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EARLY ULTRASOUND PRENATAL DIAGNOSIS OF LOWER OCCIPITAL CRANIOCEREBRAL HERNIA, AT 13/3 WEEKS OF PREGNANCY

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We describe a case of early ultrasound prenatal diagnosis of a rare disabling malformation of the fetal central nervous system, lower occipital encephalocele, at 13/3 weeks of pregnancy. The article describes the characteristic ultrasound markers, stages of prenatal ultrasound diagnostics. The prenatal diagnosis was established on the basis of the signs characteristic of this pathology of the central nervous system. The pregnancy was terminated at 17 weeks by the decision of the family. Pathomorphological examination of abortus confirmed the echographic changes detected in the prenatal period.

Keywords: fetus, lower occipital encephalocele, ultrasound prenatal diagnosis, pathomorphological examination.

Introduction. Craniocerebral hernia (encephalocele) is the formation of a hernial sac, with various anatomical contents, localized in the area of a defect in the vault of the skull bones, mainly in the cranial sutures. Frequency -1.13: 10,000 newborns [1, 2].

Encephalocele is associated with a disorder of the early embryonic development of the cranial vault, during the laying of the brain plate and its closure into the neural tube [3].

By localization, craniocerebral hernias (hereinafter CCH) are divided into anterior (frontal), nasopharyngeal, basal and posterior (occipital). Depending on the location above or below the occipital protuberance, upper or lower CCH is distinguished [2].

According to the contents of the hernial sac, a cranial hernia is divided into meningocele, encephalocele and encephalocystocele. In the first case, a craniocerebral hernia contains dura mater covered with skin; large defects, when the hernial sac includes brain tissue with arachnoid (arachnoidea encephali) and soft (pia mater) membranes, is an encephalocele. Encephalocystocele is a type of extremely pronounced form of CCH, when a part of the ventricular system of the brain also enters the hernia. There are also "detached" cerebral hernias, the most favorable form of CCH, when during the development period the hernia loses its connection with the cranial cavity.[4]

The most frequent (up to 80-90%) and prognostically unfavorable are occipital hernias [1]

An echographic marker of CCH is the detection of a defect in the bones of the cranial vault and paracranial formation of various localization and contents, depending on which brain tissue enters the hernial sac [5,6] Early prenatal diagnosis of CCH in the first trimester of pregnancy is possible after 11 weeks of pregnancy, when the ossification of the bones of the cranial vault is completed [1]

The fetal MRI method can supplement and refine the ultrasound data, which is important for choosing tactics for further pregnancy management and long-term prognoses. In 80% of cases of CCH, various combined anomalies are observed: microcephaly, ventriculomegaly, agenesis of the corpus callosum, holoprosencephaly, congenital heart defects and abnormalities of the musculoskeletal system. During prenatal examination, it is also important to take into account that CCH can be included in a number of syndromic conditions, such as Meckel-Gruber syndrome, amniotic cords syndrome, Walker-Warburg syndrome and frontonasal dysplasia [1] Treatment of CCH hernias is operative, using plastic surgery methods, especially with anterior localization. Postoperative losses in CCH reach 44%. Intellectual deficiency cases in surviving children – vary from 40 to 91% [7] Fetal surgery experience around the world shows the possibility of intrauterine surgical treatment of CCH in the period from 24 to 26 weeks of gestation, with minimal neurological symptoms in the postpartum catamnesis [8,9]

Materials and methods. Patient K., 25 years old. Healthy. This pregnancy is the second, the first ended less than a year ago with premature operative delivery at 33 weeks. The first pregnancy proceeded with severe anemia, weight deficiency, chronic fetoplacental insufficiency, fetal growth retardation syndrome of the 1st degree. The child died at the 6th week of life due to fungal bacterial sepsis.

With the current pregnancy, the patient registered with the hospital for regular screening at 6-7 weeks of pregnancy. This pregnancy proceeded with early toxicosis of moderate severity, for which she received treatment in a hospital. Chronic nicotine intoxication is noted, smoking experience is 2 years before the onset of the first pregnancy. The effect of other teratogenic factors are denied by the patient. Civil marriage, not related. Husband is 27 years old, smokes. Spouses have no industrial hazards at work. The genealogical anamnesis of the husband is not burdened. The first planned screening examination of the fetus was carried out at the time of 13/3 weeks of pregnancy on an Accuvix A-30 ultrasound device, Samsung Medison, with sensors: volumetric 4-8 Mhz and convex 4-9 Mhz.

Results. Fetometrial data during the screening study at 13/3 weeks of pregnancy corresponded to the gestational norm (Fig.1) When examining the bones of the fetal cranial vault, in the lower part of the occipital region, on the left, a thin-walled hernial formation, rounded in shape, measuring 7 x 6 mm was determined paracranially (Fig.2) The echostructure of the hernial formation is heterogeneous (most likely the brain membranes in the cerebrospinal fluid) (Fig.3)

In the occipital region of the fetal head, a defect of the skull bones with a width of 1.7 mm was clearly defined in the arch. In the CDU (Color Doppler Ultrasound) mode – avascular. There were no other echographic markers of chromosomal

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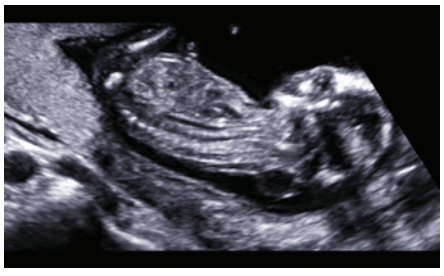

Fig.1. Sagittal section of the fetus

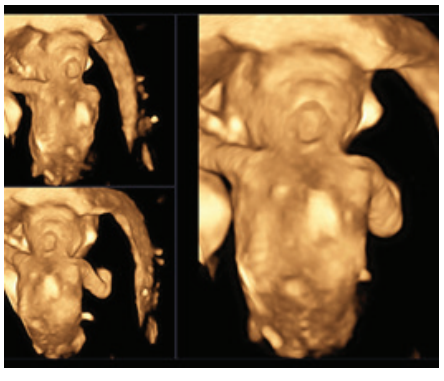

Fig.2. 3D- fetal reconstruction

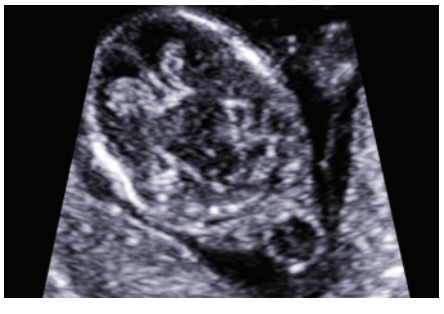

Fig.2. 3D- fetal reconstruction

abnormality and markers pathognomonic for the syndromic diagnosis. The patient was referred to the second stage of examination at the MGC, where the diagnosis was fully confirmed. Prenatal karyotyping was not performed. At the age of 17 weeks, with the consent of the family and following the results of the collegial conclusion of specialists, a medical termination of pregnancy was performed.

A pathoanatomic examination of the fetus revealed: female abortus, weighing 250 g, height 21 cm. Examination of the cervical-occipital region revealed a hernial protrusion with a diameter of 15 mm. The contents of the hernial sac are edematous membranes of the brain, a hernial defect of 6 mm. The cerebellar hemisphere was closely attached to the bottom of the hernial opening. The internal organs are formed correctly, without visible malformations.

Discussion. The described case of early prenatal diagnosis of occipital encephalocele at 13/3 weeks of gestation is the first of its kind in Yakutia, in which all stages of prenatal examination, counseling and postmortem verification are observed. Conducting the first prenatal screening study at more than 11 weeks of gestation made it possible to timely identify a disabling malformation and, with less moral and material losses, prevent the birth of a child with a severe, disabling pathology of the central nervous system.

Around the world, in specialized clinics, most cases of encephalocele are diagnosed during screening ultrasound studies [2]

In the described case, the cause of isolated folate-dependent malformation of the central nervous system in the fetus is probably multifactorial: recent pregnancy, hormonal discorrelation, chronic hypoxia, malnutrition and weight deficiency (the woman's BMI when registering with the hospital - 17.3 kg / m^2)

Taking into account the latter fact, in order to reduce the number of folate-dependent malformations, in women's consultations - it is advisable to work more actively in the family planning office with maternity hospitals with unfavorable pregnancy outcomes, prescribing folic acid, patronage of families in the first year after the loss of a child, offering psychological counseling and regular preventive talks.

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