Congenital renal tract abnormalities: a neonatal perspective

Congenital renal and urinary system malformations are among the commonest congenital anomalies. They represent a wide spectrum of disorders with varying degrees of postnatal clinical significance. Many of these conditions can be detected on antenatal ultrasonography as part of the 18-20 week anomaly scan. This article will cover the postnatal management of renal dilatation and other commonly seen anomalies.

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Key points

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1. The kidneys and urinary tract are the most common sites for congenital abnormalities.
2. Detection of renal anomalies during an antenatal scan enables prompt diagnosis and management in the postnatal period.
3. A robust screening pathway is essential to ensure early detection, avoid unnecessary investigations and reduce parental anxiety.

The kidneys and urinary tract are the most common sites for congenital abnormalities and can account for 15-20% of all prenatally detected congenital anomalies.[1] Congenital renal and urinary system malformations are seen in 3-4% of the population.[2]

Various conditions can result in renal malformations seen on an antenatal anomaly scan. Neonatal healthcare professionals should be aware of congenital renal abnormalities as early postnatal intervention and appropriate long-term follow-up will ensure prompt diagnosis and management, avoid unnecessary investigations and minimise renal damage. This article will review the postnatal management of the renal anomalies seen in **FIGURE 1**.

- Renal pelvic dilatation
- Congenital renal anomalies
  - Renal agenesis
  - Ectopic kidney
    - Pelvic kidney
    - Crossed renal ectopia
    - Horseshoe kidney
  - Duplex kidney
- Congenital cystic kidney disease
  - Multicystic dysplastic kidney
  - Dysplastic kidney (renal dysplasia)
- Posterior urethral valve (PUV)

FIGURE 1 Renal anomalies that are commonly encountered in neonates.

Anatomy of the urinary system

The urinary system produces, stores and removes urine from the body. The kidneys make urine by filtering the blood to remove waste products. From the kidneys the urine flows through the ureters to the urinary bladder and then to the outside of the body along the urethra during urination. The basic anatomy of the urinary tract can be seen in **FIGURE 2**.

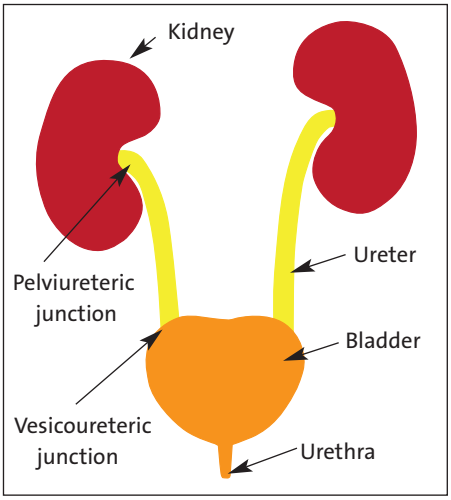

FIGURE 2 Anatomy of the urinary system.

Renal pelvic dilatation

Fetal renal pelvic dilatation is a common anomaly detected on antenatal scanning, with an incidence of 0.5-1% in pregnancy.[3] The presence of renal pelvic dilatation can indicate diseases such as structural anomalies, obstruction or vesicoureteric reflux (VUR – an abnormal backward flow of urine from the bladder to the upper urinary tract).[3] Although mostly a benign condition, it can in some cases cause significant morbidity. Renal pelvic dilatation can be classified according to the NHS fetal anomaly scan national standards (**FIGURE 3**).[3]

Renal pelvic dilatation is usually detected on the 20-week anomaly scan with some centres offering a further serial scan at 30-36 weeks' gestation.[5,7] Evidence suggests

that there is uncertainty in how fetal renal pelvic dilatation reflects postnatal abnormalities but it is generally agreed that there is a greater likelihood of postnatal problems with increasing pelvic dilatation.[3,8] It is therefore essential to have a robust screening pathway to identify those infants with significant renal problems to ensure early intervention and long-term follow-up, yet avoid unnecessary investigations. An example of a screening pathway is shown in **FIGURE 4**.

Renal pelvic dilatation should be managed in the following way:

- **Uncomplicated renal dilatation ≤7mm.** If there are no other associated renal abnormalities (such as duplex kidney) no postnatal investigations are required.
- **Uncomplicated renal dilatation 7-10mm.** The infant does not require antibiotics. The first renal ultrasound scan should take place at one month of age; it is important the scan is not delayed. Follow-up is necessary after the one-month scan if the abnormalities persist.

Grade	Renal pelvic diameter
Normal	<7mm
Mild	7-10mm
Moderate	10-15mm
Severe	>15mm

FIGURE 3 Renal dilatation grading by anteroposterior pelvic diameter.[3-6]

- **Renal dilatation >10mm with or without other abnormalities.** The infant will require prophylactic antibiotics (trimethoprim) from birth. Ideally ultrasound will be performed before discharge. Follow-up is required in the neonatal or urology clinic.

Congenital renal anomalies

Unilateral agenesis

Unilateral agenesis – the absence of a single kidney – has an incidence of approximately one in 1,000 to one in 3,200 live births.[9,10] Renal agenesis occurs after a major disruption in the metanephric stage in the first few weeks of gestation. An important differential diagnosis for unilateral agenesis is an undiagnosed ectopic kidney.[11] Renal agenesis can be diagnosed on the 20-week antenatal anomaly scan and/or during investigations for urinary tract infections. In approximately 50% of children with unilateral agenesis there is an association with other congenital abnormalities of the urinary tract:

- VUR occurs in 28% of children with unilateral agenesis
- pelviureteric junction obstruction (PUJ – blockage at the junction where the ureter attaches to the kidney) and/or vesicoureteric junction obstruction (VUJ – blockage where the ureter meets the bladder) occur in 20% of children with unilateral agenesis.[10,11]

Seventy per cent of children with unilateral agenesis have associated genital anomalies that can be either minor or major, such as an absent gonad on the ipsilateral side.[6,12] Long-term outcomes can be varied and dependent on the quality of

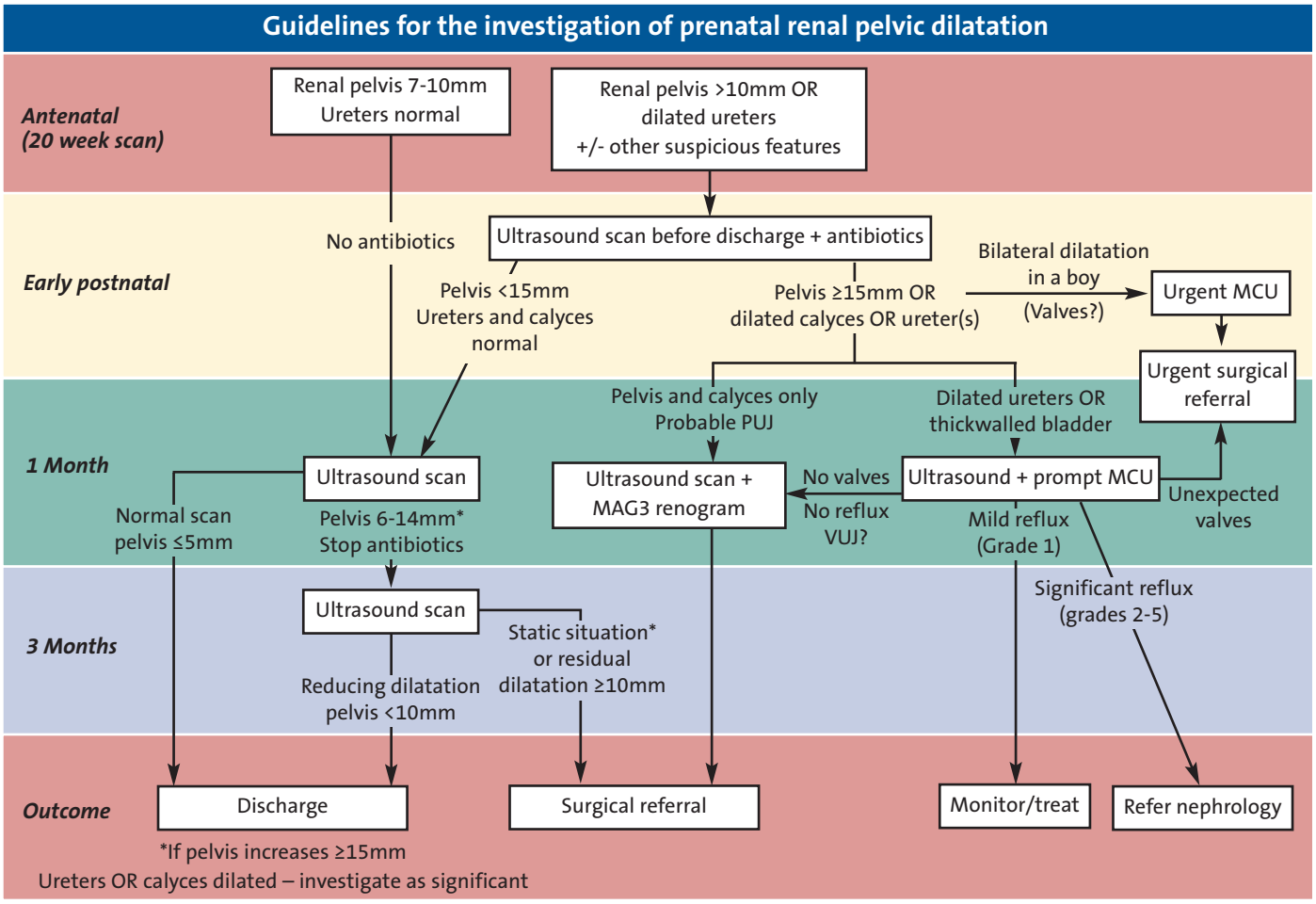

FIGURE 4 The Leeds Teaching Hospital's neonatal guideline for investigation of prenatal renal pelvic dilatation.[6] Key: MCU = micturating cystourethrography (used to examine whether reflux is occurring during urination), PUJ = pelvicureteric junction obstruction, MAG3 = MAG3 renogram (a scan to assess whether the kidneys are draining properly into the bladder), VUJ = vesicoureteric junction obstruction.

the existing kidney. If there is normal renal function and no other associated abnormalities the outcome is excellent.[5,10,12]

Action

Infants should have a postnatal ultrasound scan as an out-patient within the first month of life to measure the size of the single kidney and to look for other renal abnormalities. Small ectopic kidneys and pelvic kidneys can be difficult to visualise on antenatal scans, therefore a postnatal scan is indicated to verify the antenatal findings. There may also be evidence of compensatory renal hypertrophy (>50th centile for age and height) on the contralateral normal kidney, which is a reassuring sign.

There is no need for antibiotic prophylaxis or other investigations provided the ultrasound shows a normal single kidney.

Follow-up

Infants should have serial ultrasound scans at six months and 12 months to monitor for compensatory renal hypertrophy. If present, yearly blood pressure monitoring and urine analysis to check for proteinuria is recommended (in primary care). If there is any evidence of raised blood pressure or proteinuria with a raised creatinine level, referral to a paediatric nephrologist is advised. If absent, glomerular filtration rate measurement is recommended at one year of age by the nephrology team. In the presence of abnormal renal function or size, long-term follow-up is advised.[10]

Advise

Parents should be advised about early detection and management of urinary tract infections.

There is now evidence to show that children with a single kidney need not avoid contact sports.[13]

Ectopic kidney

The incidence of an ectopic kidney – any kidney not located in its normal anatomical position – is one in 1,000.[6,14] Ectopic kidneys can be found anywhere along the embryological ascending pathway within the abdomen and thorax, although the majority are found in the pelvis.[12,14] They are mostly unilateral, very rarely bilateral and not of reniform shape[5,10,15] and can lead to obstruction, urinary tract infections (UTI) or renal calculi. There is also a high incidence of urological complications, particularly VUR. They can be associated with non-renal anomalies (cardiac, skeletal) or part of syndromes such as in CHARGE syndrome and VACTERL association.[15,16]

Pelvic kidneys are the commonest form of renal ectopia (90%) with a left-sided predominance.[5] The majority of patients with this are asymptomatic.

In crossed renal ectopia, one kidney crosses the midline and its ureter re-crosses the midline to fuse with the bladder on the correct side. In the majority of cases (85-90%) the two kidneys are fused, most commonly the lower pole of the normal kidney with the upper pole of the ectopic kidney as it sits inferiorly, medially and abnormally rotated.[12,17] This condition is associated with ureteric and lower tract lesions and with cloacal and anorectal malformations.[10]

A horseshoe kidney (**FIGURE 5**) is the commonest form of fusion anomaly with an incidence of one in 400-800 babies, with a male to female prevalence of 2:1.[5,14] A horseshoe kidney usually involves fusion of the lower poles of the kidneys by a narrow isthmus of renal tissue.[17] The kidneys are also more caudally situated than normal.[15] There is a strong association with chromosomal abnormalities, such as in Turner syndrome, and females with a horseshoe kidney should have karyotyping.[12]

Horseshoes kidneys are usually asymptomatic (90%) and found incidentally. Occasionally, however, they can cause problems such as UTIs, abdominal pain and haematuria.[12] There are recognised complications of horseshoe kidneys: UTIs secondary to VUR occur in up to half of cases.[10] Renal calculi can also occur in approximately 20-60% of patients due to the presence of hydronephrosis (a build-up of urine leading to distension and dilation of the kidney) or obstruction causing urinary stasis and infection. PUJ obstruction is seen due to the high insertion of the ureter into the renal pelvis and displacement of the isthmus.[14] Other complications include: trauma to the isthmus, an increased risk of Wilms' tumour and hypertension secondary to renal cortical scarring.[10]

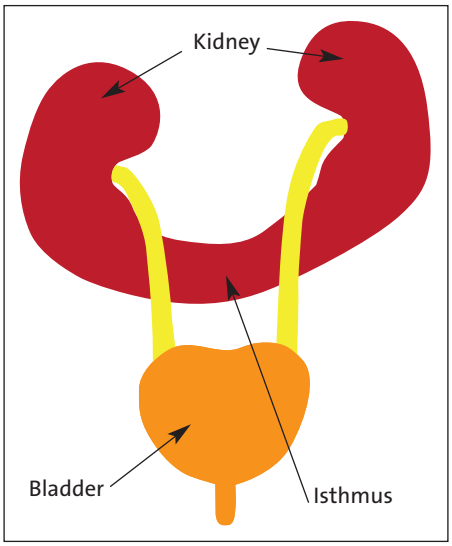

FIGURE 5 The horseshoe kidney usually involves fusion of the lower poles of the kidneys by a narrow isthmus of renal tissue.

Action

Infants should have a postnatal ultrasound to confirm the ectopic kidney and to determine if there is any associated hydronephrosis and obstruction in either kidney. If there is evidence of hydronephrosis the baby will require further investigations in the first six weeks including:

- a micturating cystourethrogram (MCU) to detect reflux
- a MAG3 renogram (technetium 99m-mercaptotriglycylglycine, Tc99m) to exclude obstruction
- a 99mTc-DMSA (dimercaptosuccinic acid) scan to detect ectopic renal tissue.

Follow-up

Infants will need follow-up with urology or with a nephrologist depending on the findings and the renal function.

Advise

Parents should be advised of the need for prophylactic antibiotics if there is evidence of obstruction or recurrent UTIs.

Duplex kidney

This is a common anomaly, often familial and occurs in 1% of the population.[10] It arises by the duplication of the ureteric bud resulting in two collecting systems with two renal pelves and two ureters (**FIGURE 6**). Duplex kidneys are mostly benign and asymptomatic but can be associated with obstruction and reflux.[5]

In complete duplication the kidney has two moieties and each of these has its own ureter that enters the bladder separately. The upper pole ureter enters the bladder medially and below that of the lower pole ureter. It can also insert ectopically into the urethra or vagina causing dribbling and the development of an ureterocoele (a cystic dilatation of the ureter), which can lead to obstruction and hydronephrosis of the upper pole. The lower pole ureter is at

risk of reflux and scarring. It can also occasionally lead to a PUJ obstruction or dysplasia of the lower pole.[10]

Action

If there is evidence of hydronephrosis, ureteric dilatation or other suspicious features, follow the renal tract dilatation guideline (**FIGURE 4**).

Follow-up

If the infant is asymptomatic with no abnormal signs, further imaging is generally not required. Some centres do offer a routine ultrasound scan at 1-3 months.[5]

Advise

Parents should be advised about early detection and management of UTIs.

Cystic kidney disease

Multicystic dysplastic kidney (MCDK)

This is the commonest and severest form of cystic kidney disease. It is twice as common in males as females and is mostly diagnosed on antenatal scans. The incidence is one in 45,000 for unilateral MCDK with the left kidney being more affected, and one in 20,000 for bilateral MCDK, which is fatal.[6,18] This condition is believed to be secondary to dysfunctional genetics, abnormal differentiation of the metanephros or *in utero* ureteral obstruction.

Within the kidney there are numerous non-communicating cysts separated by dysplastic tissue with very limited functional renal tissue.[18] If the contralateral kidney is normal there should be evidence of compensatory hypertrophy. The contralateral kidney is hydronephrotic in 5-10% of cases, and can be associated with other problems such as VUR, rotational and positional anomalies and dysplastic areas.[6,18]

Action

Babies with an antenatal diagnosis should have an ultrasound scan in the first two weeks of life; a DMSA study at three months to confirm non-function of the affected kidney; and a MAG3 renogram at three months if obstruction in the other kidney is seen on an ultrasound scan.

Follow-up

Usually MCDK disappears (involutes) by the time a child reaches school age. An annual ultrasound scan under the care of the renal team is recommended until regression and then every 2-3 years. Regular blood pressure monitoring and urine analysis are also recommended. Once regression has developed, this could be performed in primary care.

There is a life-time risk of malignancy of one in 2,500. A nephrectomy may be performed to prevent Wilms' tumour.[6]

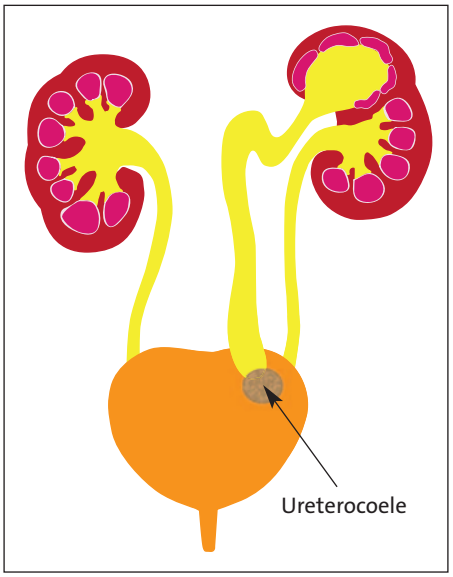

FIGURE 6 A left duplex kidney. The kidney has two moieties, each with its own ureter.

Advise

Parents should be advised that there is no need for antibiotics unless an ultrasound scan shows evidence of hydronephrosis.

Dysplastic kidney (renal dysplasia)

A dysplastic kidney is usually small with unusual architecture on histology with malformed renal parenchymal tissue such as the presence of immature tubules, cysts and non-renal tissue (eg cartilage and fat).[6,18] The incidence is two to four in 1,000 live births.[18] Renal dysplasia can be unilateral or bilateral; for bilateral, the male to female ratio is 1.3:1 and for unilateral 1.9:1. Genetic syndromes that affect multiple body systems can cause kidney dysplasia, however it can also be non-syndromic. There may be an association with posterior urethral valves.

Action

A postnatal ultrasound scan is required to confirm the size of the kidney(s).

Follow-up

The infant should be referred to the renal team for further investigation. If an ultrasound scan shows signs of obstruction further studies will be necessary (MCU/MAG3).

Advise

Antibiotics are not necessary unless the ultrasound scan shows evidence of hydronephrosis.

Posterior urethral valves (PUV)

A PUV is an obstructing membranous fold in the posterior male urethra leading to bladder outlet obstruction in newborn baby boys. Often diagnosed on antenatal scans, this is a serious condition presenting with bilateral renal pelvic dilatations and oligohydramnios. The incidence is one in 5,000-8,000 male live births.[18] PUV is the most common cause of urinary tract obstruction in males.[1] The severity of the obstruction varies; one third of babies will have end stage renal failure.[6] Milder cases of PUV can present in infancy with infection, poor urinary stream and/or a palpable bladder.

Action

Antenatal counselling is essential. A postnatal ultrasound scan and MCU should be performed in the first few days of life. Early catheterisation (per urethra or supra-pubic catheter) by a paediatric surgeon/urologist may be necessary. Refer to a nephrologist especially in the presence of abnormal renal function and hypertension.

Follow-up

Ongoing input from the urology and nephrology teams is necessary. Further ultrasound, MCU and DMSA studies will be needed. The infant may require long-term management for chronic kidney disease.[6]

Conclusions

Various conditions can result in kidney malformations. Fetuses presenting with abnormalities at an antenatal scan should be investigated early in the postnatal period. Identifying these conditions promptly will ensure timely management and can prevent or reduce renal damage, however, a balance is needed between over investigation and treatment to identify high risk babies yet minimise parental anxiety.

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