

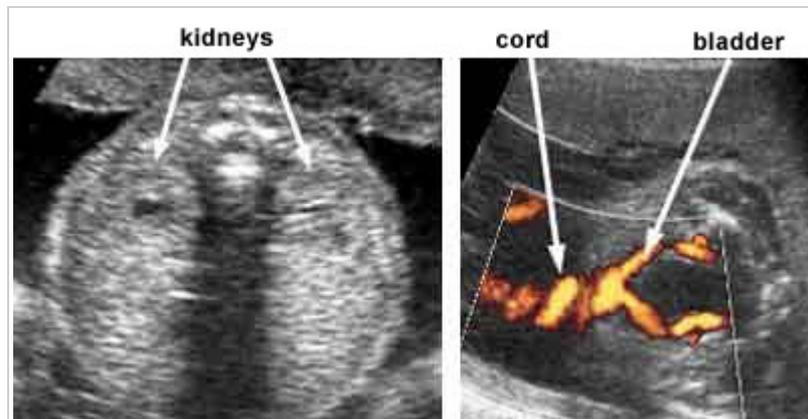
Chapter 8

Kidneys and urinary tract

NORMAL SONOGRAPHIC ANATOMY

The kidneys and adrenals, located below the level of the stomach, on either side and anterior to the spine, are visible by ultrasonography from as early as 9 weeks of gestation and in all cases from 12 weeks. The renal echogenicity is high at 9 weeks but decreases with gestation; the adrenals appear as translucent structures with an echodense cortex.

Longitudinal and transverse sections of the abdomen can be used to study the kidneys. In a longitudinal scan, kidneys appear as elliptical areas, while on transverse scan they appear as roundish structures at both sides of the spine. The kidneys appear slightly hypoechoic, compared to the liver and bowel loops. At 20 weeks, the kidneys show a hyperechoic capsule and the cortical area is slightly more echogenic than the medulla. With progressing gestation, fat tissue accumulates around the kidneys, enhancing the borders of the kidneys in contrast with the other splanchnic organs. At 26–28 weeks, renal pyramids can be detected, and the arcuate arteries can be seen pulsating in their proximity. Both the renal length and circumference increase with gestation, but the ratio of renal to abdominal circumference remains approximately 30% throughout pregnancy. The anteroposterior diameter of the renal pelvis should be < 5 mm at 15–19 weeks, < 6 mm at 20–29 weeks and < 8 mm at 30–40 weeks. The normal ureters are rarely seen in the absence of distal obstruction or reflux. The fetal bladder can be visualized from the first trimester (in about 80% of fetuses at 11 weeks and more than 90% by 13 weeks); changes in volume over time help to differentiate it from other cystic pelvic structures.



RENAL AGENESIS

Renal agenesis is the consequence of failure of differentiation of the metanephric blastema during the 25–28th day of development and both ureters and kidneys and renal arteries are absent.

Prevalence

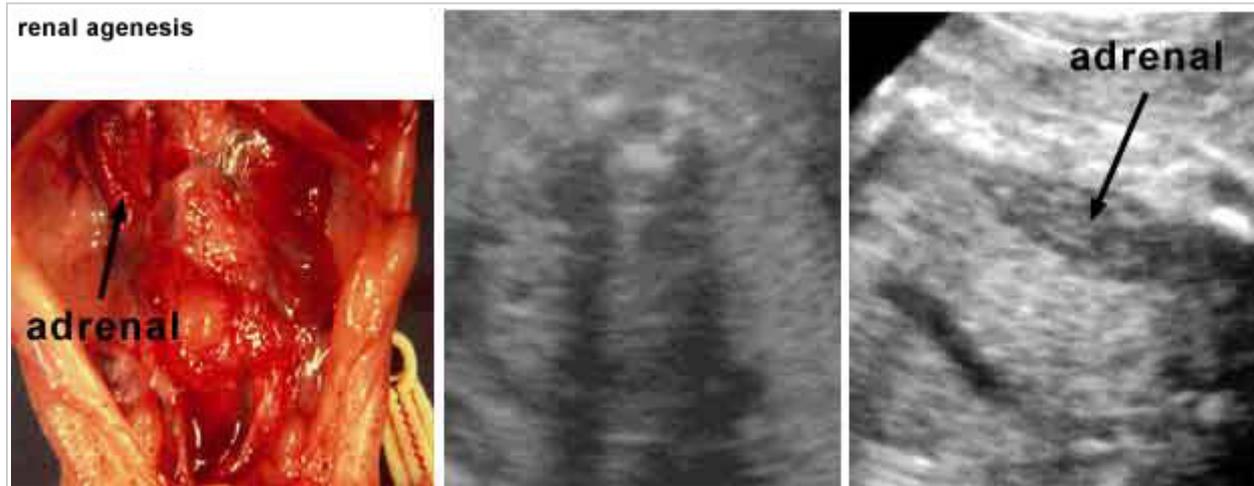
Bilateral renal agenesis is found in 1 per 5000 births, while unilateral disease is found in 1 per 2000 births.

Etiology

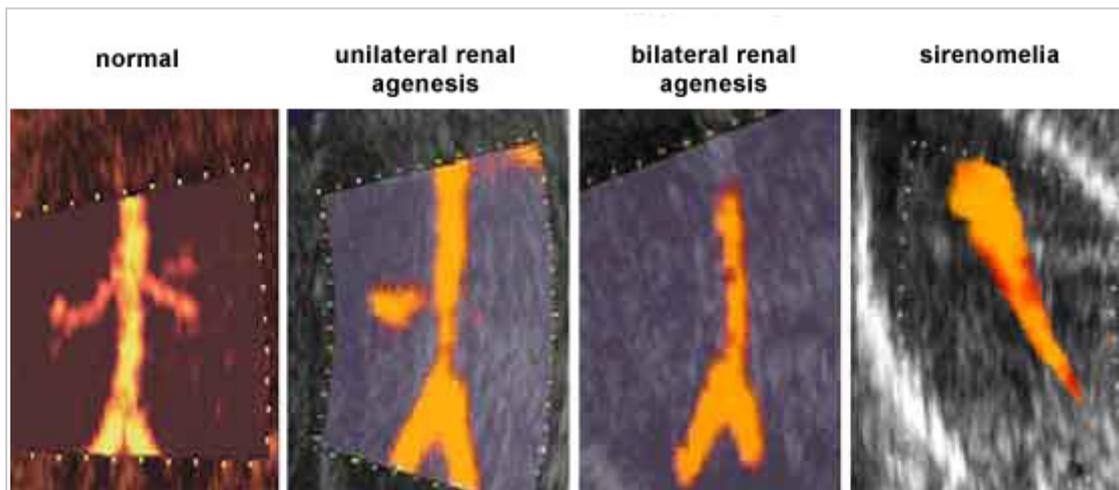
Renal agenesis is usually an isolated sporadic abnormality but, in a few cases, it may be secondary to a chromosomal abnormality or part of a genetic syndrome (such as Fraser syndrome), or a developmental defect (such as VACTERL association). In non-syndromic cases, the risk of recurrence is approximately 3%. However, in about 15% of cases, one of the parents has unilateral renal agenesis and in these families the risk of recurrence is increased.

Diagnosis

Antenatally, the condition is suspected by the combination of anhydramnios (from 17 weeks) and empty fetal bladder (from as early as 14 weeks). Examination of the renal areas is often hampered by the oligohydramnios and the 'crumpled' position adopted by these fetuses, and care should be taken to avoid the mistaken diagnosis of perirenal fat and large fetal adrenals for the absent kidneys. The differential diagnosis is from preterm rupture of membranes, severe uteroplacental insufficiency and obstructive uropathy or bilateral multicystic or polycystic kidneys. Vaginal sonography with high-frequency, high-resolution probes is useful in these cases.



Failure to visualize the renal arteries with color Doppler is another important clue to the diagnosis in dubious cases, both with bilateral and unilateral agenesis. Prenatal diagnosis of unilateral renal agenesis is difficult because there are no major features, such as anhydramnios and empty bladder, to alert the ultrasonographer to the fact that one of the kidneys is absent.



Prognosis

Bilateral renal agenesis is a lethal condition, usually in the neonatal period due to pulmonary hypoplasia. The prognosis with unilateral agenesis is normal.

INFANTILE POLYCYSTIC KIDNEY DISEASE (POTTER TYPE I)

In this condition, the markedly enlarged kidneys are filled with numerous cortical cysts and dilated collecting ducts. The disease has a wide spectrum of renal and hepatic involvement and it is subdivided into perinatal (this is the most common), neonatal, infantile and juvenile types on the basis of the age of onset of the clinical presentation and the degree of renal tubular involvement. Although recurrences tend to be group-specific, we have seen one family in which the four subdivisions were each represented in the four affected infants.

Prevalence

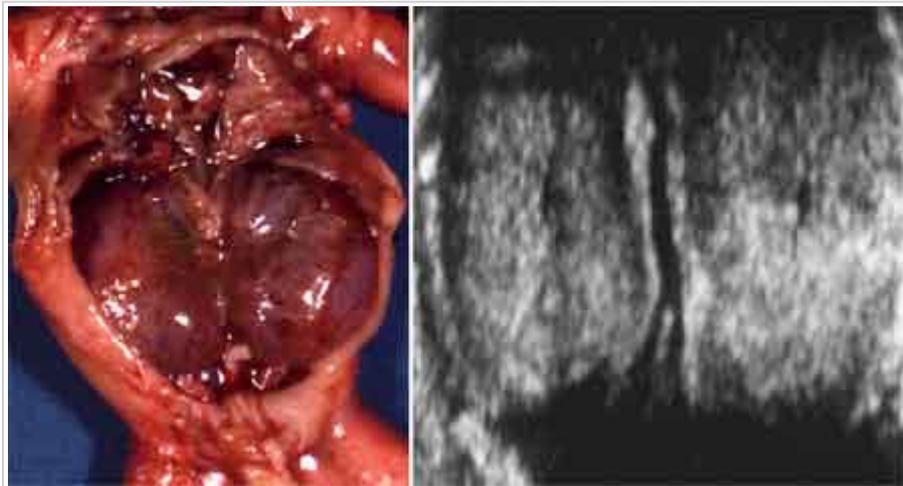
Infantile polycystic kidney disease is found in about 1 per 30 000 births.

Etiology

This is an autosomal recessive condition. The responsible gene is in the short arm of chromosome 6 and prenatal diagnosis in families at risk can be carried out by first-trimester chorion villous sampling.

Diagnosis

Prenatal diagnosis is confined to the types with earlier onset (perinatal and probably the neonatal types) and is based on the demonstration of bilaterally enlarged and homogeneously hyperechogenic kidneys. There is often associated oligohydramnios, but this is not invariably so. These sonographic appearances, however, may not become apparent before 24 weeks of gestation and, therefore, serial scans should be performed for exclusion of the diagnosis.



Prognosis

The perinatal type is lethal either in utero or in the neonatal period due to pulmonary hypoplasia. The neonatal type results in death due to renal failure within the 1st year of life. The infantile and juvenile types result in chronic renal failure, hepatic fibrosis and portal hypertension; many cases survive into their teens and require renal transplantation.

MULTICYSTIC DYSPLASTIC KIDNEY DISEASE (POTTER TYPE II)

Multicystic dysplastic kidney disease is thought to be a consequence of either developmental failure of the mesonephric blastema to form nephrons or early obstruction due to urethral or ureteric atresia. The collecting tubules become cystic and the diameter of the cysts determines the size of the kidneys, which may be enlarged or small. Exploration of the renal fossa in some cases reveals no renal artery, renal vein, ureter or cysts, suggesting that renal agenesis and dysplastic kidneys may be at different ends of a spectrum of renal malformation. This is further supported by the finding that, in about 15% of cases with multicystic kidneys, there is contralateral renal agenesis.

Prevalence

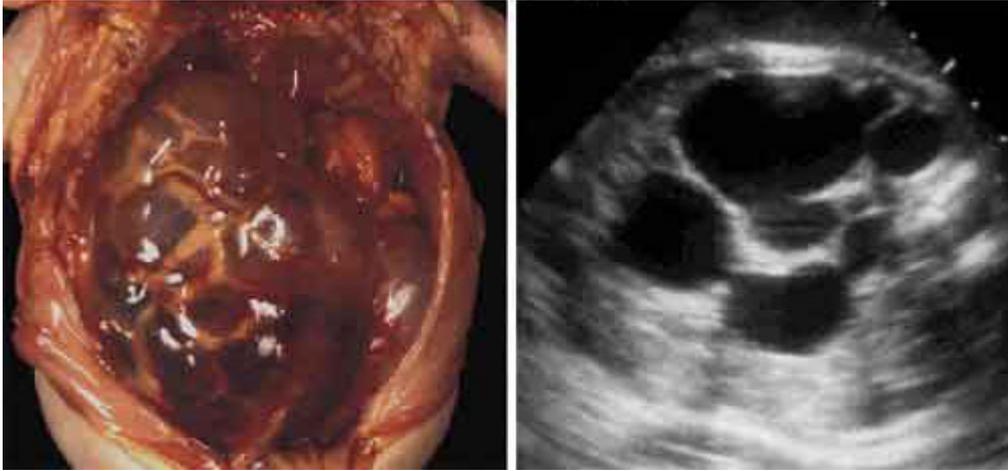
Multicystic dysplastic kidney disease is found in about 1 per 1000 births.

Etiology

In the majority of cases, this is a sporadic abnormality but chromosomal abnormalities (mainly trisomy 18), genetic syndromes and other defects (mainly cardiac) are present in about 50% of the cases.

Diagnosis

Ultrasonographically, the kidneys are replaced by multiple irregular cysts of variable size with intervening hyperechogenic stroma. The disorder can be bilateral, unilateral or segmental; if bilateral, there is associated anhydramnios and the bladder is 'absent'.



Prognosis

Bilateral multicystic dysplastic kidney disease is fatal before or soon after birth, due to pulmonary hypoplasia. Unilateral disease is associated with a normal prognosis. There is still controversy in the postnatal management of patients with a multicystic kidney; some urologists advocate prophylactic nephrectomy, but the majority adopt an expectant approach because the kidney gradually shrinks and may disappear. The parents and family should also be scanned to exclude autosomal dominant branchio-to-renal syndrome.

POTTER TYPE III RENAL DYSPLASIA

Potter type III renal dysplasia is characterized by markedly enlarged irregular kidneys with innumerable cysts of variable sizes interspersed among normal or compressed renal parenchyma. It is the common morphological expression of autosomal dominant adult polycystic kidney disease (APKD) and of other Mendelian disorders such as tuberous sclerosis, Jeune syndrome, Sturge-Weber syndrome, Zellweger syndrome, Lawrence Moon Biedl syndrome and Meckel-Gruber syndrome. Both kidneys are generally equally enlarged and only rarely is one involved so slightly that it remains of normal size. One-third of the cases have cysts in the liver, pancreas, spleen or lungs and one-fifth are found to have cerebral aneurysms.

Adult polycystic kidney disease (APKD)

One in 1000 people carry the APKD mutant gene. Adult polycystic kidney disease is usually asymptomatic until the third or fourth decade of life, and, although histological evidence of the disease is likely to be present from intrauterine life, the age of onset of gross morphological changes that are potentially detectable by ultrasonography is uncertain. Rarely, however, kidneys that are anatomically similar may cause death in infancy or early childhood and the condition has been designated as 'adult variety occurring in infancy'.

Prenatal diagnosis by ultrasonography is confined to a few case reports and the kidneys have been described as enlarged and hyperechogenic with or without multiple cysts. Unlike infantile polycystic kidneys, where there is a loss of the corticomedullary junction, in APKD there is accentuation of this junction. The amniotic fluid volume is either normal or reduced.



The kidney size is usually smaller than that of the infant polycystic kidneys. In counselling affected parents with APKD, it should be emphasized that the prenatal demonstration of sonographically normal kidneys does not necessarily exclude the possibility of developing polycystic kidneys in adult life. Nevertheless, prenatal diagnosis can now be made from chorion villous sampling and DNA analysis.

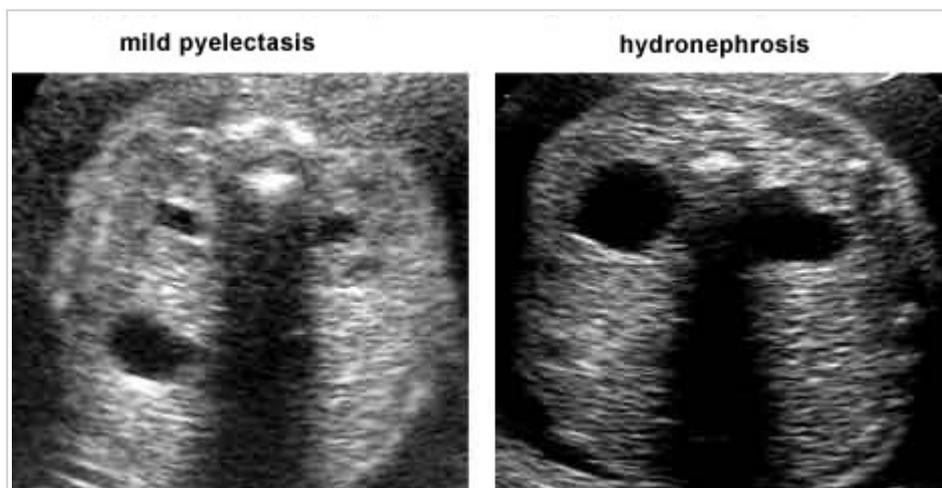
OBSTRUCTIVE UROPATHIES

The term 'obstructive uropathy' encompasses a wide variety of different pathological conditions characterized by dilatation of part or all of the urinary tract. When the obstruction is complete and occurs early in fetal life, renal hypoplasia (deficiency in total nephron population) and dysplasia (Potter type II; formation of abnormal nephrons and mesenchymal stroma) ensue. On the other hand, where intermittent obstruction allows for normal renal development, or when it occurs in the second half of pregnancy, hydronephrosis will result and the severity of the renal damage will depend on the degree and duration of the obstruction. Dilatation of the fetal urinary tract frequently, but not absolutely, signifies obstruction. Conversely, a fetus with obstruction may not have any urinary tract dilatation.

Hydronephrosis

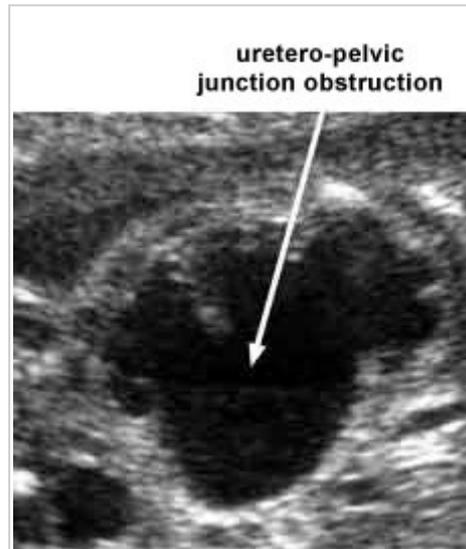
Varying degrees of pelvicalyceal dilatation are found in about 1% of fetuses. Mild hydronephrosis or pyelectasia is defined by the presence of an anteroposterior diameter of the pelvis of > 4 mm at 15–19 weeks, > 5 mm at 20–29 weeks and > 7 mm at 30–40 weeks. Transient hydronephrosis may be due to relaxation of smooth muscle of the urinary tract by the high levels of circulating maternal hormones, or maternal–fetal overhydration. In the majority of cases, the condition remains stable or resolves in the neonatal period. In about 20% of cases, there may be an underlying ureteropelvic junction obstruction or vesicoureteric reflux that requires postnatal follow-up and possible surgery.

Moderate hydronephrosis, characterized by an anteroposterior pelvic diameter of more than 10 mm and pelvicalyceal dilatation, is usually progressive and in more than 50% of cases surgery is necessary during the first 2 years of life.



Ureteropelvic junction obstruction

This is usually sporadic and, although in some cases there is an anatomic cause, such as ureteral valves, in most instances the underlying cause is thought to be functional. In 80% of cases, the condition is unilateral. Prenatal diagnosis is based on the demonstration of hydronephrosis in the absence of dilated ureters and bladder. The degree of pelvicalyceal dilatation is variable and, occasionally, perinephric urinomas and urinary ascites may be present. Postnatally, renal function is assessed by serial isotope imaging studies and, if there is deterioration, pyeloplasty is performed. However, the majority of infants have moderate or good function and can be managed expectantly.

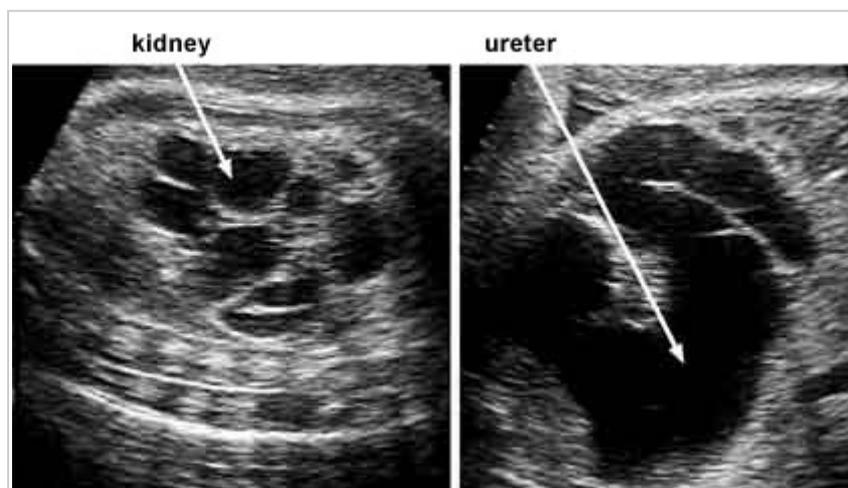


Ureterovesical junction obstruction

This is a sporadic abnormality characterized by hydronephrosis and hydroureter in the presence of a normal bladder. The dilated ureter is tortuous, and on ultrasound appears as a collection of cysts of variable size, localized between the renal pelvis, which is variably dilated, and the bladder, which is of normal morphology and dimensions. The etiology is diverse, including ureteric stricture or atresia, retrocaval ureter, vascular obstruction, valves, diverticulum, ureterocele, and vesicoureteral reflux. Ureteroceles (visible as a thin-walled and fluid-filled small circular area inside the bladder) are usually found in association with duplication of the collecting system. In ureteral duplication, the upper pole moiety characteristically obstructs and the lower one refluxes. The dilated upper pole may enlarge to displace the non-dilated lower pole inferiorly and laterally.

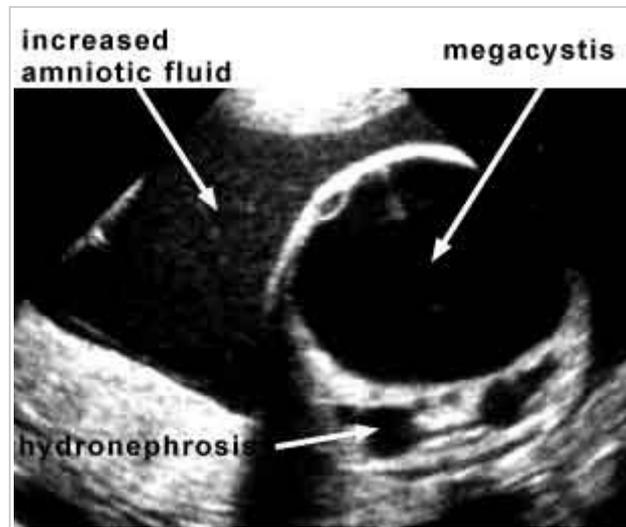
Vesicoureteric reflux

This sporadic abnormality is suspected when intermittent dilatation of the upper urinary tract over a short period of time is seen on ultrasound scanning. Occasionally, in massive vesicoureteric reflux without obstruction, the bladder appears persistently dilated because it empties but rapidly refills with refluxed urine. Primary megaureter can be distinguished from ureterovesical junction obstruction by the absence of significant hydronephrosis.



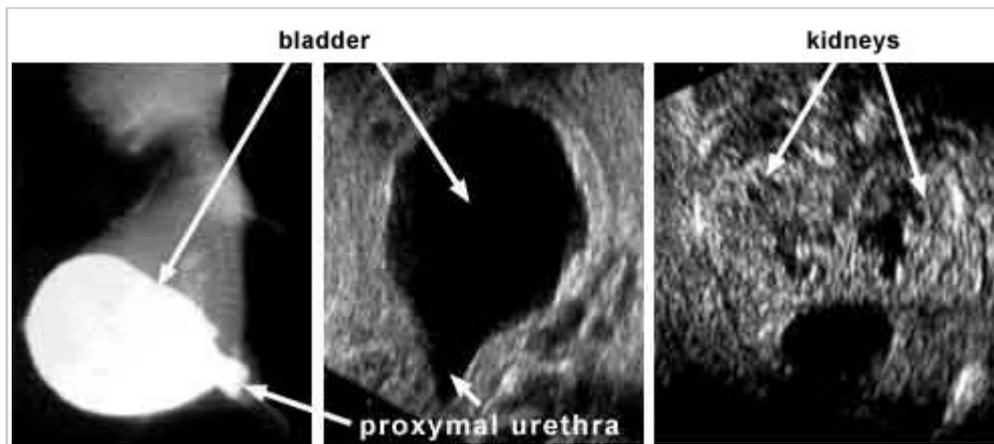
Megacystis–microcolon–intestinal hypoperistalsis syndrome (MMIHS)

This is a sporadic abnormality characterized by a massively dilated bladder and hydronephrosis in the presence of normal or increased amniotic fluid; the fetuses are usually female. There is associated shortening and dilatation of the proximal small bowel, and microcolon with absent or ineffective peristalsis. The condition is usually lethal due to bowel and renal dysfunction.



Urethral obstruction

Urethral obstruction can be caused by urethral agenesis, persistence of the cloaca, urethral stricture or posterior urethral valves. Posterior urethral valves occur only in males and are the commonest cause of bladder outlet obstruction. The condition is sporadic and is found in about 1 in 3000 male fetuses. With posterior urethral valves, there is usually incomplete or intermittent obstruction of the urethra, resulting in an enlarged and hypertrophied bladder with varying degrees of hydroureters, hydronephrosis, a spectrum of renal hypoplasia and dysplasia, oligohydramnios and pulmonary hypoplasia. In some cases, there is associated urinary ascites from rupture of the bladder or transudation of urine into the peritoneal cavity.



Fetal therapy for obstructive uropathy

In fetal lamb, ureteric ligation during the first half of gestation results in dysplastic kidneys, whereas, in the second half of pregnancy, ureteric ligation is associated with the development of hydronephrosis but preservation of renal architecture. Ligation of the urethra and urachus in fetal lambs at 100 days of gestation causes severe hydronephrosis and pulmonary hypoplasia; decompression by suprapubic cystostomy at 120 days' gestation reduces the urinary tract dilatation and improves the survival rate. Similarly, ureteric ligation at 65 days of gestation produces renal dysplasia, and subsequent decompression prior to term prevents renal dysplasia and produces reversible postobstructive changes; the degree of renal damage is proportional to the length of time for which the obstruction existed.

Encouraged by the results of these animal studies, and on the assumption that unrelieved obstruction causes progressive renal and pulmonary damage, several investigators in the 1980s performed in utero decompression of the urinary tract in the human, either by open surgical diversion or by the ultrasound-guided insertion of suprapubic

vesico-amniotic catheters. Although these techniques demonstrated the feasibility of intrauterine surgery, they did not provide conclusive evidence that such intervention improves renal or pulmonary function beyond what can be achieved by postnatal surgery. It is possible that, in a few selected cases, intrauterine intervention may be beneficial.

Assessment of fetal renal function

Antenatal evaluation of renal function relies on a combination of ultrasonographic findings and analysis of fetal urine obtained by urodochocentesis or pyelocentesis. Poor prognostic signs are:

- (1) The presence of bilateral multicystic or severely hydronephrotic kidneys with echogenic kidneys, suggestive of renal dysplasia;
- (2) Anhydramnios implying complete urethral obstruction; and
- (3) High urinary sodium, calcium and $\beta 2$ microglobulin levels.

Potential candidates for intrauterine surgery are fetuses with bilateral moderately severe pelvicalyceal dilatation and normal cortical echogenicity, or severe megacystis and oligohydramnios, or normal levels of urinary sodium, calcium and $\beta 2$ microglobulin.