THE PROBLEM OF CONGENITAL DEVELOPMENTAL DEVELOPMENTS IN CHILDREN, DEFORMATIONS AND CHROMOSOME DISORDERS IN MARIUPOL

Abstract. Congenital malformations (congenital malformations) of the fetus are considered the most important medical and social problem, since they occupy a leading place in the structure of the causes of perinatal, neonatal and infant morbidity, mortality and disability. The study is devoted to the analysis of this problem in Mariupol.

Keywords: births, congenital malformations, risk factors, diagnosis.

The issue of planning and birth of a child is one of the most important in human life, with the right approach it is possible to avoid adverse consequences and take care of the health of the unborn child. The share of hereditary pathology makes up a significant part in the structure of childhood morbidity, mortality and disability: 5-7% of newborns have various hereditary pathologies. Congenital developmental anomalies rank first among the causes of childhood disability. The study is devoted to the analysis of this problem.
**Aim:** to compare the birth rate associated with congenital malformations (CHD) in Mariupol for 2019-2020, to identify risk factors and the possibility of timely diagnosis.

**Material and methods.** A retrospective analysis of 41 cards of children's development histories of the neonatal department of MTMO "Child and Women's Health" in Mariupol for 2019-2020 was conducted.

**Results and discussion.** In 2020, 1,363 children were born in the maternity ward of MTMO "Child and Women's Health" in Mariupol, of which 14 children were diagnosed with IBD, which is 1.02% of the total number of births, compared to the previous year 1.1% less.

This year, the structure of congenital heart disease - 5, which is 2 cases less than in 2019.

Congenital heart defects are structural abnormalities and deformities of valves, openings or septa between the chambers of the heart or vessels departing from it, disrupting intracardiac and systemic hemodynamics, leading to the development of acute or chronic circulatory failure [1, 2]. 1 case was detected.

Atrial septal defect - an opening in the atrial septum, which leads to shunting from left to right and overloading the volume of the right atrium and right ventricle. The development of the anomaly can be caused by TORCH infections transmitted during gestation (toxoplasmosis, hepatitis B, syphilis, chickenpox, HIV, rubella, cytomegalovirus infection, herpes simplex virus), diabetes mellitus and other diseases of the endocrine system, drinking alcohol taking medication, complicated pregnancy (toxicosis, threat of miscarriage) [3]. Children are not often symptomatic, but long-term complications after age 20 include pulmonary hypertension, heart failure, and arrhythmias. Adults and, less frequently, adolescents may suffer from exercise intolerance, shortness of breath, fatigue and arrhythmias. Mild mesosystolic murmur in the upper left edge of the sternum with a sharp and constantly bifurcated 2nd heart tone (S2) is common. The diagnosis is established by echocardiography. Treatment consists of the use of a transcatheter device for closure or surgery. 2 cases were revealed.

A ventricular septal defect is an opening in the interventricular septum that results in communication between the ventricles. Large defects lead to a significant
discharge of blood from left to right and are the cause of shortness of breath during feeding and low growth rates during childhood. The reasons include: genetic factor, pathogenic effects of environmental conditions, ionizing radiation, exposure to alcohol and toxic chemicals [4, 5]. Loud sharp holosystolic murmur at the lower left edge of the sternum is common. Recurrent respiratory infections and heart failure may develop. The diagnosis is established by echocardiography. Defects may close spontaneously in childhood or require surgery. 2 cases were revealed.

Congenital anomalies of the genital area - 4, which is 4 cases less.

Cryptorchidism - not lowering one or both testicles into the scrotum; usually accompanied by inguinal hernia. The reason for the development of cryptorchidism can be chromosomal defects, abnormalities in the development of the inguinal canal, prematurity, the negative impact of external factors on a pregnant woman, due to which there are pathological changes in the fetus. Negative factors include: medication during pregnancy, viral diseases, hormonal disorders, smoking, alcoholism, drug addiction, exposure to toxic substances, hereditary factors [6]. The diagnosis is established at inspection, sometimes with the subsequent laparoscopy. Treatment - surgical orchypexia. Not dropping the testicles can cause reproductive failure and is interrelated with testicular cancer. 3 cases were revealed.

Hypospadias - this anomaly is caused by violation of tubularization and fusion of the urethral sulcus. The true causes of the pathology are unknown. Possible causes include: genetic disorders, intrauterine infections, multiple pregnancy, hormonal disorders in the mother during pregnancy, the effects of stress. It almost always occurs in boys in whom the urethra opens on the underside of the body of the penis, on the scrotal-stem joints, between the folds of the scrotum, or in the perineum. The foreskin cannot become circular and looks like a hood. Operative hypospadiology is one of the most difficult areas of pediatric urology, with a significant level of complications, even in the experienced hands of a surgeon. 1 case was detected.

Anomalies of the upper and lower extremities - 2, which is 5 cases less.

Postaxial polydactyly is the presence of an additional finger on the limb. This is the most common form of such deformation. Usually, the extra finger is
rudimentary, but can be fully functional. Its formation is associated with the syndrome of multiple congenital anomalies or chromosomal defects.

A simple form of syndactyly is the membranous or fused fingers or toes. In most cases, it is transmitted by autosomal dominant type of inheritance. Simple syndactyly involves only the fusion of soft tissues.

For diagnosis, radiography is usually performed to determine which bones are affected by the pathology. Treatment of polydactyly and syndactyly is performed surgically.

Oral clefts, in particular the cleft palate is bilateral - 1, which is 1 case less.

Oral clefts are the most common congenital anomalies of the head and neck. The reasons are both environmental and genetic factors. Maternal smoking and alcohol use in the prenatal period may increase the risk. Having one sick child increases the risk of giving birth to another with a defect. Supplemental folic acid intake during pregnancy planning and during the first trimester reduces the risk of developing defects. Splitting of the palate complicates feeding and interferes with the development of speech, increases the risk of ear infections. The goals of treatment are to ensure normal feeding, speech and development of the facial skeleton, as well as to prevent postoperative defects. Treatment is surgical closure of the gorge.

Agenesis of the corpus callosum - 1 - new cases that were not recorded last year.

Agenesis of the corpus callosum (AMT) is manifested in cerebral dysgenesis associated with various hereditary syndromes. The causes of abnormalities in the development of the corpus callosum can be genetic, infectious, vascular or toxic. It is traditionally divided into total (no commissural fibers) and partial (agenesis of the rostral and caudal parts of the corpus callosum). AMT can occur in isolation or in combination with other malformations of the brain. Isolated disorders of the corpus callosum may not appear clinically, which greatly complicates the timely diagnosis of this pathology. The presence of AMT can be confirmed by various methods of neuroimaging, including prenatal ultrasound of the brain. 1 case was detected.

Down syndrome - 1 - new cases that were not recorded last year.
Down syndrome is an abnormality of chromosome 21, can be manifested by impaired mental development, microcephaly, small growth and a characteristic appearance. It is believed that the probability of having a child with Down syndrome increases with age. The causes of this disorder are not fully understood, there is an assumption that chromosome imbalance in somatic cells leads to dysfunction of the genotype \[4, 5\]. The diagnosis is assumed on the basis of the presence of physical abnormalities and developmental abnormalities and confirmed by cytogenetic analysis. Management depends on specific manifestations and anomalies. 1 case was detected.

Also in 2019, cases of spina bifida - 2 were detected.

Spinal cleavage is one of the most serious neural tube defects compatible with longevity. It is most common in the lower thoracic, lumbar or sacral region and usually lasts for 3-6 vertebral segments. The severity of clinical manifestations varies from occult, in which the spinal cleft is not accompanied by a spinal hernia, to the variant of the spinal fissure with obvious external manifestations in the form of a spinal cord defect (myelomeningocele) with severe neurological disability and death \[7\]. Folic acid deficiency is an important factor in the development of the defect \[8\]. There is paralysis of varying severity and impaired sensitivity below the lesion. Lack of muscle innervation contributes to the growth of atrophic changes in the leg muscles and the development of orthopedic deformities. Prenatal screening is performed using fetal ultrasound and determination of \(\alpha\)-fetoprotein levels in the mother's serum. Shunting is performed as a symptomatic treatment to eliminate spinal cord injury, as well as orthopedic and urological disorders.

Esophageal atresia was detected in 1 case.

This is the most common gastrointestinal atresia. Two syndromes are specifically associated with esophageal atresia: VACTERL (from English Vertebral anomalies, Anal atresia, Cardiovascular malformations, Tracheoesophageal fistula, Esophageal atresia, Renal anomalies, Radial aplasia, Limb anomalies - anomalies of the spine, pores, and pore radial bone hypoplasia, limb defects)

CHARGE symptom complex (from English Coloboma - coloboma, Heart defects - heart defects, Atresia of the choanae - choan atresia, Retardation of mental
and/or physical development - growth retardation and development, Genital hypoplasia - external genital hypoplasia organs, Ear abnormalities - ear abnormalities). Clinical manifestations include excessive secretion, cough and cyanosis after feeding attempts and aspiration pneumonia. Prenatal diagnosis is an ultrasonography. It is treated by surgery.

The survey data identified the leading regional socio-economic, medical-biological and environmental risks of VVR (with a probability of p <0.05): young mother from 15 to 24 years, first pregnancy, history of miscarriage, threat of termination, the presence of toxicosis in 1 trimester, medication and infectious diseases during this pregnancy, smoking, unbalanced diet, low educational qualifications, the presence of occupational hazards in the anamnesis (including the father), lack of work, negative living conditions, living in the area location of industrial enterprises and major highways. For early diagnosis of chromosomal mutations and some common congenital pathologies, the doctor provides information on the feasibility of the first ultrasound (11 weeks + 1 day - 13 weeks + 6 days) and a double biochemical test (free-HCG, PAP-A). Also ultrasound-2 during pregnancy from 18 weeks to 20 weeks + 6 days to detect structural abnormalities of the fetus. At detection of low placentation, marginal attachment of a placenta, in addition carried out ultrasound at 32 weeks of pregnancy.

**Conclusions.** According to the report diseases in 2020 - 10.3% of the total number of sick newborns had congenital malformations of varying severity, some of which will have to undergo complex operations or had severe malformations that can not be treated. To avoid this, parents should take the issue of pregnancy as seriously as possible, try to protect themselves as much as possible from risk factors, consult a perinatologist or geneticist. Significantly increases the chance of giving birth to a healthy child, the absence of abortion and miscarriage in the history of women, the absence of cardiovascular disease, the period of more than 2 years between previous and actual pregnancy, a balanced diet, good living conditions. Detection of chromosomal pathology based on screening tests in the 1st trimester of pregnancy is 90%. Research is conducted only with the permission of a woman!
References:


