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Congenital Anomalies of Urinary Tract and Anomalies of Fetal Genitalia

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Abstract

Congenital anomalies of the kidney, urinary tract and genitalia anomalies are among the most frequent types of congenital malformations. Many can be diagnosed by means of ultrasound examination during pregnancy. Some will be discovered after birth. Kidney and urinary malformations represent 20% of all birth defects, appearing in 3–7 cases at 1000 live births. Environmental factors (maternal diabetes or intrauterine exposure to angiotensin-converting enzyme inhibitors) and genetic factors (inherited types of diseases) seem to be among causes that lead to the disturbance of normal nephrogenesis and generate anomalies of the reno-urinary tract. It is very important to diagnose and differentiate between the abnormalities incompatible with life and those that are asymptomatic in the newborn. The former requires interruption of pregnancy, whereas the latter could lead to saving the renal function if diagnosed antenatally. In many cases, the congenital anomalies of the urinary and genital tract may remain asymptomatic for a long time, even up until adulthood, and can be at times the only manifestation of a complex systemic disease. Some can manifest in more than one member in the family. This is the reason why the accurate genetic characterization is needed; it can help give not only the patient but also her family the appropriate genetic counseling, and also, in some cases, the management may prevent severe complications.

Keywords: genital, urinary, kidney, anomalies, fetal, malformation

1. Introduction

The congenital anomalies of the urinary tract include a large number of diseases caused by anomalies in the morphogenesis of the urinary system. These anomalies include obstructive

and nonobstructive dilatation of the urinary tract that can be associated with alterations in the number, size and/or position of the kidneys [1, 2].

We will discuss the congenital anomalies of the urinary and genital tract, with a short review of kidney abnormalities. These malformations may coexist within the same case, and this is due to their common embryonic origin [3].

The reno-urinary anomalies occur more frequently in males than in females, the ratio being 2.5:1 (M-F), and there are many cases with family aggregation. The incidence is 3–4 at 1000 lives or 1–5% of all pregnancies [4, 5]. If including all cases detected at post-mortem fetal autopsies, the prevalence of these malformations is much higher [2].

2. Congenital anomalies of urinary tract

2.1. Relevant embryology

The urogenital system is represented by two major components: the urinary system and the genital system. Embryological and fetal kidneys, genital tract and the urinary system develop from intermediate mesoderm. The rhythm of growth and development of the collector tubes differs greatly during pregnancy. Until 15 weeks of gestation, the rhythm of development is very rapid and later decreases [1, 2].

The urinary tract is almost entirely developed from the intermediate mesoderm. The evolution implies different stages in development: pronephros, mesonephros and metanephros. These stages will appear successively in the craniocaudal direction and coexist over time. Intermediate mesoderm is divided in the upper cervical and thoracic region, resulting in nephrotomes [2, 3].

The mesonephric duct will give rise to the ureteral bud after the formation of the pronephros. This, after 15 generations of divisions, will lead to the formation of the ureter, the calves, the collector tubes and the kidney pelvis. The lack of formation or agenesis of the pronephros or of the mesonephric duct can lead to the total or partial absence of the kidney or to other anomalies of the reno-urinary tract [4, 5].

CAKUT (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 [5, 6].

The genitalia differentiation, which leads to female or male gender, starts at 7–8 weeks of pregnancy and finalizes at 12–13 weeks. Every fetus will develop female characteristics, because at the beginning of gestational period, the Mullerian and Wolffian structures coexist. If the Mullerian structure will suffer atrophy, due to testosterone and anti-Mullerian hormone effects, male sex structures will develop from the Wolffian structure, and the fetus will

become a male. In the opposite circumstance, the fetus will become a female. In conclusion, from the embryological point of view, an individual could be a female if the masculine features will not develop [7, 8].

2.2. Classification

Congenital urinary abnormalities are often associated with the kidney anomalies, and there is a wide range of malformations resulting from disorders in the normal development process [9].

Malformations of renal parenchyma may occur due to the abnormal nephron development—in cases of renal dysplasia, renal agenesis and renal polycystic disease. The migration abnormalities of kidney embryo buds are found in renal ectopy and in the mismatched malformations [10].

Abnormalities in the development of the urinary tract system cause duplicated collective systems, posterior urethral valves and obstructions of the pyeloureteral junction. Defects may be unilateral or bilateral, and several types of defects may be associated [1–3, 6, 7].

The anomalies of the urinary system can be divided into nephropathies and uropathies:

- a. Nephropathy involves the renal parenchyma and refers to multicystic kidney disease (MCDK), renal dysplasia disease, renal agenesis defect, congenital polycystic kidney disease and other anomalies (nephromegaly, trisomy 13, Meckel syndrome, Beckwith-Wiedemann syndrome) [6–8].
- b. Uropathies represent the pathology of the urinary tract, and by the site of the defect, they can be pyeloureteral and ureterovesical, or they can refer to the vesicoureteral reflux and to the posterior urethral valve. Many authors also consider here the urachal fistula, the urachal cyst and the exstrophy of the urinary bladder [10–12].

For a better understanding of urinary tract abnormalities, we will summarize the classification of kidney malformations that may accompany these anomalies:

I. Anomalies in number

- a. *Complete bilateral renal agenesis* is a rare condition, not compatible with extrauterine life. The physical appearance of the babies is very characteristic (the so-called Potter syndrome). The absence of the kidneys may be suspected before conventional postmortem autopsy. Fetal ultrasound examination is very difficult, due to the absence of amniotic fluid.
- b. *Unilateral renal agenesis* or the ‘congenital solitary kidney’ is probably the most difficult diagnosis of renal malformations. There is a consensus in terms of a definitive diagnosis, autopsy being the only conclusive method.
- c. *Renal aplasia*—there is a fetal bud, but it does not develop into a normal functioning organ. The pelvis and ureter are usually absent or are rudimentary.

- d. *Supernumerary kidney* is the rarest anomaly and consists of a third kidney with excretory cavities and its own vascularity, completely separated from the other kidney [1–8].

II. Renal size abnormalities

- a. *Renal hypoplasia*. There are small congenital kidneys, located next to the median line and excretory cavities. The renal function is normal or low.
- b. *Kidney hyperplasia*. There is a large kidney, after ruling out any other cause [5–7].

III. Kidney's shape abnormalities

- a. *Lobular kidney*. There is a persistence of fetal lobulation.
- b. *Kidney fusion*. There are bilateral symmetrical or asymmetrical mergers or unilateral asymmetry.
- c. *The kidney in the horseshoe*. In 90% of the cases, kidneys join at the lower pole through a parenchymal or fibrous bridge.
- d. *Bilateral asymmetric mergers*. One of the kidneys is smaller than the other.
- e. *Unilateral asymmetric fusions*. There is a single kidney mass with a crossed ectopic kidney, known as 'the sigmoid kidney', usually located in the pelvis [7–10].

VI. Renal position abnormalities

- a. *Ectopic kidney*. A birth defect given by an abnormal (unilateral or bilateral) kidney position. The ectopic kidney is more often located on the left side. There are several ectopic kidney types: the caudal (lumbar, iliac, sacral) kidney, the cranial kidney and the crossed kidney.
- b. *Renal dystopia*. Initially, in embryonic life, the hill and pelvis are located in the anterior part, and then they undergo a rotation around the longitudinal axis, until they reach the median side [1–4, 7].

V. Multicystic renal dysplasia

These are large and hyperechogenic kidneys, including renal ciliopathies. These conditions may be divided into autosomal dominant diseases and autosomal recessive disorders, including the polycystic kidney disease, renal dysplasia, glomerulocystic kidney disease (trisomy 13 and trisomy 18, Beemer syndrome), multicystic dysplastic kidneys (MCDK), medullary cystic dysplasia (Meckel-Gruber and Beckwith-Wiedemann syndromes, as well as congenital infections) [10–13].

2.3. Diagnosis

Nowadays, the fetal bladder and kidneys may be visualized starting with 11 weeks (by transvaginal echography) or 12 weeks (by means of transabdominal echography). The fetal bladder and kidneys are located near the spine, having an elliptical shape [1, 2].

In the first trimester of pregnancy, the kidneys appear as an ovoid structure located on both sides of the spine (**Figure 1**) [1–4].

Renal urine production begins during week 9 of embryonic life, making it possible to visualize the bladder, as a fluid collection in the fetal pelvis. The bladder appears spherical and transonic and is located between the iliac bone centers of ossification, in the lower pelvis. It can be visualized starting with the ninth week of pregnancy, and the umbilical arteries can be visualized laterally next to the bladder. Beyond the first trimester, the bladder will fill and empty in an intermittent manner, every 25–30 minutes, due to the influence of hormonal factors on the bladder (**Figure 2**) [2, 3].

Normal ureters cannot be visualized by means of echography. The ratio of renal circumference to abdominal circumference is 0.27–0.30 and remains constant during pregnancy [8].

Evaluation of the urinary tract also requires the evaluation of the amniotic fluid volume. After the 14th week of intrauterine life, the amniotic fluid comes mainly from the production of fetal urine and only one-third of its quantity comes from the pulmonary fluid [2, 5].

For a correct diagnosis, the clinician has to perform a complete examination in all of the three planes, the coronal, the sagittal and the longitudinal plane, using 2D grayscale and color Doppler. The examination should be complete. Sometimes, the ultrasound examination has to be completed by another screening method. Due to the association between renal malformations and other congenital defects or chromosomal anomalies, performing an invasive diagnosis method may be required [14].

The renal pelvic dilatation (RPD) is the most common of the abnormalities that can be detected during antenatal ultrasonography and is probably the most frequent sign of a reno-urinary

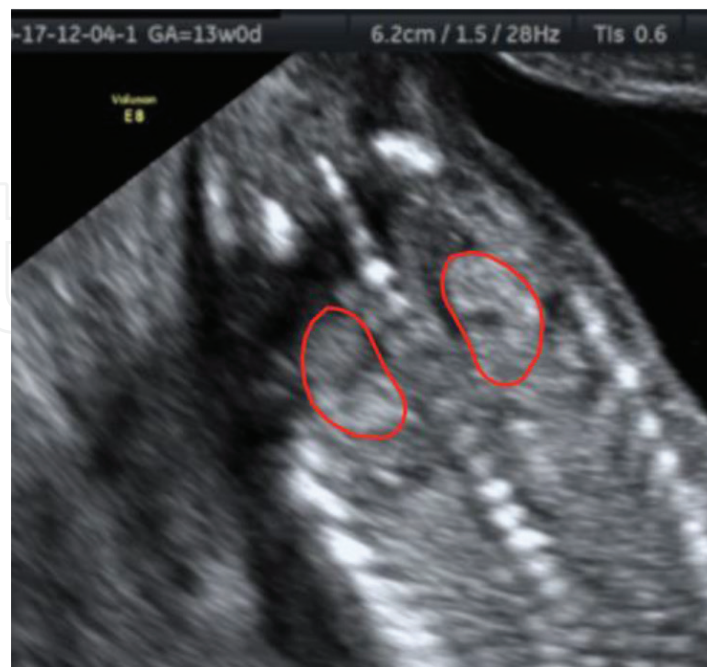


Figure 1. Fetal kidneys in the first trimester of pregnancy.

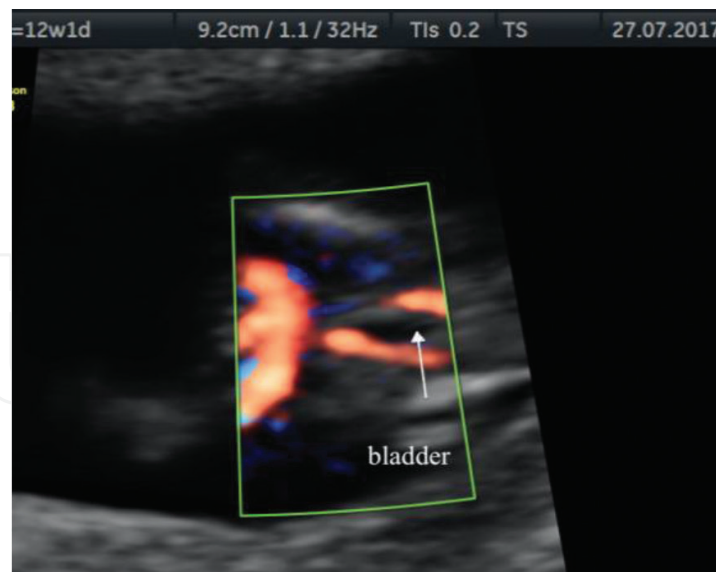


Figure 2. The bladder in the first trimester of pregnancy.

anomaly. The diagnosis of the RPD is based on an increased anteroposterior diameter of the renal pelvis in the transverse plane, and the value allows classification: severe, moderate or mild RPD. Normal values of anteroposterior diameter of the kidney pelvis are up to 4 mm at week 16 of pregnancy, less than 7 mm at 28 weeks of pregnancy and less than 10 mm postnatal [15].

In many cases a transient dilatation occurs. This situation is caused by narrowing or natural folds of the urinary tract that may occur during the early stages of development. Transient dilatation is usually less than 6 mm in the second trimester and less than 8 mm in the third trimester. It usually resolves spontaneously or disappear postnatally [16].

- Many authors concluded that for an accurate diagnosis, the assessment of the renal and urinary function may be performed by ultrasound [5, 6]. The following statements gained acceptance:
- The amount of amniotic fluid is an indirect indicator of the kidney function.
- The bladder filling indicates the normal functioning of at least one kidney.
- The absence of the bladder filling may indicate renal agenesis and bilateral ureteral obstruction.
- The bladder distension may hide a urethral reflux problem.
- The ureteral dilatation may be caused by a lower obstruction of the inferior urinary tract.
- The echogenicity of renal parenchyma cannot be considered for the renal function assessment.
- The increasing rebound pressure in the renal artery may cause renal function [5–12].

2.4. Uropathies

Uropathies are the most common reno-urinary abnormalities diagnosed during the prenatal period and may be caused by obstructive or nonobstructive factors [10–13]. The most common

urinary malformations encountered in children are vesicoureteral reflux (VUR), obstructive megaureter, posterior urethral valve and megacystis [11].

Fetal hydronephrosis may result in a number of conditions, like pelvic or vesicoureteral junction obstruction, posterior urethral valves, vesicoureteral reflux, pelvic-ureteric junction obstruction and other rare congenital anomalies [12, 13]. A clear definition of hydronephrosis would be that it is a dilatation of the calyces and the renal pelvis of over 10 mm or more than 50% of the anteroposterior diameter of the kidney. This dilatation of the urinary system may occur at the upper urethral segment, ureters, bladder or kidney pelvis. Pyelectasis is the dilatation of the renal pelvis only (**Figure 3**) [6, 15].

By some authors, the clinician can diagnose hydronephrosis if the renal pelvic AP diameter is increased over 4 mm in the second trimester and 7 mm in the third trimester [3, 11, 15] (**Figure 4**). A renal pelvic AP diameter > 15 mm is strongly associated with a pathology of the urinary tract that requires treatment after birth. However, in the majority of cases, the renal pelvis is mildly or moderately dilated, and no cause is identified [16].

There is no clear consensus on the follow-up and management of mild or moderate hydronephrosis observed on antenatal ultrasound, although it is generally accepted that a postnatal assessment should be performed if the AP diameter of the renal pelvis exceeds 10 mm at any point in gestation [17].

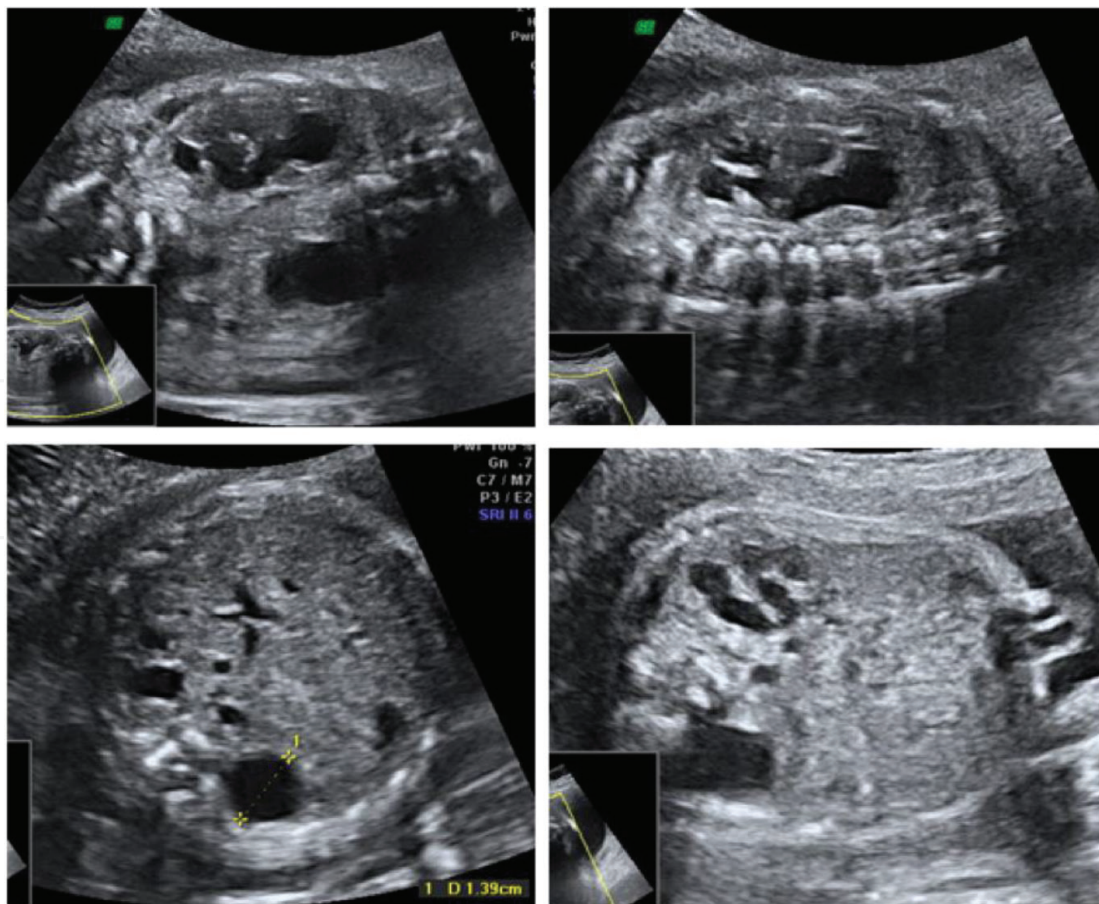


Figure 3. Second trimester: different aspects in hydronephrosis cases.

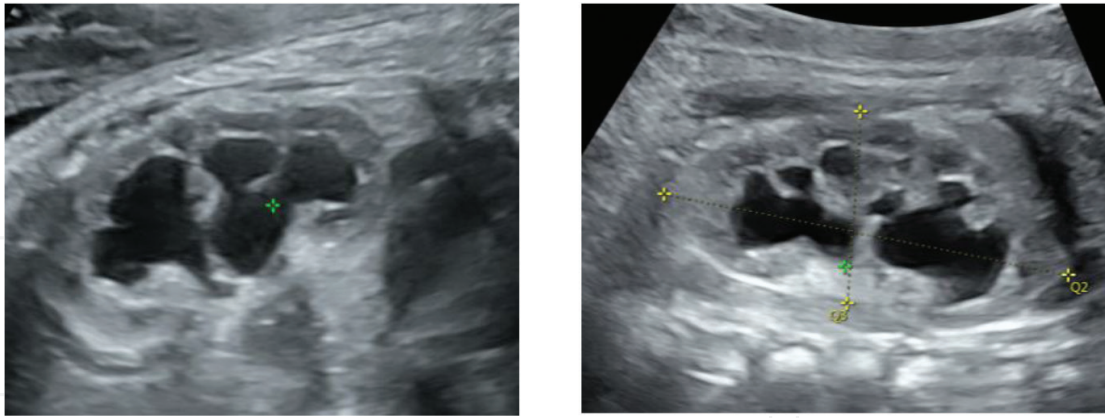


Figure 4. Hydronephrosis in the third trimester of pregnancy.

Many studies show that a right or left prenatal AP renal pelvic diameter > 4 mm is associated with a higher risk of postnatal hydronephrosis compared with a right and left prenatal AP renal pelvic diameter ≤ 4 mm [15, 16]. Male neonates have a higher risk of postnatal hydronephrosis than females. These results can assist in establishing the appropriate follow-up method and evaluation of fetuses with renal pelvic dilatation [18, 19].

2.5. Ureteral malformations

In medical practice many abnormalities can occur, like:

- Number abnormalities
- Structure abnormalities
- Calibration abnormalities
- Vesicoureteral reflux (VUR)
- Abnormalities of the pyeloureteral junction
- Position and opening abnormalities [1, 2, 14]

a. Ureteral abnormalities

- Ureteral agenesis is accompanied by the absence of the ipsilateral kidney. It may be uni- or bilateral, the latter being incompatible with life.
- Incomplete ureteral duplication (ureter fissus or ureter bifid)—a double-sided pyeloureteral system: the ureters bend before opening into the bladder through a single orifice.
- Complete ureteral duplication (duplex ureter)—a double-sided pyeloureteral collection system: the ureters open through separate boreholes in the bladder, one in orthoposition and the other in the ectopic position (**Figure 5**) [15–17].



Figure 5. Coronal section and high-definition power color Doppler, in a supernumerary kidney cases. Visualization of the double renal artery.

b. The ureteropelvic junction obstruction

Obstruction of the ureterobladder junction (obstructive megaureter) or obstruction on pyeloureteral junction is one of the most common causes of hydronephrosis found in children, its incidence being 1 case in 1000–2000 newborns. In this situation, hydronephrosis may be caused by an abnormal shape of the ureteral junction—the existence of the ureteral valves and the mucosal fold at this level. Like many other abnormalities, this pathology occurs much more frequently in males, usually unilaterally and especially on the left side [10, 14].

The ultrasound diagnosis is based on the observation of the increase of the anteroposterior diameter of the kidney, a degree of kidney pyelectasis. If the obstruction is unilateral and the filling of the bladder is normal, the normal amount of amniotic fluid will be preserved. When the damage is bilateral, the oligoamnios/oligohydramnios may appear.

In most cases this condition does not produce symptoms after birth; in 10–15% of the cases, it regresses spontaneously, but sometimes repeated evaluation is needed [1–8].

c. Obstruction of the ureterobladder junction

This condition is the cause of approximately 5–10% of all cases of dilatation of the urinary tract. Ultrasound diagnosis is based on dilated urethra, with renal pelvis dilatation and normal bladder image. The cause of the disease is the dysfunction of the lower ureter, the outcome is favorable and the pathologic aspects disappear postnatally in many cases [16] (**Figure 6**).

The normal ureter size in children is up to 5 mm. Normal ureter cannot be detected during prenatal ultrasound examination. It can only be detected if it is dilated to a size greater than 7 mm and is observed like a translucent structure. The dilated ureter should be differentiated from full intestinal loops. Pathological dilatation of the ureter is more common in male patients, usually appearing to be unilateral (**Figure 7**) [15–17].



Figure 6. Contralateral pyelectasis in a unilateral MCDK case.

The dilatation of the ureter may be partial or total, and this pathology can be classified into the following conditions:

- Obstructive-type megaureter
- Reflux-type megaureter
- Megaureter without reflux and without obstruction
- Obstructive- and reflux-type megaureter

d. Vesicoureteral reflux

Vesicoureteral reflux (VUR) is characterized by the return of urine from the bladder to the kidney which often causes hydronephrosis and sometimes even abnormalities in kidney development (renal dysplasia). Causes include transient obstruction of bladder discharge, delayed maturation of the vesicoureteral junction and high bladder evacuation pressure. Patients with vesicoureteral reflux have a high risk of pyelonephritis, hypertension and progressive renal failure. Vesicoureteral reflux is represented by the retrograde passage of urine from the bladder to the ureter and subsequently to the kidneys, due to the incompetence of antireflux mechanisms [17, 19, 20].

This condition is usually easy to diagnose prenatally, when visualizing renal pelvis and calyces dilatations that vary in size, associated with unilateral or bilateral ureteral dilatations. RVU reveals a nonprogressive hydronephrosis with normal amniotic fluid index [10–13, 18].

VUR classification:

- Grade I reflux: reflux is only present in the ureter that has various degrees of dilatation.

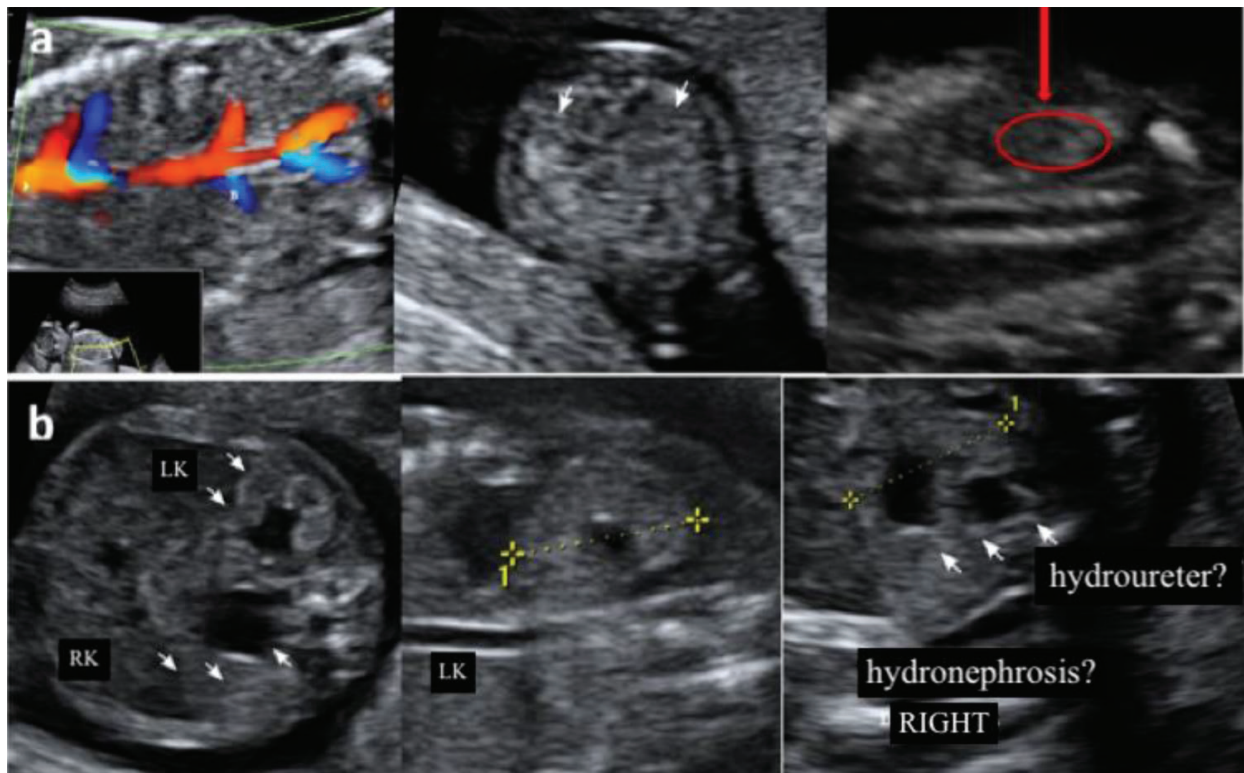


Figure 7. (a) First trimester: both renal arteries visualized in color Doppler, normal kidney images on both sides of the cross section and a kidney highlighted in the coronal section. (b) Second trimester (same case): asymmetrical bilateral pyelectasis, left kidney longitudinal diameter and hydroneurter on the right side.

- Grade II reflux: reflux reaches the renal pelvis, without dilatation of the collector system, the papillae being normal.
- Grade III reflux: moderate dilatation of the ureter, with or without sinusitis; moderate dilatation of the collector system.
- Grade IV reflux: moderate dilatation of the ureter with or without sinusity; moderate dilatation of the collector system; calyces are flattened but with the impression of the papilla still visible.
- Grade V reflux: severe dilatation with ureter sinusity and marked dilatation of the collector system; reflux in the parenchyma, with parenchymal thinning [10–15, 18].
- The prognosis is good. The prenatal diagnosis is important. Forty percent of the newborns develop severe kidney damage [2].

e. Ureterocele

As many other kidney abnormalities, ureterocele is more common in male patients. This pathology appears as a thin wall cyst or sept on the bladder wall [1, 16]. In many cases the diagnosis may be omitted when the bladder is full, the filling masking the cyst. There have been described cases of ureterocele shed in the urethra. This phenomenon may lead to acute obstruction of the lower urinary tract [1, 2]. The prognosis is favorable, but its evolution must be assessed postnatally, by residual renal function testing and reflux tests (Figure 8) [20].



Figure 8. Ureterocele and hydronephrosis.

3. Bladder anomalies

The bladder is a very important element in antenatal fetal examination. Clinicians may suspect anomalies of the bladder when not visualized on prenatal examination or when enlarged. From the pathological point of view, we may encounter various anomalies as well:

- a. Megacystis is a neurodysplastic disease, due to the anomaly of innervation. This leads to bladder dilatation (over 7 cm in diameter). It may be associated with small, short and dilated bowels, as well as a low-caliber colon. At ultrasound, the striking signs are polyhydramnios, enlarged ureters and kidneys [17].
- b. Congenital bladder diverticulitis. A septum can be observed.
- c. Bladder exstrophy. It is commonly associated with other urogenital malformations and is constantly accompanied by epispadias. This pathology is characterized by many anomalies and lesions, such as:
 - Abdominal anterior wall injuries: inguinal hernia is common.
 - Lesion of pelvic bones: shortening and defects of rotation of pubic bones.
 - Lesions of the genitalia: micropenis and cryptorchidism appear in boys; in girls, the vagina is short and has various degrees of stenosis, the clitoris is bifid and the small labia are divergent.
 - The anterior bladder wall is absent, and the posterior wall is the submuscular segment of the anterior abdominal wall.

- The bladder mucosa is exposed to the external environment, with the ureteric holes and the inner urethra opening visible.
- Vesicoureteral reflux is present in all cases.
- Some patients have rectal prolapse and anal incontinence; this phenomenon is due to the malposition of sphincter structures [1–4, 15–17, 19, 20].

4. Urethral malformations

The most common obstructive cause of the lower urinary tract is the posterior urethral valve (PUV). PUV is due to the existence of membranes in the posterior urethra. Echography shows dilated bladder and urethral dilatation, giving a classic echographic sign—‘the keyhole sign’. Dystriation of ureters and troughs also occurs [14]. In advanced forms, oligohydramnios-associated pulmonary hypoplasia and urinary ascites may also appear. These fetuses require prenatal intervention to avoid kidney damage. Some authors tried to demonstrate that creating a vesicoamniotic passage may lead to a better outcome, but this has not yet been fully demonstrated [16].

In the prenatal life, other malformations may be seen:

1. Urethral agenesis (absence of urethra) is a rare malformation, incompatible with life unless an alternative communication between the bladder and amniotic sac exists. The urine is discharged through abnormal communication with other organs (between bladder and bowels or by umbilical fistula).
2. Congenital structures.
3. Rear urethral valves: mucosal lining located in the posterior urethra originating from the seminal colonic and insertion on the anterior wall of the urethra [14–17].
4. Epispadias: abnormality characterized by opening the urethra on the dorsal face of the penis at variable distance of the gland, dorsal curvature of the penis and foreskin abnormalities. There are many forms of epispadias:
 - Balanic: distal incomplete defect.
 - Penian: incomplete proximal defect.
 - Penopubian: a complete defect, often associated with bladder exstrophy. In this situation, a distinct pathological condition appears: the exstrophy-epispadias complex [19].
 - Hypospadias: the anomaly is characterized by opening the urethra on the ventral side of the penis, at any level between the gland and the perineum, causing a ventral penis (chord). Patients with hypospadias have often abnormalities of the foreskin and cryptorchidic scrotum, inguinal hernia and hydrocele. Studies have shown a higher presence of hypospadias in patients with fetal growth restriction (**Figures 9 and 10**) [19]

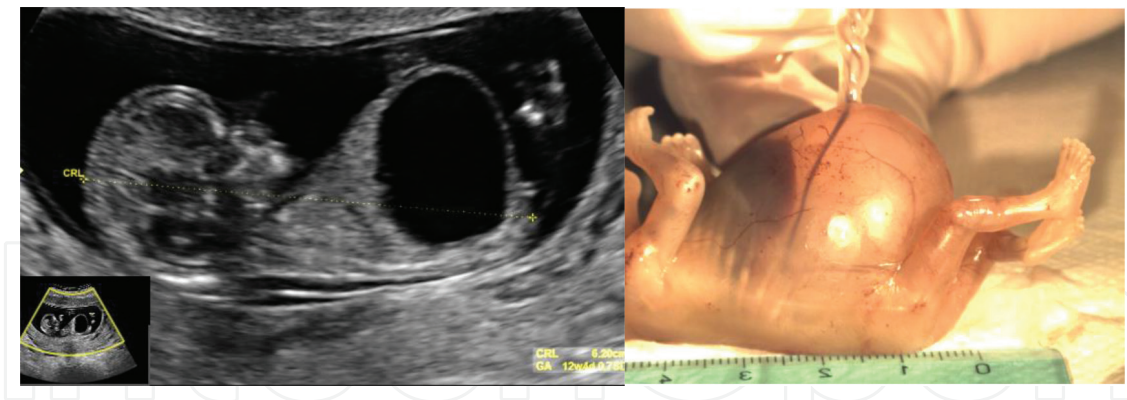


Figure 9. Isolated megacystis: the right-side image is the sagittal plane of the fetus and the CRL (crown-rump length) measurement; the left-side image (same case) is the pathological specimen after medical termination of pregnancy.

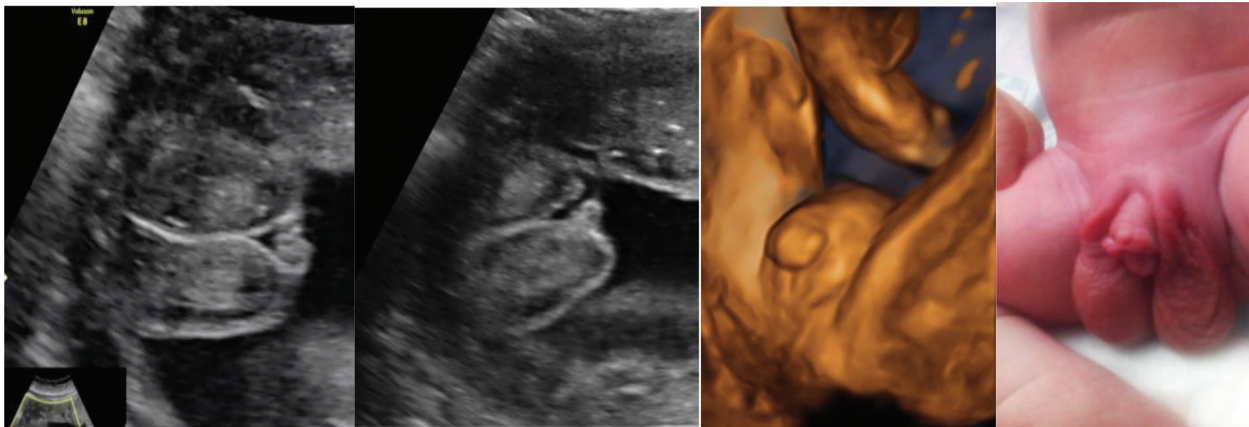


Figure 10. Grade III hypospadias (perineal) is the most severe form, with the opening of the urethra at the penoscrotal junction.

The diagnosis of hypospadias with prenatal ultrasound is based on several important criteria:

- A blunter bulbous tip to the penile shaft rather than the normal.
- A 'tulip' sign formed by the ventral-bent penis located between the two scrotal folds.
- Abnormal curvature of the penis.
- A short penile shaft.
- Ventral deflection of the urinary stream which can be studied by color Doppler [20].

5. Evolution and treatment

Renal pelvic dilatation is commonly seen during antenatal ultrasound examinations, and its management remains a clinical dilemma. Although it is proven that severe antenatal hydronephrosis requires postnatal clinical and ultrasound evaluation, there is no consensus on the

follow-up and management of mild or moderate hydronephrosis observed during antenatal ultrasound examinations [15]. The prenatal diagnosis may improve the prognosis and the outcome of the fetus. An early diagnosis and treatment of urinary obstruction may prevent the renal damage or loss of renal function [5, 21].

Most of the cases will spontaneously resolve after delivery. Thus, some assurance should be given. Many studies suggest follow-up when the AP diameter is 4–7 mm and antibiotic therapy when the AP diameter is greater than 7 mm [14, 18].

The postnatal assessment of fetal hydronephrosis may be invasive and lengthy [6–8]. Thus, the risks and the inconvenience of a protracted evaluation need to be weighed against the probability that milder degrees of renal pelvic dilatation will decrease without resulting in any renal damage [17].

6. Congenital genital anomalies

The spectrum of congenital genital malformation is very broad, and the diagnosis is usually difficult. A reliable classification reveals four main types of genital anomalies:

1. True hermaphroditism: an individual has both ovarian and testicular tissues.
2. Male pseudohermaphroditism.
3. Female pseudohermaphroditism: adrenogenital syndrome or congenital adrenal hyperplasia. The main feature is clitoris hypertrophy.
4. Gonadal dysgenesis [5, 19, 20].

Fetal gender abnormality is diagnosed by ultrasound or by finding a discrepancy between fetal phenotype and sex chromosomes. The ultrasound diagnosis is late, usually accessible in the second or third trimesters [5, 20–24].

I. Male genital malformations

Due to improved ultrasound technology and increased experience in this area, the fetal sex can be established with high accuracy beyond 13 weeks of pregnancy [5]. Determination of fetal sex is based on the 'sagittal sign'. During the first trimester, in the sagittal plane, the penis is oriented upwards and the clitoris downwards. In the third trimester, the genitalia can be described with high accuracy [21].

Penian malformations (abnormal phallic structure)

1. Micropenis or penian hypoplasia
2. Penian agenesis
3. Megalopenia or hyperplasia of the penis [1–3, 22]

Testicular malformations.

1. Number abnormalities

- Anorchia (or anorchism): the absence of both testicles in the presence of a normal male phenotype (46XY)
- Monorchism (or monorchidism): the absence of a testicle
- Polyorchism: the presence of more than two testicles
- Testicular fusion: the fusion of the two testicles in the same scrotum [2, 5, 20–24]

2. Development abnormalities

- Microorchidism: small, hypoplastic testicles.
- Macroorchidism may be secondary to contralateral testicular damage and may be bilateral in congenital syndromes (fragile X syndrome) or other disorders (hypophysis adenoma, aromatase deficiency).
- Cystic dysplasia of the testicle: benign, congenital tumor, frequently associated with other testicular malformations [5, 20–24].

3. Migration abnormalities

- Cryptorchidism: the process of descensus testis is affected by anatomical and mechanical factors, such as a poor connection between the gubernaculum and testis and endocrine factors (**Figure 11**) [5, 22].

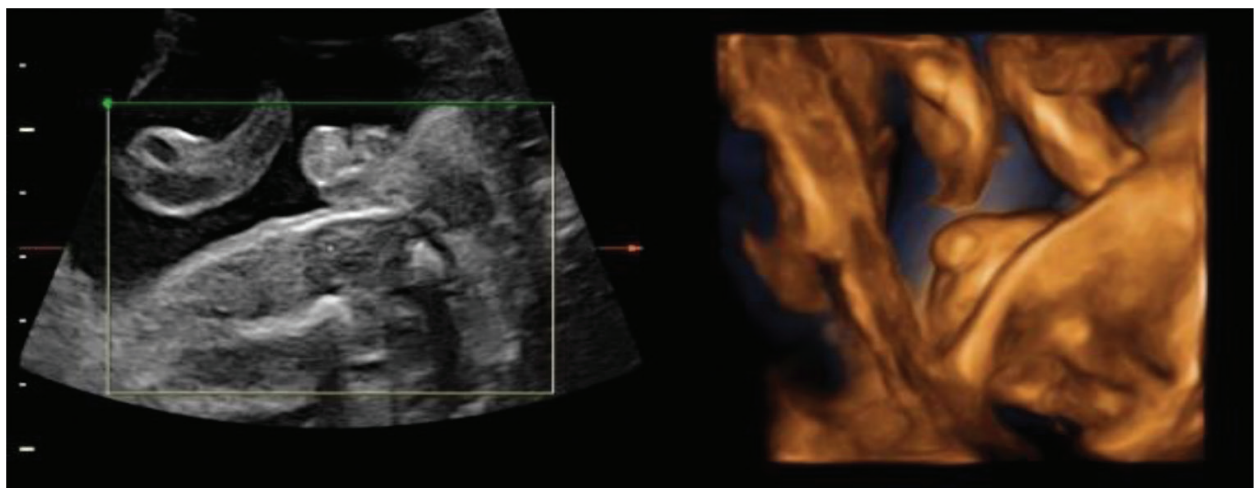


Figure 11. Hypospadias: the left-side image is the 'tulip sign' in 2D conventional ultrasound; the right-side image is a 3D reconstruction, surface rendering technique.

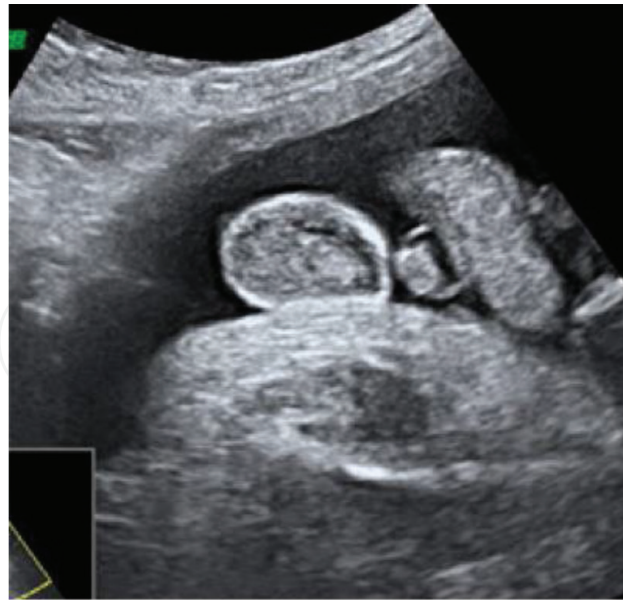


Figure 12. Unilateral cryptorchidism (in association with penian anomaly).

Classification:

- Intraabdominal: above or at the internal inguinal opening
- Intracanalicular: in the inguinal canal, between the inguinal inlet and the external groove
- Extracanalicular:
- Suprapubic: just above the external inguinal opening above the pubic symphysis level
- Infrapubic: in retroscrotal space, inferior to pubic symphysis [1, 5] (**Figure 12**)
- Ectopic testis: migration is normal to the level of the inguinal external hole, but the testicle follows an abnormal tract. Possible localizations are between aponeurosis of external oblique muscle and subcutaneous or femoral, etc. [24].

It is recommended that information about the fetal abnormalities, postnatal and prenatal options of treatment and prognosis should be presented to the parents by a multidisciplinary team that includes neonatologists, urologists, perinatologists and medical geneticists with expertise in this field [5].

7. Female genital malformations

7.1. Ovarian cysts

Ovarian cysts are the most frequently abdominal tumors that may be seen in female fetuses and newborns. The incidence of fetal ovarian cysts has increased lately, due to the improvement

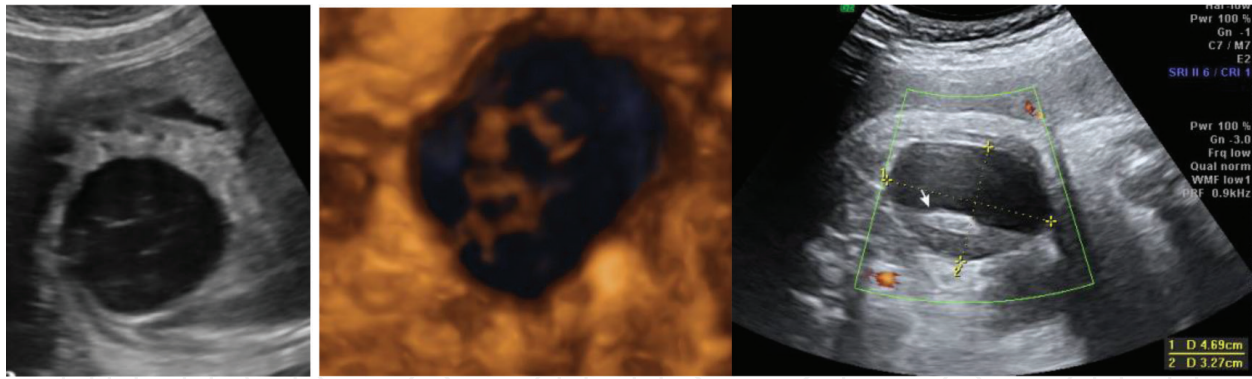


Figure 13. Fetal ovarian cyst: 2D and 3D surface rendering images. The evolution of the case led to the typical ‘fluid–fluid’ level aspect, due to the intracystic hemorrhage.

of ultrasound technology and due to the increase in the incidence of pregnancies that require hormonal treatment during the gestation period [2].

These ovarian tumors may be uni- or bilateral, sometimes multiple, and their appearance is as anechoic structures, thin-walled and having various sizes [5, 21–24]. Some authors described cases with solid tumors, teratomas or hemorrhagic cysts. To increase the accuracy of the diagnosis, MRI examination may be required (magnetic resonance imaging (MRI)) [22].

Fetal ovarian cysts have usually a good outcome; most of them progress to spontaneous resolution in the postnatal period. The most frequent complication of cysts is ovarian torsion. Other complications may be intracystic hemorrhage, rupture, dystocia during birth, etc. There are no guidelines for monitoring and treatment of this condition. The non-invasive monitoring by ultrasound seems to be the best approach in prenatal life [1, 5, 19–25] (**Figure 13**).

7.2. Hydrocolpos

Hydrocolpos is the accumulation of fluid in the vagina. When the fluid is observed on the vagina and the uterine body, the condition is called hydrometrocolpos. These diseases are caused by the persistence of the urogenital sinus or cloaca malformations [23]. The presence of a cystic mass in the presacral area, containing an anechoic fluid (suggesting urine) or sediment content, should guide the clinician to a diagnosis of urogenital sinus persistence [1, 2, 25].

8. Conclusions

The ultrasound diagnosis of renal and urinary tract abnormalities is generally based on the exclusion criteria, by comparison with the normal imaging one. In most cases, kidney or urinary tract abnormalities are diagnosed considering the appearance of amniotic fluid abnormalities, visualization of kidney size abnormalities or dilated appearance of the urinary tract. Congenital kidney abnormalities are often associated with the urinary tract malformation, and there is a wide range of anomalies resulting from disorders in the development process. It is important to differentiate abnormalities incompatible with life (as they require interruption of pregnancy) and asymptomatic/paucisymptomatic diseases in the postnatal life.

The genitalia anomalies must be treated by a multidisciplinary team that includes neonatologists, endocrinologists, urologists, perinatologists and medical geneticists with expertise in this field.

Conflict of interest

We have no conflict of interest.

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