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Case Report

MULTICYSTIC DYSPLASTIC KIDNEY AND CONTRALATERAL RENAL AGENESIS IN A TWIN GESTATION: A CASE REPORT

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Abstract: Multicystic dysplastic kidney (MCDK) is a common congenital condition in neonates caused by the abnormal formation of renal tissue into multiple fluid-filled cysts that are often dysplastic, affecting kidney function. [1] It has an incidence of 1:4300 live births and is more common in males than females, most often presenting unilaterally with a slightly higher percentage of cases affecting the left kidney. We present a case of a monochorionic diamniotic twin female, with twin A having multicystic dysplastic kidneys on the left and twin B having agenesis of the right kidney. Documented cases involving twins rarely involve both twins and, if so, infrequently in the contralateral kidneys of each twin, highlighting the complexity of renal development in utero.

Keywords: Multicystic dysplastic kidney, Monochorionic diamniotic twins, Renal abnormalities.

INTRODUCTION In MCDK, cysts are noncommunicating and abundant, scattered between dysplastic tissue. The kidneys are usually nonfunctional, and the ureter may be absent or atretic. It arises from a failure of the last step of metanephric differentiation and ureteric bud branching, although the cause is yet to be elucidated. It can be detected as early as 18-20 weeks, appearing as multiple hypoechoic noncommunicating cysts which look like a cluster of grapes [1]. More than half of the cases of unilateral MCDK are expected to involute or regress in the first five years after birth [2], with compensatory hypertrophy of the contralateral kidney, while the other half is expected to develop complications, the most common being VUR. Documented cases of twins have involved mostly one of both infants, with the other twin developing normally without any other organ system abnormalities. These cases occur mostly in males, with the left kidney affected more than 50% of the time.

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CASE PRESENTATION This is a case of monochorionic diamniotic twin girls delivered at 37 weeks of gestation via C-section for antenatal findings of multicystic kidneys on both twins and IUGR in twin B. Their mother is a 21-year-old primigravid who had a history of depression that was managed with Sertraline until four years prior to conception. She was maintained on prenatal vitamins and calcium 2500 mg daily. She had obesity with a pre-pregnancy BMI of 30.1 and anemia with a hemoglobin of 10.3 in the 3rd trimester. Antenatal infectious screening laboratory results were negative except for group B streptococcal culture. She was not in active labor on admission, the rupture of membranes was at delivery, and amniotic fluid was clear. Their APGAR scores were 8 and 8, only needed 2 minutes of blow-by oxygen at birth. The cord pH was normal at 7.27.

At birth, anthropometric measurements were as follows: Twin A with a weight of 2360 g (13.54%), length of 47.5 (37%), head circumference of 31.5 cm (15.94%); twin B with a weight of 2380 g (14.64%), length of 48 cm (40%), head circumference of 31 cm (8.98%). Physical examination of both twins showed a pink, crying baby with flat and open anterior and posterior fontanelles. Their head was normocephalic and atraumatic. Ear placement was appropriate with no malformations; nares were patent, and the palate was intact. The cardiopulmonary examination was normal; the pulses were full and equal.

The abdomen was non-distended, bowel sounds were normal, the liver and spleen were normal in size, and there were no palpable masses. They had normal female external genitalia, and their anus was patent. They moved all his extremities equally, no clavicular crepitus or fracture was palpated, and both Barlow and Ortolani maneuvers were negative. Capillary refill time was 2-3 seconds, and no skin lesions were noted. The tone was normal and primitive reflexes were +2 on all extremities. They did not exhibit any dysmorphic features. Vital signs were stable, with BP at the 24-hour mark at 77/34 for twin A and 75/50 for twin B.

The twins had a 7% discordance antenatally. Unilateral multicystic kidneys were found on both twins on their 26-week ultrasound, twin A on the left and twin B on the right. Level 2 sonogram at 29 weeks ag of gestation showed the same findings on twin A, while there was noted resolution on twin B. Renal ultrasound was repeated after delivery on the 48 hours of life. Twin A had multiple renal cysts, the largest measuring 1.8 x 1.4 x 1.1 cm on the left, mild renal pelviectasis on the right, and the right renal pelvis measuring 4.2 mm (Figure 1). Twin B had no kidney identified on the right, with a normal ultrasound appearance of the kidney on the left (Figure 2).

At eight months, both girls are growing appropriately without episodes or symptoms of UTI and are followed closely by Nephrology. The most recent ultrasound for twin A showed the persistence of the multicystic dysplastic kidney on the left with multiple renal cysts, the largest one measuring 1.9 x 1.8 x 1.6 x cm (Figure 3). The mild renal pelviectasis on the right showed some improvement compared to the one done at four months of age (Figure 3). The most recent ultrasound for twin B showed a normal left kidney measuring 5.7 x 3.0 x 2.9 cm with normal echogenicity, no stones, solid mass, renal cysts, and hydronephrosis (Figure 4).

The right kidney was again not visualized, and there were no cysts within the right renal fossa (Figure 4).

DISCUSSION The urinary system develops in 3 stages: pronephros, mesonephros, and metanephros. The pronephros is a temporary and nonfunctioning system that generates and degenerates at the beginning and the end of 4th week, respectively. This degenerative process is required for normal kidney development. The mesonephros develops into 20 paired collecting tubules and the ureter by the 5th week [3] and eventually fuses

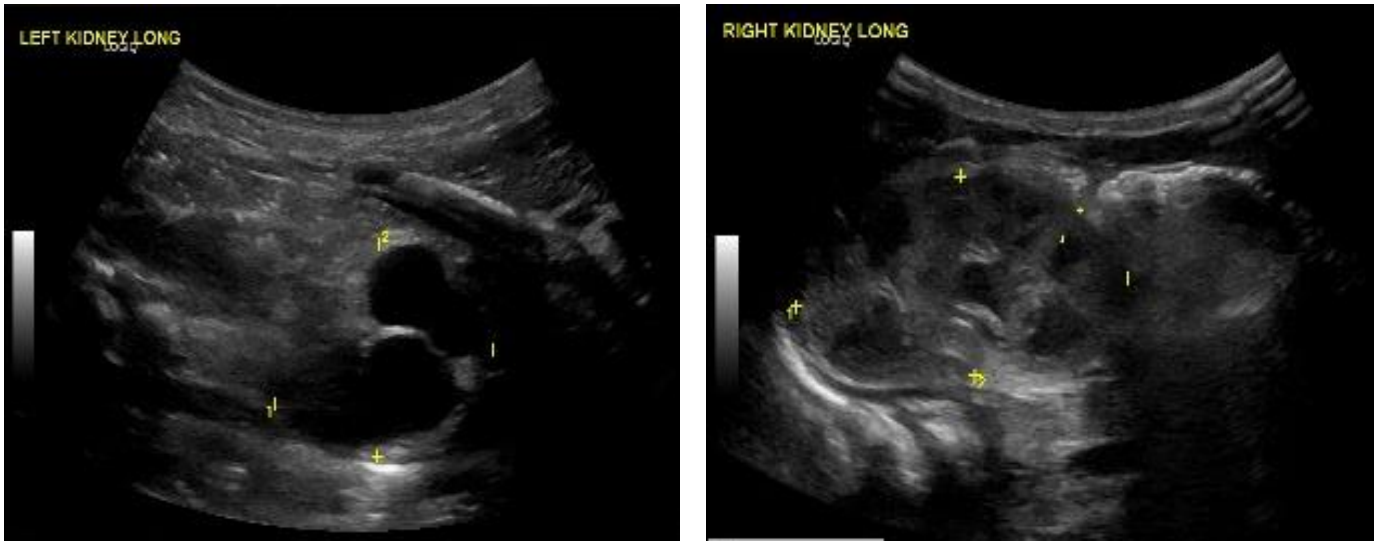

Figure 1. Longitudinal view of the left and right kidney of Twin A on the 48th hour of life. The left kidney measures 3.9 x 2.0 x 2.2 cm with no normal appearing renal parenchyma identified. There are multiple left renal cysts, the largest measuring 1.8 x 1.6 x 1.9 cm. The right kidney measures 5.3 x 2.6 x 3.3 cm with normal echogenicity. There is minimal right pelviectasis with the right renal pelvis measuring 2.6 mm transverse.

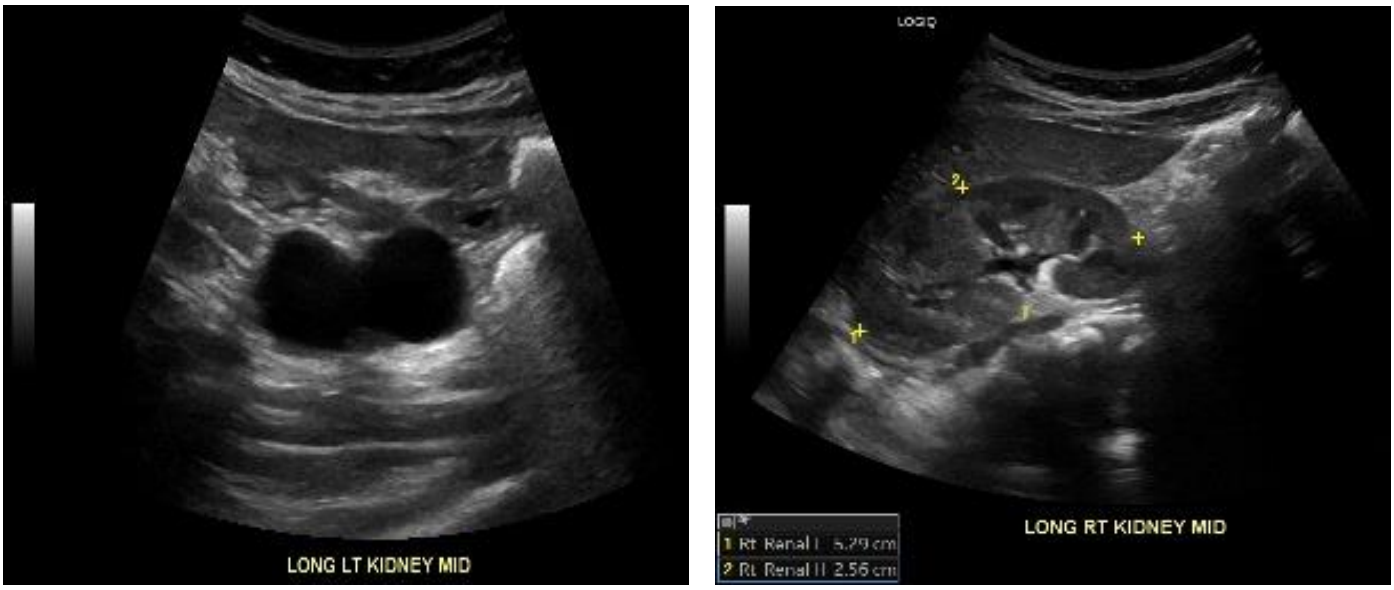

Figure 2. Longitudinal view of the left and right kidney of Twin A at seven months of age. The left kidney measures 3.9 x 2.0 x 2.2 cm with no normal-appearing renal parenchyma identified. There are multiple left renal cysts, the largest measuring 1.8 x 1.6 x 1.9 cm. The right kidney measures 5.3 x 2.6 x 3.3 cm with normal echogenicity. There is minimal right renal pelviectasis, which has improved since the prior exam. The right renal pelvis measures 2.6 mm transverse.

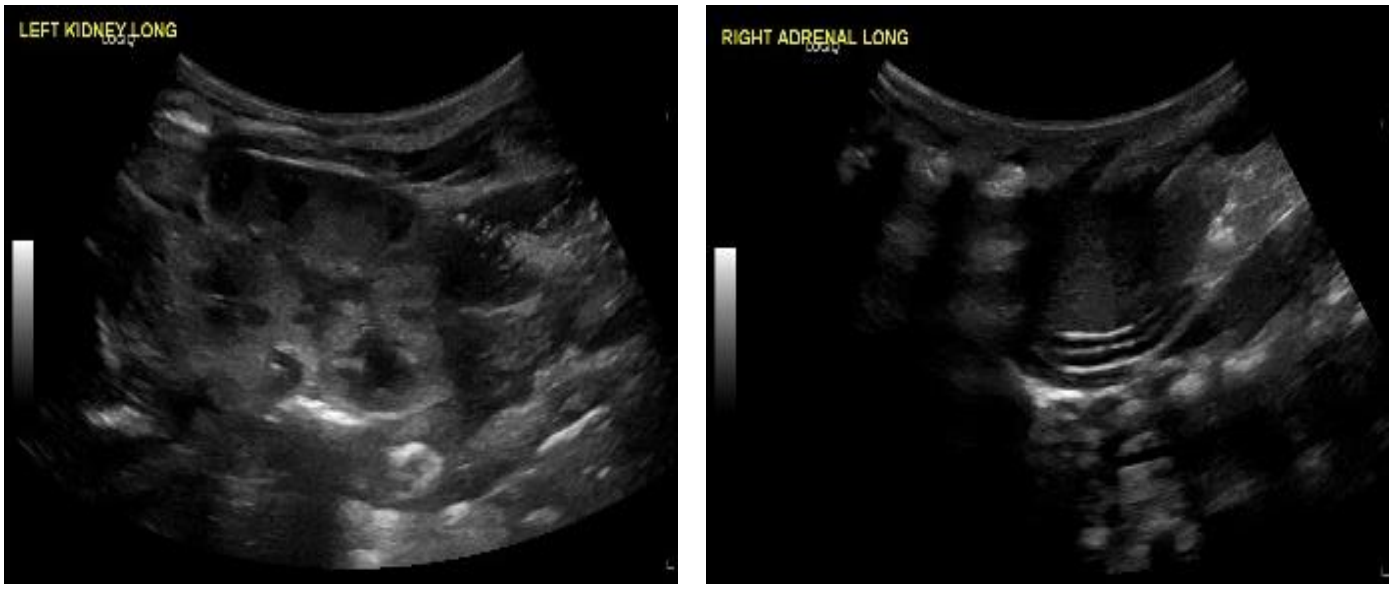

Figure 3. Longitudinal view of the left and right kidney of Twin B on the 48th hour of life. The left kidney measures 4.8 x 2.1 x 1.7 cm with normal echogenicity, no stones, no solid mass, no renal cysts, and no hydronephrosis. Only a normal right adrenal gland was visualized.

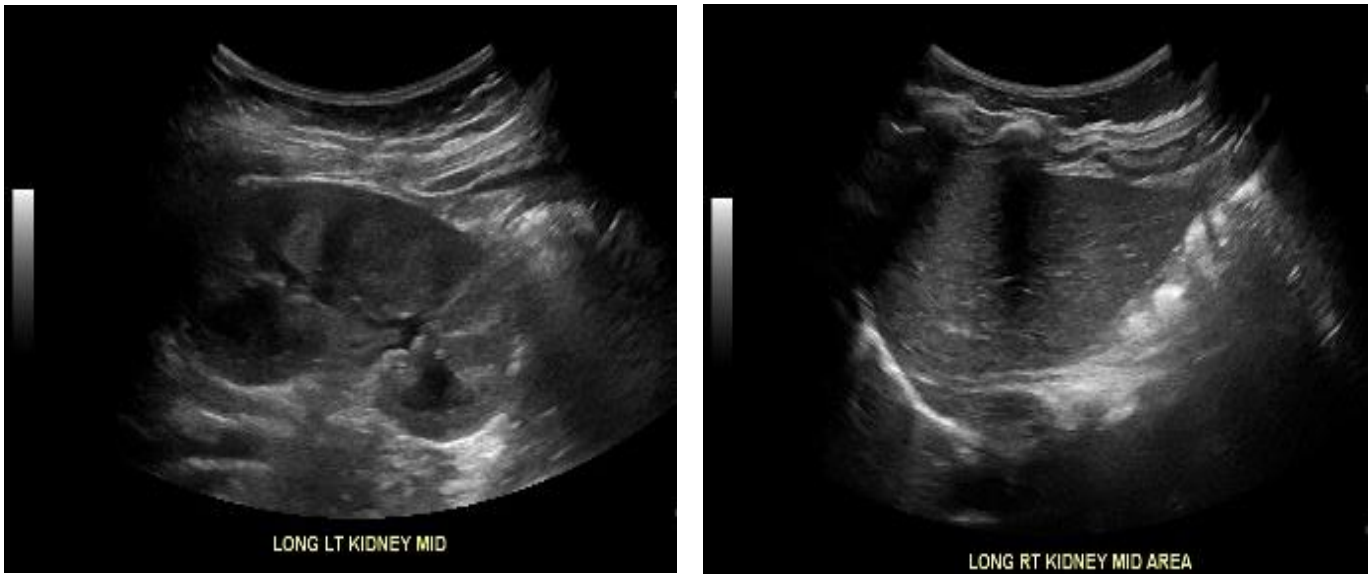

Figure 4. Longitudinal view of the left and right kidney of Twin B at four months of age. The left kidney measures 5.7 x 3.0 x 2.9 cm with normal echogenicity, no stones, no solid mass, no renal cysts, and no hydronephrosis. The right kidney was not visualized, and there were no cysts within the right renal fossa.

with the cloaca to form the urinary bladder. The metanephros is the final stage that produces the permanent kidney from the 5th week of embryonic development. Failure of metanephric differentiation, as well as the branching of the ureteric bud into the collecting ducts and ureter at this stage [4], leads to the sporadic congenital malformation called multicystic dysplastic kidney (MCDK) consisting of multiple, noncommunicating cysts along with immature, undifferentiated and primitive or dysplastic tissue [3, 5].

Multicystic dysplastic kidney (MCDK) is a common congenital condition in neonates caused by the abnormal formation of renal tissue into multiple fluid-filled cysts that are often dysplastic, affecting kidney function. The affected kidney is non-functional in 86% of cases and functions at 7-18% of normal in those that are partly functional [1]. MCDK has an incidence of 1:4300 live births and is more common in males than females, most often presenting unilaterally with a slightly higher percentage of cases affecting the left kidney. It can present as unilateral, bilateral, or segmental. Unilateral and segmental MCDK leads to hypertrophy of the contralateral kidney for compensation. 20-50% of MCDK cases are associated with abnormalities of the contralateral kidney, including vesicoureteral reflux, ureteropelvic junction obstruction, or renal agenesis. Bilateral involvement is incompatible with life as it leads to oligohydramnios, pulmonary hypoplasia, and fetal death [1, 2]. MCDK can be further divided into simple and complex. Simple MCDK is defined as unilateral renal dysplasia without additional genitourinary (GU) abnormalities detected by ultrasound and physical exam aside from hypertrophy of the contralateral kidney, resulting in a very low risk for developing chronic renal insufficiency. Complex MCDK is defined as bilateral renal dysplasia or unilateral renal dysplasia with other GU abnormalities and, therefore, has a worse prognosis. Complex MCDK has a 29% risk of progressing to chronic renal insufficiency and a 21% risk of progressing to end-stage renal disease (ESRD) within seven years [2, 3].

Karapinar et al. described a case in dichorionic diamniotic male twins where one fetus had bilaterally enlarged kidneys with hyperechoic parenchyma surrounded by non-communicating cysts and anhydramniosis. In contrast, the other fetus had normal kidney size and amniotic fluid volume [7]. They were delivered at 37 weeks, but the newborn with renal abnormalities only survived for 12 hours [7].

Campbell described a case of a dichorionic diamniotic twin that delivered spontaneously at 38 weeks.

Twin A, a female, had normal ultrasound findings [8]. Twin B, a male, had a multicystic, left-sided abdominal mass measuring 25 mm in diameter and 35 mm in length with marked oligohydramnios [8]. Each weighed 3100 grams at

birth, but twin B was meconium-stained, required extensive resuscitation, and expired 5 hours later. The autopsy revealed a small dysplastic left kidney with a contralateral right renal aplasia [8].

The cause of MCDK is not clearly understood, with most cases attributed to sporadic malformations, although there can be a genetic component. The classic cluster of multiple non-communicating cysts seen in MCDK directly results from atresia of the ureteral bud, which prevents it from branching into metanephros during embryogenesis, resulting in dysplastic renal parenchyma. As the ureteral bud is crucial to many parts of the GU system, this abnormal development leads to defects in the development of genitalia, hindgut, and cloacal derivatives [1, 2]. Studies analyzing coding exons of genes associated with Congenital Anomalies of the Kidney and Urinary Tract (CAKUT) revealed a correlation with MCDKs and mutations in CHD 1L genes ROBO2, HNF 1B, and SALL1 [4]. MCDK can also be diagnosed as part of a chromosomal abnormality or syndrome, including Hereditary Renal Dysplasia, Potter Sequence, VATER, Williams Syndrome, and branchio-oto-renal syndrome, although most of these causes present with extrarenal pathology [1].

MCDK is routinely diagnosed at the 18–20-week ultrasound, appearing as multiple hypoechoic non-communicating cysts which look like a cluster of grapes. The kidneys can be normal in size, hypoplastic, or grossly enlarged. The affected kidney often lacks a discernable pelvis and calyces and is associated with ureteropelvic junction atresia [1]. Most unilateral MCDKs (50 – 60%) will involute or regress within the first 3-5 years of life, with 10% of prenatally detected MCDKs involuting before initial postnatal ultrasound [2]. Once the diagnosis of MCDK is made, additional testing can be performed, including high-resolution anatomy ultrasound to examine kidney and GU system function, MRI to assess fetal anatomy and look for extrarenal pathology, fetal echocardiography, and amniocentesis or chromosomal analysis to look for anomalies [1].

The prognosis for MCDK depends on the function of the contralateral kidney [1]. Studies have shown that 48% of affected kidneys will involute with hypertrophy of the contralateral kidney in 93% of unilateral MCDK cases, allowing for preserved renal function [2]. Prenatal factors like kidney size, location, and amniotic fluid volume have not shown a correlation to prognosis for MCDK [1].

The most common complication of MCDK is vesicoureteral reflux in the contralateral kidney, which has been found in up to 30% of unilateral MCDK cases. This can lead to renal scarring, which can affect renal function. Other GU complications include increased risk of UTIs, ureterocele, pyeloureteric stenosis, genital abnormalities, and cryptorchidism. Non-GU complications include fetal hydronephrosis, proteinuria, hypertension, malignancy, chronic renal insufficiency, and end-stage renal disease. Pathology associated with syndromes includes bilateral cleft lip, microcephaly, micrognathia, and complicated congenital heart disease [1][2]. More rarely, MCDK can cause respiratory distress, gastrointestinal obstruction, and contralateral ureteral obstruction due to mass effect [3].

Monsoor *et al.* conducted a single-center, retrospective study of children with unilateral multicystic dysplastic kidney to describe the long-term renal outcome of children diagnosed between 1985 and 2009 in Holtz Children's Hospital in Miami [9]. Among 121 patients with MCDK, complete involution was observed in 60% within five years, while 20% underwent nephrectomy [9]. The contralateral kidney had vesicoureteral reflux in 16.8% of patients, with 6 having high-grade VUR (grade 4-5). One patient had a hypoplastic contralateral kidney [9]. Schreuder *et al.* had similar findings when they conducted a meta-analysis of 67 cohorts with over 3500 patients with unilateral MCDK [10]. VUR was found in 19.7%, with 40% in the contralateral kidney, while 77% had hypertrophy of the contralateral kidney after a ten-year follow-up [10].

While there is no consensus on treatment, the options are conservative management with frequent follow-up or nephrectomy based on an individualized approach. Antibiotic prophylaxis for UTIs is given until a thorough physical exam, and postnatal renal function testing can be done. Most cases of MCDK are treated from a multi-disciplinary approach, including Maternal Fetal Medicine, pediatric nephrology and urology, and genetic counselors and neonatologists. Conservative management requires long-term follow-up every six months with pediatric nephrology for regular renal ultrasounds and renal function tests, in addition to monitoring for hypertension and malignancy. While MCDKs often regress on their own, if they grow or present with more complications, a nephrectomy can be pursued to avoid adverse outcomes. Although previously performed at the time of diagnosis, nephrectomy is no longer practiced unless the affected kidney is large enough to cause symptoms or if obstruction at the ureteropelvic junction with no renal function is a

concern [9]. Bilateral MCDK would require long-term dialysis until a kidney transplant is possible [1, 2].

In a retrospective review of 53 cases of prenatally diagnosed MCDK; Balasundaram *et al*. found that unilateral disease in the absence of significantly associated findings has a good prognosis and outcome. In contrast, those with concomitant extrarenal or contralateral anomalies, bilateral disease, or anhydramnios have a higher predisposition to needing dialysis in the future [13].

CONCLUSION This report highlights a rare occurrence of multicystic dysplastic kidneys on the opposing sides of term twin females. Prior cases that have been documented and described rarely involve both twins and, if so, infrequently in the contralateral kidneys of each twin, highlighting the complexity of renal development in utero. Female gender defies the male predominance of this disease entity. Close follow-up, parental counseling, and guidance are keys to preventing complications and long-term effects of kidney disease.

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