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SIRENOMELIA AS EXAMPLE OF LIMB MALFORMATION

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Abstract. *The article is devoted for syndrome mermaid - very severe syndrome of caudal regression (complex malformation the caudal portion of the embryo), which is a rare severe congenital malformation of the distal spine and spinal cord (his clinical picture is accompanied by hypoplasia of the lower half of the trunk and extremities, a fusion of the lower limbs). We discuss the etiology, classification, pathological anatomy, the prognosis of this disease.*

Keywords: *fetus, sirenomelia, congenital defect*

Congenital limb malformations rank behind congenital heart disease as the most common birth defects observed in infants [8]. A well know example of such pathology is the wave of thalidomide-induced malformations that occurred in middle of XX century. The term «sirenomelia» («mermaid syndrome») is derived from the physical similarity of the affected fetus to mythical creatures mermaids - charming women with the lower part of the body in the form of a fish tail, where there is a fusion of the lower extremities and partial or complete fusion of the feet [7]. It is a lethal developmental defect characterized by different degrees of fusion of the lower extremities in association with lumbosacral and pelvic bone abnormalities, blind-ending colon, absent external genitalia, renal agenesis and SUA [6]. The same applies regarding its relationship with narrow pelvis syndrome and VATER (vertebral defect, anal atresia, interauricular communication; interventricular communication, tracheal and esophageal atresia, and renal or radial agenesis) syndrome [2]. The diagnostic and management aspects of sirenomelia in twin pregnancies as extremely rare case was described by V. Nisenblat and coauthors [4].

The syndrome of the mermaid (fig.1) - a very severe form of the syndrome of caudal regression (complex malformation the caudal portion of the embryo), which, in turn, is a rare severe congenital malformation of the distal spine and spinal cord (his clinical picture is accompanied by hypoplasia of the lower half of the trunk and

extremities, fusion of the lower limbs). Fusion can be within the bone or soft tissues only. There are renal agenesis, blindly ending colon, the lack of external and internal genitalia, single umbilical artery atresia of the anus in most cases of sirenomelia.

Fig.1. Antenatal died fetus with sirenomelia from collection of Pathology Anatomy Department of Kharkiv National Medical University. A 16-year-old pregnant with addictive injections in anamnesis. She reported no familial history of congenital anomalies or diabetes.

The incidence of this fatal defect - 1 for 60 thousand newborns; sex ratio - 2.7:1 (male:female). Type of inheritance - presumably autosomal dominant. Sirenomelia is almost always a fatal disease because birth defects named above. Approximately 50% of infants with this diagnosis are stillborn. However, babies who are born alive, usually don't live long: after a few minutes or a couple of days later, rarely - a few months due to renal anomaly or anomalies of the urinary bladder.

Children with sirenomelia have no chromosomal abnormalities. Defect occurs in the blastula stage, gestation 1 to 28 days. The reason for this violation of the main blood vessels of the body with abnormal circulation of blood in the distal parts of the embryo, followed by violation of development of the child's lower body. Violation of the blood supply leads to the disturbance of tissue differentiation in that area and so severe defects.

The etiology of the syndrome of caudal regression is not finally clarified. Animal experiments have shown that the syndrome of caudal regression may occur when exposed to retinoic acid, diethylpropion, lithium, sulfonamides, cadmium, ochratoxin A, vitamin A deficiency, radiation, hyperthermia, organic solvents, 6

aminonicotinamide. Stevenson reported in 1986 that the etiology of sirenomelia was explained by vascular steal theory [3]. Most authors causative factors is considered maternal diabetes, genetic predisposition, and insufficient blood supply to the lower half of the body of the fetus in the genesis of this pathology. In a normal embryo develops two umbilical arteries that pump blood from the fetus to the placenta and one umbilical vein, which returns blood from the placenta to the fetus. Most babies with the syndrome of the mermaid have only one umbilical artery and one vein. Rarely embryos with this syndrome develop typical two arteries and one vein. One functional artery is more developed than usual and it is a branched from the aorta in high level in the abdominal part. At the same time the aorta becomes narrow wrong. Due to the fact that the lower extremities are not supplied sufficiently and they lack the normal blood flow, they are not formed as separate extremities also kidney are natformed, colon ending blindly into the abdominal cavity, the internal and external genitalia are absent or are developed with disabilities in such disorders. Currently, scientists have not yet determined why only one umbilical artery can cause such changes.

The clinical picture and pathological anatomy: caudal regression syndrome in its extreme form (sirenomelia) fatal. The complex malformations is incompatible with life. Children are missing kidneys and renal arteries, there is hypoplasia of the lungs, causing them to death in the first hours and days after birth. At the birth of a child found fused lower limbs, with hypoplastic pelvis, narrowing, there is the absence or hypoplasia of the caudal spine (sacrococcygeal vertebrae). In general, when viewed from a child marked narrowing of the lower part of the body and shortening it by the length of the lower extremities.

Fusion of the lower extremities can be involving the bones may be skeletal anomalies of the lower limbs in the form of lack of fibula synostosis and hypoplasia of other bones, congenital dislocation of the hips, deformed or non stop. The feet are fairly well formed, divided, resembling a mermaid's tail (hence the term - sirenomelia). There may be a lack of feet or having one foot only. When the feet are formed they

are deformed by the type of clubfeet usually. Flexion contractures and abnormal development of the joints of the lower extremities are typical.

Two separate systems of classification of sirenomelia have been proposed, one based on the number of feet and the other based on the fused bones (Stocker-Heifetz classification) [1,2]. We support that sirenomelia sequence is classified into three groups according to the number of feet present (Sarpong & Headings 1992). The most common of the three conditions is symelia apus, in which both legs are merged completely into a single lower extremity. In this condition both feet are absent or rudimentary. Symelia unipus shows a presentation of one foot, two femora, tibiae and fibulae. In symelia dipus, two distinct feet are present but are malrotated and resemble fins [3]. In the late second and third trimester, symelia unipus and dipus are difficult to diagnose with ultrasound because the bones of the thighs and legs are fully formed. In addition, severe oligohydramnios makes prenatal sonographic visualization of the lower extremities very difficult. Color Doppler imaging can be useful in identifying aortic bifurcation and renal arterial flow to assist in the prenatal diagnosis of sirenomelia [1, 3].

Genitals may be present, but almost always there is a problem of sex determination of the fetus; the child may have a cesspool. As a rule, in all cases, the anus is absent; there is atresia of the anus and rectum. There are disturbance of innervation of the caudal part of the body, characterized by paresis and paralysis.

Diagnosis: When a child is born the diagnosis is obvious. Prenatal diagnosis is important because the presence of such a malformation of the fetus is an indication for abortion. Since the defect is formed very early, it can be diagnosed at the first ultrasound screening for the term of 11-12 weeks. Ultrasound examination of such fetus is always characterized by oligoamnios; kidney are not detectable a in the fetus, and there is a characteristic abnormality of the lower extremities. It can be considered a marker of the syndrome, the presence of this anomaly is reason enough for a diagnosis of "sirenomelia" and abortion.

A diagnosis of sirenomelia may be easier during the first trimester because the amniotic fluid volume is relatively normal, since amniotic fluid is secreted by the

amniotic membrane covering the placenta and the umbilical cord in the first trimester. A diagnosis of sirenomelia is made in early pregnancy by confirmation of the existence of a single lower limb, but few ultrasonographs confirm all of the limbs in early pregnancy because sirenomelia as so rare[5].

Sirenomelia is fatal in most cases because of the characteristic pulmonary hypoplasia and renal agenesia [2]. About 50% of the children are born alive after eight or nine months of pregnancy. Death occurs in the five days following birth. Post-natal management requires the presence of kidneys, even if they are dysgenesic [2]. Prognosis is very poor because of the condition involves variable major anomalies, including bilateral renal agenesis, sacral agenesis and imperforate anus. Only four cases of a surviving infant with sirenomelia have been reported [3]. The death of a child usually occurs during the first hours and days of life. The cause of death is lung hypoplasia and renal failure.

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Сиреномелія як приклад пороку розвитку кінцівок

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Резюме. Стаття присвячена синдрому русалки - вкрай важкій формі синдрому каудальної регресії (комплекс вад розвитку каудальної частини ембріона), який, у свою чергу, є рідкісним важким уродженим пороком розвитку дистального відділу хребта та спинного мозку (його клінічна картина захворювання супроводжується гіпоплазією нижньої половини тулуба і кінцівок, злиття нижніх кінцівок). Обговорюються етіологія, класифікація, патологічна анатомії, прогноз даної патології.

Ключові слова: плід, сиреномелія, вада розвитку

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Сиреномелия как пример порока развития конечностей

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Резюме. Статья посвящена синдрому русалки – крайне тяжелой форме синдрома каудальной регрессии (комплекс пороков развития каудальной части эмбриона), который, в свою очередь, является редким тяжелым врожденным пороком развития дистального отдела позвоночника и спинного мозга (его клиническая картина заболевания сопровождается гипоплазией нижней половины туловища и конечностей, слияние нижних конечностей). Обсуждаются этиология, классификация, патологическая анатомии, прогноз данной патологии.

Ключевые слова: плод, сиреномелия, врожденный дефект

Received: 25.04.2015 *Accepted: 3.07.2015*