Upper Urinary Tract Anomalies and Perinatal Renal Tumors

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KEYWORDS

• Renal agenesis • Ectopia and fusion anomalies • Renal hypoplasia and dysplasia
• Congenital mesoblastic nephroma • Wilms' tumor

KEY POINTS

- Unilateral renal agenesis occurs in 1100 births, is associated with Müllerian duct (MD) anomalies in 25% to 50% of females with unilateral renal agenesis, and requires lifelong follow-up for hypertension and microalbuminuria.
- Renal ectopia, although usually asymptomatic, may have associated ureteropelvic or ureterovesical junction obstruction, reflux, or genital anomalies.
- Crossed renal ectopia occurs when a kidney is located on the side opposite from that in which its ureter inserts into the bladder. Ninety percent are fused, with the lower pole of the normal kidney fusing with the upper pole of the ectopic kidney, and they may have associated MD anomalies.
- Horseshoe kidneys occur in about 1 in 400 persons, are malrotated with calyces pointing posteriorly, have a variable blood supply, and are found in association with other congenital anomalies, including ureteropelvic junction obstruction and reflux.
- Congenital mesoblastic nephroma, the most common renal tumor in the first 3 months of life, can be diagnosed prenatally or may present as an abdominal mass, and is curable in most cases with only primary surgical excision.

RENAL EMBRYOLOGY

The mesonephric duct or Wolffian duct (WD) elongates caudally and fuses with the anterior cloaca. The metanephric blastema, which becomes the definitive kidney, differentiates from a region of intermediate mesoderm termed the metanephric mesenchyme. Signaling from the metanephric blastema leads to ureteral bud branching from the WD at the 28th day of gestation and reciprocal induction proceeds between the ureteral bud and metanephric blastema. Invasion of the metanephric blastema by

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the ureteral bud results in repetitive branching that forms the collecting duct system. In turn, the metanephros is complete with the induction of nephron differentiation in the surrounding mesenchyme.[1,2] When the nephric ridge fails to form, or there is failure of ureteral bud formation, a kidney will not develop, resulting in renal agenesis.

Specific anomalies of the Müllerian ducts (MD) are often seen in association with renal agenesis. This is explained by a 3-phase model of MD development: (1) Coelomic epithelium in the cervical region of the intermediate mesoderm, which are specified to become MD cells, (2) a WD-independent phase with these cells invaginating to become the MDs, which is complete when the MD extends caudally and contacts the WD, and (3) a WD-dependent phase involving elongation of the MDs posteriorly until they are joined at the urogenital sinus.[3–8] This final phase requires the MD epithelium at its posterior end to be in close physical contact with the WD epithelium, while the MDs are separated from the coelomic epithelium by only a basement membrane.[4] Because normal MD formation depends in part on a normal WD, abnormalities of the WDs result in an high incidence of anomalies of the MD structures including the fallopian tube, uterine horn and body, and proximal vagina.[9]

UNILATERAL RENAL AGENESIS

Incidence, Diagnosis, and Associated Anomalies

Unilateral renal agenesis (URA) occurs once in 1100 births in autopsy series.[10] URA may be an incidental finding on a sonogram performed after an abnormal physical examination of the external genitalia or female pelvis and/or after an abnormal radiographic evaluation of the female or male pelvis (**Fig. 1**). URA is detected with increasing frequency. Approximately 5% of URA may have been a dysplastic or multicystic dysplastic kidney (MCDK; incidence 1 in 4300) that involuted during gestation.[11–13] Renal aplasia is found in 1 in ~1300 births and is considered the most common cause of congenital solitary kidney. A renal unit with renal aplasia has rudimentary parenchyma and ureter (absent in 60%) owing to early regression of the ureteral bud, altered metanephric differentiation, or defects in the branching ureteral bud and/or the metanephric blastema, affecting reciprocal communication.[14]

URA is observed in X-linked and sporadic cases of Kallmann syndrome, Turner syndrome, Poland syndrome, Frazer syndrome, brachio-oto-renal syndrome, and maternal diabetes.[15–19] URA is seen in about 30% of neonates with VACTERL association (*V*ertebral, imperforate *A*nus, *C*ardiac, *T*racheo-*E*sophageal atresia, *R*enal, and *L*imb anomalies).[20] Associated anomalies of other organ systems involve primarily the cardiovascular, gastrointestinal, hematologic, neurologic, and musculoskeletal systems.[21] Because the ipsilateral gonad is derived from nearby mesenchyme and is usually present in its normal position, the metanephric blastema is not thought to be responsible for URA in most cases.[14] In addition, the adrenal gland is almost always found in its normal position owing to its dual origin from primitive mesoderm and ectodermal neural crests cells, but it may seem to be flattened ("lying down" sign) on ultrasonography.[22]

The contralateral ureter may be abnormal, with associated vesicoureteral reflux in 30% and obstruction at the ureteropelvic or ureterovesical junction in 11% and 7%, respectively.[23] Reproductive tract malformations in females are found in 25% to 50% despite URA being more common in males (1.8:1) in whom reproductive tract anomalies occur in only 10% to 15%.[24] About 40% of females with genital anomalies have URA. As noted, these anomalies involve the MD owing to its close developmental relationship with the WD. In males, an anomalous WD affects its derivatives, including the vas deferens, seminal vesicles, ampulla, ejaculatory duct, and the body and tail of