ETIOPATHOGENETIC FEATURES OF THE ANOMALIES OF THE URINARY SYSTEM

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Introduction. Anomalies of development of the genitourinary system usually appeare due to a genetic mutation and can be expressed both in the underdevelopment of individual organs and in hyperdevelopment, up to doubling. They are widespread, make up about 40% of all congenital anomaly and are capable of leading to serious complications of a psychological and physiological nature. Specific causes of the most of anomalies are unknown. Renal agenesis, hydronephrosis, poly- and multicystosis of the kidneys, stenosis of the urethra are the most common anomalies of the urinary system. Due to the close interaction between the development of the urinary and reproductive system in 33% of cases, abnormalities of the organs of the urinary system are combined with anomalies of the genital organs, and the developmental defects of the genitourinary system are often associated with the developmental defects of other organs and systems.

Aim. To analyze the etiology and pathogenesis of agenesis and polycystic kidneys based on the study of modern scientific literature.

Results and discussion. The polycystic kidney is a genetically determined pathological process, which is associated with the formation and progression of cysts in the kidneys originating from tubule epithelial cells and (or) collecting tubules, represented by two types of disease - autosomal dominant and autosomal recessive. This is a difficult bilateral process leading to chronic pyelonephritis, arterial hypertension and chronic renal failure. Frequency of occurrence is 1 case per 400 autopsies. This anomaly is divided into polycystic kidney disease in children and adults. For polycystic childhood autosomal recessive inheritance is characteristic, for polycystic adults – autosomal dominant. In children, this anomaly is severe, most of them do not live to adulthood. Polycystic in adults has a more favorable course, manifested in young or middle age, is more common in women, and for many years has been compensated.

The autosomal dominant type of the polycystic kidney is caused by mutations in the genes PKD1 (chromosome 16p13.3) and PKD2 (chromosome 4q21). These genes code of proteins are called polycystins 1 and 2 (PC1 and PC2). The mutation PKD1 is main (85-90 %). Autosomal recessive polycystic kidney disease is caused by a mutation of the PKHD1 gene (chromosome 6p21), with a

25% risk of disease in the offspring. The protein product of the PKHD1 gene is fibrocystin found in the primary cilia and centrosomes. Allelic heterogeneity of the gene determines significant differences in phenotypic concordance both within the same family and between different families. The mechanism of development consists in the dissonance of the connection of the primary tubules of the metanephrogenic blastema with the methanephros flow, which leads to an incorrect development of the secretory and excretory segments of the nephron, i.e. straight and convoluted tubules. As a result, urine outflow from the proximal parts of the nephron is disturbed, an expansion of the blindly terminating tubules and the formation of cysts from them. The growth of cysts causes ischemia of unchanged renal tubules and death of kidney tissue. This process is facilitated by the adhering chronic pyelonephritis and nephrosclerosis.

Unlike polycystosis, renal agenesis is attributed to congenital diseases, which is formed in the first six weeks of fetal development, and refers to anomaly of quantity. This anomaly can be combined with a one- or two-sided absence of the ureters and bladder. The etiology is heterogeneous, possibly its multifactorial origin. In most cases, renal agenesis occurs sporadically. The causes of development include the effect on the mother's body of exogenous factors during pregnancy: viruses (measles, rubella, herpes), venereal diseases (especially syphilis), alcohol abuse, exposure to radiation or toxic substances (bilirubin, ammonia, phenol). A special risk group is women with diabetes. It is established that 4-8 % of children are born with renal agenesis. In boys, the disease occurs twice as often. At the basis of pathogenesis lies the absence or stoppage in the development of the Wolfov duct on the corresponding half of the urinary tract. A disturbance in the formation of the ureter of the embryo subsequently prevents the full development of the organ and is accompanied by the absence of the ureter, its mouth and the corresponding half of the urinary bladder.

Conclusions.

1. It is determined that polycystosis belongs to hereditary diseases, and agenesis – to congenital

2. There are gender differences in the frequency of occurrence of the anomalies studied – polycystosis is more common in women, and agenesis in men.

3. Polycystic is a defect in the structure, and agenesis is a defect in quantity.