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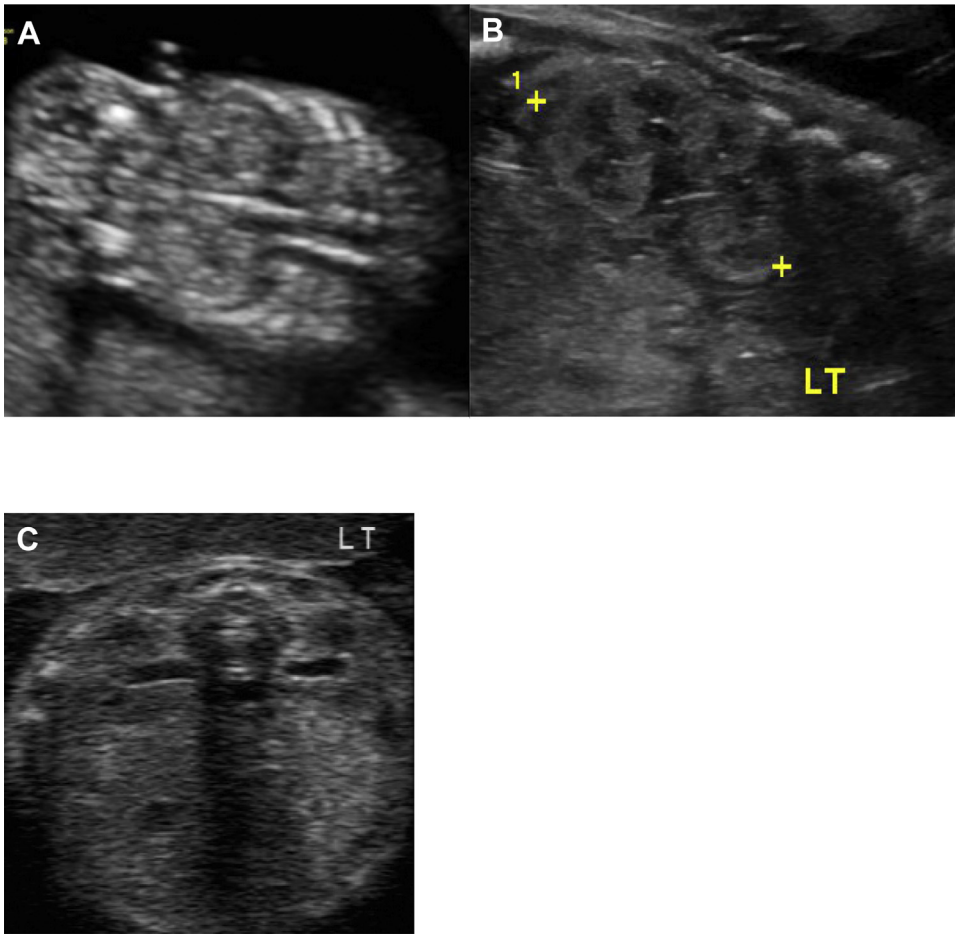
SMFM Fetal Anomalies Consult Series #4: Genitourinary anomalies

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Introduction

The fetal genitourinary system includes the fetal kidneys, ureters, and bladder and internal and external genitalia. The standard ultrasound examination of the fetal genitourinary system after the first trimester of pregnancy includes visualization of the fetal kidneys and bladder (Figures 1, A and B and 2). The renal pelvis should be assessed for dilation in an axial view of the anteroposterior diameter in the second and third trimesters of pregnancy (Figure 1, C). The fetal genitalia should be examined in multiple gestations, as

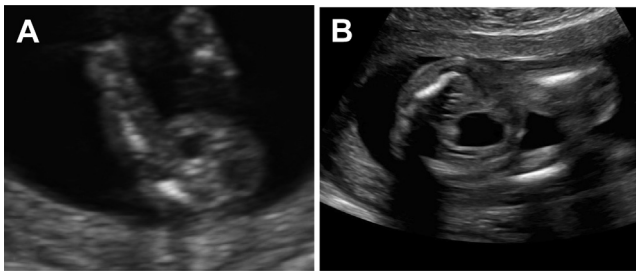
FIGURE 1
Evaluation of the fetal kidneys



A, Coronal view of the fetal kidneys in the first trimester of pregnancy. **B,** Sagittal view of the fetal kidneys in the second trimester of pregnancy. **C,** Anteroposterior assessment of the fetal pelvis in the second trimester of pregnancy.

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FIGURE 2
Fetal bladder



Fetal bladder in the (**A**) first and (**B**) second trimesters of pregnancy.

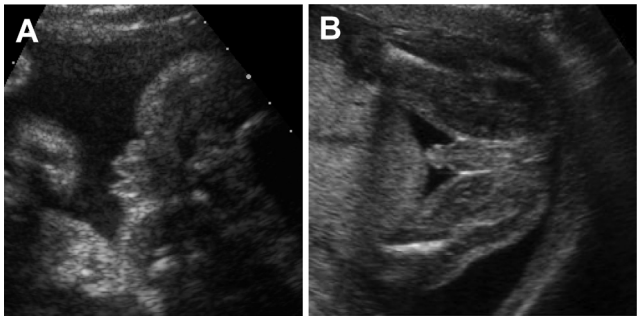
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this can aid in determination of chorionicity, or when medically indicated as when a patient is at risk of an X-linked genetic disorder (Figure 3). In addition, measurement of the amniotic fluid volume should be performed (Figure 4), as it provides a functional assessment of the fetal kidneys, which produce amniotic fluid after 16 to 17 weeks of gestation. Renal pathology can therefore result in both increased and decreased amniotic fluid volume.

When indicated, a detailed ultrasound examination (76811) may include an examination of the adrenal glands and interrogation of the renal arteries (Figure 5). Examination of the fetal genitalia is also included.[1]

Abnormalities of the genitourinary system are among the most common fetal structural malformations. Such anomalies range from mild (eg, mild urinary tract dilation) to severe life-threatening anomalies (eg, bilateral renal agenesis).

FIGURE 3
Female and male genitalia in the second trimester of pregnancy



A, Female genitalia in the second trimester of pregnancy. **B,** Male genitalia in the second trimester of pregnancy.

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FIGURE 4
Amniotic fluid assessment



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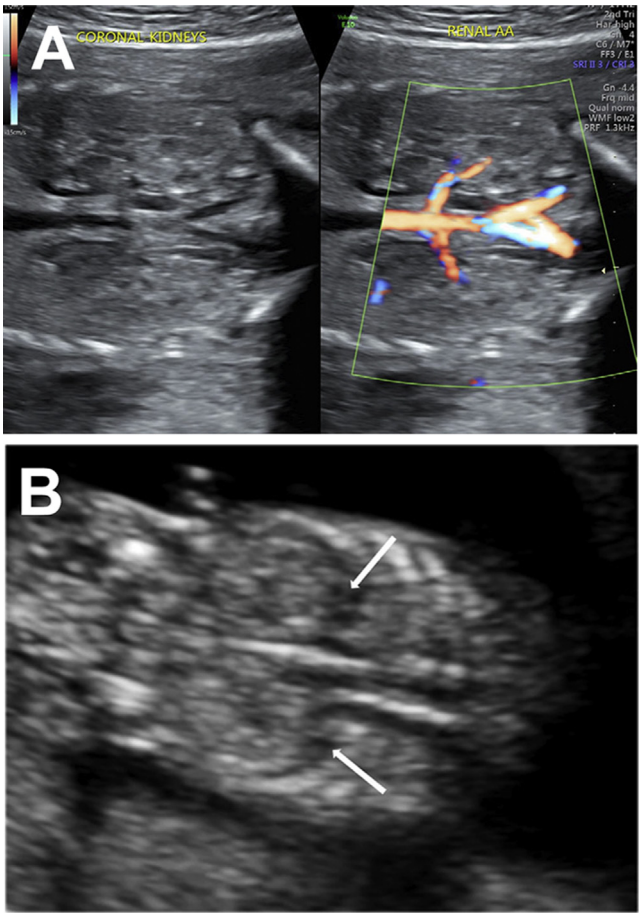
Because the kidneys are responsible for the production of amniotic fluid, serious renal abnormalities that impair the production or excretion of urine (eg, urinary tract obstruction) can result in severe oligohydramnios, which can lead to pulmonary hypoplasia and can be life-threatening. The fetal bladder and renal pelvis are relatively easily visualized, and therefore, renal abnormalities are usually readily detected. Those abnormalities that are more difficult to identify, such as unilateral renal agenesis or a pelvic kidney, are less likely to be life-threatening or harmful to the fetus.

Most renal anomalies are isolated, and as a general rule, the risk of underlying aneuploidy or genetic syndromes is low. The exception is polycystic kidneys, which can be associated with autosomal recessive or dominant disorders. Other genetic syndromes can be characterized by various forms of multicystic or polycystic kidneys. Therefore, cystic kidney disease should prompt careful assessment to look for other anomalies and inherited diseases.

This Consult reviews the ultrasonographic diagnosis, genetic evaluation, and potential treatment and outcome of the following genitourinary abnormalities:

- Adrenal neuroblastoma
- Autosomal recessive polycystic kidney disease
- Bladder outlet obstruction
- Duplicated collecting system
- Ectopic ureterocele
- Hydroureter
- Hypospadias
- Multicystic dysplastic kidney
- Ovarian cyst
- Pelvic kidney

FIGURE 5
Coronal image of the kidneys



A, Coronal image demonstrating interrogation of the renal arteries. **B,** First trimester of pregnancy image of adrenal glands superior to the kidneys (*arrows*).

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- Renal agenesis
- Renal pelvic dilation
- Urinoma

Coding

When coding for fetal genitourinary anomalies, the Society for Maternal-Fetal Medicine Coding Committee recommends utilizing the *International Classification of Diseases*, Tenth Revision, code series O35.8XX.

ACKNOWLEDGMENTS

The authors wish to acknowledge Mary E. Norton, MD; Jeffrey A. Kuller, MD; and Angie C. Jelin, MD, for providing a review of the genetics content and Joseph Wax, MD, for providing a general review of this Consult.

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Adrenal neuroblastoma

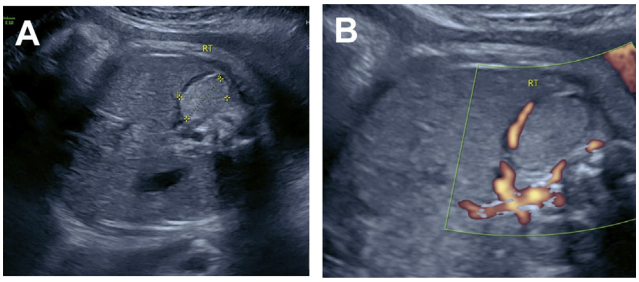
Society for Maternal-Fetal Medicine (SMFM); Jeffrey Sperling, MD

Introduction

The fetal adrenal glands can be seen by ultrasonography by the end of the first trimester of pregnancy.[1] They appear as pyramidal hypoechoic structures superior to the hyperechoic kidney. During the second trimester of pregnancy, corticomedullary differentiation can be observed with a hypoechoic cortex and hyperechoic medulla. The size of the gland increases throughout gestation but remains smaller than the kidney. During the third trimester of pregnancy, the appearance of the fetal adrenal glands is similar to that of the neonatal adrenal glands.[1]

Neuroblastomas account for 50% of fetal adrenal masses.[2,3] They are more common in White infants and slightly more common in males than in females.[2] Neuroblastomas originate in the neural crest cells of the sympathetic nervous system. Although most cases arise from the adrenal gland, they can also occur in the posterior mediastinum.[2]

FIGURE
Fetal neuroblastoma



A, Solid suprarenal well-encapsulated mass. **B,** Doppler ultrasound showing limited vascular flow.

RT, right.

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Definition

The term neuroblastoma refers to a spectrum of neuroblastic tumors (eg, neuroblastomas, ganglioneuroblastomas, and ganglioneuromas) that arise from sympathetic ganglion cells.[2] Although neuroblastomas are malignant tumors, and some can metastasize, the prognosis is excellent, and many cases regress spontaneously.[4]

Ultrasound Findings

Adrenal neuroblastoma generally appears as a well-encapsulated cystic or solid mass adjacent to but separate from the kidney and other retroperitoneal structures (Figure).[3] Assessment of the contralateral adrenal gland to rule out normal but prominent adrenal glands is recommended. Findings on color Doppler interrogation are variably reported and include peripheral flow, no flow, or internal vascularization of the hyperechogenic aspects of the mass.[3,5] A single feeding artery is not typically present and would suggest the more common subdiaphragmatic bronchopulmonary sequestration (BPS).[6]

Associated Abnormalities

Postnatally, neuroblastoma has been associated with Li-Fraumeni syndrome, Hirschsprung disease, and neurofibromatosis type 1, but these associations have not been reported in prenatal cases. If a neuroblastoma markedly enlarges and compresses the gastrointestinal tract, polyhydramnios may develop.[7] Elevated catecholamines have been reported and may cause maternal symptoms, such as tachycardia, hypertension, nausea, and vomiting.[8] In addition, catecholamine release has been associated with fetal cardiomyopathy, tachycardia, and hydrops.[9] Rarely, fetal neuroblastomas may metastasize to the liver.[10]

Differential Diagnosis

The differential diagnosis of a mass in this region includes adrenal cysts[11] (isolated or associated with multicystic dysplastic kidneys or Beckwith-Wiedemann syndrome), adrenal hemorrhage,[12] subdiaphragmatic BPS,[13] hepatic tumor, and adrenogenital syndrome (secondary to congenital adrenal hyperplasia).[14] Hemorrhage in adrenal cysts associated with Beckwith-Wiedemann syndrome has been reported, in which case the appearance is that of a complex mass.[15]

Genetic Evaluation

Fetal neuroblastoma is typically a sporadic finding. If no further abnormalities are noted on ultrasound and the family history is unremarkable, no genetic evaluation beyond standard aneuploidy screening is typically recommended.

Pregnancy and Delivery Management

Hydrops fetalis, polyhydramnios, or both may develop in rare circumstances, typically with large lesions or in the setting of metastatic disease; therefore, serial ultrasound examinations should be performed. Serial ultrasound assessment can help rule out adrenal hemorrhage, which can evolve over time.[4,16] A consultation with pediatric oncology, neonatology, and surgery should be obtained to plan and coordinate prenatal and postnatal management. In general, pregnancy termination is an option that should be offered to patients when a major fetal anomaly is detected.

However, in most cases of neuroblastoma, the prognosis is favorable, and the outcome is good. Shared patient decision-making requires a thorough evaluation and multidisciplinary counseling regarding prognosis. If pregnancy termination is pursued, histologic evaluation can confirm the diagnosis.

In most cases, vaginal delivery is appropriate, and cesarean delivery should be reserved for the usual obstetrical indications. Cesarean delivery has been suggested as potentially preferable for very large cystic adrenal masses to prevent rupture or soft tissue dystocia. Adrenal cyst aspiration before delivery is controversial because this may cause bleeding, malignancy seeding, preterm labor, or infection. Delivery in a tertiary care center is recommended with consideration of early delivery if there is evidence of fetal compromise. Postnatal investigation, including ultrasound, magnetic resonance imaging, or other imaging modalities, is recommended. Expectant management, needle biopsy, or surgical exploration may be needed on the basis of the results of the above imaging, final diagnosis, and neonatal condition.

Prognosis

In general, the prognosis is good, and neuroblastomas may even resolve in utero or shortly after birth.[3,13] Survival rates of infants with low-stage disease are excellent, even for those with metastatic disease.[17] Cases of spontaneous involution have been reported.[18] The recurrence risk of this lesion is unknown but likely low.

Summary

Fetal adrenal neuroblastomas, derived from neural crest cells, are the most frequently diagnosed extracranial solid tumor of childhood. These rare tumors are associated with an excellent postnatal prognosis. Close surveillance with serial ultrasound examinations evaluating for signs of hydrops fetalis and polyhydramnios is recommended. Antenatal consultation with pediatric oncology, neonatology, and surgery is advised.

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Autosomal recessive polycystic kidney disease

Society for Maternal-Fetal Medicine (SMFM); Kate Swanson, MD

Introduction

Autosomal recessive polycystic kidney disease (ARPKD) is a rare genetic disorder with an estimated incidence of 1 in 20,000 live births.[1] Although there is variability in presentation and new advances changing the life expectancy of affected individuals, it remains a disease with high morbidity and mortality, particularly when diagnosed prenatally.

Definition

Individuals affected with ARPKD typically have biallelic pathogenic variants in the *PKHD1* gene on chromosome 6p21. These variants affect the production of fibrocystin, a protein found in the primary cilium or basal body complex of epithelial cells in the renal tubules and hepatic bile ducts.[2] These variations result in the elongation and dilation of the collecting ducts, formation of microcysts, and diffuse enlargement of the kidneys.[3] As a result, affected individuals develop end-stage renal disease and hepatobiliary disease.

Ultrasound Findings

There is wide variability in the prenatal presentation of ARPKD. In some cases, enlarged hyperechoic kidneys with poor corticomedullary differentiation can be identified in the second trimester of pregnancy. Frequently, the kidneys are quite enlarged, ranging from 4 to 15 standard deviations above normal in size. In addition, oligohydramnios or anhydramnios are frequently present in ARPKD. In more subtle cases, mild enlargement and a hyperechoic cortical rim may be the only findings. In the third trimester of pregnancy, larger cysts >3 mm in size may develop. These cysts can be bilateral or unilateral. In some cases, no abnormality is identified prenatally (Figure).[4]

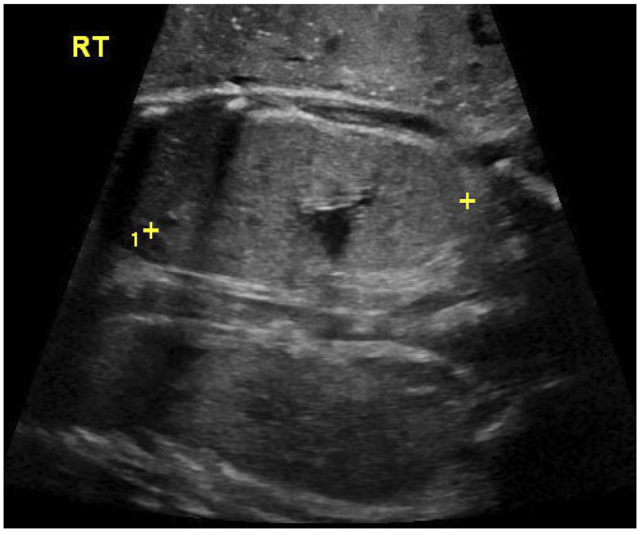
Associated Abnormalities

Aside from oligohydramnios or anhydramnios and the resultant pulmonary hypoplasia, which can sometimes be suspected with ultrasonography, additional ultrasonographic anomalies are uncommon. Although hepatic fibrosis and biliary dysgenesis are common in affected patients, these findings are rarely identified prenatally.[4,5]

Differential Diagnosis

Bardet-Biedl syndrome is another ciliopathy that can present prenatally with enlarged cystic kidneys. Postaxial polydactyly is common in individuals with Bardet-Biedl syndrome and can help differentiate it from ARPKD. In addition, the renal parenchyma is typically more homogenous in individuals with Bardet-Biedl syndrome.[6] Meckel-Gruber syndrome is another rare autosomal recessive condition that can present with enlarged cystic kidneys; the cysts often appear earlier than in ARPKD. Postaxial polydactyly and central nervous system malformations are common with Meckel-Gruber syndrome.[5]

FIGURE
Autosomal recessive polycystic kidney disease



Kidneys are enlarged, measuring 6.0 cm, and demonstrate typical loss of corticomedullary differentiation. Note the absence of amniotic fluid surrounding the fetus, which is consistent with oligohydramnios.

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Genetic Evaluation

Prenatal diagnostic testing should be offered when ARPKD is suspected. In addition, chromosomal microarray analysis can be offered when prenatal diagnosis is performed, particularly in the presence of additional ultrasound findings, although it will not detect single-gene disorders, including ARPKD. Molecular genetic testing on tissue obtained by chorionic villus sampling or amniocentesis can be used to identify pathogenic variants in the *PKHD1* gene. The presence of two pathogenic variants in this gene confirms the diagnosis of ARPKD. However, even in cases with strong histopathologic and clinical support for the diagnosis, variant detection rates range from 80% to 85%.[7,8] If there are additional anomalies, consanguinity, or a family history of a specific condition, gene panel testing or exome sequencing is sometimes useful.

Pregnancy and Delivery Management

Given the poor prognosis of individuals with prenatally identified ARPKD, particularly in the setting of early oligohydramnios or anhydramnios, patients with an affected fetus should be offered pregnancy termination. Parents who choose to continue their pregnancy should be offered comfort care for the neonate at the time of delivery. During pregnancy, serial ultrasound examinations can be useful to assess kidney size and amniotic fluid volume.[9] Parents who desire full resuscitation should deliver at a center with a level IV neonatal intensive care unit. Because massive enlargement of the kidneys can result in an abdominal circumference that may preclude a vaginal delivery at term, cesarean delivery may be necessary.[10]

Prognosis

There exists marked variability in the presentation and prognosis of ARPKD. Prenatally diagnosed ARPKD is associated with poorer outcomes than ARPKD identified in the neonatal period or childhood. Those with oligohydramnios or anhydramnios identified on prenatal ultrasound are at considerable risk of pulmonary hypoplasia and have an approximately 30% mortality rate within the first year of life.[11] In addition, it is reported that the genotype may have some association with phenotype and that individuals with truncating variants are more likely to have poor outcomes than those with missense variants.[3]

Individuals who survive beyond the first month of life have a better prognosis, with reported survival rates at 10 years as high as 82%. However, these patients typically require dialysis and renal transplantation and frequently develop hypertension, hepatic fibrosis, and portal hypertension.[12]

Summary

ARPKD is infrequently diagnosed on prenatal ultrasound. However, when it is identified, it is associated with significant morbidity and mortality. As an autosomal recessive disease with variable expressivity, parents and siblings of patients with affected pregnancies should be evaluated and counseled on their risks of also having an affected pregnancy.

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Bladder outlet obstruction

Society for Maternal-Fetal Medicine (SMFM); Anne Mardy, MD

Introduction

Fetal bladder outlet obstruction (or lower urinary tract obstruction [LUTO]) is most commonly caused by posterior urethral valves and urethral atresia and can lead to abnormal renal development and pulmonary hypoplasia. It is associated with a high rate of perinatal morbidity and mortality.

Definition

Prenatally detected LUTO occurs because of a blockage in the lower urinary tract (the bladder outlet) of the developing fetus and leads to megacystis, a thickened bladder wall, and bilateral hydronephrosis with or without cystic dysplasia of the renal parenchyma.

Ultrasound Findings

In the first trimester of pregnancy, megacystis, or an enlarged bladder, is commonly defined as a sagittal length >7 mm.[1–3] After the first trimester of pregnancy, there is no single definition of megacystis, with many different definitions found in the literature.[4] One study defined the normal sagittal length as the gestational age in weeks minus 5 mm (±95% upper or lower confidence interval [CI]=7); megacystis was defined as greater than the upper limit of the 95% CI for the gestational age.[5] A thickened bladder wall is defined as one that measures >3 mm. Hydronephrosis is defined as dilation of the renal pelvis, as measured in the anteroposterior diameter, of ≥4 mm in the second trimester of pregnancy and ≥7 mm in the third trimester of pregnancy (Figure 1). A dilated posterior urethra, also known as the "keyhole" sign, is commonly associated with posterior urethral valves (Figure 2). In addition, ureteral dilation may be seen because of the reflux from high bladder pressure. The kidneys may develop cystic dysplasia or become echogenic and atrophied.

FIGURE 1
Marked bladder dilation with dilation of the renal pelves



Dilation of renal pelvis consistent with bladder outlet obstruction.

LK, left kidney; *RK*, right kidney.

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Associated Abnormalities

Most cases (78%) of LUTO are isolated.[6] LUTO is often associated with oligohydramnios, possibly leading to clubbed feet or pulmonary hypoplasia. Urinary ascites and perinephric urinomas can occur as a result of bladder or kidney rupture.

Differential Diagnosis

The most common etiology of LUTO is posterior urethral valves (63%), which are congenital membranes in the posterior urethra that act as valves to block micturition. Classic features include megacystis, thickened bladder wall, dilated posterior urethra ("keyhole" sign), bilateral hydronephrosis or cortical cysts, and oligohydramnios. Urethral atresia is the second most common etiology of LUTO (10%) and may have the same appearance of a greatly enlarged bladder, although without a dilated urethra or keyhole appearance. Unlike posterior urethral valves, which occur only in males, urethral atresia can occur in both male and female fetuses.[6]

Other conditions in the differential diagnosis of a dilated bladder include Prune-Belly syndrome (the triad of lax or absent abdominal musculature; a thin-walled, dilated bladder; and cryptorchidism), aneuploidy (most commonly trisomy 13,18, or 21), megacystis-megaureter syndrome (severe vesicoureteral reflux), and megacystis-microcolon syndrome (thin-walled bladder without dilated posterior urethra; normal or increased amniotic fluid).[6,7] In a female fetus, a dilated vagina caused by a septal anomaly can mimic a dilated bladder. A persistent cloaca (convergence of bladder, rectum, and vagina with a single perineal opening) should also be considered. A large case series

FIGURE 2
Markedly dilated bladder



The *asterisk* indicates a distended proximal urethra, giving the characteristic keyhole appearance of the proximal urethral valves. Oligohydramnios is present.

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found that 26.9% of prenatal diagnoses of LUTO were falsely positive. The most common final postnatal diagnoses in these cases were vesicoureteral reflux (24.5%), cloacal dystrophy (18.9%), and hydronephrosis (11.3%). In 5 cases, the obstruction resolved during the pregnancy.[6]

Genetic Evaluation

Diagnostic testing with amniocentesis or chorionic villus sampling and chromosomal microarray analysis (CMA) should be offered when bladder outlet obstruction is detected. If a dilated bladder and severe oligohydramnios make amniocentesis not feasible, testing can be done by placental biopsy or on fluid obtained by vesicocentesis. If ultrasound findings or screening test results suggest a common aneuploidy, it is reasonable to initially perform karyotype analysis or fluorescence in situ hybridization, with reflex to CMA if these test results are normal. If there are additional anomalies, consanguinity, or a family history of a specific condition, gene panel testing or exome sequencing is sometimes useful because CMA does not detect single-gene (Mendelian) disorders. If exome sequencing is pursued, appropriate pretest and posttest genetic counseling by a provider experienced in the complexities of genomic sequencing is recommended. After appropriate counseling, cell-free DNA screening is an option for patients who decline diagnostic evaluation particularly if a common aneuploidy is suspected.

Pregnancy and Delivery Management

Given the poor prognosis associated with LUTO, pregnancy termination should be offered. Shared patient decision-making requires a thorough evaluation and multidisciplinary counseling regarding prognosis. For patients who continue their pregnancy, serial vesicocenteses have been suggested to assess fetal renal function to help determine whether fetal intervention should be pursued, although there is controversy regarding their benefit as prognostic markers.[7] Fluid from the bladder is completely removed two or three times sequentially to measure urinary electrolytes and the degree of bladder refilling. Normal values for fetal urine are as follows: sodium<100 mg/dL, chloride<90 mg/dL, osmolarity<200 mOsm/L, calcium<8 mg/dL, total protein<20 mg/dL, and beta-2-microglobulin<4 mg/dL.[8] After the first vesicocentesis, a subsequent ultrasound examination can determine whether the bladder refills. The absence of bladder refill usually indicates severe renal dysfunction, and no further vesicocenteses are recommended.[7]

A staging system for LUTO with recommended fetal therapies has been established.[7] Possible fetal interventions include cystoscopy, vesicoamniotic shunt, or amnioinfusion. Cystoscopy can allow for both diagnosis and therapy by guidewire passage through the urethra or laser ablation of posterior urethral valves. In the PLUTO (Percutaneous vesicoamniotic shunting for fetal Lower Urinary Tract Obstruction) trial, a vesicoamniotic shunt did not increase survival to 28 days compared with conservative management in an intention-to-treat analysis but did increase survival based on actual treatment. Morbidity and mortality were very high in both groups, and there was a high rate of shunt complications.[9] Serial amnioinfusions have been proposed as an option to allow survival in severe cases by reducing the risk of pulmonary hypoplasia, although data are limited, and further research is needed to determine the appropriate role for this intervention.[10]

Parents who desire full resuscitation should deliver at a center with a level IV neonatal intensive care unit (NICU). In general, mode of delivery should be based on usual obstetrical indications and parents' preferences regarding resuscitation in severe cases. Planned preterm delivery to shunt the bladder has not been demonstrated to be of benefit.

Prognosis

LUTO is associated with high fetal and perinatal morbidity and mortality. The worst prognosis is seen in the presence of early, severe, prolonged oligohydramnios with associated pulmonary hypoplasia.[11,12] Other poor prognostic features include renal parenchymal abnormalities and abnormal fetal urinalysis. A significant percentage of patients with posterior urethral valves will develop end-stage renal disease and require dialysis and transplantation.[13] These patients require a prolonged NICU stay, often require a gastrotomy tube for several years, and are prone to infections and mechanical dialysis failures. In addition, posterior urethral valves may cause damage to the bladder, and the child may require clean intermittent catheterization or bladder surgeries to achieve continence after birth and throughout life.

Summary

Fetal LUTO is characterized by an enlarged bladder, thickened bladder wall, and hydronephrosis. It is most commonly caused by posterior urethral valves. Fetal LUTO can lead to abnormal renal development and pulmonary hypoplasia and is associated with a high perinatal morbidity and mortality rate. Vesicocentesis and genetic testing should be offered to evaluate for the possibility of fetal intervention, although the optimal intervention and outcomes are unclear. Interventions that have been reported include cystoscopy with or without ablation of the valve, vesicoamniotic shunting, or amnioinfusion. Despite intervention, the prognosis is often poor, with high rates of pulmonary hypoplasia, end-stage renal disease, and bladder dysfunction.

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Duplicated collecting system

Society for Maternal-Fetal Medicine (SMFM); Linda M. Hopkins, MD

Introduction

The normal urinary collecting system develops from a single ureteric bud arising from the mesonephric (Wolffian) duct. The ureteric bud migrates to meet the primitive kidney (metanephros), where it develops into the collecting system. Abnormalities in the signaling pathways are thought to result in a variety of collecting system variants, including two ureteric buds arising from the mesonephric duct (complete duplication) or one bud that bifurcates during development (incomplete duplication).[1] The incidence of duplication in adults is thought to be approximately 0.7% to 4%, with incomplete duplication more common than complete duplication.[2]

Definition

The term duplicated (or duplex) collecting system is defined as one kidney with two separate, noncommunicating renal pelves. In complete duplication, each pelvis has an associated ureter that inserts independently into the bladder. In incomplete duplication, two pelves are present but share a single (bifid) ureter that inserts into the bladder but bifurcates at some point before reaching the two pelves.

Ultrasound Findings

A single kidney measuring slightly larger than normal (above the 95th percentile for gestational age) with two renal pelves is seen longitudinally.[3] One renal pelvis is seen in the lower pole of the kidney and the second in the upper pole (Figure). Although a bifid ureter or two ureters are present, both are difficult to visualize unless dilated. A duplicated collecting system is estimated to be bilateral in 30% of cases.[3]

The bladder should be inspected for a ureterocele, which appears as a cystic structure at the bladder wall. Of note, if the bladder is full, the ureterocele may be compressed and therefore not seen.

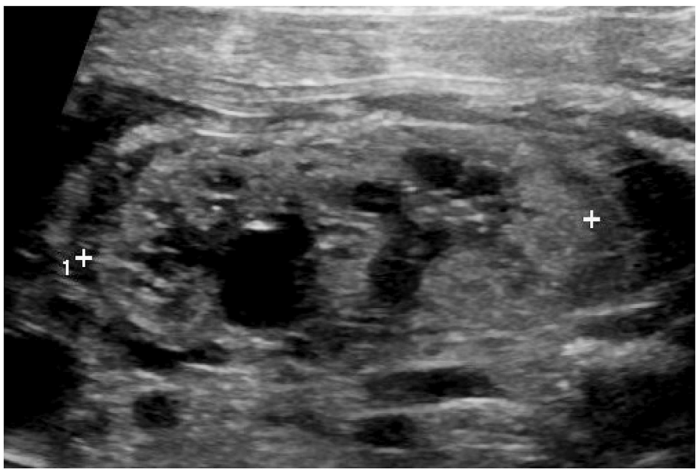
In complete duplication, the ureter draining the upper pole moiety typically has an ectopic insertion site into the bladder located inferior and medial to the normal bladder insertion site. If a ureterocele is present, it is usually associated with the ectopic insertion site.

Associated Abnormalities

In complete duplication, the upper pole moiety is more prone to abnormalities than the lower pole moiety. The abnormality is usually hydronephrosis because of an obstructive process. The lower pole moiety typically has a normal appearance, although vesicoureteral reflux can occur.[4]

A duplicated collecting system is usually an isolated finding and only rarely seen in aneuploidy or genetic syndromes. A detailed evaluation of the fetal anatomy is indicated if a duplicated system is seen, although additional anomalies are uncommon.

FIGURE
Longitudinal view of a fetal kidney with a duplicated collecting system



The kidney has two separate (upper and lower) renal pelves that do not communicate.

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Differential Diagnosis

The fetus with a duplicated collecting system typically has a normal-sized bladder. When hydronephrosis is the initial finding, the differential diagnoses include both physiological or transient hydronephrosis and other congenital anomalies of the kidney and urinary tract. Assessment of bladder size and number of pelves using a longitudinal view is recommended. The findings of a normally sized kidney with a single pelvis rule out a duplicated system as the cause of hydronephrosis. All other causes of hydronephrosis, including ureteropelvic junction obstruction and vesicoureteral reflux, should be considered. An abnormally sized kidney may also be present with urethral atresia, Prune-Belly syndrome, and posterior urethral valves. A ureterocele can occasionally be seen in single collecting systems; therefore, a longitudinal view of the kidney is helpful in differentiating between single and duplex systems.

Finally, an enlarged kidney that lacks a second pelvis can be seen in the presence of a renal tumor. Overall, tumors of the kidney are rare, with mesoblastic nephroma the most commonly detected. This tumor arises exclusively in the third trimester of pregnancy and demonstrates rapid growth.[5]

Genetic Evaluation

If a duplicated collecting system is seen as an isolated anomaly and the family history is unremarkable, the chance of a genetic syndrome is low. A genetic component of this anomaly is suspected because duplicated systems seem to have an autosomal dominant inheritance in some families.[6] If no further abnormalities are noted on ultrasound, no genetic evaluation beyond standard aneuploidy screening is typically recommended.

Pregnancy and Delivery Management

An ultrasound examination during the third trimester of pregnancy is recommended to assess for hydronephrosis and monitor the amniotic fluid volume. Otherwise, standard obstetrical care and delivery are warranted.

Prognosis

If neither renal pelvis demonstrates hydronephrosis prenatally, the duplicated system is considered a normal variant, and the prognosis is excellent. However, if hydronephrosis is present, postnatal evaluation is indicated. Obstruction, reflux, incontinence, and infection can occur, which can lead to renal scarring or dysplasia if left untreated.

Summary

A duplicated collecting system in the fetus is a relatively common finding that is typically isolated in nature and not significantly associated with chromosomal abnormalities or genetic syndromes. On ultrasound, a longitudinal view of the kidney is usually necessary to appreciate this anomaly, which is defined by two noncommunicating renal pelves in a single kidney. A duplicated collecting system detected antenatally should be followed up with an ultrasound examination in the third trimester to assess for hydronephrosis in one or both pelves. If hydronephrosis does occur, postnatal follow-up is indicated.

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Ectopic ureterocele

Society for Maternal-Fetal Medicine; Neda Ghaffari, MD

Introduction

An ectopic ureterocele is commonly associated with the ectopically inserted ureter of a duplicated collecting system and is more common in female fetuses.[1,2] It can be detected on prenatal imaging as an incidental finding or during the evaluation of hydronephrosis.

Definition

A ureterocele is a cystic dilation of the terminal ureter. The American Academy of Pediatrics classifies ureteroceles based on their location. Intravesical ureteroceles are located entirely in the bladder and are more common in adults than in fetuses and children.[3] Ectopic ureteroceles are located distal to the trigone, inserting into the bladder neck, urethra (cecoureterocele), or elsewhere in the pelvis, and are more likely to be detected during prenatal or pediatric imaging.[4] Approximately 80% of ureteroceles are associated with the upper pole of a duplicated collecting system; moreover, 60% of these are ectopic.[3,5]

Ultrasound Findings

On ultrasound, a ureterocele appears as a thin-walled cystic mass seen in the fetal bladder. It is best visualized when the bladder is partially filled. A ureterocele can be confused with the bladder when the bladder is completely empty. Conversely, the ureterocele can become compressed or prolapse if the bladder is completely full.[6] Therefore, the bladder should be visualized more than once throughout the ultrasound examination.

When an ectopic ureterocele is suspected, the bladder, bilateral kidneys, bilateral ureters, and amniotic fluid should be carefully evaluated. The fetal kidneys should be visualized both axially and longitudinally. Obstruction of the ureter by the ureterocele at the ureterovesical junction can lead to a dilated ureter and hydronephrosis. Renal duplication is often associated with the visualization of two separate renal poles, with the ectopic ureter most commonly draining the upper renal pole moiety.[7] Obstruction from the ureterocele is often seen in the upper pole moiety. In addition, the lower renal pole moiety can be dilated, generally from reflux. The duplicated kidney may be larger than the contralateral kidney. The lower pole moiety may also be displaced inferiorly because of mass effect.

If a ureterocele is sufficiently large, it can lead to obstruction of the contralateral kidney or bladder outlet obstruction, resulting in oligohydramnios.

Associated Abnormalities

Ureteroceles are generally isolated anomalies and are not associated with anomalies outside of the urologic system. The contralateral kidney and ureter should be visualized carefully to assess for contralateral duplication.

Differential Diagnosis

When a cystic mass is seen in the fetal bladder, it most commonly represents an ectopic ureterocele. The kidneys should be carefully examined to look for a duplicated collecting system. The differential diagnoses of ectopic ureterocele include anomalies associated with hydronephrosis, such as posterior urethral valves, ureteropelvic junction obstruction, vesicoureteral reflux, and congenital megaureter. However, in these conditions, the bladder appears normal. A bladder "hutch" diverticulum, or periureteral bladder diverticulum, can have a similar appearance to that of a ureterocele, but it is extrinsic and does not prolapse into the bladder lumen. The visualization of a crescentic membrane in the fetal bladder is considered pathognomonic for ectopic ureterocele (Figure).

Genetic Evaluation

An ectopic ureterocele is generally considered a sporadic finding. In the absence of other anomalies or a concerning family history, genetic evaluation beyond routine aneuploidy screening is not generally recommended.

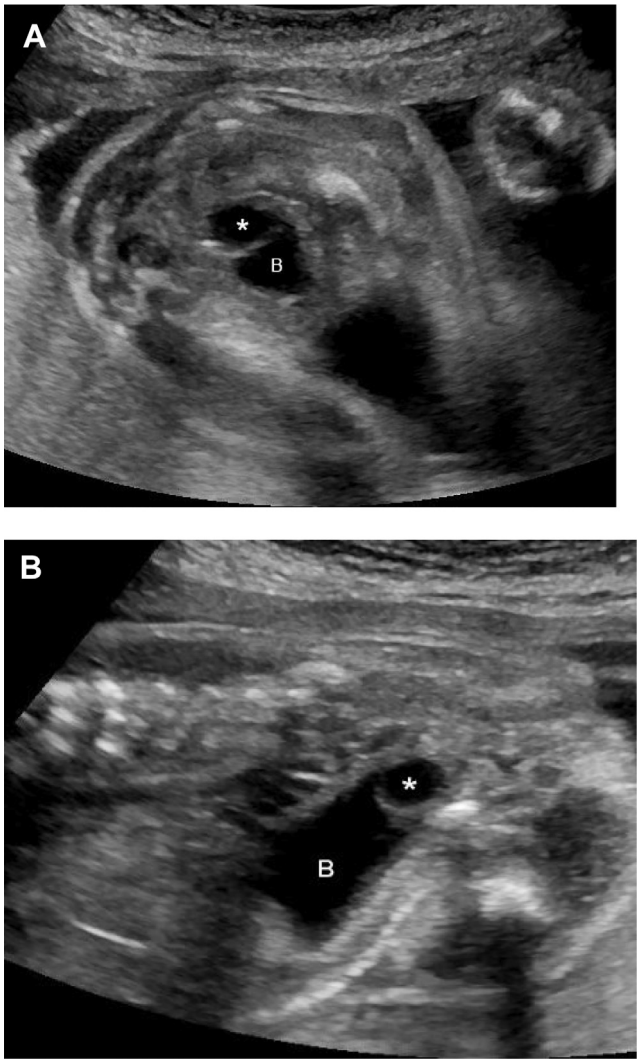
Pregnancy and Delivery Management

Follow-up ultrasound examination is recommended to follow fetal growth, measure amniotic fluid volume, and assess for development of renal obstruction. In utero decompression with laser incision or needle puncture has been described in cases of ectopic ureterocele with bladder outlet obstruction and oligohydramnios[8] and may be considered in specialty centers under a research protocol. Prenatal pediatric urology and neonatology consultations should be considered to provide counseling about prognosis and postnatal management. Obstetrical management, delivery timing, and mode of delivery should be based on obstetrical indications.

Prognosis

The prognosis of ectopic ureterocele is largely dependent on the degree of renal obstruction. The prognosis is excellent in the absence of obstruction or reflux.[9,10] If not detected antenatally, ectopic ureterocele may present as a urinary tract infection in the first months of infancy[3] and can lead to urosepsis. The prognosis is improved with prenatal diagnosis and earlier initial intervention when indicated. In one retrospective study of 95 patients with prenatal or postnatal diagnosis, the incidence of urinary tract infection was decreased with prenatal diagnosis, as was the need for secondary procedures (20% vs 46%).[11]

FIGURE
Axial and longitudinal views of the bladder



Axial (**A**) and longitudinal (**B**) views of the bladder showing a ureterocele (marked with *asterisk*).

B, bladder.

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The postnatal workup generally includes an ultrasound examination of the bladder and kidneys and a voiding cystourethrogram. A radionucleotide renal scan can help assess kidney function. Pediatric urology consultation is recommended. Surgery can be considered, especially in the case of vesicoureteral reflux.[9,10] Surgical options include endoscopic ureterocele decompression, heminephrectomy, bladder reconstruction, and ureteral reimplantation.

Summary

An ectopic ureterocele appears ultrasonographically as a thin-walled cystic mass seen in the fetal bladder and represents a cystic dilation of the terminal ureter. It is most commonly associated with a duplicated collecting system and can lead to varying degrees of renal obstruction. Serial follow-up ultrasound examinations are recommended to assess fetal growth, amniotic fluid volume, and progression of renal obstruction. The prognosis of ectopic ureterocele is largely dependent on the degree of renal obstruction and can be excellent in the absence of obstruction or reflux. Prenatal pediatric urology and neonatology consultations should be considered, and postnatal pediatric urology consultation is recommended. Postnatal urologic surgery may be necessary, especially in cases of severe obstruction or reflux.

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Hydroureter

Society for Maternal-Fetal Medicine (SMFM); Sarah S. Osmundson, MD, MS

Introduction

A hydroureter can be an isolated finding, but it is usually associated with other genitourinary tract anomalies. The finding of hydroureter can be useful in identifying associated anomalies and determining the location of obstructive lesions in the genitourinary tract.

Definition

Persistent visualization of the ureter on prenatal ultrasound is abnormal and warrants further evaluation. Megaureter is defined as a ureter of ≥7 mm in diameter in a fetus with a gestational age of >30 weeks.[1]

Ultrasound Findings

Hydroureter appears as a tortuous, tubular structure in the fetal pelvis communicating with the kidney, bladder, or both. (Figure). Observation of bowel peristalsis can help differentiate hydroureter from a dilated loop of bowel. Hydroureter is commonly found in conjunction with renal pelvis and calyceal dilation and, when observed, warrants a follow-up ultrasound examination and postnatal evaluation.[2]

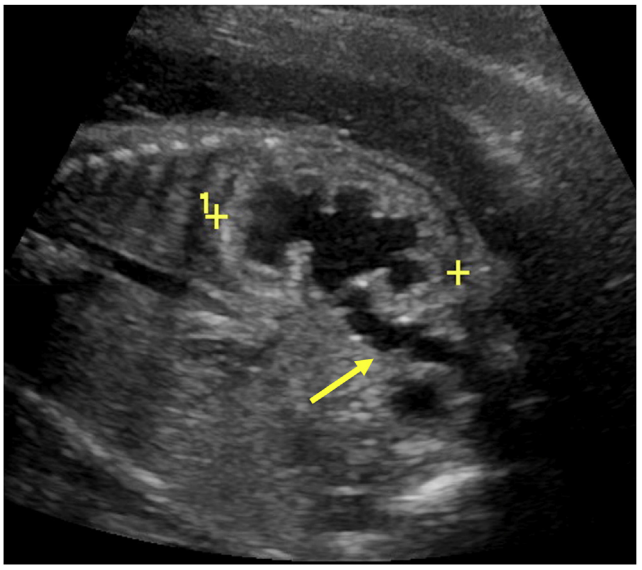
Associated Anomalies

Fetal hydronephrosis and hydroureter are often associated with other urinary tract or nonrenal congenital abnormalities, and hydroureteronephrosis is a feature of many genetic syndromes; these can include anomalies of any organ system. Hydroureter is commonly associated with vesicoureteral reflux or lower urinary tract obstruction, including a duplicated collecting system or bladder outlet obstruction. Fetuses with complete bladder outlet obstruction or significantly impaired fetal renal function will have associated oligohydramnios or anhydramnios.

Differential Diagnosis

Hydroureter can result from an anomaly intrinsic to the ureter in which abnormal development results in an aperistaltic segment and functional obstruction.[3] More commonly, a hydroureter is due to either (1) vesicoureteral reflux (VUR) or (2) obstructive uropathy. VUR causes retrograde passage of urine from the bladder to the upper urinary tract, which can lead to pyelonephritis and end-stage renal disease after birth.[4] VUR is a common cause of urinary tract dilation and can be accompanied by a hydroureter. Fluctuation in the size of the ureter and renal pelvis as the bladder contracts can be a marker for VUR.[5] Obstruction can occur at various points along the urinary tract, and the presence or absence of a hydroureter is useful in identifying the location and type of renal anomaly, as outlined below:

FIGURE
Kidney with severe hydronephrosis and hydroureter



Kidney (between *calipers*) with severe hydronephrosis and hydroureter (*arrow*), seen as a tubular structure communicating with the kidney.

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1. Ureteropelvic junction obstruction: Obstruction at the junction between the kidney and ureter causes dilation of the renal pelvis and calyces but usually is not associated with a hydroureter, as urine is prevented from filling or dilating the ureter.[6]
2. Ureterovesical junction (UVJ) obstruction: Obstruction at the junction where the ureter enters the bladder causes a hydroureter and usually hydronephrosis without an enlarged bladder.[7] A common cause of obstruction at the UVJ is an ectopic ureter, which inserts in a location other than the normal position at the trigone of the bladder. Insertion into the bladder can be associated with ureterocele, a cystic dilation of the terminal ureter within the bladder.[8] Approximately 90% of cases of ectopic ureter are associated with a duplicated collecting system.[9]
3. Bladder outlet obstruction: Bladder outlet obstruction should be suspected when bilateral hydroureter and hydronephrosis are seen in combination with a thick-walled, trabeculated bladder. The most common cause is posterior urethral valves in males and urethral atresia in females.[10]

Genetic Evaluation

Diagnostic testing with amniocentesis or chorionic villus sampling and chromosomal microarray analysis (CMA) should be offered when a hydroureter is detected, particularly with other associated anomalies. If ultrasound findings or screening test results are suggestive of a common aneuploidy, it is reasonable to initially perform karyotype analysis or fluorescence in situ hybridization, with reflex to CMA if these test results are normal. If there are additional anomalies, consanguinity, or a family history of a specific condition, gene panel testing or exome sequencing is sometimes useful because CMA does not detect single-gene (Mendelian) disorders. If exome sequencing is pursued, appropriate pretest and posttest genetic counseling by a provider experienced in the complexities of genomic sequencing is recommended. After appropriate counseling, cell-free DNA screening is an option for patients who decline diagnostic evaluation particularly if a common aneuploidy is suspected.

Pregnancy and Delivery Management

When a unilateral hydroureter is detected, careful examination of the contralateral collecting system and bladder should be undertaken. Associated urinary tract anomalies occur frequently and affect the prognosis for both normal amniotic fluid and pulmonary development during fetal life and renal function after birth. Fetuses with moderate or severe urinary tract dilation, and those with additional findings to suggest CAKUT, warrant surveillance with follow-up ultrasound examinations. These patients may also benefit from prenatal consultation with a pediatric urology or nephrology specialist to formulate a plan for postnatal evaluation. Timing of delivery should not be affected by the presence of hydronephrosis, as preterm delivery has not been demonstrated to improve outcomes. The presence of hydronephrosis should not alter the mode of delivery, which should be based on the usual obstetrical or medical indications.

Prognosis

The long-term outcome of hydroureter depends on the underlying etiology. The prognosis for primary megaureter is generally excellent and can be managed expectantly without surgical intervention in almost three-quarters of infants.[11,12] Infants with a hydroureter due to severe obstructive uropathy may present with renal failure requiring dialysis after birth.[7]

Summary

A hydroureter is the visualization of the ureter on ultrasound, which is considered abnormal. It is commonly seen in association with hydronephrosis, and its presence can identify associated anomalies and determine the location of obstructive lesions in the genitourinary tract. ■

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Hypospadias

Society for Maternal-Fetal Medicine (SMFM); Teresa N. Sparks, MD

Introduction

Hypospadias occurs in approximately 0.2 to 4.1 in 1000 live births.[1,2] This anomaly of the male genitourinary system occurs when the embryologic urethral folds fail to fuse completely between the 7th and 14th weeks of gestation.[1,3,4] Therefore, the position of the urethral meatus becomes abnormally located along the length of the ventral shaft of the penis, scrotum, or perineum. Chordee (curvature of the penis with ventral shortening) is often present as well.[1–3]

Definition

Hypospadias is diagnosed when the urethral meatal position is abnormally located along the length of the ventral shaft of the penis, scrotum, or perineum.[1–3,5] It has been classified as proximal (urethral meatus between the perineum and midshaft), distal (between the midshaft and the glans of the penis), and glandular (within the glans of the penis).[1] Alternatively, hypospadias has also been described as anterior (urethral meatus glandular or coronal), middle (penile shaft), and posterior (penoscrotal, scrotal, perineal). Approximately 50% of cases are anterior, 30% are middle, and 20% are posterior.[4]

Ultrasound Findings

Hypospadias can be diagnosed with ultrasound as a "blunt tip" appearance of the penis on ultrasound, which indicates abnormal tapering of the distal phallus (Figure 1). Ventral shortening and curvature of the penis represents chordee. A "buried" appearance of the penis has been described, in which the penis is significantly foreshortened. The "tulip sign" describes severe hypospadias, in which there is penoscrotal transposition, and the penis is curved and located between the folds of a bifid scrotum. Finally, ventral deflection of the urinary stream in a fan shape has been described; power or color Doppler may be used to illustrate an abnormal origin of the urinary stream.[1,2,5,6] Normal amniotic fluid volume is reported in most cases of hypospadias, and polyhydramnios is rarely seen.[1] The positive predictive value of ultrasound for the prenatal detection of hypospadias has been reported as 72%.[1] However, as most cases are mild, many cases are not detected at all before birth. Some data have suggested that 3-dimensional ultrasound may give a more precise depiction of the urogenital structures with higher diagnostic yield (Figure 2).[3,4]

Associated Abnormalities

Although hypospadias is often an isolated finding, it is associated with additional anomalies in 7% to 40% of cases.[1,3,5] A variety of genetic, endocrine, and environmental etiologies have been proposed that may increase the risk of hypospadias. A two-hit hypothesis has been

FIGURE 1
Fetal hypospadias



Fetal hypospadias demonstrating a small "blunt-ended" penis (*arrow*) with two echogenic lines at the tip representing prepuce lateral folds.

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FIGURE 2
A 3-dimensional image demonstrating the "tulip sign" in severe hypospadias



The tulip is formed by a ventrally oriented penis located between two scrotal folds.

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suggested, in which an environmental insult combines with an underlying genetic predisposition.[4]

When additional anomalies are present, 40% have been reported to also affect the genitourinary system.[3–5] Bifid scrotum has been described in 33% of hypospadias cases, and cryptorchidism in 17%.[1] Inguinal hernia, vesicoureteral reflux, and ureteropelvic junction obstruction may also be seen.[2] Less common concurrent genitourinary anomalies include unilateral renal agenesis, bladder exstrophy, and perineal lipoma.[1] Moreover, hypospadias may be present as part of an underlying genetic syndrome, and additional nongenitourinary anomalies occur in 7% to 10% of cases.[3,4] Anomalies of the craniofacial structures as well as the cardiac, pulmonary, gastrointestinal, and central nervous systems have been described in association with hypospadias.[1,4,7] Additionally, an increased risk of fetal growth restriction has been reported in cases of hypospadias.[8,9]

Differential Diagnosis

The differential diagnosis when hypospadias is seen on prenatal ultrasound includes micropenis, epispadias, and cloacal anomalies.[10] In addition, it is important to distinguish between hypospadias and abnormal female genitalia, such as clitoromegaly. One method that has been described to differentiate between male and female genitalia is to measure the angle of the genital tubercle from a horizontal line through the lumbosacral skin surface in the midsagittal plane; if this angle is $>30^{\circ}$, the sex is likely to be male.[5] Testicular descent or the "dome sign" of the penis and scrotum also indicate male sex, and parallel lines of the labial folds or visualization of the uterus indicate female sex.[3,5]

Hypospadias is an isolated finding in most cases. Familial inheritance of isolated hypospadias has been documented in an autosomal dominant or X-linked manner in some cases. Recurrence risk may be as high as 14% for a sibling of an affected child, and approximately 4% to 10% of affected boys have an affected father.[7] Hypospadias also can be associated with underlying genetic syndromes, such as Smith-Lemli-Opitz, Wolf-Hirschhorn, multiple lentigines, Opitz G/BBB, Schilbach-Rott, hand-foot-genital, Elsahy-Waters, Pallister-Hall, Bardet-Biedl, Mowat-Wilson, trisomies 13 and 18, triploidy, and many others.[7,10] Microdeletion syndromes, such as 19q13.11 and 9p24.3, have been associated with hypospadias. Finally, androgen insensitivity, adrenal hyperplasia, and other endocrine abnormalities have been reported in conjunction with hypospadias, many as part of an underlying syndrome.[7]

Genetic Evaluation

Diagnostic testing (amniocentesis) should be offered when hypospadias is detected, including specifically chromosomal microarray analysis (CMA). If there are additional anomalies, consanguinity, or a family history of a specific condition, gene panel testing or exome sequencing is sometimes useful because CMA does not detect single-gene (Mendelian) disorders. If exome sequencing is pursued, appropriate pretest and posttest genetic counseling by a provider experienced in the complexities of genomic sequencing is recommended. After appropriate counseling, cell-free DNA screening is an option for patients who decline diagnostic evaluation and can be helpful in determining biologic sex with severe hypospadias or ambiguous genitalia.

Pregnancy and Delivery Management

Expectant families may be advised that hypospadias is often an isolated finding, but additional anomalies or underlying genetic syndromes are possible. Insufficient evidence exists regarding any potential benefit of antenatal testing, although this is not typically recommended with isolated hypospadias. Timing and mode of delivery should be based on usual obstetrical indications.

Prognosis

The most frequent complications of hypospadias are stenosis of the urethral meatus and difficulty controlling the urinary stream.[10] Hypospadias is treated with surgery.

Prognosis is very favorable among cases of isolated hypospadias. However, outcomes can be more variable in the setting of multiple anomalies or an underlying genetic syndrome.

Summary

Hypospadias is an anomaly of the male genitourinary system that results in the abnormal location of the urethral meatus along the length of the ventral shaft of the penis, scrotum, or perineum. Characteristic ultrasound findings, such as "blunt tip," chordee, or the "tulip sign," may lead to the diagnosis. A large number of genetic, endocrine, and environmental etiologies have been described in association with hypospadias. Hypospadias is often an isolated finding, although it can be transmitted through families. In addition, it can be associated with additional anomalies in 7% to 40% of cases, most of which affect the genitourinary system but may affect other organ systems as well. Diagnostic testing with amniocentesis should be offered when hypospadias is detected, and standard obstetrical principles should guide timing and mode of delivery. Prognosis is very favorable among isolated cases of hypospadias, although it is more variable in the setting of multiple anomalies or an underlying genetic syndrome.

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Multicystic dysplastic kidney

Society for Maternal-Fetal Medicine (SMFM); Shilpa Chetty, MD

Introduction

Multicystic dysplastic kidney disease (MCDK) is one of the most common renal abnormalities that can be identified during fetal ultrasound evaluation. It occurs in approximately 1 in 4300 live births.[1] Frequently, MCDK is unilateral and isolated with overall favorable outcomes. However, when it occurs bilaterally or with other anomalies, the prognosis may be considerably worse.[2] Bilateral MCDK occurs less frequently, with a reported incidence of 1 in 10,000. It is frequently found in male fetuses.

Definition

Multicystic dysplastic kidneys are defined by echogenic renal parenchyma with multiple noncommunicating cysts of variable sizes with no evidence of obstructive nephropathy.[2] The affected kidney is often enlarged with loss of the typical reniform shape and tends to increase in size over fetal life.[3]

Ultrasound Findings

The commonly identified ultrasonographic appearance of MCDK is a cystic paraspinal mass with cysts of various sizes distributed along the periphery of the kidney that do not communicate with each other.[4] When unilateral MCDK is present, a normal fluid-filled bladder should be present, and the level of amniotic fluid should also be normal. Absence of a fluid-filled bladder or decreased amniotic fluid should prompt an evaluation for an anomaly in the contralateral kidney, which will affect outcomes (Figure). A contralateral renal anomaly is present in approximately 40% to 50% of cases.[3] When the contralateral kidney is normal, it may be enlarged due to compensatory hypertrophy.

Associated Abnormalities

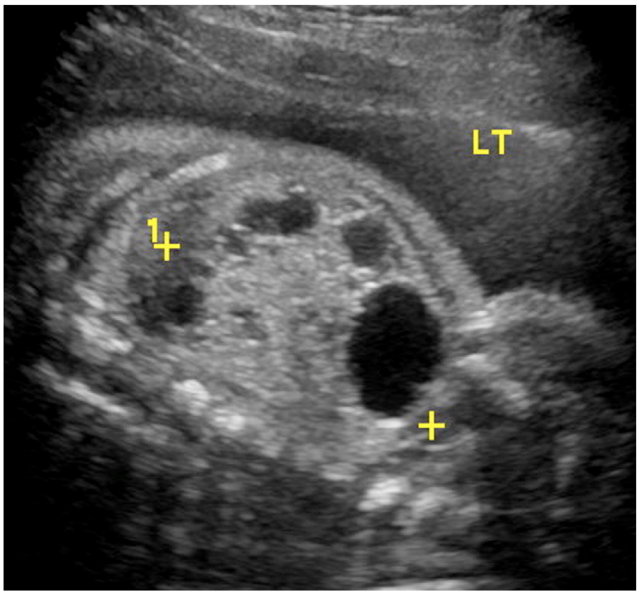
MCDK is isolated in approximately 70% to 75% of cases. Many cases include associated anomalies of the genitourinary tract, including contralateral renal agenesis, which occurs in approximately 15% of cases.[1] Other genitourinary abnormalities observed in the contralateral kidney include urinary tract obstruction and vesicoureteral reflux; abnormalities of the genitalia can also be present.[2]

Extrarenal malformations can be seen in approximately 25% of cases. These may include cardiac, central nervous system, and, gastrointestinal anomalies; omphalocele; and vertebral and skeletal malformations.[2] The presence of additional anomalies raises the concern for a possible genetic etiology.

Differential Diagnosis

MCDK may be mistaken for hydronephrosis with calyceal involvement.[3] Obstructive renal dysplasia due to ongoing severe urinary tract obstruction may cause cystic dysplasia with cysts of various sizes and an appearance similar to MCDK, but these cysts tend to develop over time and are noted in the late second and third trimesters of pregnancy. Autosomal recessive kidney disease results in enlarged hyperechoic kidneys with small uniform cysts, and with this disorder, the renal cortex is often spared and can appear hypoechoic. Many other genetic syndromes are characterized by cystic renal disease with cysts present in the medulla or glomeruli of the kidney. The appearance of these cysts may differ from those seen in MCDK, in which cysts occur throughout the kidney and the kidney parenchyma is also dysplastic.

FIGURE
Multicystic dysplastic kidney



Multicystic dysplastic kidney demonstrating a typical appearance of echogenic renal parenchyma with multiple noncommunicating cysts of variable sizes (between calipers).

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Genetic Evaluation

Chromosomal abnormalities and syndromes are present in approximately 7% to 14% of pregnancies with suspected MCDK.[1,5] The chance of an underlying chromosomal or genetic etiology increases with the presence of extrarenal anomalies.

Diagnostic testing (chorionic villus sampling or amniocentesis) with chromosomal microarray analysis (CMA)

should be offered when MCDK is detected. Xi et al[5] found that when CMA was performed in cases of isolated MCDK, a pathogenic copy number variant was identified in 14% of cases. In cases of nonisolated MCDK, specific molecular testing may be indicated depending on the syndrome being considered. Although cystic kidneys may occur as an isolated abnormality, a detailed ultrasound examination to identify associated anomalies may inform further prenatal testing.[6] If there are additional anomalies, consanguinity, or a family history of a specific condition, gene panel testing or exome sequencing is sometimes useful because CMA does not detect single-gene (Mendelian) disorders. If exome sequencing is pursued, appropriate pretest and posttest genetic counseling by a provider experienced in the complexities of genomic sequencing is recommended. After appropriate counseling, cell-free DNA screening is an option for patients who decline diagnostic evaluation, particularly if a common aneuploidy is suspected.

Pregnancy and Delivery Management

Follow-up ultrasonography is recommended at 32 weeks of gestation to evaluate the contralateral kidney, fetal growth, and amniotic fluid volume. Fetal echocardiography should be considered given the association of MCDK with other anomalies. Consultation with pediatric nephrology and urology specialists may be considered, particularly when there is concern for an anomaly in the contralateral kidney. Insufficient evidence exists regarding any potential benefit of antenatal testing, and timing and mode of delivery should be based on usual obstetrical indications.

Prognosis

Unilateral isolated MCDK with a normal contralateral kidney is associated with an excellent prognosis. Aslam and colleagues[7] reported outcomes in a cohort of children with isolated unilateral MCDK diagnosed prenatally and found that there was complete involution of MCDKs by 10 years in more than 50% of those affected. There was no hypertension, malignancy, or significant proteinuria noted long term. Surgical removal is indicated when the affected kidney is enlarging or if the mass effect from the MCDK impacts respiration or feeding in the neonate.[8]

In contrast, bilateral MCDK results in severe impairment of renal function that will result in oligohydramnios or anhydramnios. Early-onset anhydramnios and oligohydramnios will result in varying degrees of pulmonary hypoplasia and significant morbidity and mortality. The combination of both renal failure and pulmonary hypoplasia is lethal. Postnatal evaluation by both pediatric nephrology and urology specialists is recommended as ongoing surveillance may be needed.

Summary

MCDK is one of the most commonly seen genitourinary abnormalities that can be noted in midgestation. Prognosis is dependent on whether the lesion is unilateral or bilateral, and the presence of associated abnormalities in the contralateral kidney or extrarenal abnormalities impact outcomes. The presence of extrarenal findings raises concern for a possible genetic etiology. Isolated unilateral MCDK has an excellent prognosis both in utero and postnatally. Most cases resolve over time without the need for surgical intervention.

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Ovarian cysts

Society for Maternal-Fetal Medicine (SMFM); Yvonne Cheng, MD, PhD

Introduction

Ovarian cysts are the most common intra-abdominal mass diagnosed in female fetuses.[1] Although they are most often benign and often undergo spontaneous resolution, fetal ovarian cysts can cause ovarian torsion with the potential risk of loss of an ovary or ovarian function. Because ovarian torsion often occurs before birth, management during pregnancy remains controversial. Some experts advocate in utero decompression via fetal intervention, whereas other experts recommend expectant management.[1–5] Postnatal surgical treatment of ovarian cysts may be indicated, usually with the goal of ovarian preservation.[6]

Definition

An ovarian cyst of <20 mm in diameter is considered physiological and represents a maturing follicle. A cyst of >20 mm in diameter is considered abnormal.[1] With improved ultrasound resolution, the incidence of ovarian cysts detected in a fetus has increased and is currently estimated to be approximately 1 in 2500 live births.[1,7]

Ultrasound Findings

The ovaries arise mainly from the genital ridge and mesonephros. The gubernaculum becomes the ovarian ligament and connects the ovary to the uterine fundus. The ovary descends no further than the level of the uterine fundus. Because the fetal pelvis is shallow and the ovary originates as an abdominal organ, fetal ovarian cysts are most often seen as an abdominal mass.[1]

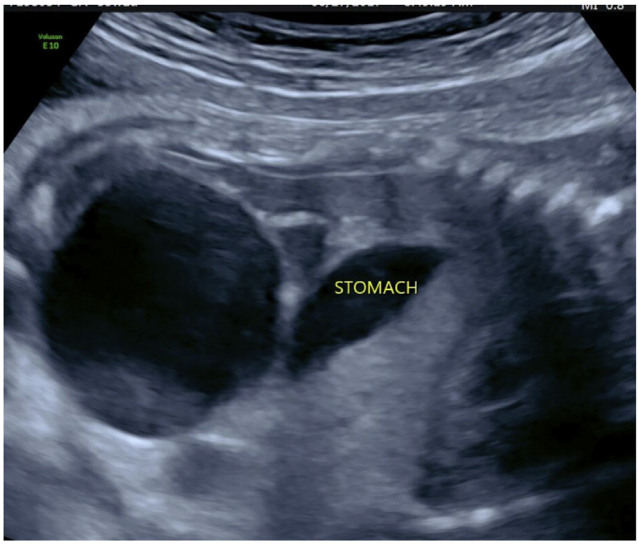
The fetal ovary is normally dormant, but follicular cysts may develop, likely secondary to exposure to fetal pituitary gonadotropins, placental human chorionic gonadotropins, and maternal estrogens.[3] Typically diagnosed in the third trimester of pregnancy, fetal ovarian cysts are often unilateral and simple, although complex cysts can occur.[1,8] A simple cyst may contain a single septation. A small, round anechoic structure within a cyst, or the "daughter cyst sign," is considered pathognomonic for an ovarian cyst (Figures 1 and 2).[9,10]

Complex ovarian cysts can be thick walled, with heterogeneous echogenicity.[1] Postnatal surgical specimens and histologic analysis suggest that complex ovarian cysts may result from primitive gonadal dysgenesis caused by vascular compromise.[11] In addition, complex cysts representing ovarian teratomas or hemorrhagic cysts have been reported.[1,5] Other ultrasonographic findings include polyhydramnios or fetal ascites, which may result from transudate or cyst rupture.[12,13]

Associated Abnormalities

Fetal ovarian cysts are typically rare, incidental findings in the third trimester of pregnancy and are not associated with genetic or structural anomalies. Secondary obstruction of the fetal bowel or kidneys can occur and is thought to result from adhesions in the presence of ovarian torsion, causing ovarian necrosis.[14]

FIGURE 1
Large fetal ovarian cyst inferior to and separate from the fetal stomach



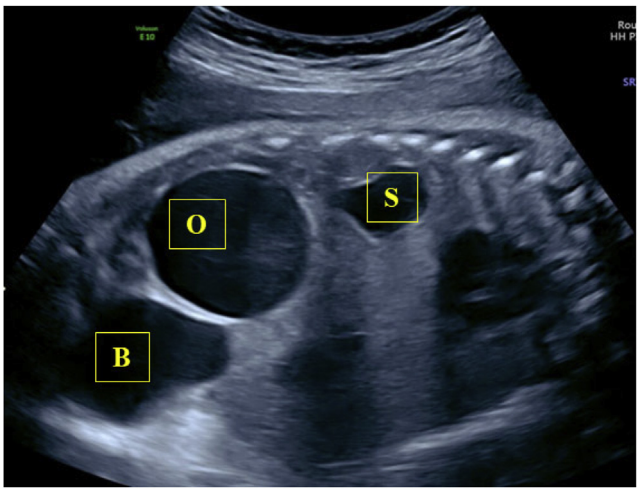
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Differential Diagnosis

The differential diagnosis of a fetal intra-abdominal cyst is broad and can be narrowed by determining the organ of origin and the location and appearance of the surrounding structures. As discussed in other anomalies presented in this Consult, several genitourinary abnormalities can present with a fetal abdominal or pelvic cystic mass. These include simple renal cysts, multicystic dysplastic kidneys, hydronephrosis, urachal cysts, and hydrocolpos.[1,15–21]

Other potential etiologies for an abdominal cyst include gastrointestinal findings, such as enteric duplication cysts, meconium pseudocysts, or choledochal cysts. Enteric duplication cysts are cystic structures consisting of all three bowel wall layers that often share a common wall with normal small bowel. A meconium pseudocyst occurs after bowel perforation and can be seen as an irregular, thick-walled mass that conforms to the peritoneal contours and liver surfaces. In addition, bowel dilation, intraperitoneal calcifications, and ascites may be seen. Choledochal cysts,

FIGURE 2
Fetal ovarian cyst between and separate from the stomach and bladder



B, bladder; *O*, ovarian cyst; *S*, *stomach*.

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which are dilations of the common bile duct, are most commonly unilocular cysts that are located in the right upper quadrant of the fetal abdomen adjacent to the liver. If these become very large, they can extend into the abdomen and displace other organs.[1,22–24]

In addition, lymphatic malformations can present as intra-abdominal masses, although the most common sites are the fetal neck, head, and axilla. Intra-abdominal lymphatic malformations often present as large, thin-walled, multilocular cysts with multiple septations, commonly located in the small bowel mesentery.[1,25] Cystic sacrococcygeal teratoma, which is more likely to occur in female fetuses, can present as a purely cystic, midline pelvic structure; however, it usually contains cystic and solid components and grows in the sacrococcygeal region with external extension.[26]

Genetic Evaluation

Fetal ovarian cysts are typically sporadic findings. If no further abnormalities are noted on ultrasound and the family history is unremarkable, no genetic evaluation beyond standard aneuploidy screening is typically recommended.

Pregnancy and Delivery Management

Once a fetal ovarian cyst is diagnosed, serial ultrasound examinations have been recommended to assess changes in size or complexity of the cyst. The most common complication of an ovarian cyst is torsion, which has been reported in as many as 15% to 34% of fetuses with cysts measuring 30 to 59 mm.[3,14,27,28] Because of the high risk of torsion, prenatal cyst decompression by percutaneous aspiration has been suggested, particularly for cysts of ≥40 mm in diameter or for those with rapid growth, defined as ≥10 mm per week.[14,29,30] However, some experts argue that spontaneous resolution of ovarian cysts, particularly small, simple cysts, can occur both prenatally and postnatally. Thus, conservative management is a reasonable approach.[3,5,11] To date, optimal management of fetal ovarian cysts is uncertain.

Timing of delivery should not be affected by the presence of a fetal ovarian cyst, as the risk, timing, and duration of torsion cannot be predicted, and early delivery would not necessarily preserve ovarian function.[1] The presence of an ovarian cyst should not alter the mode of delivery; cesarean delivery should be performed for the usual obstetrical or medical indications.

Prognosis

Most small, simple fetal ovarian cysts resolve spontaneously after birth because of cessation of exposure to maternal and placental hormones.[1,31] Conservative management of fetal and neonatal ovarian cysts is generally advocated,[6] although there is a risk of loss of ovarian function with ultrasonographic evidence of intracystic hemorrhage.[31,32] Postnatal surgical treatment is often recommended for large cysts. Most operations can be performed in the newborn period laparoscopically with minimal morbidity, with the goal of ovarian preservation. However, one study with long-term follow-up reports that a follicular ovary was detected only in 39% of cases, with a higher likelihood of preservation of function in simple cysts (85%) vs hemorrhagic cysts (16%).[31]

Summary

Fetal ovarian cysts typically present as unilateral intra-abdominal cystic structures in the third trimester of pregnancy, likely because of follicular stimulation from maternal and placental hormones. Spontaneous resolution of fetal and neonatal ovarian cysts can occur. The most common complication is ovarian torsion. The risk of torsion is higher in the prenatal period than after birth. Management includes serial ultrasound examinations to assess changes in size and complexity. Some experts advocate for decompression for cysts of ≥40 mm in diameter or those that demonstrate rapid growth of ≥10 mm per week to minimize the risk of torsion. However, the risks and benefits of prenatal intervention remain controversial. The diagnosis of fetal ovarian cysts should not alter the timing and mode of delivery. If postnatal surgical intervention is indicated, the goal of treatment is ovarian preservation. Simple cysts tend to have more favorable long-term outcomes than complex cysts. ■

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Pelvic kidney

Society for Maternal-Fetal Medicine (SMFM); Jacquelyn K. Chyu, MD

Introduction

A pelvic kidney is the most common type of renal ectopia and occurs in 1 in 700 live births.[1] Pelvic kidneys are usually smaller than expected for gestational age and have an aberrant blood supply. Most of these kidneys are malrotated on their vertical axis, with the hilum facing anteriorly instead of medially toward the spine.

Definition

A pelvic kidney is one located below the aortic bifurcation in the presacral area. The pelvic location results from failure of renal ascent in early embryonic development.

Ultrasound Findings

A pelvic kidney is usually discovered when an empty renal fossa prompts a thorough search for the absent kidney. Demonstration of a kidney within the pelvis can be challenging because of bowel and acoustic shadowing from the iliac wings.[2] Furthermore, the small kidney size, diagonal or horizontal orientation, and anteriorly facing the hilum make the visualization of the kidney more challenging. Frequently, dilation of the renal collecting system and cystic dysplasia further confound the ultrasonographic diagnosis because of distortion of normal anatomy. However, a reniform structure between the iliac vessels on the coronal view or adjacent to the bladder suggests a renal pelvic location, even if the appearance is somewhat atypical (Figure 1). Color Doppler tracing of the blood supply from the distal aorta, aortic bifurcation, or an aberrant pelvic vessel to the renal hilum may provide further supportive evidence for the presence of a pelvic kidney. Not infrequently, a follow-up examination is helpful in visualizing the ectopic kidney, as bowel echogenicity changes with gestational age.

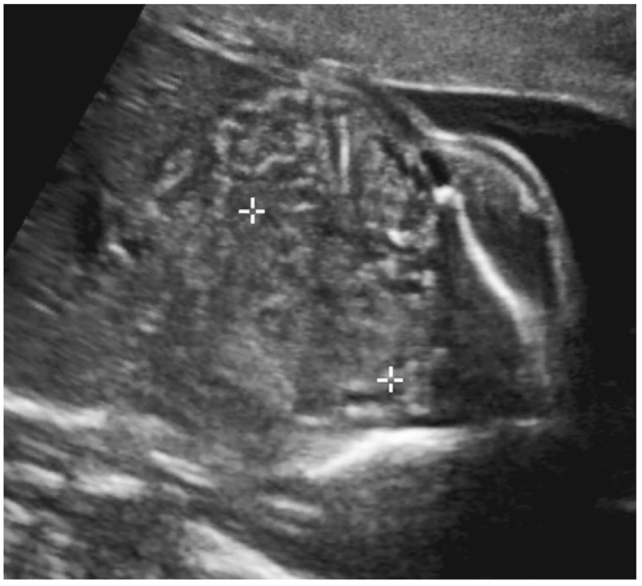
Occasionally, the absence of the kidney from the renal fossa is overlooked because loops of bowel can be mistaken for a kidney. However, close inspection in the transverse and parasagittal views will reveal the absence of the slit-like renal pelvis and the lack of hypoechoic medullary pyramids converging toward the hilum. Other clues of an empty renal fossa are loss of the boomerang shape of the adrenal gland, which may appear flattened, elongated, and "lying down" in the parasagittal plane (Figure 2), and the inability to demonstrate a renal artery in the coronal plane using color Doppler.[3,4]

The contralateral kidney, bladder, and genitalia should be carefully assessed as well. Rarely, both kidneys are located in the pelvis.

Associated Abnormalities

Nearly 50% of pelvic kidneys are affected by hypoplasia, dysplasia, or hydronephrosis.[5] However, these conditions rarely correlate with an adverse prognosis for the fetus.[5]

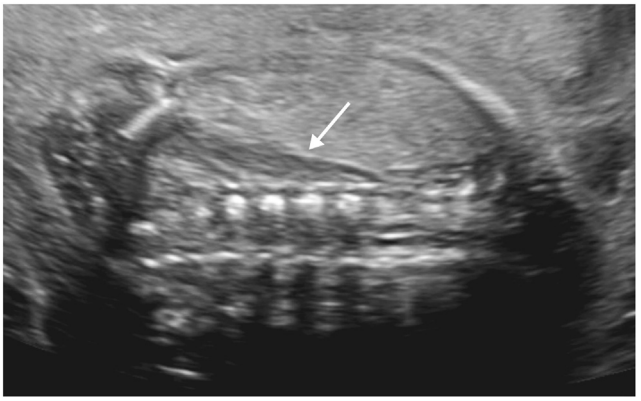
FIGURE 1
Pelvic kidney between iliac bones (calipers)



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Although vesicoureteral reflux is identified in 80% of contralateral kidneys during childhood, it usually resolves over time with conservative management.[5,6] Dysplasia affects the contralateral kidney in 4% of cases.[7] Contralateral hydronephrosis requiring surgical intervention, such as that

FIGURE 2
Empty renal fossa with a "lying-down" adrenal sign (*arrow*)



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caused by ureteropelvic junction (UPJ) obstruction, is uncommon, occurring in 5% of cases.[6] Coexisting single umbilical artery and cardiac and intracranial anomalies have been reported.[5] Hypospadias and cryptorchidism in males and genital anomalies affecting females seem to be slightly increased in pediatric series.[5–7]

Differential Diagnosis

Other pelvic masses, including teratoma, neuroblastoma, supernumerary pelvic kidney, and ovarian or mesenteric cysts, should be considered. An empty renal fossa is associated with renal agenesis in 47% of cases and an ectopic kidney in 42% of cases, usually in the pelvis.[8] In some cases, the missing kidney may be fused to the contralateral kidney or in an unusual location if other anomalies (eg, diaphragmatic hernia) are present.[8]

Genetic Evaluation

Chromosomal anomalies are rare with an isolated pelvic kidney. If no further abnormalities are noted on ultrasound and the family history is unremarkable, no genetic evaluation beyond standard aneuploidy screening is typically recommended. If additional nongenitourinary anomalies are present, amniocentesis and chromosomal microarray analysis (CMA) should be offered. Syndromic conditions, such as VACTERL (**v**ertebral defects, **a**nal atresia, **c**ardiac defects, **t**racheo**e**sophageal fistula, **r**enal anomalies, and **l**imb abnormalities) and CHARGE (**c**oloboma, **h**eart defects, **a**tresia choanae [choanal atresia], **g**rowth restriction, **g**enital anomalies, and **e**ar anomalies), should be considered in the presence of multiple anomalies.[9,10]

If there are additional anomalies, consanguinity, or a family history of a specific condition, gene panel testing or exome sequencing is sometimes useful because CMA does not detect single-gene (Mendelian) disorders. If exome sequencing is pursued, appropriate pretest and posttest genetic counseling by a provider experienced in the complexities of genomic sequencing is recommended. After appropriate counseling, cell-free DNA screening is an option for patients who decline diagnostic evaluation, particularly if a common aneuploidy is suspected.

Pregnancy and Delivery Management

A detailed anatomic survey focusing on the genitourinary and cardiovascular systems should be performed to exclude associated anomalies. A fetal echocardiogram should be considered if a cardiac anomaly is suspected. A follow-up ultrasound examination in the third trimester of pregnancy to assess for late development of hydronephrosis is warranted, and consultation with pediatric urology or nephrology specialists can be helpful to review childhood follow-up. The finding of a pelvic kidney in the absence of other anomalies should not affect timing and mode of delivery, which should be based on the usual obstetrical indications.

Prognosis

The prognosis of a pelvic kidney is favorable in most cases, although renal function in the affected kidney is usually reduced.[11] Generally, vesicoureteral reflux involving either the pelvic or contralateral kidney improves with conservative management.[5] Surgical intervention for hydronephrosis in the contralateral kidney (eg, UPJ obstruction) is needed in 5% of cases.[6] Although there is no evidence for an increased incidence of hypertension, follow-up until young adulthood is recommended.[11] An increased incidence of urinary tract infections and nephrolithiasis is reported in adults.[12]

Summary

The diagnosis of pelvic kidney relies on a high index of suspicion on recognizing an empty renal fossa. Direct visualization of a kidney within the pelvis requires focused gray scale and color Doppler examination due to challenges posed by the fetal bowel and pelvic bones and, occasionally, atypical anatomic appearance. Amniocentesis and CMA are warranted only when additional, extragenitourinary malformations are present. Obstetrical management should not be altered, and the prognosis is generally good. ■

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Renal agenesis

Society for Maternal-Fetal Medicine (SMFM); Angie Jelin, MD

Introduction

Renal agenesis occurs when the ureteric bud fails to fuse with the metanephric blastema during embryogenesis, resulting in the absence of the nephron and often the ureter. Unilateral renal agenesis has an incidence of 1 in 1000 live births, whereas bilateral renal agenesis is less common, occurring in 1 in 3000 to 4000 pregnancies. Unilateral renal agenesis generally carries a very favorable prognosis, whereas bilateral renal agenesis is associated with a high rate of perinatal morbidity and mortality because of the absence of amniotic fluid leading to lethal pulmonary hypoplasia.

Definition

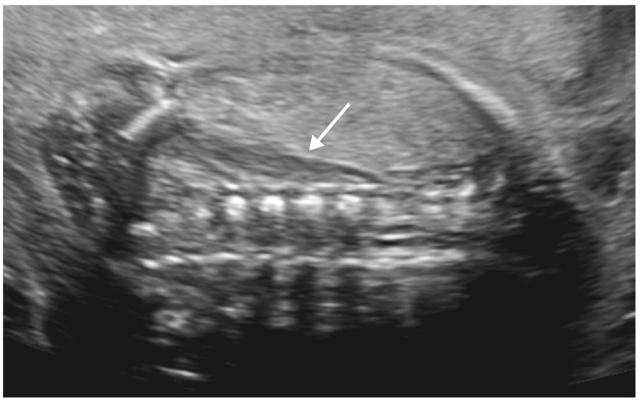
Renal agenesis is the absence of renal tissue due to a defect in early embryologic development. Both unilateral and bilateral renal agenesis are commonly associated with other congenital anomalies.

Ultrasound Findings

Although fetal kidneys can be visualized by ultrasound, inferior to the adrenals, by approximately 12 weeks of gestation,[1] this requires a detailed examination. The diagnosis of renal agenesis is typically made during an anatomy ultrasound examination in midgestation. The presence of amniotic fluid in the first trimester of pregnancy is not predictive of bilateral renal agenesis because early in gestation, amniotic fluid is placental in origin, and relatively normal amniotic volume can be present until approximately 16 to 18 weeks of gestation even in the absence of kidneys or renal function.

The initial sign of renal agenesis is an empty renal fossa. The absence of kidneys should be documented in the axial, sagittal, and coronal planes at the level of the spine below the stomach.[2] If unilateral renal agenesis is suspected, the possibility of a pelvic or ectopic kidney should be assessed. Hypertrophy of the visualized kidney is suggestive of unilateral renal agenesis.[3] The presence of bilateral renal agenesis is suspected in the setting of additional findings of a nonvisualized bladder and anhydramnios.[2] The lack of amniotic fluid impedes ultrasonographic imaging; thus, diagnostic amnioinfusion has been reported as one technique to confirm the diagnosis. Additional ultrasound findings of renal agenesis include the "lying-down" adrenal sign (Figure 1), in which the adrenals appear large and flat instead of maintaining the normal "Y" configuration. The echogenic adrenal medulla between the cortex creates a layered appearance in the adrenal glands; they lack corticomedullary pyramidal differentiation and should not be confused with renal tissue.

FIGURE 1
Empty renal fossa with a "lying-down" adrenal sign (*arrow*)



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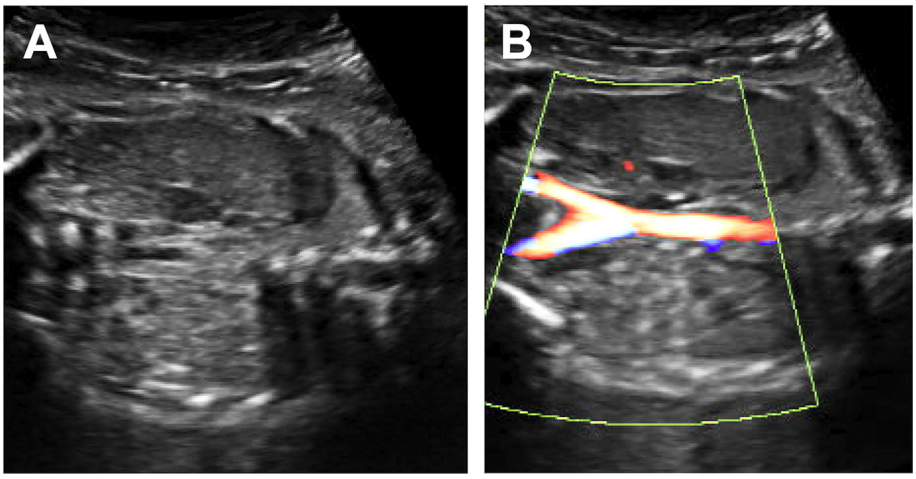
Color Doppler should be utilized to visualize the renal arteries originating from the aorta (Figure 2). The absence of renal artery flow at a 90° angle in the coronal plane[1] is confirmative and should not be confused with flow through the lumbar and adrenal arteries. Confirmation of the absence of a bladder is assisted by evaluating flow through the umbilical arteries. Parental renal ultrasonographic imaging is recommended because of the potential for an inherited etiology.

Associated Abnormalities

Approximately 30% of cases of renal agenesis are associated with a genetic syndrome or other anomalies.[4] A careful assessment should include a full evaluation of the urogenital system, including the genitalia. Bilateral renal agenesis with anhydramnios results in Potter syndrome. Features include a flat face with micrognathia, beaked nose, upslanted palpebral fissures, low-set ears, and joint contractures, such as talipes equinovarus. These deformations are secondary to anhydramnios and, in the absence of additional organ system anomalies, consistent with isolated bilateral renal agenesis. Clinically significant variants in multiple genes, including *GREB1L*,[5] *GFRA1*,[6] *ITA8*, and *FGF20*, have been identified in cases of isolated renal agenesis and present with variable expressivity and penetrance.

Ultrasound findings that demonstrate cerebral, cardiovascular, gastrointestinal, or muscular systems can aid in the diagnosis of a chromosomal or syndromic etiology.

FIGURE 2
Color Doppler imaging



A, Coronal view of the empty renal fossa. **B,** Absence of the renal artery flow on coronal color Doppler imaging in a case of bilateral renal agenesis.
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Single-gene disorders include acro-renal-ocular syndrome (*SALL4*), branchio-oto-renal syndrome (*EYA1*), Pallister-Hall syndrome (*GLI3*), and Fraser syndrome (*FRAS1*, *FEB2*, and *GRIP1*).[7,8] A molecular genetic diagnosis is particularly useful for prognostic and recurrence risk counseling. Moreover, renal agenesis is associated with maternal environmental factors, including diabetes mellitus, smoking, and alcohol consumption. Diabetes mellitus as the etiology should be considered if identified anomalies suggest VACTERL (**v**ertebral defects, **a**nal atresia, **c**ardiac defects, **t**racheo**e**sophageal fistula, **r**enal anomalies, and **li**mb abnormalities).

Differential Diagnosis

When renal agenesis is suspected, the presence of other normal or abnormal ectopic or pelvic kidneys should be excluded. It is important to rule out the possibility of preterm premature rupture of membranes and assess for structural anomalies of other organ systems. Fetal growth restriction is another etiology of early-onset anhydramnios, but if that is the primary reason for anhydramnios, kidneys are present. In the setting of unilateral renal agenesis, a second nonfunctioning kidney, such as a multicystic dysplastic kidney, could also result in anhydramnios and the misdiagnosis of bilateral renal agenesis.

Genetic Evaluation

Diagnostic testing with amniocentesis or chorionic villus sampling and chromosomal microarray analysis (CMA) should be offered when bilateral renal agenesis (or unilateral renal agenesis in the setting of other anomalies) is detected. In the setting of anhydramnios, in which amniocentesis is not feasible, testing can be done by placental biopsy. If ultrasound findings or screening test results are suggestive of a common aneuploidy, it is reasonable to initially perform karyotype analysis or fluorescence in situ hybridization, with reflex to CMA if these test results are normal. If there are additional anomalies, consanguinity, or a family history of a specific condition, gene panel testing or exome sequencing is sometimes useful because CMA does not detect single-gene (Mendelian) disorders. If exome sequencing is pursued, appropriate pretest and posttest genetic counseling by a provider experienced in the complexities of genomic sequencing is recommended. After appropriate counseling, cell-free DNA screening is an option for patients who decline diagnostic evaluation, particularly if a common aneuploidy is suspected and anhydramnios is present.

Pregnancy and Delivery Management

Isolated unilateral renal agenesis does not require any alteration in obstetrical management aside from appropriate genetic counseling. If associated anomalies are suggestive of a syndromic etiology or in the setting of bilateral renal agenesis, pregnancy termination should be offered. Shared patient decision-making requires a thorough evaluation and multidisciplinary counseling regarding prognosis.[9] Fetal therapy with serial amnioinfusions has been reported with a goal of allowing survival by reducing the risk of in utero fetal demise (IUFD) and neonatal pulmonary hypoplasia.[10,11] However, the benefit of this procedure in improving survival or morbidity is unknown, and this intervention should be considered investigational and offered only under research protocols, such as the prospective *Eunice Kennedy Shriver* National

Institute of Child Health and Human Development-funded Renal Anhydramnios Fetal Therapy (RAFT) trial. This study is examining the natural history of bilateral renal agenesis and the safety, feasibility, and efficacy of serial amnioinfusions.[12] It is important to recognize that even if serial amnioinfusion allows improved survival because of better respiratory function, survivors will need to contend with the considerable challenges of absent renal function.

Prognosis

The prognosis for unilateral renal agenesis is favorable in the setting of normal amniotic fluid; however, the long-term prognosis of a hypertrophied unilateral kidney can include adult-onset renal dysfunction and hypertension. Bilateral renal agenesis is considered a life-limiting condition because of anhydramnios associated with IUFD and nonsurvivable neonatal respiratory failure. The RAFT trial may elucidate whether serial amnioinfusions are efficacious in improving outcomes.

Summary

Fetal renal agenesis is characterized by unilateral or bilateral absence of renal tissue. It is commonly associated with additional congenital anomalies as part of a chromosomal or syndromic condition. Targeted ultrasound examinations are recommended to improve counseling regarding an inherited condition. Appropriate genetic and prognostic counseling are recommended, including the likely lethality of bilateral renal agenesis.

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Renal pelvic dilation

Society for Maternal-Fetal Medicine (SMFM); Lisa C. Zuckerwise, MD

Introduction

Renal pelvic dilation refers to excessive dilation of the fetal intrarenal collecting system. Although a different terminology is used, pelviectasis or pyelectasis most often refers to mild dilation, and hydronephrosis is reserved for severe cases that are more likely to be clinically significant. Normal measurements of the anteroposterior renal pelvis diameter are <4 mm at 16 to 27 weeks of gestation and <7 mm at ≥28 weeks of gestation.[1,2] Although fetal urinary tract dilation is often mild and benign, occurring in 1% to 5% of all pregnancies, it also can be associated with genetic and structural disorders. Outcomes range from normal to significant structural anomalies requiring surgical repair.[3–5]

Definition

Fetal urinary tract dilation is defined as an anteroposterior renal pelvis diameter of ≥4 mm at 16 to 27 weeks of gestation and ≥7 mm at ≥28 weeks of gestation. The severity of renal dilation can be categorized on the basis of gestational age-specific criteria. At 16 to 27 weeks of gestation, dilation is categorized as mild (4 to <7 mm), moderate (7 to ≤10 mm), and severe (>10 mm). Beyond 28 weeks of gestation, dilation is categorized as mild (7 to < 9 mm), moderate (9 to ≤15 mm), and severe (>15 mm) (Table).[6]

Ultrasound Findings

The anterior-posterior (AP) renal pelvis diameter is measured in a transverse plane, with fetal spine ideally positioned at the 12-o'clock position (Figure 1). Proper caliper placement occurs at the interior margin of the renal parenchyma, measuring across the widest portion of the fluid-containing renal pelvis (Figure 2). Renal pelvic dilation can also be assessed in a coronal plane, which aids in the identification of caliectasis (Figure 3). Additional ultrasound findings that contribute to prognosis and should be reported with a finding of renal pelvic dilation include those indicative of associated congenital anomaly of the kidney and urinary tract (CAKUT): peripheral calyceal dilation, abnormal appearance of the renal parenchyma (thickness, echogenicity, and the presence of cysts), dilated ureters, abnormal bladder appearance, and oligohydramnios.[7] These findings have been described and attempts made to standardize prenatal and postnatal classifications of renal tract dilation based on these criteria.[8]

TABLE
Classification of fetal urinary tract dilation[6]

Classification	AP diameter at 16–27 wk (mm)	AP diameter at >28 wk (mm)
Mild	4 to <7	7 to <9
Moderate	7 to ≤10	9 to ≤15
Severe	>10	>15

AP, anterior-posterior.

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FIGURE 1
Assessment of anterior-posterior renal pelvis diameter



Axial image of the fetal kidneys with the fetal spine at the 12 o'clock position.

LT, left.

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Associated Abnormalities

Although mild fetal pelviectasis is most often incidental and benign, it is a common and nonspecific finding that can also be associated with genetic and structural abnormalities. Isolated mild urinary tract dilation in the second trimester of pregnancy has been reported to carry a likelihood ratio of 1.6 to 2.78 for trisomy 21, although the strength of this association is debated.[9–12] Progressive, moderate, or severe urinary tract dilation is more commonly associated with vesicoureteral reflux or CAKUT, such as ureteropelvic junction (UPJ) obstruction, duplicated collecting system

FIGURE 2
Measurement of the renal pelvis diameter



Caliper placement occurs at the interior margin of the renal parenchyma, measuring across the widest portion of the fluid-containing renal pelvis.

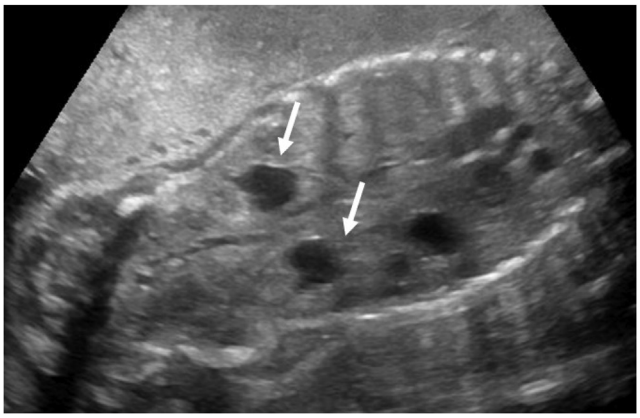
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with obstruction, and bladder outlet obstruction (most commonly due to posterior urethral valves).[5,13] Fetuses with complete bladder outlet obstruction or significantly impaired fetal renal function will have associated oligohydramnios or anhydramnios.

Differential Diagnosis

The differential diagnosis of fetal pelvic dilation or hydronephrosis includes a normal variant or physiological and transient dilation as well as pathologic causes, such as vesicoureteral reflux and obstructive uropathy. Obstructive causes include those at the level of the ureter, such as UPJ obstruction, and obstruction of a prolapsed ureterocele in a duplicated collecting system, which occurs more often in female fetuses. Obstruction at the bladder outlet is most commonly caused by posterior urethral valves, which occur in males and cause a keyhole-shaped bladder. Rarer causes of fetal urinary tract dilation include urethral atresia and megacystis-microcolon-intestinal hypoperistalsis syndrome, both of which have a markedly enlarged bladder as a dominant ultrasound finding.

FIGURE 3
Coronal view of the fetal kidneys



The *arrows* indicate renal pelvic dilation without caliectasis.

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Genetic Evaluation

Fetal hydronephrosis is most commonly an isolated finding. If no further abnormalities are noted on ultrasound and the family history is unremarkable, it is reasonable to perform no genetic evaluation beyond standard aneuploidy screening. Because of the association with Down syndrome, counseling should occur in the context of an individual's a priori risk of trisomy 21 based on maternal age, previous screening results, and additional ultrasound findings. If there are additional anomalies, consanguinity, or a family history suggestive of a specific condition, diagnostic testing with chromosomal microarray analysis (CMA) should be offered; gene panel testing or exome sequencing is sometimes useful in cases of significant hydronephrosis if CMA is normal, as CMA does not detect single-gene disorders. If exome sequencing is pursued, appropriate pretest and posttest genetic counseling by a provider experienced in the complexities of genomic sequencing are recommended.

Pregnancy and Delivery Management

Given the high likelihood of resolution when mild urinary tract dilation is noted in the second trimester of pregnancy, a single follow-up ultrasound examination in the third trimester of

pregnancy at approximately 32 weeks of gestation is recommended. Normalization of the AP renal pelvic diameter suggests no further need for follow-up beyond routine obstetrical care. Fetuses with moderate or severe urinary tract dilation, and those with additional findings to suggest CAKUT, warrant closer surveillance with follow-up ultrasound examinations at 4- to 6-week intervals or sooner if clinically indicated. These patients may also benefit from prenatal consultation with a pediatric urology or nephrology specialist to formulate a plan for postnatal evaluation. Timing of delivery should not be affected by the presence of hydronephrosis, as preterm delivery has not been demonstrated to improve outcomes. The presence of hydronephrosis should not alter the mode of delivery, which should be based on the usual obstetrical or medical indications.

Prognosis

For fetuses with mild antenatal hydronephrosis, the risk of postnatal pathology is reported to be 11% to 15%, increasing to 27% to 45% and 53% to 88% for moderate and severe hydronephrosis, respectively.[5,14] Although the incidence of postnatal vesicoureteral reflux appears constant among fetuses with any degree of hydronephrosis, the likelihood of postnatal obstructive uropathy or abnormal renal function and the need for subsequent surgery increases with the degree of dilation and with the presence of additional findings suggestive of CAKUT.[15]

Summary

Fetal renal pelvic dilation is a relatively common finding on prenatal ultrasound. Enlargement of the AP renal pelvis diameter beyond the gestational age-specific norms of <4 mm at 16 to 27 weeks of gestation and <7 mm at ≥ 28 weeks of gestation indicates a need for a thorough evaluation of the fetal genitourinary tract for evidence of CAKUT and review of screening results and genetic counseling related to an increased risk of trisomy 21. Follow-up ultrasound examination should be performed to assess for persistent or progressive renal dilation. Isolated mild pelviectasis in the second trimester of pregnancy warrants a single follow-up ultrasound examination at approximately 32 weeks of gestation. These patients can be reassured that the prognosis is favorable, with 85% to 90% having normal postnatal outcomes. In cases of moderate or severe hydronephrosis, or when there are associated findings suggestive of CAKUT, serial assessments are warranted, and pediatric urology or nephrology consultation should be offered. Obstetrical management and timing and mode of delivery should be based on standard obstetrical indications.

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Urinoma

Society for Maternal-Fetal Medicine (SMFM); Katherine Connolly, MD; and Mary E. Norton, MD

Introduction

Severe urinary tract obstruction, particularly bladder outlet obstruction, can lead to extravasation of urine and development of urinary ascites or a urinoma. These occur as complications of urinary tract obstruction either through rupture of the bladder or a calyceal fornix or tear in the renal parenchyma, which can be identified in the second or third trimester of labor. In one study, urinomas or urinary ascites were reported in 20% of cases of posterior urethral valves.[1]

Definition

Urinomas are encapsulated collections of urine around the kidney.[2] Some speculate that the kidney rupture that gives rise to a urinoma serves as a protective mechanism for the renal parenchyma in the setting of increased pressure due to obstruction.[1] Others interpret the presence of a urinoma as evidence of an already compromised renal parenchyma.[3]

Ultrasound Findings

On ultrasound, a urinoma appears as an ellipsoid or crescent-shaped anechoic cystic structure that is adjacent to the kidney (Figure). Depending on its location in the perinephric space, a large urinoma can displace the kidney anteriorly or posteriorly.[2] In addition to the presence of a fluid collection, an underlying urinary tract obstruction should also be observed, such as bladder outlet obstruction or bilateral hydroureteronephrosis. There may be evidence of a decompressed urinary tract obstruction because of rupture, with findings of a thickened bladder wall or renal dysplasia.

Associated Anomalies

Urinoma is strongly associated with underlying urinary tract obstruction, most commonly because of posterior urethral valves or ureteropelvic junction (UPJ) obstruction.[1,3]

Differential Diagnosis

The differential diagnoses for a cystic structure adjacent to the kidney include lymphangioma, hemorrhagic neuroblastoma, mesenteric cyst, enteric duplication cyst, multicystic kidney disease, polycystic kidney disease, cystic renal tumor, and ureteric duplication. It is important to identify other intra-abdominal structures, such as the stomach and bladder, to determine the exact location of the fluid collection and accurately classify it. In most cases where the cystic structure is adjacent to the lumbar spine, the etiology is genitourinary.[3] Small urinomas may be mistaken for a dilated calyx in the presence of hydronephrosis; identifying a mass effect on the renal cortex can aid in distinguishing these two diagnoses.[2,3] In addition, intra-abdominal ascites can mimic a urinoma; a thorough evaluation of fluid in other compartments is warranted. There have also been cases of urinoma formation after trauma sustained during amniocentesis.[4] Relevant patient history and clinical setting should be reviewed in conjunction with ultrasound findings.

FIGURE
Urinoma with bilateral renal pelvic dilation



Urinoma seen as a cystic structure adjacent to (but not communicating with) the unilateral kidney with bilateral renal pelvic dilation.

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Genetic Evaluation

The suggested genetic evaluation is based on the recommended evaluation for the underlying urinary tract anomaly, which is usually diagnostic testing (amniocentesis or chorionic villus sampling) with chromosomal microarray analysis (CMA). If ultrasound findings or screening test results are suggestive of a common aneuploidy, it is reasonable to initially perform karyotype analysis or fluorescence in situ hybridization, with reflex to CMA if these test results are normal. If there are additional anomalies, consanguinity, or a family history of a specific condition, gene panel testing or exome sequencing is sometimes useful because CMA does not detect single-gene (Mendelian) disorders. If exome sequencing is pursued, appropriate pretest and posttest genetic counseling by a provider experienced in the complexities of genomic sequencing is recommended. After appropriate counseling, cell-free DNA screening is an

option for patients who decline diagnostic evaluation particularly if a common aneuploidy is suspected.

Pregnancy and Delivery Management

A detailed ultrasound examination that focuses on identifying an underlying urinary tract obstruction should be performed when a urinoma is diagnosed. Serial ultrasound surveillance to monitor the size, evidence of progression of the underlying obstruction, and amount of amniotic fluid should be initiated. Magnetic resonance imaging may be helpful in cases where it is difficult to assess the relationship between the kidney and the mass and has been reported to aid in evaluating the renal parenchyma.[3]

Attempts at puncture and drainage have not been shown to improve renal function.[5] However, drainage has been successful in preventing abdominal dystocia at the time of vaginal delivery.[3]

Prognosis

Prognosis depends on the cause of obstruction and the gestational age at presentation. A review of case reports suggests a worse prognosis with upper urinary tract obstruction compared with lower urinary tract obstruction. Preserved renal function was seen in 75% of cases associated with posterior urethral valves, but only in 7% of cases due to UPJ obstruction.[6] Overall, the presence of a urinoma is associated with irreversible ipsilateral renal dysfunction in 70% to 80% of cases.[5,6] Associated anhydramnios portends an extremely poor prognosis, especially if seen in the second trimester of pregnancy.

Spontaneous resolution is a possible, although uncommon, outcome. In a series of 25 cases, 2 cases associated with UPJ obstruction spontaneously resolved.[3] A review of the literature showed no improvement in the renal prognosis when the urinoma remained stable in size, resolved, or drained.[5] The lack of improvement may be because the formation of a urinoma requires a functioning kidney, and thus, its resolution or lack of growth may indicate renal compromise. Other cases of spontaneous resolution in the setting of posterior urethral valves have been reported with normal postnatal kidney function.[7]

Summary

Although a rare prenatal finding, the diagnosis of urinoma is an important one, as it indicates likely urinary tract obstruction with irreversible ipsilateral renal damage in most cases. This finding should prompt a thorough diagnostic evaluation, surveillance, and postnatal follow-up.

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All authors and committee members have filed a conflict of interest disclosure delineating personal, professional, business, or other relevant financial or nonfinancial interests in relation to this publication. Any substantial conflicts have been addressed through a process approved by the Society for Maternal-Fetal Medicine (SMFM) Board of Directors. Moreover, the SMFM has neither solicited nor accepted any commercial involvement in the specific content development of this publication.

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