CAUSES OF PATAU SYNDROME

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ABSTRACT

Clinical symptoms of Patau's disease were identified in the 17th century. 13th of the disease connection with the increase in the number of paired chromosomes in 1960 K. Patau was identified by, therefore, this disease is associated with the name of this scientist. 13-chromosomal trisomy occurs in approximately 1/10,000 births; about 80% cases are complete trisomy on the 13th chromosome. Mother's age (average 31 age), the higher the probability of developing this syndrome will be Babies are usually too small for their gestational age. Medium anomalies are frequent and include holoprosencephaly (correct forebrain not splitting), anomalies of the face, for example, clefts of the upper lip and palate, microphthalmia, retinal colobomas (cracks) and retinal dysplasia enters. Brow systems are small, eyes are located far from each other. Ears irregularly shaped and usually located lower. Skull defects and dermal sinuses are also common. Single transverse palmar fold, polydactyly and visible narrow fingernails are common. About 80% in cases, severe congenital anomalies of the cardiovascular system are detected; dextrocardia is common. Genital organs are often atypical in both sexes there is: cryptorchidism in boys, bicornuate uterus in girls. There was PS special complications in pregnancy - this frequency occurs in 50% of PS.

GENETIC CHARACTERISTICS

Basically, there are two genetic forms of trisomy on the thirteenth chromosome: Simple trisomy – three chromosomes of the same type exist freely and are genetic Fulfills the potential. Robertsonian translocation – two chromosomes remain empty,

the third is long Joins hands with another acrocentric chromosome (for example, 14 or 21With). Acrocentric chromosomes on the short arms only multiple times in the karyotype. Because they have repeated rRNA genes, they have many functions.Cases are preserved. In any case, a patient with Patau syndrome Karyotype is determined by the formula 47 XX (XY) ,13+. Sometimes in rare cases: Nerobertson translocation, isochromosomal and mosaic forms of trisomy 13 Occurs. 75% have simple trisomy, 25% have Robertsonian translocation.

CAUSES OF PATAU SYNDROME

The basis for the development of Patau syndrome is the 13th chromosome in the karyotype. Is the availability of an additional copy. In most cases (75-80%) one of the parents (most often in the mother) simple complete due to nondissociation of chromosome 13 in meiosis. Trisomy is present. All fetal cells are either 47, XX 13+ or 47, XY 13+ karyotype. Will have Exact causes of triplication of chromosome 13. Not defined. It is known that a genetic defect occurs during the formation of gametes or Can occur during the zygote formation stage. Patau's syndrome in the fetus.There is a correlation between the frequency of development and the age of the mother, but this is a correlation. Less noticeable than Down syndrome. Other factors (infections, mother's. Somatic diseases, bad habits, environmental problems, etc.) and Marriage between relatives, high-dose radiation to a pregnant woman, chemical. Effects of substances, toxins, serious changes in the work of the mother's endocrine system, PS. Can cause. Gametogenesis or genetic mutation in the germ cell. Mostly occurs de novo as a random event. Hereditary forms of Patau syndrome. Due to the presence of a Robertson (balanced) translocation in the parents. A novel Robertsonian translocation causing Patau syndrome in a child. Can be inherited without issue, but certain in subsequent generations. Increases the risk of having a child with an abnormality. Patau syndrome is often fetal.With the formation of several serious defects that lead to intrauterine death.Comes together. Children are usually born on time, but relative to the gestation period. With a small weight of about 2500 g (so-called prenatal hypotrophy). Purpose of the study: during ultrasound examination of the fetus at 11-14 weeks Determination of echographic manifestations of Patau's syndrome.

MATERIALS AND METHODS

9 pregnant women diagnosed with Patau syndrome at 11-14 weeks the results of ultrasound examination were analyzed.

Echographic studies Voluson 730 Expert and Voluson E8(GE) ultrasound was carried out on devices, transanominal and transvaginal sensors were used. Ultrasound examination of each screening performed at 11-14 weeks of pregnancy the results show the main indicators of fetometry (biparietal size and head girth, abdominal girth, femur lengths) and chromosomal abnormalities

exiographic signs (thickness of the collar cavity, length of the nasal bone, venous speed of blood flow in the curves of the canal), ultrasound of the fetus to assess the anatomy and its heart rate, the thickness of the chorion and structure, as well as structural features of uterine appendages and walls is determined. Evaluation of chromosomal ultrasound markers of abnormalities in the fetus.

Examination of the fetus organized by the international medical fund in accordance with the rules.

It was carried out at the Fetal Medicine Foundation. Fetal anatomy study to the methodology proposed by M.V. Medvedev and N.A. Altinnik done according to.

RESEARCH RESULTS

Our research showed that in 9 fetuses with Patau syndrome Various chromosomal anomalies as a result of ultrasound examination at 11-14 weeks various malformations and hisiographic signs were determined. In 5 out of 9 fetuses (55.5%) several birth defects were detected. Prenatal ultrasound in Patau syndrome congenital when the diagnosis is made at 11-14 weeks of pregnancy spectrum of defects is as follows: alobaric holoprosencephaly (3), Dandy-Walker malformation (4), facial cleft (4), congenital heart defects (5), omphalocele (6), polydactyly (4), a single artery in the umbilical cord (3). He had Patau syndrome the listed defects in fetuses were detected during the first ultrasound examination. Echographic signs of chromosomal anomalies are 9 with Patau syndrome found in 7 fetuses (77.8%). Of all the fetuses recorded in the follow-up expansion of the collar space (more than 95%) was found. Hypoplasia and nose bone underdevelopment was noted in 6 (66.6%) fetuses. Most an interesting reference is the curve of the venous blood vessel of a fetus with Patau syndrome speed of blood flow in chzig. Abnormal curves of arterial contraction venous blood in a fetus with Patau syndrome It is distinguished by the appearance of reverse values of blood flow in the vein and 9 found in 5 fetuses (55.5%). It should be noted that in all cases there were abnormal curves of blood flow in the venous blood vessels of the fetus.

Congenital heart defects were diagnosed, in our opinion, this prenatal echographic marker is not only chromosomal abnormalities, but also congenital heart defects allows you to find it important to distinguish high risk groups.

CONCLUSION

In such cases, screening ultrasound at 11-14 weeks of pregnancy Many symptoms of Patau’s syndrome can be determined by examination. This to identify only echographic signs to determine the earliest diagnosis of the syndrome

not necessary, but the chromosome at the end of the first trimester of pregnancy anomalies, as well as ultrasound anomalies of the fetus should be determined.

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