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IMPORTANCE OF PRENATAL FETAL SCREENING IN DIAGNOSING ANEUPLOIDYA: THE CLINICAL CASE OF EDWARDS SYNDROME

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Edwards syndrome (SE) or trisomy of chromosome 18, according to pathogenetic classification refers to chromosomal diseases. Manifested from birth, characterized by multiple congenital malformations (MCM). The first signs of trisomy 18 of chromosome can be detected by prenatal screening of the fetus. The prevalence of SE is 1: 3000-8000 newborns. The ratio of girls / boys is 3: 1. SE is the cause of miscarriage in 1% of all cases. Children with SE die in utero in about 60% of cases. Despite this, among genetic diseases, this syndrome in surviving infants is quite common, the frequency of occurrence second only to Down syndrome. The most common cause of the syndrome is considered to be the older age of the mother [1, 2].

In 95% of all cases of SE development in cells there is an extra 18th chromosome (complete trisomy), in 2% there is a translocation variant; in 3% of cases there is a "mosaic trisomy", when an additional 47 chromosome is not found in all cells. Clinically, the first option may be more severe. Prenatal symptoms, which occur in 50% of cases, include: prolonged pregnancy (up to 42 weeks), weak fetal movements; prematurity; polyhydramnios; small placenta; agenesis of one of the umbilical arteries; intrauterine developmental delay. Signs of Edwards syndrome can be divided into two major groups. The first group includes symptoms characteristic of the child's appearance: low body weight (less than 2200 g); disproportionately small head; micrognathia; distortion of the shape of the face, occlusion; cleft palate and / or upper lip; flexor position of the fingers; low landing of ears; webbing; congenital clubfoot;

"Foot-swing"; microstomy. The second group includes signs of disorders of internal organs, motility and neuropsychological development: congenital heart disease (CHD), various hernias; congenital malformations of the gastrointestinal tract (GIT), central nervous system (CNS), urinary tract (UT), musculoskeletal system (MSS). The most common congenital malformations (CM) include: umbilical and inguinal hernia; cryptorchidism; congenital heart defects, primarily a defect of the interventricular septum [2].

Due to MCM, the diagnosis of SE is difficult and the prognosis is unfavorable. The average life expectancy of infants is 14.5 days in 5-10%. Babies need resuscitation after birth; they may periodically stop breathing throughout the neonatal period. Suck poorly, gain weight poorly. Mortality in SE in the 1st year of life is about 90%, and the average life expectancy in severe disease in boys is 2-3 months, and girls - 10 months, only a few live to adulthood. Most often, children with SE die from complications caused by congenital malformations: asthma, pneumonia, cardiovascular failure or intestinal obstruction. Children who are a year old are mentally retarded. Older children die mainly from pneumonia due to cardiovascular insufficiency and/or urinary tract infection [1, 2].

SE can be detected before the baby is born. Prenatal diagnosis of this syndrome is performed in 2 stages. The first stage is performed at 11-13 weeks and consists of ultrasound of the fetus and biochemical screening of the pregnant woman. In the early stages of SE on ultrasound is extremely difficult to suspect, but at 12 weeks of pregnancy can detect characteristic signs: intrauterine growth retardation, bradycardia, omphalocele, lack of visualization of the nasal bones, one umbilical artery. Ultrasound can also detect cysts of the vascular plexus, which in themselves do not pose a threat to health and disappear by 26 weeks of pregnancy. However, such cysts are often accompanied by various genetic diseases, such as SE (in this case, cysts are found in 1/3 of children suffering from this pathology). At 11-13 weeks of gestation in a woman's blood is determined by the level of blood proteins: β-HCG and plasma protein A associated with pregnancy. Taking into account these data, the age of the pregnant woman, the risk of giving birth to a child with SE is calculated and the risk group of pregnant women is formed. Pregnant women at risk are recommended to determine the karyotype of the fetus. Material in the fetus for karyotype examination can be obtained by different methods, depending on the week of gestation: 8-12 weeks - a biopsy of chorionic villi, 14-18 weeks - amniocentesis, 20 weeks - cordocentesis with ultrasound control. If the pregnant woman has not undergone a genetic screening examination, then at a later date, the preliminary diagnosis of SE is carried out using ultrasound of the fetus [1-4].

In most cases, the prognosis for SE is unfavorable. Units of children with SE who live to adulthood have severe mental disorders and constantly need outside care and monitoring [2].

Clinical case. Here is an example of SE in a newborn girl. A child from the first pregnancy, which took place against the background of anemia, toxicosis and chronic pyelonephritis. Biophysical profile at birth 7 points. The pregnant woman has been registered since the 15th week, but refused to perform prenatal screening and examination for TORCH infection, prenatal ultrasound of the fetus was performed for

the first time at 30 weeks of pregnancy. Conclusion Ultrasound of the fetus: Congenital Malformations. Ventricular septal defect (VSD), double exit of the main vessels from the right ventricle, transposition type, tricuspid valve dysplasia, cardiomegaly. Ultrasound signs of intrauterine growth retardation of the fetus of late form, the middle location of the liver. The risk of giving birth to a child with congenital pathology is considered high; pregnant woman was sent to the Research Institute of Obstetrics and Gynecology named after O.M. Lukyanova, where a repeat ultrasound of the fetus was performed. Conclusion Ultrasound: pregnancy 38 weeks +5 days (for mensis). MCM of fetus: the combined heart defect. Fetal developmental delay syndrome (symmetrical shape), III degree. Hepatosplenomegaly. Acentric output of the umbilical cord. Ultrasound markers of intrauterine infection of the fetus. Ultrasound markers of chromosomal pathology of the fetus. Ultrasound markers of fetal hypoxia.

The girl was born in the 40-41st week of gestation. Birth weight: 1480 g, length 40 cm; head circumference 28 cm, bust circumference 4 cm; Apgar score 4/4 points. Primary resuscitation of the newborn was performed in the delivery room. At the 5th minute she was transferred to artificial lung ventilation. Later she was admitted urgently to the Intensive Care Unit with the main diagnosis: Edwards Syndrome; main complications: heart failure, respiratory failure, cerebral depression; concomitant diagnosis: small before gestation, the risk of intrauterine infection. The girl was promptly consulted by a pediatric neurologist and geneticist. Examinations were performed: radiography of the chest and abdominal organs, ultrasound of the heart of the abdominal cavity, neurosonography, as well as cytogenetic examination (47, XX, +18). Child phenotype: multiple dysmorphic features, including small eye slits, low ears, microstomy, micrognathia. Examination of the musculoskeletal system revealed shortened limbs, wrists were in a flexion position, bilateral 4-finger furrows on the palms and "rocker's foot". From the first day, a neurologist diagnosed a cerebral coma (5 points on the Glasgow Scale); from the second week - cerebral neonatal depression; signs of cerebral edema. Despite the treatment, which was carried out in a specialized hospital for a month, the child's condition progressively deteriorated. Pathological examination revealed that the main disease and cause of death of a premature and morphologically immature girl aged 30 days was a chromosomal abnormality – trisomy of chromosome 18 – Edwards syndrome, which was complicated by the development of multiple organ failure, which was the direct cause of death.

This clinical case illustrates the importance of disseminating information about current opportunities for prenatal diagnosis of chromosomal diseases among families who are planning a child or are already expecting a child.

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