processes), pronounced spinal deformity develops. Most often (up to 70% of cases) spina bifida is localized in the lumbosacral region, in 21% - in the thoracic region and in 9% - in other localization.On July 17, 2018, the Ministry of Health of the Republic of Uzbekistan adopted the "Regulation on the procedure for preventing the birth of children with birth defects that hinder the health and survival of the fetus." According to the annex to this statute, spina bifida in the cervical, thoracic and lumbosacral region with hydrocephalus is determined by ultrasound screening in the second trimester of pregnancy and is considered an uncorrectable defect. Spina bifida in the cervical, thoracic and lumbosacral region without hydrocephalus is determined by ultrasound screening in the second trimester of pregnancy and is considered a correctable defect.

There are three variants of spina bifida: Spina bifida occulta – "hidden" spina bifida. In this variant, there is no visible external defect. Latent non-inflammation of the spine is usually localized in the lumbosacral region and, as a rule, does not manifest clinically in any way. Often they are an accidental "find" during an X-ray examination of the spine or MRI. The anatomical essence of the hidden cleft of the spine consists in incomplete overgrowth of the vertebral arch. This is the most favorable variant of spina bifida. Sometimes there are hyperpigmentation and tufts of hair in the area of the defect. In such cases, the child has spinal cord abnormalities below the affected area: lipomas and abnormal fixation of the spinal cord.

Meningomyelocele – meningomyelocele is the most severe form of spina bifida, involving the membranes, spinal cord and its roots in the hernial sac. The bone defect is usually wide and extended, captures from 3 to 6-8 vertebrae. The degree of neurological defect is always severe paraplegia of the lower extremities, sensory disorders, neurogenic bladder and intestinal paresis. It is this form of spinal hernias that is most common - about 75% of all forms . In almost all cases, meningomyelocele is combined with Arnold-Chiari II syndrome. Thus, the detection of signs of Arnold-Chiari II anomaly in the fetus is a marker of the presence of spina

bifida. In addition, in 70-80% of cases, the fetus develops hydrocephalus (Fig. 1).

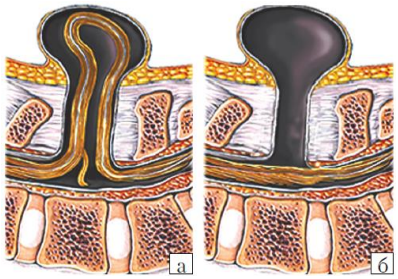
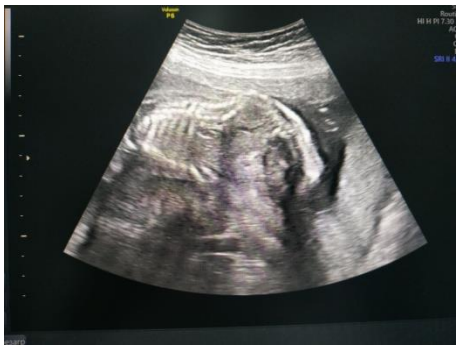


Fig. 1. Diagram of fetal spine defects.
Diagram of fetal spine defects: a - meningomyelocele, b - meningocele

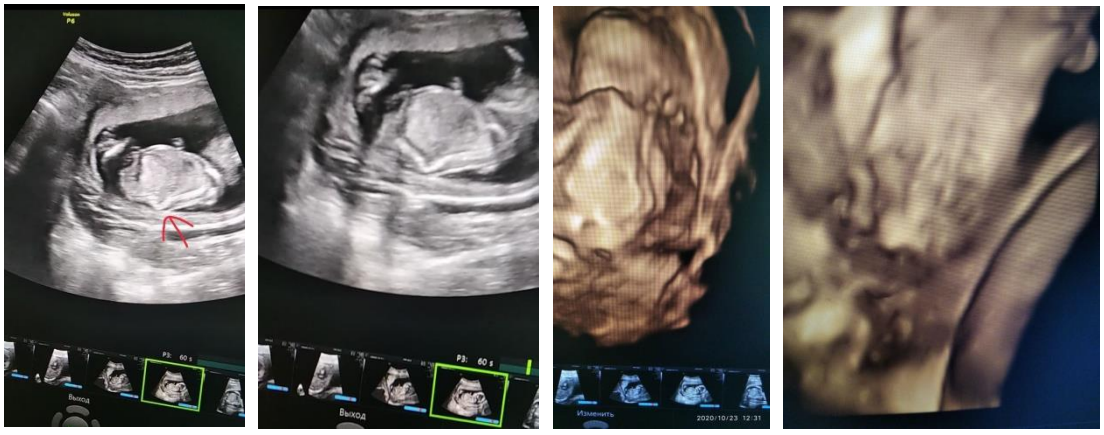
We present clinical observations demonstrating the possibilities of ultrasound in the diagnosis of fetal spinal defects.

Clinical observation 1

Patient G., 27 years old, turned to the prenatal office at 16 weeks of pregnancy. The study was conducted on the GE Voluson P6 device. The indicators of fetometry fully corresponded to the period of pregnancy. In the process of scanning the spine in the frontal and sagittal planes, angular deformation of the spine in the lumbar region was revealed. Violations of tissue structures and integrity were detected in the transverse scanning plane (Fig. 2).



Clinical observation 2. Patient M., 35 years old, turned to the prenatal office at 14-15 weeks of pregnancy. The study was carried out on the GE Voluson P6 apparatus using the 3D/4D surface volume reconstruction mode. In the process of scanning the spine in the frontal and sagittal planes, angular deformation of the spine in the lumbar region was revealed. Violations of tissue structures and integrity were detected in the transverse scanning plane (Fig. 3).



Conclusions: For 9 months of 2019, 23 children with this diagnosis were born in the Fergana region. Comparative statistics show that in 9 months of 2020, the number of births with this diagnosis decreased, and 5 children were born with this diagnosis. The frequency of bifid spin is, according to various data, 1-2 cases per 1 thousand newborns. The frequency of repeated

births with this defect is 6-8%, which confirms the role of genetic factors in the appearance of the disease. Most often, the diagnosis in question occurs in children from mothers of advanced age. 95% of newborns with spina bifida are born to parents without such a diagnosis. Localization area: lip-57.6%, cheek 8%, tongue 26.3%. Complications of DSO: anatomical disorders, edema, bleeding, pain, infection, respiratory disorders occurred in 83.7% of patients.

Currently, the detection of this defect in the first trimester of pregnancy is of the greatest interest. Ultrasound criteria such as the absence of intracranial translucency, smoothness of the brain stem angle, reduction of the biparietal size below the 5th percentile, etc. are studied. Thus, the diagnosis of spina bifida remains an important task of prenatal diagnosis.

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