MODERN APPROACHES TO PRENATAL DIAGNOSIS OF CONGENITAL MALFORMATIONS OF THE URINARY SYSTEM. LITERATURE REVIEW

Lastivka Iryna,
PhD, Associate Professor
Bukovinian State Medical University

Antsupova Vita,
PhD, Associate Professor,
Bogomolets National Medical University

Brisevac Ljudmila,
Assistant Professor
Shupyk National Medical Academy
of Postgraduate Education

Diagnosis of congenital malformations (CM) before the birth of a child is an important aspect of modern prenatal medicine. Disorders of the urinary system among all antenatally diagnosed defects is 27%. Qualitative diagnosis of the fetus can significantly increase the reliability of medical and genetic prognosis, and reduce the likelihood of birth of children with severe incurable US. Recent studies show that the formation of complications of fetal development occurs within 12 weeks. Therefore, the first screening at 11-14 weeks of gestation is optimal for the diagnosis of most lethal and disabling US and chromosomal abnormalities [1-4].

For the first trimester of gestation, the manifest ultrasound marker of CM urinary system is Megacystis syndrome (MS). MS is an increase in the size of the bladder of the fetus more than 10 mm in the first trimester of pregnancy. The frequency of detection of MS in the first trimester of pregnancy is 0.02-0.19%. In the second and third trimesters of gestation it is possible to diagnose the symptom of "keyhole", pathognomonic for valvular obstruction of the urethra; intestinal dilatation, which indicates in favor of megacystis-megaureter-microcolon syndrome; thinning of the anterior abdominal wall, characteristic of Prun-Bella syndrome. Detection of these signs at a later date complicates the prognosis [1, 4-6].

The most unfavorable consequence of the obstructive condition of the bladder is the formation of cystic dysplasia of the renal parenchyma and is the cause of renal decompensation. The complexity of prognostic decisions in the diagnosis of MS in early gestation is hampered by the lack of such an important feature as dehydration, because by the time of functioning of the fetal urinary system amniotic fluid is the result of amniotic membrane production. Only from 14 weeks the process of formation and excretion of urine by the fetus begins, which gradually becomes a priority in the formation of amniotic fluid [1, 2, 4].

Given that MS in 25-40% of cases is combined with chromosomal pathology, a significant role in deciding whether to prolong or terminate a pregnancy is played by the results of genetic research, karyotyping of fertile material. If the enlargement of the bladder is caused by obstruction, the fetuses are more common trisomies on 13, 18, 21 chromosomes, monosomy 21 chromosomes, deletion of chromosome 13, translacation [1, 4, 6].

Recommendations of management of the pregnant woman are defined by the sizes of a bladder: at the sizes of 7-15 mm in all cases karyotyping of a fruitful material in connection with existence of high percent of the combined pathology is carried out. If the test for trisomy is positive, abortion is recommended. With a normal karyotype, it is reasonable to monitor the pregnancy to determine the size of the bladder. With the normalization of the latter, the pregnancy is prolonged. When increasing the size of the bladder from 15 to 30 mm, this situation is treated as an obstructive condition of anatomical or functional nature and abortion is performed, followed by karyotyping of fetal material, which is important for predicting the next pregnancy. Indications for prolongation or termination of pregnancy in the presence of MS in the fetus require active discussion, analysis of pregnancies, the development of clear prenatal objective criteria that confirm the prognosis, which will justify the unity of tactics in solving this problem at the antenatal stage [2-6].

The reasons for the development of MS are: 1) urethral abnormalities; 2) syndrome of posterior urethral valves (SPUV); 3) vesicoureteral reflux (VCR); 4) prune belly syndrome (PBS); 5) megacystis-microcolon-interstitial hypoperistaltic syndrome (MMIHS). Obstructive are the valves of the posterior urethra, functional – PBS, MMIHS. The most common cause is SPUV, MMIHS, as a rule, occurs in females, and PBS and obstructive conditions – in males [1, 2, 5].

SPUV – violation of urethral patency with retrograde changes of urinary system. SPUV accounts for 38% of all obstructive uropathies at a low level and is observed in males. Very rarely is in females, a similar pattern can be caused by agenesis/atresia of the urethra. During prenatal ultrasound in SPUV there is a stable dilatation of the bladder, which does not take place in the dynamics. The walls of the bladder are thickened to 2 mm, may be calcified. There is an expansion of the proximal urethra, with the progression of the process – the expansion of the higher departments of the urinary system. Urinary ascites, urine may be observed. Dehydration (50-60%) worsens the prognosis because it leads to pulmonary hypoplasia. Vesico-amniotic shunting allows to achieve survival of 70% of fetuses. Prognostic ultrasound criteria are the amount of amniotic fluid, the timing of detection of severe dehydration, duration and severity of renal impairment. Before shunting, prenatal karyotyping should be performed [1, 2, 4].

PBS – hypotension or complete absence of muscles of the anterior abdominal wall, megacystis with wall hypertrophy in combination with megaureter and hydronephrosis and bilateral cryptorchidism in males. Frequency – 1: 35-50 thousand babies. The type of inheritance is unknown. It is 20 times more common in boys. Gradual accumulation of urine in the abdominal cavity from 7-8 weeks of fetal development, leads to the development of the syndrome. Almost all patients have prostate hypoplasia, which probably triggers a chain of disorders. A swollen bladder prevents the testicles from

sinking, prevents the intestines from turning, and can constrict the iliac arteries, inhibiting the development of the lower extremities. Cystic degeneration of the kidneys can occur due to high urine pressure in the urinary system [1, 3-5].

Because the fetus does not excrete urine into amniotic fluid, dehydration develops, which entails a complex of consequences – Potter's syndrome. Severe obstruction of the urethra is incompatible with life, fetal death can occur in the II-III trimester of pregnancy. Urine can flow through the open urethra (urachus) or through a rupture in the valve of the bladder or ureter. When the pressure in the abdominal cavity drops, the stretched anterior abdominal wall falls and gathers in folds ("plum belly"). Most babies die after birth due to severe kidney damage. Survivors are operated on to restore urine flow and prevent infectious complications. The probability of rebirth of a sick child depends on the cause of the obstruction. Prenatal diagnosis is made at 10 weeks of pregnancy, when a distended bladder is detected. At the beginning of the second trimester of pregnancy the main ultrasound sign is megacystis, in the second-third – a sharply enlarged bladder with hypertrophy of its walls and thinning of the anterior abdominal wall. At the expressed process the bilateral megaureter, hydronephrosis and dehydration, urinary ascites join. Prenatal examination should include karyotyping and thorough ultrasound to examine the anatomy of the face and internal organs of the fetus. In case of death of the fetus or newborn, a pathological examination is indicated. At detection of PBS which is combined with the expressed low water, it is necessary to terminate pregnancy in connection with seriousness of the forecast. Urethral obstruction leads to the development of severe dehydration and pulmonary hypoplasia. Antenatal death occurs in 20%, 50% of children die in the first two years of life. The success of surgical correction depends on the degree of involvement in the pathological process of organs and systems. Intrauterine decompression of the bladder can prevent the development of a detailed clinical picture of the syndrome [1, 3, 5].

MMIHS - consists of incomplete bowel rotation, functional intestinal obstruction, distal microcolon and dilated non-obstructive bladder. About 200 cases have been described in the literature. Autosomal recessive type of inheritance should be excluded during medical and genetic counseling. The cause of the disease is an immune inflammatory process with damage to the gastrointestinal tract, which leads to the deposition of fibrous connective tissue between the muscular layers with the destruction of intramural nerve plexuses, manifested by hypoperistalsis. Such changes that develop in the bladder are manifested by its overstretching, the development of secondary retention disorders in the upper urinary tract. The prognosis is disappointing – life expectancy does not exceed several months. The main cause of mortality is the complication of total parenteral nutrition. Prenatal MMIHS is diagnosed by identifying non-obstructive urinary excretory tract dilatation: bladder dilation, renal hydronephrosis in a female fetus with normal or mild enlargement of amniotic fluid. Bowel changes are diagnosed prenatally in about 19% of cases and more often in the third trimester. Polyhydramnios is not expressed and occurs in 50% [1, 5-7].

Thus, visualization of the kidneys and bladder of the fetus is possible and necessary, given the unfavorable prognosis for life, from early pregnancy. Differential diagnosis of obstructive and non-obstructive nature of the lesion and dynamic observation are required in the detection of urinary system. Due to the fact that early

screening does not allow to determine the likelihood of the presence in the fetus of CM urinary system, the risk group should be formed on the basis of the diagnosis of increased longitudinal size of the bladder. MS – is the basis of severe obstructive disorders of urodynamics and renal dysplasia with a serious prognosis. The set of diagnostic procedures to clarify the prognosis should include karyotyping of fetal material (chorionic villi). Genetic studies should be performed in all cases of detection of MS of the first trimester of gestation. Decide to terminate a pregnancy in the presence of combined genetic pathology and continue fetal monitoring in the absence of such in the hope of spontaneous self-resolution of the process or detection of additional diagnostic markers of urinary system pathology, determine decision-making tactics.

References:

1. Kenneth L. Jones. Hereditary syndromes according to David Smith. Atlas-reference, 2011: 746-747.

2. Bonilla Musoles F., Raga F., Bonilla F.Jr., Machado L.E., Osborn N., Ruiz F., Castillo Jc. Prune-belly Syndrome versus Posterior Urethral Valve. Donald School Journal of Ultrasound in Obstetrics and Gynecology, 2010; 4(4): 405-417

3. Спахи О.В., Лятуринская О.В., Макарова М.А. Врожденные пороки развития передней брюшной стенки (синдром внешних деформаций). Омфалоцеле. Гастрошизис. Синдром Вильяма-Ослера. Запорожье, 2016: 70.

4. Toshihiko Nakamura. Isolated congenital megacystis with spontaneous resolution: a case report. Fukushima J. Med.Sci., 2017; 63(2): 116-120

5. Gloor, J.M., Torres V.E. Reflux and Obstructive Nephropathy. Atlas of diseases of the kidney, 2008; 2(8).

6. Manop Jr., Chamnanvanakij S., Wattanasarn Ch. Megacystis Microcolon Intestinal Hypoperistalsis Syndrome (MMIHS): a case report in Thailand. J Med Assoc Thai 2004; 87(11): 1385-8

7. Halim D., Hofstra R., Signorile L. et al . Alves, ACTG2 variants impair actin polymerization in sporadic Megacystis Microcolon Intestinal Hypoperistalsis Syndrome Human Molecular Genetics, 2016; 25(3): 571–583

INNOVATIVE TECHNOLOGIES IN SCIENCE AND EDUCATION

Scientific publications

Materials of the IX – the International Science Conference «Innovative technologies in science and education», Jerusalem, Israel. 327 p. (March 04 – 06, 2021)

UDC 01.1

ISBN – 978-1-63732-147-8

DOI - 10.46299/ISG.2021.I. IX

The content and reliability of the articles are the responsibility of the authors. When using and borrowing materials reference to the publication is required. Collection of scientific articles published is the scientific and practical publication, which contains scientific articles of students, graduate students, Candidates and Doctors of Sciences, research workers and practitioners from Europe, Ukraine, Russia and from neighboring countries and beyond. The articles contain the study, reflecting the processes and changes in the structure of modern science. The collection of scientific articles is for students, postgraduate students, doctoral candidates, teachers, researchers, practitioners and people interested in the trends of modern science development.

The recommended citation for this publication is: Liubych V.,
Baking properties of spelt wheat // Innovative technologies in science and education. Abstracts of IX International Scientific and Practical Conference. Jerusalem, Israel 2021. Pp. 12-14.

URL: https://isg-konf.com.