Chapter 13

Renal Anomalies

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Additional information is available at the end of the chapter

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Abstract

This chapter is dedicated to the main renal anomalies detectable by ultrasound. Anomalies of the lower urinary tract will be addressed in a separate chapter. The anomalies presented are renal agenesis, renal development variants, autosomal recessive polycystic kidney disease, multicystic dysplastic kidney disease, autosomal dominant polycystic kidney disease, obstructive cystic dysplasia, pelvis dilatation, renal tumors, and non-chromosomal syndromes associated with renal anomalies. All chapters are structured similar into definition, incidence, pathology, ultrasound findings, differential diagnosis, and clinical facts.

Keywords: kidney, anomaly, ultrasound

1. Introduction

The present chapter addresses the main renal anomalies. It will be structured on nine subjects made to help the reader orientate easily when facing an anomaly in everyday practice. Information regarding the moment an anomaly is visible has taken into account midrange ultrasound machines that are responsible for most of the anomaly screening.

Kidneys are visible at 12–14 weeks of gestational age, easier with transvaginal examination, and the renal architecture is seen first at 16–18 weeks. Current protocols advise documenting the presence of the normal kidneys at the second and third trimester ultrasound. A special attention must be given not to confuse them with "lying-down" adrenal structures. We recommend using both transversal and longitudinal views; coronal views are helpful in the diagnosis of the horseshoe kidneys. Color Doppler ultrasound can be used to identify the renal

Weeks of gestation	Fetal renal mean longitudinal length (cm) (±SD)
16	1.7 (0.3)
17	1.8 (0.1)
18	2.0 (0.0)
19	2.3 (0.3)
20	2.1 (0.1)
21	2.1 (0.1)
22	2.4 (0.3)
23	2.5 (0.3)
24	2.8 (0.1)
25	2.9 (0.2)
26	2.8 (0.1)
27	3.0 (0.1)
28	3.3 (0.3)
29	3.5 (0.2)
30	3.4 (0.3)
31	3.6 (0.1)
32	3.7 (0.2)
33	3.7 (0.2)
34	3.8 (0.2)
35	3.9 (0.3)
36	4.1 (0.3)
37	4.3 (0.3)
38	4.2 (0.3)
39	4.2 (0.2)
40	4.3 (0.2)
41	4.1 (0.2)

Table 1. Mean renal length by gestational age.

arteries. Normal measurements for renal length are shown in **Table 1** [1]. The renal circumference to abdominal circumference is about one-third. The anterior-posterior renal pelvis is usually less than 4 mm before 22 weeks and less than 7 mm in the third trimester.

2. Renal agenesis

Definition: this chapter will address only bilateral renal agenesis, a condition defined as the absence of both kidneys which is invariably lethal.

Incidence: 1:2000–1:5000.

Pathology: it results from failure of development of the ureteric bud. The consequence for the pregnancy is Potter sequence: oligohydramnios, Potter face, clubbed hands and feet, and pulmonary hypoplasia which leads to death in the cases that reach birth [2].

Ultrasound findings: we notice severe oligohydramnios and fail to see the kidneys and the bladder. Sometimes, lying-down adrenals may be confused with kidneys in the conditions of poor visibility associated with low amniotic fluid/absence of amniotic fluid. Color Doppler interrogation fails to demonstrate the renal arteries. A small thorax is noticed, especially if we take the time to measure the heart/chest ratio.

Differential diagnosis:

- PROM (patient history and the presence of kidneys and bladder point us the right diagnosis).
- Severe IUGR (kidneys are present, and there are abnormal Doppler values).

Clinical facts:

- Risk of chromosomal anomalies is low (though there have been described cases of trisomy 7, 10, 21, 22).
- It may be part of a nonchromosomal syndrome (COF syndrome, VACTERL).
- Oligohydramnios is an associated sign only after 16 weeks.
- You should always examine carefully not to confuse adrenal glands with kidneys; keep in mind that adrenal arteries can also mimic renal arteries, so Doppler is not always a solution.
- Bilateral agenesis is always lethal (one-third stillbirth, the rest die at birth from pulmonary hypoplasia) (**Figure 1**).

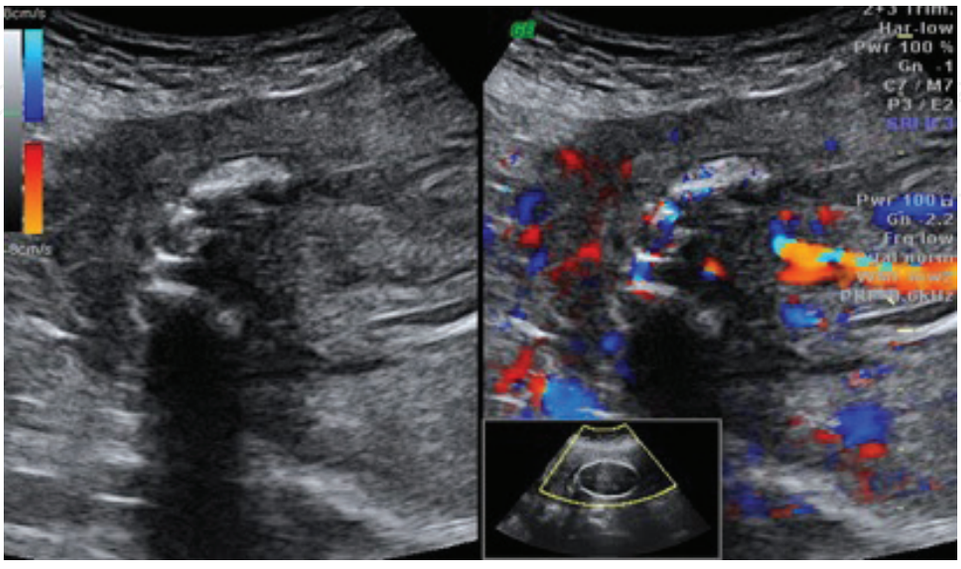

Figure 1. Renal agenesis (absence of renal arteries).

3. Renal development variants

3.1. Unilateral agenesis

Definition: one kidney does not form resulting one present kidney and one renal artery.

Incidence: 1:1000 [2].

Pathology: failure of development of only one ureteric bud with normal development on the other side.

Ultrasound findings: we notice an empty renal fossa on axial view; this view should be completed with longitudinal and coronal views. The contralateral kidney is increased in size (>95 percentile)—compensatory hypertrophy. The use of color Doppler shows only one renal artery. Some structures may mimic the second kidney—one is the adrenal gland, and the other is the colon.

Differential diagnosis: an empty renal fossa may be present in:

- Pelvic kidney.
- Unilateral renal agenesis.
- Crossed renal ectopia.
- Horseshoe kidney (graph).

Clinical facts:

- Careful scanning of the fetal abdomen (do not confuse with renal ectopia/do not confuse kidney with adrenal glands).
- Isolated unilateral kidney has good prognosis and associates rarely with chromosomal anomalies (**Figure 2**).

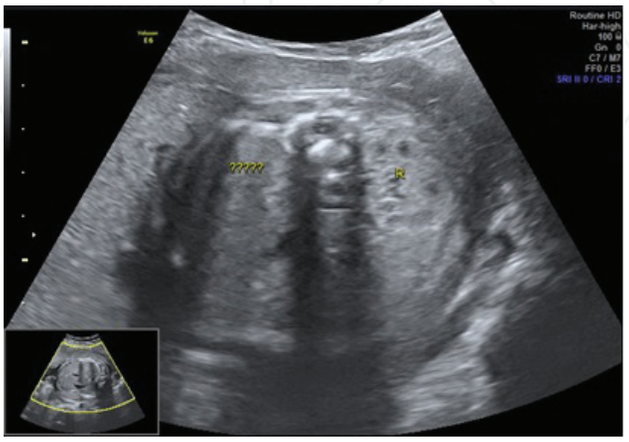

Figure 2. Unilateral renal agenesis.

3.2. Pelvic kidney

Definition: the presence of one kidney in the pelvis; the most common location for ectopic kidney.

Incidence: 1:700–1:1200 [2–4].

Pathology: the kidney forms normally but fails to ascend to the lumbar area. This normally happens between 6 and 10 weeks of gestational age.

Ultrasound findings: the first thing we notice is an empty renal fossa; careful scanning reveals the kidney adjacent to the bladder. The normally positioned kidney shows no compensatory hypertrophy. Amniotic fluid is within a normal range. The use of color Doppler can be helpful—sometimes, you can follow the renal artery to the ectopic kidney, but sometimes a pelvic kidney can have vascularization from the iliac arteries.

Differential diagnosis: empty renal fossa (see above).

Clinical facts:

- Pelvic kidney should be the first thing to search in an empty renal fossa.
- Visualization can sometimes be difficult due to bowel loops or interposed iliac wing.
- May be associated with genital, gastrointestinal, or cardiac anomalies.
- Risk of chromosomal anomalies is low and so is the risk of nonchromosomal syndromes.
- May be a family group so parents should be scanned.
- Vesicoureteral reflux is frequently present so postnatal ultrasound monitoring is recommended (**Figure 3**).

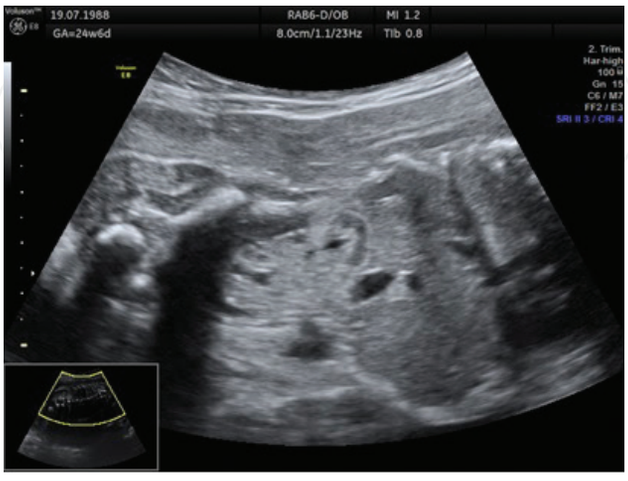

Figure 3. Pelvic kidney.

3.3. Horseshoe kidney

Definition: the kidneys are fused in their lower poles, with equal amount of renal tissue bilaterally. The fused portion may be renal parenchyma or fibrous tissue.

Incidence: 1:400.

Pathology: the fusion takes place before the ascent of the kidney which is partially impeded by the emergency of the inferior mesenteric arteries, causing also alteration of the kidneys' axis.

Ultrasound findings: on the standard axial scan, we can see renal tissue in front of the descending aorta. On coronal sections we can see the kidneys fused in the region of the inferior poles (other variants are possible but extremely rare). We also notice a medial rotation of the inferior poles and a lower position than normal kidneys.

Differential diagnosis: includes empty renal fossa (see above), but also severe oligoamnios may suggest pathology due to lack of visibility.

Clinical facts:

- It is frequently associated with hydronephrosis and genital anomalies.
- 33% of cases have CNS and cardiac or skeletal malformations [5].
- Risk of chromosomal anomalies—horseshoe kidney may be found in fetuses with Turner's syndrome or trisomy 18.
- Risk of nonchromosomal syndrome (caudal regression syndrome, otocephaly, Oro-facial digital syndrome).
- Recurrence risk—low in isolated forms.
- Careful anatomy scan to exclude other anomalies.
- Karyotyping should be offered (especially if other anomalies or soft markers are present).
- Postnatal monitoring for vesicoureteral reflux, hydronephrosis, and urinary tract infections is recommended.
- Prognosis is considered good in isolated forms (**Figure 4**).

3.4. Crossed renal ectopia

Definition: both kidneys are on the same side of the abdomen; a significant number (95%) are fused.

Incidence: 1:7000.

Ultrasound findings: at the anatomy scan, we notice one empty renal fossa and one abnormally large, frequently bilobed contralateral kidney. Statistically, it is more likely to find the kidney/kidneys on the right side. Color Doppler study shows two renal arteries on the same side (one in the normal position and one lower).

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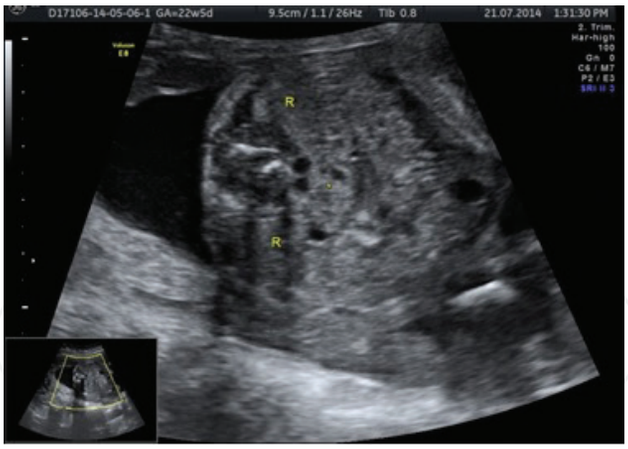

Figure 4. Horseshoe kidney.

Differential diagnosis: empty renal fossa (see above).

Clinical facts:

- May be associated with renal anomalies, spina bifida, and sacral agenesis, so attentive evaluation of the spine should be conducted.
- As all renal development variants, it may be associated with infections, obstructions, and vesicoureteral reflux so postnatal monitoring is recommended.
- Postnatal evaluation of genital organs—uterine anomalies may be associated.

4. Autosomal recessive polycystic kidney disease (Potter type I)

Definition: autosomal recessive polycystic kidney disease (ARPKD) is a bilateral renal anomaly caused by a gene disorder.

Incidence: 1:20,000–1:45,000.

Pathology: the PKHD1 gene on chromosome p21 [6] is generally accepted as a primary cause though the specific mechanism is not completely understood. Mutations are specific for individual families. The anomaly is characterized by convoluted tubes and collecting ducts often associated with liver fibrosis [4].

Ultrasound findings: ARPKD is characterized by kidney enlargement (>2SD above the mean for that gestational age) [4], increased echogenicity (resulting from the interference of the microcysts) [3], absent bladder, and oligoamnios (present from 16 weeks).

Differential diagnosis:

- Autosomal dominant polycystic kidney disease (ADPKD)—normal quantity of amniotic fluid and a normal bladder.
- Trisomy 13 (holoprosencephaly, polydactyly, facial anomalies).

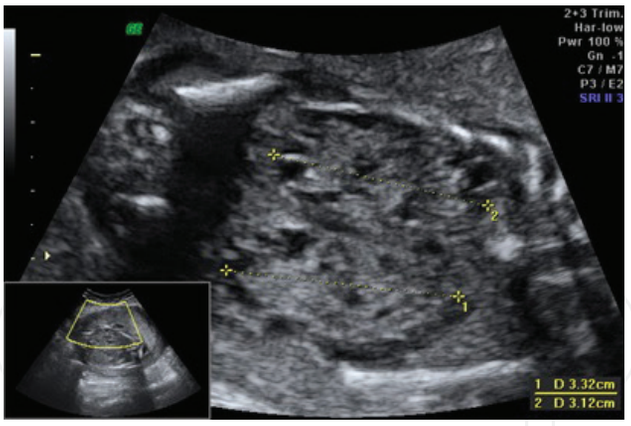

Figure 5. Autosomal recessive polycystic kidney disease.

Clinical facts:

- Not associated with chromosomal anomalies.
- Enlarged, hyperechogenic kidneys may be present in many syndromes (Meckel-Gruber, Bardet-Biedl, Beckwith-Wiedmann, Perlman, Elejade).
- Most cases are diagnosed by 24 weeks, but you must keep in mind that kidneys may look normal until 20 weeks.
- ARPKD is classified in perinatal, neonatal, infantile, and juvenile form.
- Cases diagnosed in utero end with stillbirth or neonatal death.
- Thirty to fifty percent die in the neonatal period.
- Juvenile form has less renal involvement but marked hepatic fibrosis.
- Survivors develop systemic hypertension (75%) and portal hypertension (44%).
- Recurrence risk is 25%.
- When diagnosed prenatally, termination should be offered (**Figure 5**).

5. Multicystic dysplastic kidney disease (Potter type II)

Definition: Multicystic dysplastic kidney (MCDK) presents with unilateral/bilateral enlarged kidneys with parenchyma replaced by multiple, noncommunicating cysts [3].

Incidence: 1:1000–1:5000; more common in males (2:1), but females have a worse prognosis (twice more likely to have bilateral forms and four times more likely to have aneuploidy).

Pathology: in normal kidney embryology, the ureteric bud signals the metanephros to form nephrons. Early ureter obstruction or atresia prevents the signaling so the metanephric tissue does not form nephrons, resulting in dysplastic cystic tissue. Segmental/partial MCDK may result from a duplex ureter [2].

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Ultrasound findings:

Unilateral (75–80%): the diagnostic is made in the presence of multiple cyst structure in the renal fossa, significantly larger than normal kidneys. The bladder is normal. Amniotic fluid is within the normal range [3].

Bilateral (20%): both kidneys are multicystic; the bladder cannot be visualized, and severe oligoamnios is associated.

Partial (rare): in rare cases of duplex kidney, only part of the kidney may be involved, more frequently the superior pole.

Differential diagnosis:

- Hydronephrosis (distended calyces appear as cysts, but at attentive scrutiny communication with the renal pelvis can be proved).
- Obstructive cystic dysplasia (more normal renal tissue visible).
- Ureteral dilatation.

Clinical facts:

- Risk of chromosomal anomalies is relatively low in unilateral forms (2–4%).
- The risk for nonchromosomal syndromes is about 5–10% (branchio-oto-renal syndrome, cerebro-reno-digital syndrome, VACTERL).
- Careful examination of the contralateral kidney (40% have an associated anomaly).
- Genetic counseling and karyotyping are advised if associated anomalies are present.
- Antenatal kidney monitoring is recommended.
- Conservative management is standard as most cases involute in the first years of life.
- Postnatal ultrasound evaluation is recommended every 6 months (**Figure 6**).

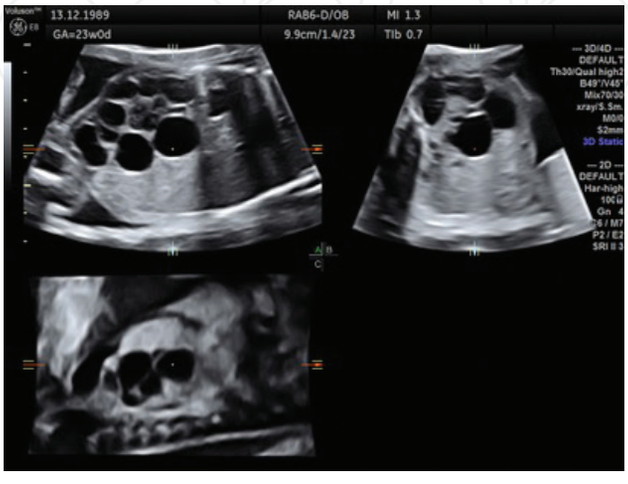

Figure 6. Multicystic kidney (unilateral).

6. Autosomal dominant polycystic kidney disease (Potter type III)

Definition: ADPKD is a bilateral renal anomaly where cysts arise from all areas of the nephron or collecting ducts. It commonly appears in adults but can rarely be seen prenatally, especially when screening is targeted to families at risk.

Incidence: 1:1000.

Pathology: the genetic mechanism involves two genes PKD1 and PKD2 on chromosome 16. The condition is associated with multiple renal cysts, hypertension, and renal failure. Cysts are also present in the liver, spleen, and pancreas.

Ultrasound findings: the kidneys are hyperechoic, in some cases only in the cortical region. Amniotic fluid and the bladder are usually normal.

Differential diagnosis: ARPKD (autosomal recessive polycystic kidney disease). Normal fluid, bladder, and family history help us make the difference.

Clinical facts:

- Once diagnosed, serial monitoring is recommended.
- Examination of parent's kidneys is indicated due to the autosomal dominant nature of the disease.
- The disease manifests in the third to fifth decade, most patients needing dialysis and transplant.
- Normal ultrasound cannot exclude the disease later in life!

7. Obstructive cystic dysplasia (Potter type IV)

Definition: obstructive cystic dysplasia results from early and severe obstruction of the collecting system causing the formation of renal cysts [5].

Pathology: most cases result from early urethral obstruction, but vesicourethral junction obstruction and upper urinary tract obstruction are also a possible cause. Obstruction leads to ascension of fluid in the upper tract, with fluid retention in the nephron, with secondary cyst formation, and with a decrease in the number of normal nephrons.

Ultrasound findings: sonographic examination reveals renal macrocysts and signs of urinary tract obstruction (hydronephrosis, hydroureter, bladder distension). In cases of urethral obstruction, thickening of the bladder wall and severe oligoamnios are met.

Differential diagnosis:

- MCDK.
- Hydronephrosis.
- ARPKD.

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Clinical facts:

- Risk of chromosomal anomalies (5–10%).
- Risk of nonchromosomal syndromes may be found in VACTERL, cerebro-reno-digital syndrome, and tuberous sclerosis.
- Look for renal cysts when urinary tract obstruction is diagnosed.
- Unilateral: renal cysts + hydronephrosis/hydroureter.
- Bilateral: oligoamnios + distended bladder + bilateral renal cysts.
- Perform careful follow-up.
- Amniocentesis is indicated when associated anomalies are present.
- May be impossible to differentiate from MCDK.
- Termination should be offered for bilateral form.

8. Pelvis dilatation

Definition: the dilatation of the pelvis is the most common anomaly detected by ultrasound. It can present as a mild pelviectasis or as hydronephrosis. Though numbers may vary in different sources, generally values are around these figures:

- Mild pelviectasis [2]: above 4 mm in the second trimester and above 7 mm in the third trimester.
- Hydronephrosis [4]: above 7 mm between 16 and 20 weeks and above 10 mm after 20 weeks.

Limits of normal size for gestational age have also been described [2]:

- 3 mm in the first trimester.
- 4 mm between 14 and 22 weeks.
- 5 mm between 22 and 32 weeks.
- 7 mm after 32 weeks.
- Above 10 mm always pathology.

Incidence: 1–5:500 newborns.

Pathology: mild pelviectasis has been associated with aneuploidy (minor marker), especially trisomy 21. The mechanism for unilateral hydronephrosis may be obstruction of the ureteropelvic junction, vesicoureteral reflux, and obstruction of the vesicourethral junction. Bilateral hydronephrosis may be caused by bilateral vesicoureteral reflux or by urethral obstruction.

Ultrasound findings: renal scanning reveals a dilated renal pelvis above the cutoff for the respective gestational age. Frequently, when hydronephrosis is installed, the calyces are also

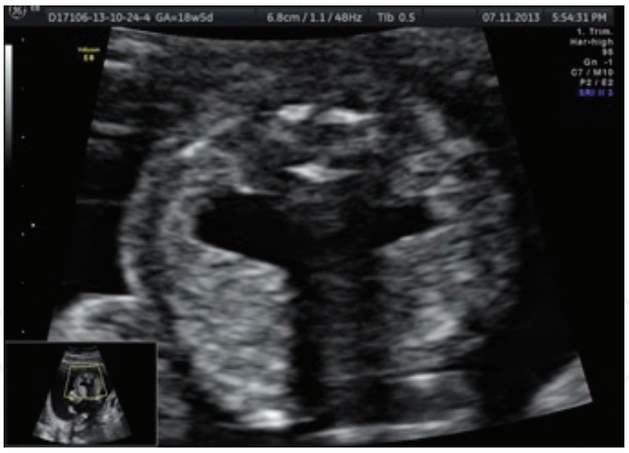

Figure 7. Bilateral hydronephrosis.

dilated. Sometimes, the dilatation is isolated (as in ureteropelvic junction stenosis) or includes dilatation of the ureters. In rare cases dilatation may lead to urinoma (only in cases of severe obstruction). Amniotic fluid is usually normal and in one-third of the cases may even be increased (impaired concentration ability).

Clinical facts:

- Risk of chromosomal anomalies is low, though mild pelviectasis has been associated with trisomy 21.
- Risk of nonchromosomal syndromes (VACTERL, Schinzel-Giedion syndrome, camptomelic dysplasia).
- In the presence of mild pelviectasis, screening for T21 markers is recommended.
- Eighty percent of mild pelviectasis resolve antenatally, and half of the rest resolve postnatally [2].
- Pelviectasis that is slowly progressing to hydronephrosis usually has an underlying pathology that would have to be addressed postnatally.
- Even with hydronephrosis the prognosis is excellent if there is no renal impairment.
- Poor prognosis may appear in cases of bilateral renal pathology or associated anomalies (syndromic or not).
- Postnatal following is recommended with scans and evaluation of the renal function.
- Prenatal intervention is rarely needed (**Figure 7**).

9. Renal tumors

Definition: renal tumors in the fetus are more commonly mesoblastic nephroma (a benign tumor) with rare occurrence of Wilms' tumor (which is malignant).

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Pathology: mesoblastic nephroma is a benign mesenchymal tumor with spindle-shaped cells. It is frequently associated with polyhydramnios through mechanisms that are not yet fully understood; polyuria caused by hypercalcemia and bowel obstruction by mass effect are among the most accepted theories.

Ultrasound findings: examination usually reveals a tumor/mass that occupies part or the entire kidney. Mesoblastic nephromas have ill-defined margins and may present on color Doppler ultrasound as a vascular mass. When there are arteriovenous shunts, fetus may present hydrops.

Differential diagnosis:

- Adrenal mass (tumor or hemorrhage).
- Crossed fused ectopia.
- Renal collecting system duplication.

Clinical facts:

- Risk of chromosomal anomalies is very low.
- Risk of nonchromosomal anomalies: Wilms' tumor may be associated with Beckwith-Wiedemann or Denys-Drash syndrome [5].
- The first sign may be polyhydramnios.
- Tumor may have rapid growth.
- You should look for Beckwith-Wiedmann signs.
- May have a–v shunts and hydrops, or cardiac failure may appear.
- Surgical removal of the tumor or nephrectomy is indicated in the neonatal period (**Figure 8**).

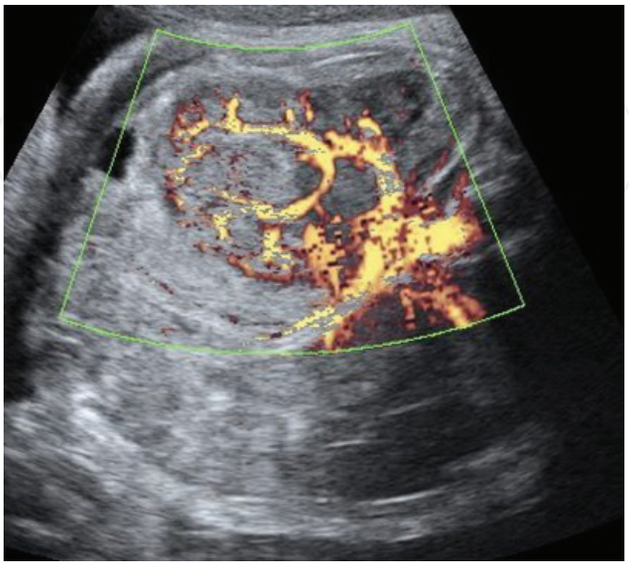

Figure 8. Nephroblastoma.

10. Nonchromosomal syndromes associated with renal anomalies

Nonchromosomal syndromes associated with abnormal kidneys on ultrasound that have been mentioned throughout this chapter have been included in **Table 2**.

Syndrome	Short description of the syndrome
COF skeletal syndrome	Renal agenesis + microcephaly, micrognathia, and joint contractures
VACTERL	Renal agenesis + vertebral anomalies, anal atresia, CHD, tracheoesophageal fistula, and limb anomalies
Meckel-Gruber syndrome	Polycystic kidney + cephalocele, microcephaly, and polydactyly
Bardet-Biedl syndrome	Polycystic kidney + polydactyly and genital anomalies
Beckwith-Wiedmann syndrome	Polycystic kidney + macroglossia, omphalocele, and hemihypertrophy
Perlman syndrome	Polycystic kidney + diaphragmatic hernia, macrosomia, cleft palate, and dextrocardia
Elejade syndrome	Polycystic kidney + omphalocele, corpus callosum agenesis, macrosomia, craniosynostosis, and skeletal dysplasia
Brachio-oto-renal syndrome	Multicystic kidney + preauricular tags and brachial cleft fistulas
Cerebro-reno-digital syndrome	Multicystic kidney + digital and limb anomalies and CNS malformations
Schinzel-Giedion syndrome	Hydronephrosis + midface retraction, skull anomalies, talipes, and cardiac anomalies
Camptomelic dysplasia	Hydronephrosis + bowed tibiae/femurs, scapular hypoplasia, micrognathia, and sex reversal in males
Denis-Drash syndrome	Nephroblastoma + ambiguous genitalia and diaphragmatic hernia (rare)

Table 2. Nonchromosomal syndromes associated with renal anomalies.

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