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Prenatal Ultrasound Diagnosis of Agenesis of the Corpus Callosum: Analysis of 34 Cases

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Abstract.

The purpose of our trial was to study indirect prenatal echographic signs of agenesis of the corpus callosum in screening axial planes of scanning depending on the form of pathology. The analysis of agenesis of the corpus callosum (ACC) was carried out in 34 cases: 14 fetuses with complete ACC and 20 fetuses with partial ACC in terms of 19 to 34 weeks of pregnancy. Final prenatal diagnosis of ACC was established in the middle sagittal plane obtained by volumetric echography. In cases of complete ACC, its image was absent in this plane, while the length of the corpus callosum was measured on the background of partial ACC. An abnormal image of the cavity of the septum pellucidum was recorded in all 14 (100%) fetuses with complete ACC, and 9 (45%) fetuses with partial ACC. The parallel course of the frontal horns of the lateral ventricles was recorded in 19 (55.8%) of 34 cases. Displacement and dilation of the third ventricle was diagnosed in 5 (14.7%) of 34 fetuses, ventriculomegaly was detected in 25 (73.5%) of 34 fetuses - in 11 (78.5%) of 14 fetuses with complete ACC, and in 14 (70%) of 20 fetuses with partial ACC. The teardrop form of the lateral ventricles was recorded only in case of complete ACC in 71.4% of fetuses. Identification of indirect signs of ACC in the axial planes of scanning allows to suspect this pathology. In order to make the final diagnosis, it is necessary to assess corpus callosum in the middle sagittal plane, which in some cases can be obtained by volumetric echography.

Keywords: fetus, brain, corpus callosum agenesis, ultrasound examination, prenatal diagnosis.

INTRODUCTION

Corpus callosum is an extremely important commissure connecting the hemispheres of the brain. It contains conductive pathways that connect various zones of the cortex in order to coordinate functions of the cerebral hemispheres. Corpus callosum consists of several anatomical parts: genu, rostrum, truncus and splenium. The development of the corpus callosum begins at 11 weeks of pregnancy. Its anterior parts develop first and grow from the anterior part to posterior one [1]. Although corpus callosum can be considered fully developed to the child's age of about 4 years, like most brain structures, it continues to change throughout life [2]. There are various classifications of the developmental anomalies of the corpus callosum, but according to the most common one, all congenital anomalies of the corpus callosum can be classified as agenesis (complete or partial), and hypoplasia of the corpus callosum when its thickness decreases along all its parts [3]. Quite often, in case of partial absence of the corpus callosum, the term dysgenesis [4] is also used, which can be considered synonymous with partial agenesis. Agenesis of the corpus callosum (ACC) occurs both in isolated form and in combination with other anomalies of brain development, as well as other congenital malformations, chromosomal abnormalities and genetic syndromes [5, 6]. The prevalence of isolated ACC is about 50% of the diagnosed cases. However, the prognosis for neurological and mental development is more favorable in case of isolated ACC [7]. Nevertheless, even with partial ACC, the frequency of pronounced mental retardation may reach 62%, and the incidence of epilepsy is 46% [8]. It should be noted that in case of isolated ACC, clinical manifestations after childbirth may be absent [9]. In case of combination of an abnormal development of the corpus callosum with other congenital malformations of the brain, the clinical picture usually manifests itself immediately after birth and depends on the severity of the concomitant pathology.

Therefore, early prenatal diagnosis of anomalies of the development of the corpus callosum is extremely important, as it will allow to inform the family about the prognosis for the development and health of the child in time.

Ultrasound examination of the fetus is currently the main method of prenatal diagnosis of congenital malformations, including the developmental defects of the brain. Until recently, according to the domestic experts, antenatal diagnosis of ACC did not exceed 40% [10], which can be explained by the methodology of screening brain structures, namely, using only axial scanning planes in the study of the fetal brain. However, in a number of centers in recent years, there has been a significant improvement in the prenatal diagnosis of ACC, which is explained by the use of multiplane evaluation of fetal brain structures, including primarily the study of the average sagittal plane [11, 12, 13]. However, obtaining this plane is probably not always possible with the use of two-dimensional echography, since with an "uncomfortable" position of the fetal head, obtaining the middle sagittal plane is often difficult.

The application of the methods of volumetric echography allows to analyze the structures of the brain in any scan plane within the limits of the collected volume in real time. The sagittal plane of the scan of the fetal brain becomes easily accessible for examination. The study of the fetal brain with the help of volumetric echography is optimally carried out in the static (3D) mode with the use of the maximum quality of the volume. It is this approach that makes it possible to get the clearest picture of the structures studied. Among the numerous regimens of volumetric echography, multiplanar (multiplanar) reconstruction was the most widely used method for evaluating fetal brain structures, when the study of the structures of the fetal brain is possible simultaneously in three mutually perpendicular planes. In this case, the analysis can be carried out at any level, and in any direction.

Multiplanar reconstruction of the fetal brain is used to evaluate midline structures, often inaccessible for examination by screening two-dimensional echography, including the corpus callosum. If a more informative image is needed, VCI (Volume Contrast Imaging) mode is additionally used, which allows obtaining a thicker section. Omni View technology allows to build images according to a specialist assigned direction. This mode will contribute to obtaining sagittal section of the fetal brain from the transverse section. This can help in evaluation of the corpus callosum in cases of "uncomfortable" position of the fetus when the cross section of its spine is within 3 or 9 hours. The possibility of assessing the corpus callosum when using the multiplanar regimen of volumetric echography within the period of 16-20 weeks of pregnancy reaches 84%, and at the time of 21-24 weeks of pregnancy the visualization of the corpus callosum is available in 97% of cases [14]. The percentage of successful visualization

of the corpus callosum in the second trimester of pregnancy according to different authors is from 88% to 100% [15, 16].

To date, the normative values of the length of the corpus callosum have been developed, which can be used to assess its development [17, 18, 19]. It should be emphasized that only in the middle sagittal plane of scanning it is possible to establish a final prenatal ultrasound diagnosis of agenesis of the corpus callosum. Given that today screening assessment of the brain is carried out only in axial scanning planes, the diagnosis of ACC during screening neurosonography can be suspected only on the basis of indirect signs. These include the following: the lack of visualization of the cavity of the septum pellucidum or changes in its shape and size, ventriculomegaly, changes in the shape of the lateral ventricles [20].

Additional information in studying the corpus callosum in the fetus in the middle sagittal plane of scanning can be obtained using the color Doppler mapping (CDC) regime, which allows to establish the absence of visualization of the arteries of the corpus callosum in cases of the complete form of ACC and its abnormal course with partial ACC. The abnormal course of the pericullic artery means that it repeats the contour of the preserved parts of the corpus callosum (more often the knee and trunk) and rises upwards at the level of the missing corpus callosum [21, 22]. DCC mode for the identification of the pericullic artery can be used in cases of difficult visualization of the corpus callosum, especially when there are doubts about its true boundaries.

Purpose of the study is to study indirect prenatal echographic signs of ACC in screening axial planes, depending on the form of pathology.

Materials And Methods

The analysis of 34 cases of complete and partial ACC found in the fetus in the period from 19 to 34 weeks of pregnancy was conducted. The final prenatal diagnosis of ACC was established in the middle sagittal plane obtained by volumetric echography. In this case, the diagnosis of complete ACC in our studies was established in the absence of its visualization, and at numerical values of the length of the corpus callosum less than the 5th percentile of normative indices, a diagnosis of partial ACC was established.

Ultrasound studies were carried out on the Voluson E8 (GE) using volume scanning sensors. The age of the examined patients with ACC in the fetus ranged from 22 to 39 years, and averaged 30 years.

Results

The duration of prenatal diagnosis of ACC in our studies ranged from 19 to 34 weeks, and averaged to 24.4 weeks of pregnancy. At the same time, there were no significant differences in the groups of patients with complete and partial ACC - 25.3 and 24.2 weeks of pregnancy, respectively. It should be noted that in the period up to 22 weeks of pregnancy prenatal diagnosis of ACC was established in 10 (29.4%) of 34 cases, and up to 24 weeks in 15 (44.1%) cases.

Complete ACC was diagnosed in 14 fetuses, partial ACC is in 20 fetuses. In case of complete ACC, an abnormal image of the septum pellucidum cavity was recorded in all 14 (100%) fetuses, and mainly consisted in the absence of its image, only in one case there was a significant dilation of septum pellucidum cavity. In partial ACC, an abnormal image of the cavity of the septum pellucidum was recorded in 9 (45%) of the fetuses, in 4 of them septum pellucidum cavity was not visualized, and in 5 fetuses its extension and anomalous form were recorded. In 11 (55%) of 20 fetuses with partial ACC, the width of septum pellucidum cavity was within normal limits.

Another indirect sign of ACC is the parallel course of the anterior horns of the lateral ventricles. This change was recorded in more than half of our observations - 55.8% (19/34). It should be noted that, with the complete form of ACC, this symptom was registered in 71.4% (10/14) of cases, while in partial ACC - in 9 (45%) of 20 fetuses.

The previously considered classical sign of ACC - displacement and dilation of the third ventricle - was found only in 5 (14.7%) of 34 fetuses, and in all of these observations complete ACC was detected. Thus, the displacement and dilation of the third ventricle is not effective in providing prenatal diagnosis of ACC.

The elongated drop-shaped form of the lateral ventricles as an indirect sign of ACC proved to be effective in 4 (20%) of 20 fetuses with partial ACC and in 10 (71.4%) of 14 fetuses with complete ACC.

The most frequent of the indirect signs of ACC due to the obtained results was ventriculomegaly. This feature was found in 25 (73.5%) of 34 fetuses, including 11 (78.5%) of 14 complete ACC and 14 (70%) of 20 fetuses with partial ACC.

Discussion

Despite the fact that the screening evaluation of the fetal brain is carried out in the axial planes of the scan and does not include a direct evaluation of the corpus callosum, diagnosis of ACC during screening ultrasonography may be suspected on the basis of indirect signs' detection. In our studies, there were no cases of prenatal ultrasound diagnosis of ACC without the presence of indirect signs of this pathology in the axial scanning planes.

The obtained results correlate with the previously published studies. Not all the signs, previously considered classic for ACC detection, have high informativeness. Thus, displacement and dilation of the third ventricle in our study was noted only in 14.7% (5/34) of the fetuses. Similar data were provided by other researchers. According to O. Shen et al. [23], an abnormal third ventricle was observed only in 7 (10%) of 71 cases.

Ventriculomegaly, being the most common indirect symptom in our study, was recorded in 25 (73.5%) of 34 fetuses. According to D. Paladini et al. [24], ventriculomegaly was noted in 73.5% of the fetuses in the period up to 24 weeks of pregnancy and in 25.7% of fetuses after 24 weeks. During the ultrasonic examination of the fetal brain, it is difficult to assess corpus callosum in a number of cases. However, the use of volumetric echography can significantly alleviate this problem and thereby ensure timely ACC diagnosis.

Conclusions

ACC refers to congenital malformations of the brain, the diagnosis of which is possible in the prenatal period. In order to ensure ultrasound diagnosis of ACC in screening studies using axial planes of the fetal brain, a thorough evaluation of the cavity of the septum pellucidum and the lateral ventricles is necessary. When revealing such signs as anomalous image of the septum pellucidum cavity, parallel course of the anterior horns of the lateral ventricles, ventriculomegaly, dilation and displacement of the third ventricle, it is necessary to perform an expanded neurosonography by using volumetric echography for direct evaluation of the corpus callosum.

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