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Prenatal diagnosis of urinary track defects

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Abstract Introduction
Congenital malformations are the main cause of miscarriage, perinatal mortality and disabil among children. According to Polish Registry of Congenital Malformations for 2005-20 congenital disorders affect 2.0-4.0% of new-borns. Heart defects, musculoskele abnormalities and urinary track defects are the most common.
Purpose
The article aims to shortly describe characteristic ultrasound findings concerning cert urinary tract defects.
Brief description
Renal abnormalities result from complexity of kidneys embryogenesis.
Obligatory ultrasound screenings during pregnancy enable early prenatal diagnosis of urin track defects and it is helpful for planning postnatal or prenatal treatment.
Oligohydramnios is characteristic for urinary tract congenital disorders. Lack of re parenchyma, increased echogenicity of renal parenchyma, difficulties in bladder visualisati cysts within kidneys, hydronephrosis always indicate abnormalities concerning urinary track.

Visualisation of urinary track defect obligate further screening for accompanyi abnormalities. Sequence of defects, plural abnormalities are indications for genetic evaluati in referral medical centres.
Postnatal treatment of congenial urinary track disorders includes watchful observation, ea surgical treatment or palliative care. The prognosis depends on the type of defect a accompanied anomalies.
Summary / conclusion
Urinary track defects in prenatal period are very common. Every child suspected for urin track defect requires detailed evaluation in referral medical centre including echocardiograp and assessment of karyotype.

Key words: urinary tract; congenital abnormalities; ultrasonography; prenatal; infant; new-bo kidney / abnormalities

Introduction

Congenital malformations are the main cause of miscarriage, perinatal mortality and disability among children. According to Polish Registry of Congenital Malformations for 2005-2006 congenital disorders are diagnosed in 2.0-4.0% of newborns[1]. Urinary tract defects occur with a frequency of 20.7 per 10,000 births and are one of the three most frequent abnormalities after heart defects and musculoskeletal system disorders.

In 2018, in the Department of Obstetrics and Pathology of Pregnancy of First Clinical Hospital in Lublin, 1571 children were born. Hydronephrosis, defined as dilatation of renal pelvis above 10 mm, was diagnosed in 6 children, what accounts for 0,4 percent of total births. One case of renal agenesis and policystic kidney disease was diagnosed, what comprises 0,06 percent of total births.

Purpose

The article shortly describes the most common urinary tract defects and characteristic prenatal ultrasound findings concerning those urinary tract malformations.

Embryology

Kidney embryogenesis is complex. Urinary system is formed from third germ layer, precisely from intermediate mesoderm. All three stages of kidney development: pronephros, mesonephros and metanephros form from intermediate mesoderm.
During embryogenesis kidneys ascends and rotate around their long axis. Primarily they are situated in sacral region. Further they ascends to lumbar region and locate beneath adrenal glands. During upward movement kidneys receive temporal vasculates from abdominal aorta. Final vessels- renal arteries rise perpendicularly from aorta in lumbar region. Additional vasculation of kidneys results from preserved temporal vasculature.

Rearrangement in mentioned processes result in kidneys or urinary tract defects like renal agenesis, posterior urethral valvae or kidney ectopia. Defects of adrenal glands do not coincidence with renal abnormalities because of their different origins: adrenal cortex forms from mesoderm, adrenal medulla forms from ectoderm.

Prenatal diagnosis

According to Polish Society of Gynecologists and Obstetricians, a pregnant woman should have at least three ultrasound screenings:11-14 weeks gestations,18-22 weeks gestations, 30-34 weeks gestations.

The aim of ultrasonography screening in first trimester is to evaluate implantation of embryo. Assessment of urinary track is difficult, however bladder is visible almost in 100 percent in 13th week of gestation[3]. It is indirect information about presence of functioning kidneys.

Ultrasound evaluation of second and third trimester aims to assess development of fetus and its organs for example kidneys size, renal parenchyma, urethers, bladder and blood flow in renal arteries[4].

Table 1. Ultrasound parameters used in prenatal assessment of urogenital system in certain week of gestation.

Weeks of gestation	Prenatal Ultrasound evaluation of urinary track system	
	Anatomy of urinary track system	additional parameters
11-14	Visualization of bladder	
18-22	visualization of bladder blood flow in renal arteries	amniotic fluid volume cardiovascular score
30-34	kidneys size urethers visualization of bladder with ureterocele or posterior urethral valvae blood flow in renal arteries renal parenchyma	amniotic fluid volume cardiovascular score lung development skeletal abnormalities in case of oligohydramnion genital defects dysmorfic features

Visualization of urinary track defect obligate further screening for accompanying abnormalities. Sequence of defects or plural abnormalities suggest genetic background and are indications for cardiovascular and genetic evaluation in referral medical centers[4].

The prognosis of prenatally detected kidneys defects depends on:

type of defect: insolated or complex, degree of renal dysplasia, degree of obstructive uropathy,

amniotic fluid volume, fetal cardiovascular profile[4].

Progression of the majority of defects is slow therefore treatment is dominated by conservative approach.

Referral medical centers should prepare parents for postnatal therapeutic treatment or palliative care.

In accordance with applicable law in Poland in case of lethal defect, termination of pregnancy is possible until fetus is able to survive outside the uterus[5]. World Health Organisation determined the cutoff on 22 weeks gestation.

Table 2. Common genetic syndromes associated with urinary track malformation and other anomalies that might be diagnosed prenatally

Syndrome/Diagnosis	Type of urinary track malformation	Congenital heart defects diagnosed prenatally	Other anomalies diagnosed prenatally	Gene	Ref
Trisomy 21	Tubular dysgenesis	VSD, AVSD	Duodenal atresia, anal atresia	46XX+21 46XY+21	6
Trisomy 18	Horseshoe kidney, obstructive utopathy	VSD	Growth retardation, Skeletal anomalies	46XX+18 46Xy+18	7
Trisomy 13	Hypoplasia, cysts	VSD, dextrocardia	Brain anomalies, cleft palate	46XX+13 46XY+13	7
Turner syndrome	Horseshoe Sidney, hypoplasia, cyst obstructive uropathy	CoA, BAV	Thicker NT Growth retardation, webbing neck,	45X0	8
DiGeorge syndrome	dysgenesis, cysts	IAA, TA, TOF	Thymus aplasia, cleft palate	Del 22q11.2	9
Smith-Lemlie-Opitz syndrome	Unilateral or bilateral renal agenesis Cystic kidneys	AVSD	Growth retardation, cerebral malformation, genital anomalies	DHCR7	10
Mecel-Gruber syndrome	renal cystic dysplasia	ASD, VSD, dextrocardia	occipital encephalocele, facial abnormalities, polydactyly	MKS1	11

VSD- ventricular septal defect, AVSD- atrio-ventricular septal defect, ASD- atrial septal defect, PDA- persistent ductus arteriosus, CoA- coarctation of aorta, BAV- Bicuspid aortic valve, IAA- interrupted aortic arch, TA- truncus arteriosus, TOF- tetralogy of Fallot, NT- nuchal transluciency

Urinary tract anomalies in the prenatal period

Renal agenesis.

Renal agenesis is lack of renal parenchyma. One-sided renal agenesis is clinically silent in contrast to bilateral renal agenesis that is lethal. Characteristic in bilateral renal agenesis is oligohydramnion seen in third trimester as a consequence of fetal anuria. The amount of amniotic fluid may be correct up to

second trimester because amniotic fluid is produced by placenta up to 16 weeks of gestation. Worth noting is correct amount of amniotic fluid in case of accompanying esophageal or duodenal atresia[12]. Absence of fetal urine production is known for causing fetal malformations described in Potter sequence including limb malformations, flattened facies, low set abnormal ears, and pulmonary hypoplasia. As the presence of amniotic fluid is vital to lung development, bilateral renal agenesis is deemed to be fatal due to pulmonary hypoplasia. If children are born alive, they die shortly after birth due to respiratory failure and end-stage kidney failure. In this case parental counselling is directed towards palliative care after birth.

Characteristics of bilateral renal agenesis in prenatal ultrasound scans[2]

oligohydramnios.

lack of renal parenchyma

lack of bladder/ hypoplastic bladder

inability to visualize renal arteries

pulmonary hypoplasia

features of Potter sequence: improper positioning of limbs, bell-shaped chest

large adrenal glands

On the contrary unilateral agenesis has good prognosis. Second kidney overgrows compensatory. The amount of amniotic fluid is correct. Due to similar embryogenesis defects of reproductive track might coexist for example: testes agenesis, cryptorchidism, ovarian defects, oviduct and uterus defects. I addition other urinary tract might be present for example vesico-ureteral reflux[17], therefore postnatal diagnosis of potentially associated defects should be planned.

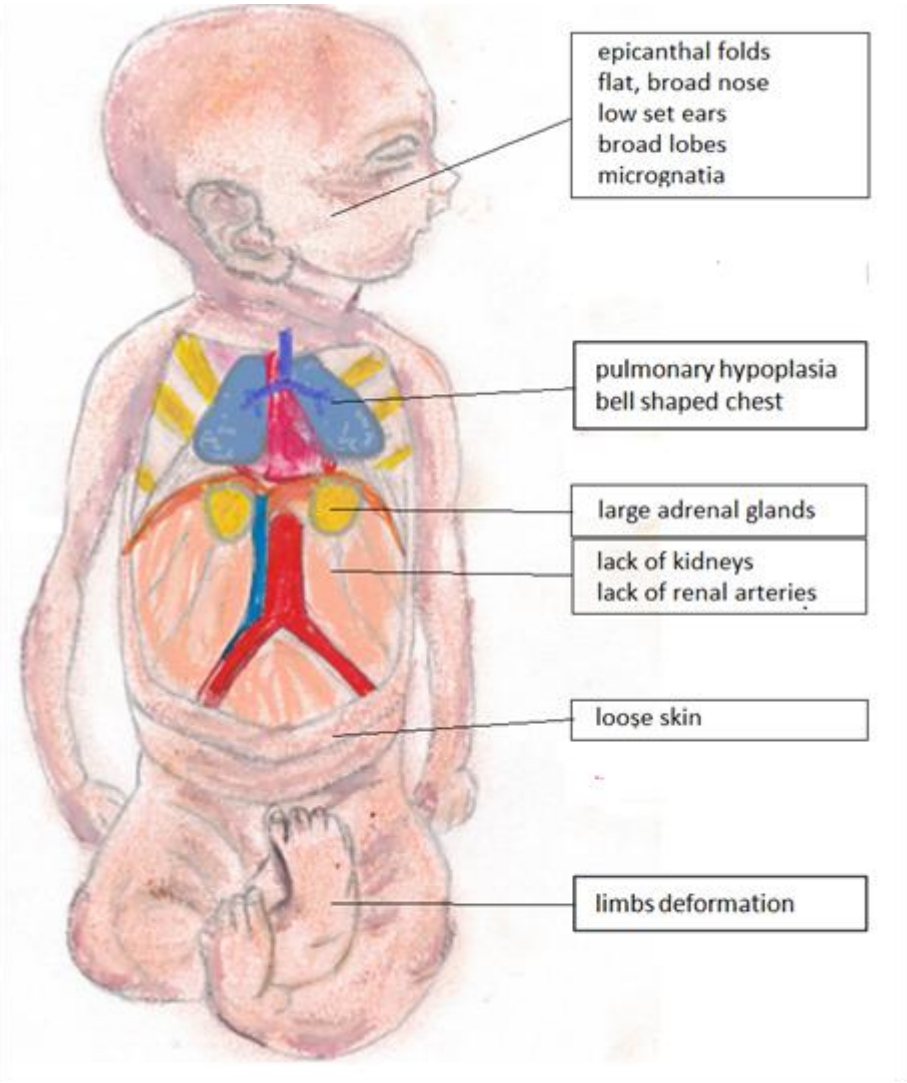

Figure 1. Potter sequence (author's drawing)

Polycystic kidney disease (PKD)

PKD is a general term for two types of the disease, autosomal dominant polycystic kidney disease (ADPKD) and autosomal recessive polycystic kidney disease (ARPKD). Both are of genetic origin. ARPKD may manifest in prenatal or neonatal period or during childhood (late form). Multiple cysts develop within kidneys and destroy renal structure. Destroyed kidneys do not produce urine, thus oligohydramos is present. Cysts may also occur in liver or pancreas. ARPKD is lethal. If children are born alive, they die shortly after birth because of lung hypoplasia[13]. Those who develop the disease later (to about 10-13 years old) are struggling with hypertension, hepatic and renal impairment[13]. Characteristic polycystic kidney disease (PKD) in prenatal ultrasound scans are

Enlarged hyperechoic kidneys with multiple cysts 1-2 mm in diameter
Difficulties in bladder visualisation because of lack of urine
Oligohydramnios, anhydramnios
Features of Potter sequence: bell-shaped chest, limbs deformities

Typically first abnormalities are visible in second trimester.

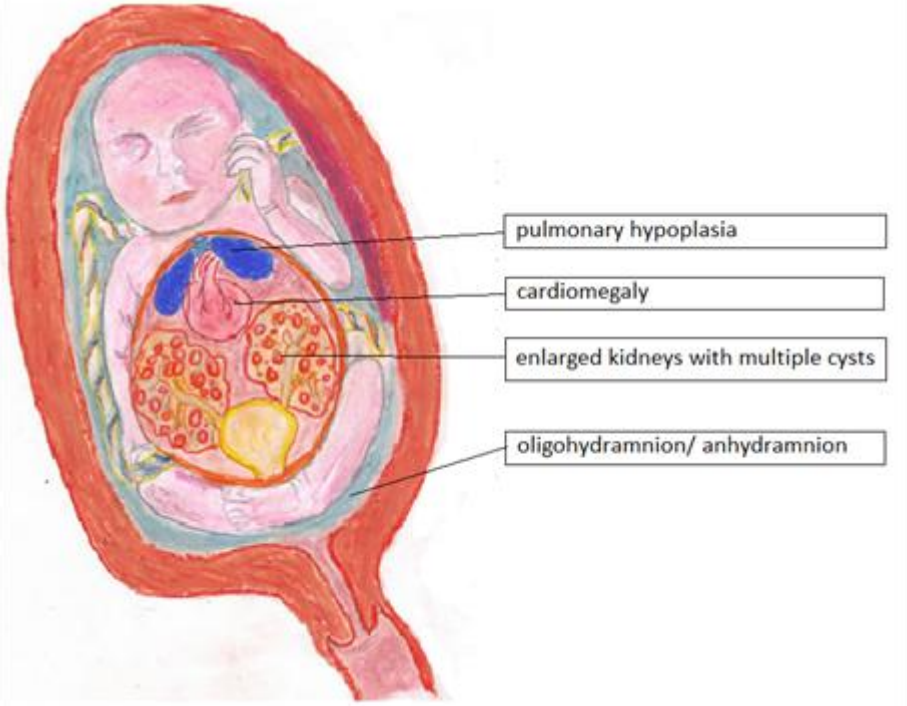

Figure 2. Characteristic features of polycystic kidney disease (PKD) in prenatal period (author's drawing)

ADPKD reveals usually after age of 30, however ultrasound scans performed prenatally or soon antenatally with positive family history may suggest presence of defect.
Ultrasound examination shows enlarged hyper-echoic kidneys, isolated different-sized cysts. Cysts may be present in other organs: liver, pancreas, spleen.

Bladder agenesis and bladder extrophy

Bladder is round, anechoic structure filled with urine, situated between two umbilical arteries. Size of fetal bladder is similar to fetus stomach[4]. In diagnosis of bladder defects, estimation of the amount of amniotic fluid is crucial. Umbilical cord should contain 2 arteries and a vein. Persisting urachus is a pathology.

Bladder agenesis should be suspected when there is no bladder between umbilical arteries and concomitant oligohydramnios. In difficulties of bladder visualization second ultrasound examination should be performed after 15-20 minutes and mother hydratation.
Usually the defect coexists with another urinary and reproductive track abnormalities for examples: agenesis of kidneys, prostate or vagina. Defect is lethal.

Characteristic of bladder agenesis in prenatal ultrasound scans

Difficulties in bladder visualisation
Oligohydramnios

Bladder extrophy can be distinguished from bladder agenesis based on amount of amniotic fluid and presence of bladder template on the lower abdominal wall.

Characteristic bladder extrophy in prenatal ultrasound scans are:

Difficulties in bladder visualisation
Mass of tissue in the lower abdomen
Genital defects: small penis, scrotum dysmorphy, doubts about sex of the child
Low set navel
Normal amount of amniotic fluid[2]

If defect is isolated, it may not affect pregnancy.

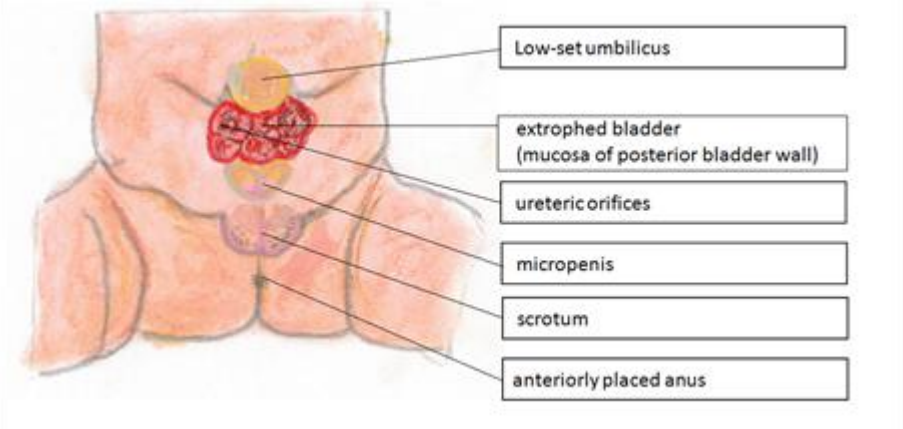

Figure 3. Male infant with bladder extrophy (author's drawing)

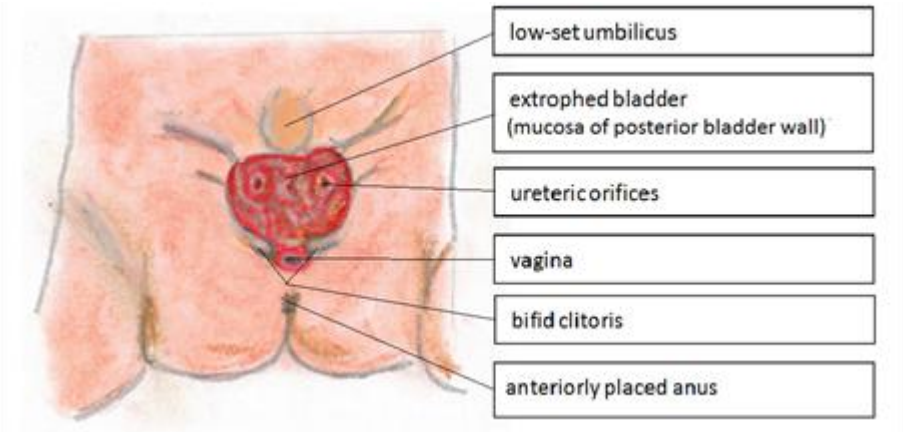

Figure 4. Female infant with bladder extrophy (author's drawing)

Obstructive uropathies

Obstructive uropathies are group of urinary track defects, where drainage of urine is blocked. Depending on localization and degree of obstruction we can distinguish: pyelectasis, hydronephrosis, megabladder, megaurether. Amount of amniotic fluid may be diminished or correct when pathology is unilateral. Usually pathology is first seen in second trimester.

Pyelectasis or functional dilatation of the renal pelvis to 4-5mm in AP projection. It requires regular monitoring throughout pregnancy. Pyelectasis involutes within first year of life.

Hydronephrosis describes dilatation of renal pelvis above 10 mm in AP projection regardless of gestational age[4].

The most common cause of hydronephrosis is uretero-pelvic junction obstruction. Others are: ureterocele , ureterovesical junction obstruction, posterior urethral valve, vesicoureteral reflux. Prognosis is variable and depends on the cause of hydronephrosis. Some of these abnormalities can be already identified prenatally for example posterior urethral valve whereas some after birth for example vesicoureteral reflux. Progresive hydronephrosis requires assessment of amniotic fluid, placenta and cardiac function. Assessment of kariotype should be considered[4]. Coincidence of defects worsen prognosis.
Regarding treatment, ultrasound monitoring is all that is necessary. Surgery is not advised because of high risk of preterm labor. Postnataly ultrasonography examinations are mandatory. Procedure should be conducted at least at 3rd day of life due to the physiological oliguria[13.]

Megaurether- dilatation of urether above7 mm[13].The most common causes are ureterovesical junction obstruction or ureterocele[4]. Often megaurether is associated with ectopic ureter and ureterocele. Megaurether implies an obstruction beneath[13]. After birth treatment is aimed at diagnosis of cause of dilatation(cystography, renoscintigraphy[13].
Characteristic in prenatal ultrasound scans are:

sausage-shaped loop in abdomen (megaurether easily can be confused with bowel loop[13]
The defect is typically detected in third trimester.

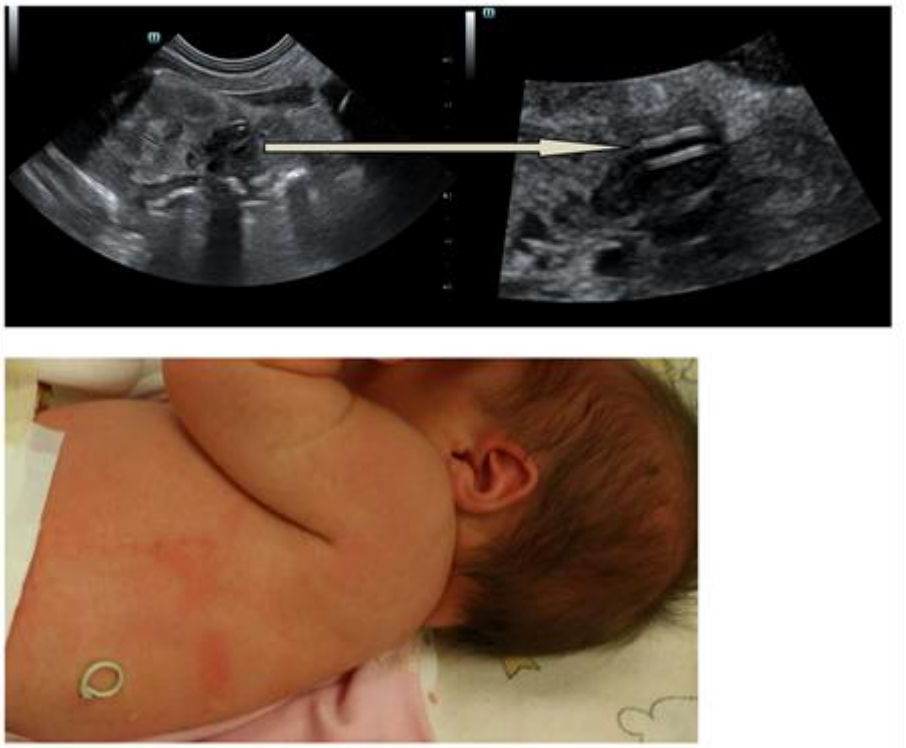

Figure 5. 3-day old girl born vaginally with pigtail shunt inserted at the 27th of gestational age because of unilateral hydronephrosis of left kidney with 23mm pelvic in diameter. Ultrasound scan shows left kidney with patent shunt inserted in kidney. No dilatation of renal pelvis was seen at the neonatal period. Shadow seen beneath the shunt.

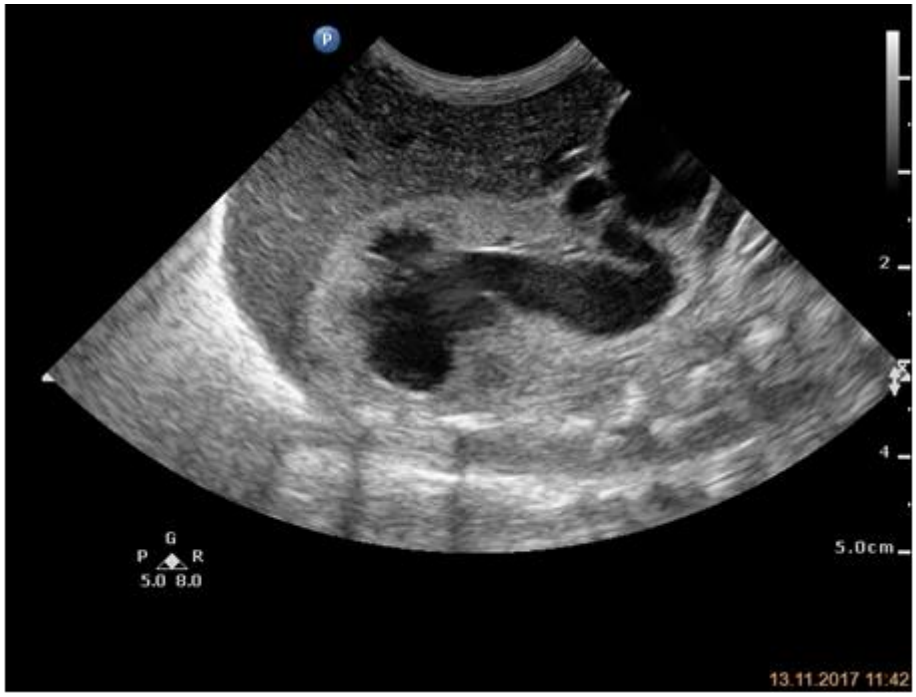

Figure 6. Hydronephrosis with megaureter in premature neonate born in 26 week of gestation (own clinic's material).

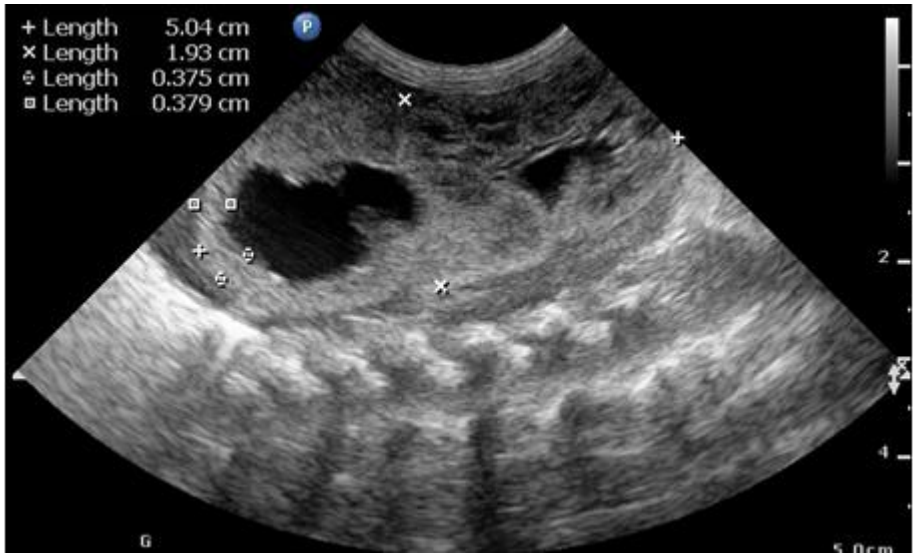

Figure 7. Duplication of urether with hydronephrosis of upper pelvis (own clinic's material).

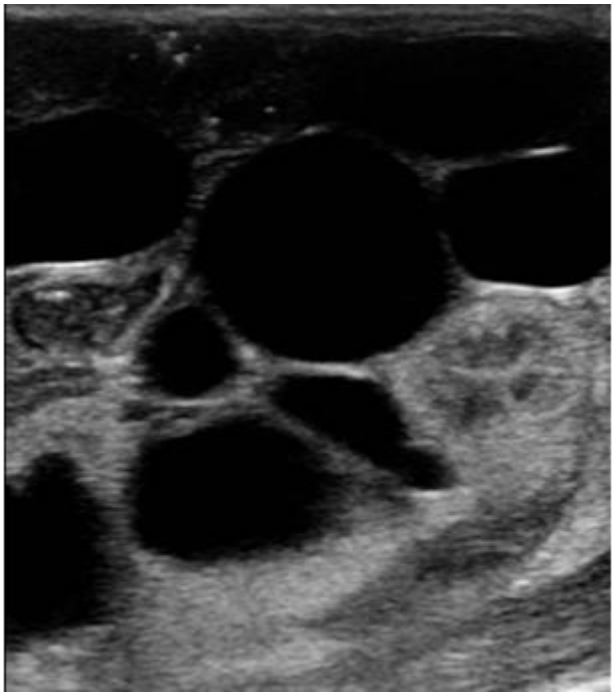

Figure 8. Megaureter seen as sausage-shaped loop in abdomen (own clinic's material).

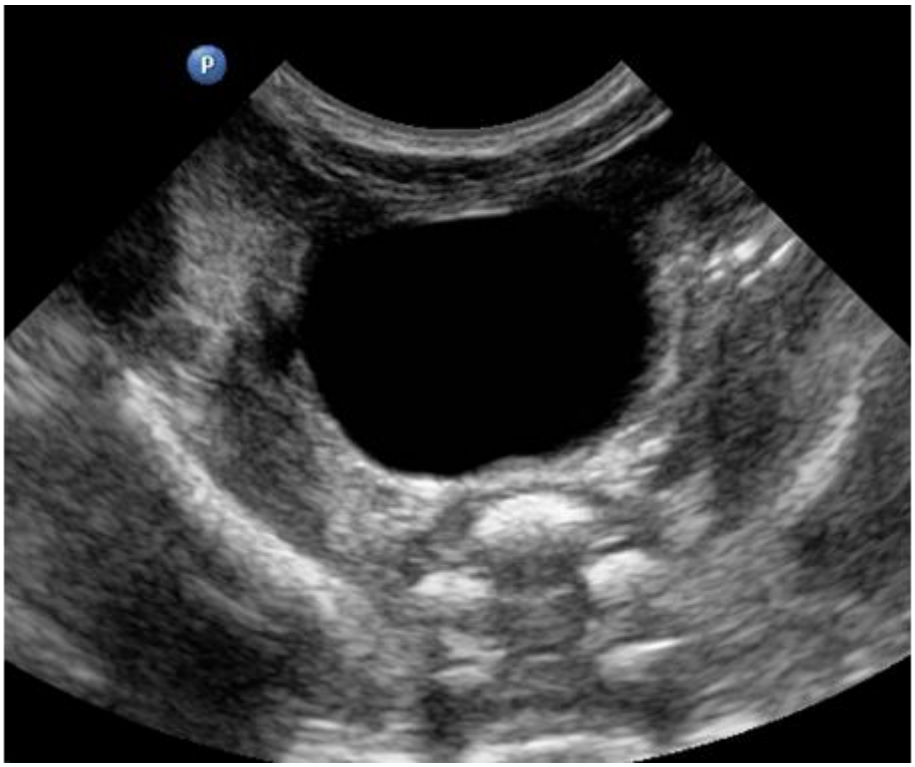

Figure 9. Ectopic insertion of megaureter in bladder (own clinic's material).

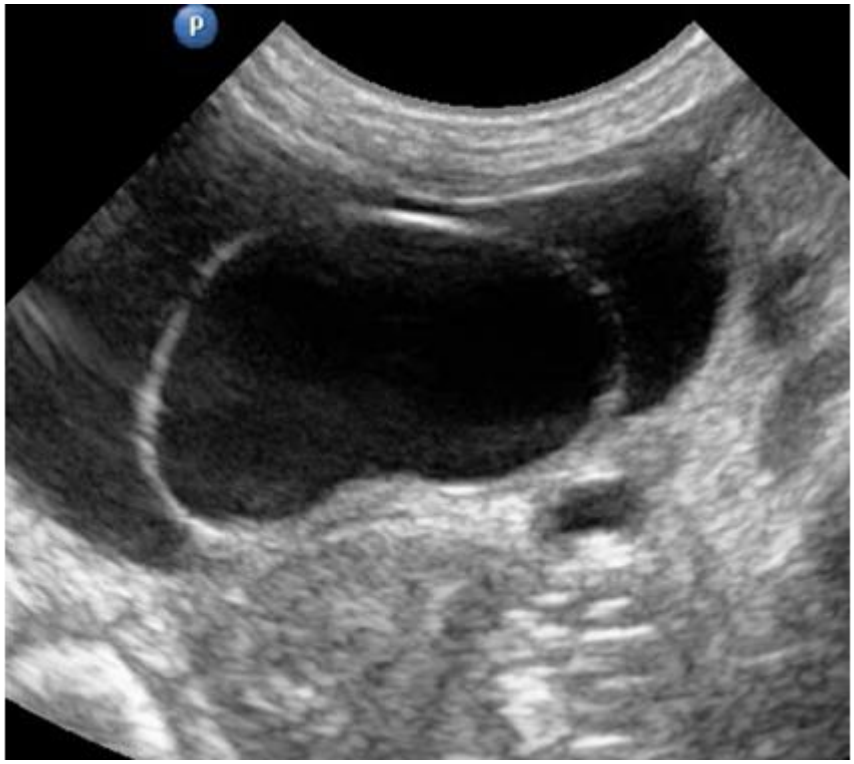
Figure 10. Ureterocele (own clinic's material).

Posterior urethral valve

Posterior urethral valves (PUV) is the most common and most severe congenital defect of the urinary tract in boys. The disorder varies in degree. Severe cases causes lung hypolasia and kidney failure resulting in severe respiratory failure. Quality of life of newborns with milder form of defect who survive is low. They are affected with end stage kidney disease (minimum 50 %), may struggle with infertility or bladder disfunction- incontinence or urine retention and urinary tract infection. Children with milder forms of defect usually struggle in later age with chronic kidney disease and are potential transplant recipient. In 2017 in Poland there were 99 patients under age of 18 on waiting list for kidney transplantation. Waiting for kidney transplantation under 18 from living donor last around 160 days, deceased donor 421 days[14.]

Characteristic in prenatal ultrasound scans are:

Bilateral hydronephrosis
Enlarged bladder
Oligohydramnios[13]
Dilated posterior urethra, key hole sign
Increased echogenicity of renal parenchyma
Diagnosis is made in first or second trimester. Mild form in third trimester[4.]

Treatment is rather conservative. There is a possibility of prenatal fetal surgery for example vesicoamniotic shunt implantation however, data concerning results of prenatal treatment are not optimistic. Defect impairs renal function on the very early stage, thus regardless whether surgery was applied or not renal function is deeply impaired Considering high risk of pregnancy loss, intrauterine infection, premature rupture of membranes, preterm delivery prenatal interventions are rather dissuaded[15,16]

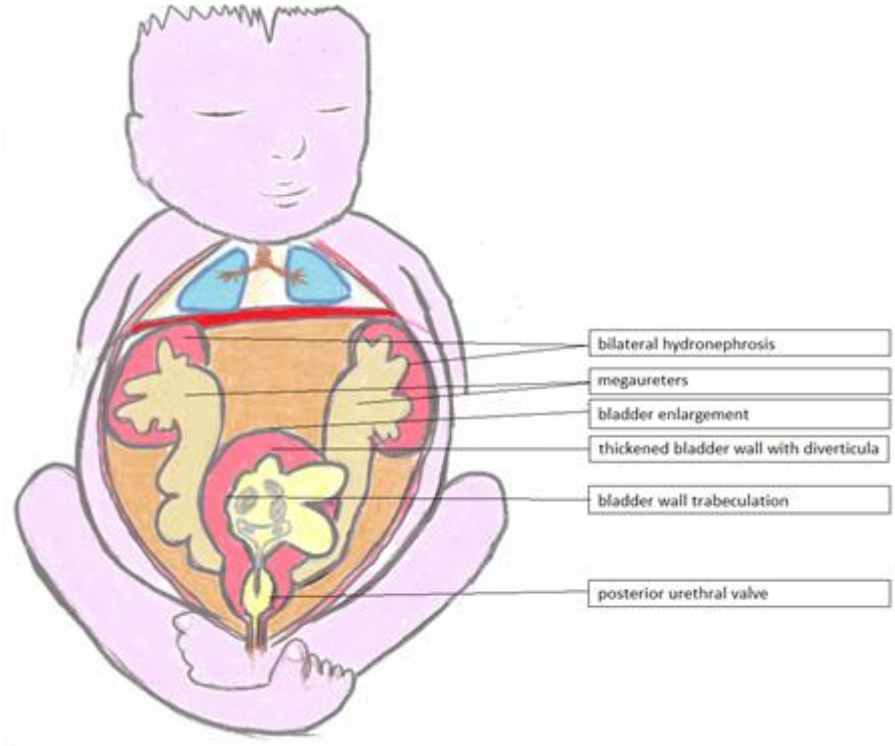

Figure 11. Male infant with posterior urethral valve (author's drawing)

Summary

1. Urinary track defects in prenatal period are very common
2. The prognosis depends on the type of defect and accompanied anomalies
3. In case of urinary track defect- further evaluation in referral center is necessarily (assessment of renal function, cardiovascular function and assessment of karyotype)
4. Assess karyotype especially when it is crucial for treatment
5. Childbirth should be performed in hospitals which cooperate with the urologist and pediatric surgeon

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