Early Diagnosing of Urinary Tract Anomalies

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Abstract

Nowadays, with advanced diagnostic technology and accessibility of patients to qualified physician, many anatomical anomalies are diagnosed in uterine life, including urinary tract anomalies in prenatal period, however, even with advanced technology, many of such anomalies continue to be diagnosed later on, in early stage of life or even later when a medical problem arise as a consequence of these anomalies. In Albania, nowadays, high technology and high definition diagnostic imaging machines are available; however, we do face lack of qualified and experienced medical specialist, especially in peripheral regions - city hospitals throughout Albania. Early diagnosing of anomalies like urinary tract ones, are possible to be diagnosed since the end of first trimester of pregnancy by 14 weeks, and later on during the anatomic ultrasound examination or otherwise called morphologic fetal ultrasound at 18 – 22 weeks of pregnancy, if examination carried out by a qualified medical specialist, should clearly visualize fetal urinary tract. However, this fetal problems undiagnosed during pregnancy, there are often diagnosed in postnatal period. In some countries, many ultrasound examinations during pregnancy are performed by ultrasound technicians and interpreted by radiologist or perinatologist, however, generally these examination are performed by fetal medicine specialist, or obstetrician and gynecologists. In Albania these examination are performed only by obstetrician and gynecologist, since we do not have a fetal medicine center. Any urinary problem during early age, it is appropriate to perform a general abdominal ultrasound examination as fast, reliable and cost-effective examination tool with no side effect.

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Introduction

Congenital anomalies of urinary tract system are among most common ones.1 Urogenital tract system normally represents two main components: urinary and genital system. Until 15-16 weeks of pregnancy rhythm of urogenital development is very rapid and later on decreases.1, 2

With meticulous examination 1 in 100 - 500 neonate may have an anomaly of urinary system, respectively congenital anomalies of kidney and urinary tract.2 Various congenital anomalies do not have signs and symptoms and represent only numerical diagnostic anomalies, some of them may be incapability with life like bilateral renal agenesis. Other urinary tract anomalies may be responsible for obstructive urinary tract pathologies like posterior valve of urethra, stenosis of pelvic-urinary segment, strictures of ureters, ureterocele. Later on, some urinary tract anomalies may be responsible for chronic urinary infections, vesicourethral reflux. In addition, we have some urinary tract anomalies that are base of development of tumors, like urinary vesicle extrophy. Congenital anomalies of kidney and urinary tract may be part of multi-organ processes in single-gene disorders, with dominant or recessive inheritance, as we can find in Fraser syndrome, the branchiootorenal syndrome, Kallmann syndrome, Ehlers-Danlos syndrome and others.4

Discussion

Causes of congenital anomalies of kidney and urinary tract are complex. It is likely that mixture of genetic and environmental factors contribute to development of kidney and urinary tract abnormalities. Genetic factors involved in most cases of congenital anomalies of kidney and urinary tract are unidentified. Syndrome of congenital anomalies of kidney and urinary tract it is thought to be caused by changes in genes associated with specific syndrome. Variations in these same genes can also underlie some cases of isolated congenital anomalies of kidney and urinary tract.

The fetal kidneys can be visualized from 10 weeks of gestation and are seen in almost all fetuses at 14 weeks using transabdominal ultrasound (ultrasound illustration figure 1).5, 6 They initially appear in the transverse plane as hypoechoic oval structures on both sides of the fetal spine. In the longitudinal axis, they appear along the paravertebral plane of the fetal spine. A detailed view of the fetal kidneys by transvaginal sonography has enabled development of normograms for the kidney diameters at the first trimester.5
Bilateral renal agenesis is usually diagnosed at the beginning of second trimester due to anhydramnios. Since most of the amniotic fluid before 11 weeks is not produced by the fetal kidneys, this absence of amniotic fluid cannot help to detect renal agenesis during the first trimester. However, prenatal diagnosis of bilateral renal agenesis has been reported after 12 weeks gestation.\textsuperscript{6,7,8}

Internationally, congenital anomalies of kidney and urinary tract are responsible for 40% to 50% of pediatric and 7% of adult end-stage renal disease.\textsuperscript{9} Pathogenic variations in genes causing congenital anomalies of kidney and urinary tract include monogenic diseases such as polycystic kidney disease (illustration figure 2) and ciliopathies, as well as syndromes that include isolated kidney disease in conjunction with other abnormalities.

Antenatal diagnosis most often follows using ultrasonography; however, further genetic diagnosis may be made employing a range of testing approaches. Family history and pathologic examination also provide information to improve ability to make a prenatal diagnosis of congenital anomalies of kidney and urinary tract.

In conclusion, diagnostic imaging is playing an important role nowadays, especially with advanced technology incorporated in such service, together with high resolution imaging and of course trained and qualified physician, makes early diagnosing, such a urinary tract anomalies possible in very early stage like intrauterine life. Albanian state authorities in charge of medical political strategies and management should emphasize the role of radiologist in order to employ their clinical skills in bigger accessibility to health care service for a better health care in general.

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