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Optimization of Fetal Ultrasound Examination for the Presence of Congenital Malformations of the Brain in Pregnant Women in the Second Trimester

Djurabekova Aziza Takhirovna
Doctor of Medical Sciences, Professor, Head of the Department of Neurology, Samarkand State Medical University

Vaseeva Umida Khamidovna
Doctoral student of the Department of Neurology, Samarkand State Medical University

Annotation: Ultrasound examination (US) remains the primary method of prenatal diagnosis of congenital fetal anomalies due to its safety, accessibility, and informativeness. The second trimester of pregnancy (18-24 weeks) is the optimal period for detecting most fetal brain anomalies, as by this time the main processes of CNS structure formation are completed while maintaining adequate visualization due to the relatively small size of the fetal head and low ossification of the skull bones.

Keywords: Prenatal diagnostics, ultrasound examination, second trimester of pregnancy, congenital brain anomalies, fetal neurosonography, volumetric echography, standardized protocol, central nervous system developmental defects, fetal CNS anomalies, prenatal screening.

Introduction. Despite significant progress in ultrasound diagnostics over recent decades, the detection rate of fetal brain developmental defects remains insufficiently high, which is associated with several factors: technical limitations of diagnostic equipment, variability in examination methods, different levels of specialist qualification, as well as objective visualization difficulties caused by the structural features of the fetal CNS and the position of the fetus in the uterus.

In this regard, improving methods of ultrasound diagnostics of congenital fetal brain anomalies represents an urgent task of modern prenatal medicine. The development of standardized examination protocols, implementation of new visualization modes, optimization of data interpretation algorithms, and application of additional technologies, such as volumetric echography and magnetic resonance imaging (MRI), can significantly increase the effectiveness of detecting congenital developmental defects of the fetal CNS.

This study aims to improve the methodology of fetal brain ultrasound examination in the second trimester of pregnancy to increase the sensitivity and specificity of diagnosing congenital developmental anomalies, which will ultimately contribute to reducing perinatal morbidity and mortality, as well as improving the quality of care for pregnant women and newborns.

Congenital brain developmental defects are diagnosed in more than three million cases annually worldwide, more than 15% of which result in infant death, and are one of the leading causes of childhood disability. The prevalence of congenital brain developmental defects ranks second among developmental anomalies, yielding only to defects associated with cardiovascular diseases. Today, there remains an increased interest in information about brain structure, to ensure diagnosis during the intrauterine period both at the level of expert activity and screening analysis among practicing physicians. The standards for diagnosing the fetal brain to confirm, clarify, or refute congenital pathologies are ultrasound examination (US) and magnetic resonance imaging (MRI).

Materials and methods. To increase the effectiveness of prenatal diagnosis of congenital brain developmental defects, pregnant women with suspected fetal pathological processes (based on history and clinical presentation) underwent rationalized and standardized ultrasound examination of the fetal

brain in the second trimester (from 18 to 28 weeks). The examination of women was conducted during the period of 2023-2024, at the departments of the Multidisciplinary Clinic of Samarkand State Medical University. During the study, the results of examinations of 31 women whose fetuses were prenatally diagnosed with various brain structure anomalies according to ultrasound data were analyzed. Ultrasound examinations were performed on Voluson E8, Voluson E6 (GE) ultrasound machines using transabdominal RAB 4–8 D probes and transvaginal RIC 6–12 D probes.

In cases where severe developmental anomalies were detected, women were offered artificial pregnancy termination for medical reasons, which was followed by pathological examination in the pathology department at the Multidisciplinary Clinic of Samarkand State Medical University, with verification of the prenatal ultrasound data against the pathological examination results.

During fetal brain ultrasound, the assessment of brain structures was conducted using a standardized approach established by domestic normative indicators, according to the gestational age. The basis for the ultrasound examination was a staged approach used in specialists' practice, consisting of three types: when there is no image of the cavity and walls of the septum pellucidum; or when it has an expansion of more than 95%.

Evaluation of results based on the absence of the septum pellucidum cavity image revealed agenesis of the corpus callosum, with partial absence detected in 7 patients (20.7%), and complete absence in 24 patients (79.3%), showing a significant predominance of complete agenesis.

In 5 women who underwent fetal ultrasound to detect expansion of the septum pellucidum (more than 95%), agenesis of the corpus callosum was identified, with complete absence found in only one case, and partial absence of the corpus callosum in the remaining 4 cases (1.25%). Additionally, the average gestational age in all five women was around 27 weeks; consequently, this indicator is determined by the end of the first trimester.

As noted above, the third examination was necessary to determine the condition of the septum pellucidum walls. Of 31 pregnant women, six showed absence of the septum pellucidum walls, with septo-optic dysplasia identified in five of them. In one case, the absence of walls in the septum pellucidum cavity was combined with hypoplasia of the optic chiasm. This pregnancy was not terminated (at the mother's insistence), and therefore after birth, neuroimaging study of the brain confirmed the absence of the wall in the septum pellucidum cavity and hypoplasia of the optic chiasm. This clinically manifested as psychomotor developmental delay, lack of response to objects (the child did not fix their gaze), and ophthalmological examination revealed a 2.5-fold reduction in optic discs. It should be noted that in addition to the main symptoms of studying changes associated with the septum pellucidum, the ultrasound method identified congenital anomalies of the anatomical defect of the septum pellucidum cavity in 65.7% of cases (20 patients).

Anomaly of the anatomical structure of the corpus callosum was detected in 44% of cases (14 patients). In 66% of cases (out of 31 pregnant women), a developmental anomaly was found in the lateral ventricles, in the form of colpocephaly or teardrop-shaped lateral ventricles, in combination with partial agenesis of the corpus callosum. With complete agenesis of the corpus callosum, a defect of the third ventricle in the form of abnormal expansion or abnormal displacement was detected in parallel in 3 patients (0.93% of cases). Signs of lissencephaly in combination with other brain defects were found in 2 fetuses (0.62%). In 6 cases (1.86%), fetal anomalies of underdevelopment were determined in the posterior cranial fossa, particularly Dandy-Walker malformation (with an increase in the depth of the cisterna magna, more than 95%). The most common and widespread finding among ultrasound examinations of pregnant women was ventriculomegaly in the fetus, detected in 84.3% of cases (local right-sided or left-sided ventriculomegaly), in combination with agenesis of the corpus callosum and retrocerebellar cyst.

In addition, out of the total number of pregnant women with suspected fetal brain developmental anomalies, three (at the insistence of the women themselves and their relatives) underwent MRI examination of the fetal brain, which confirmed the signs of disorders previously detected by

ultrasound. Thus, pronounced ventriculomegaly was revealed, where the expansion of both ventricles exceeded 11 mm, and in one case, there was an asymmetric arrangement of the ventricles (right side larger than the left) and a combination of brain developmental pathology (maximally narrowed septum pellucidum cavity or septum pellucidum cyst). According to ultrasound data, one woman had suspected agenesis of the corpus callosum, which was confirmed by MRI as complete absence of the corpus callosum.

Out of 31 cases of ultrasound examination of pregnant women at 18 to 28 weeks, 29 women were recommended artificial termination of pregnancy, three of whom refused termination. Pathological analysis of the results of the terminated pregnancies confirmed the diagnosis established by clinical and instrumental methods. Of the three children born, one child died from secondary infection at the age of 4 months.

The remaining two children were diagnosed with congenital hydrocephalus with signs of brain anomalies; both children are mentally delayed, one of them has epileptic seizures, and the other, as previously mentioned, has pathology of the visual analyzer. Thus, the work presents various forms of congenital brain developmental pathologies in the prenatal period, which can be timely detected by standard ultrasound examination, with optimization of further prediction tactics.

Conclusions

1. Ultrasound examination in the second trimester of pregnancy is an effective method for diagnosing congenital fetal brain anomalies, allowing timely detection of pathological changes in CNS structures.
2. A standardized approach to fetal brain ultrasound, including assessment of the cavity and walls of the septum pellucidum, significantly increases the diagnostic accuracy of the examination.
3. The most frequently diagnosed anomaly in our study is agenesis of the corpus callosum, with complete absence of the corpus callosum occurring more frequently (79.3%) than partial absence (20.7%).

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