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Case report

Multicystic renal dysplasia; a perinatal autopsy examination of a neonate

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Abstract

Multicystic renal dysplasia is a rare developmental anomaly of the kidney, often associated with urinary tract obstruction and incompatible with long-term survival when bilateral. Early recognition through antenatal imaging and confirmation by post-mortem examination are crucial for accurate diagnosis and genetic counselling. We present a case of a 31-year-old P2C0 mother who delivered a baby at 36 weeks of gestation. The antenatal ultrasound scan performed at 16 weeks revealed oligohydramnios, and a subsequent foetal anomaly scan showed multicystic kidneys with absent liquor. Further imaging suggested lower urinary tract obstruction, leading to progressive renal impairment and pulmonary hypoplasia. The baby died a few hours after birth. Post-mortem examination revealed a non-syndromic baby with bilateral cystic kidneys, left ureteric atresia, lung hypoplasia, and a large ostium secundum atrial septal defect. Microscopically, both kidneys showed variably sized cysts lined by cuboidal epithelial cells, surrounded by immature mesenchymal stroma with primitive tubules, glomerular structures, and focal cartilage islands. The diagnosis of bilateral multicystic renal dysplasia with left ureteric atresia was made. This case highlights the diagnostic value of foetal autopsy in elucidating the underlying pathology of congenital renal anomalies and its pivotal role in reproductive counselling for future pregnancies.

Keywords: multicystic renal dysplasia, lung hypoplasia, foetal autopsy

Introduction

Neonatal and paediatric renal cystic diseases comprise a variety of hereditary and non-hereditary conditions characterized by the presence of cysts in one or both kidneys (1). Cystic kidneys are the most recognized urinary tract anomalies found in autopsies of stillborn and neonatal deaths (1). Non-hereditary cystic diseases include simple and complex renal cysts, multicystic renal dysplasia (MCRD), obstructive cystic dysplasia and medullary sponge kidney. Hereditary cystic renal diseases include autosomal recessive polycystic kidney disease, autosomal dominant polycystic kidney disease, nephronophthisis and syndromic conditions such as tuberous sclerosis and Von Hippel-Lindau syndrome (1).

Multicystic renal dysplasia (MCRD) is the most commonly recognized cause of enlarged non-functioning cystic kidney, diagnosed on antenatal ultrasound scan (2). It is a congenital anomaly, most commonly occurring in males. It usually affects one kidney, but bilateral renal

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involvement is not rare. Bilateral multicystic dysplastic kidneys (MCDK) are not compatible with life. Two theories on the aetiology have been described. One theory is blockage in the genitourinary tract causing ballooning effect on the renal tubules resulting in the formation of multiple cysts. Functional renal tissue does not develop. The other theory is failure of differentiation of embryonal mesoderm into nephrogenic epithelium because of a signal transduction error in development. The characteristic feature is multiple non-communicating cysts in the renal parenchyma (3). Ultrasound scan plays a major role in diagnosing MCRD. It is diagnosed in 1 in 4000 neonates and early detection is very important to plan the subsequent management and follow up (3).

We report a case of multicystic renal dysplasia diagnosed at neonatal postmortem examination in a baby who was found to have multicystic kidneys in the antenatal ultrasound scan.

Case History

A 31-year-old P2C0 mother with a history of gestational diabetes mellitus delivered the baby at 36 weeks of period of amenorrhea (POA). She had no history of pregnancy induced hypertension. She had a history of a first trimester miscarriage. Regarding this pregnancy, oligohydramnios was detected during the antenatal ultrasound scan performed at 16-week of POA. The subsequent foetal anomaly scan revealed multicystic kidneys with absent liquor. Further imaging suggested lower urinary tract obstruction, leading to progressive renal impairment and pulmonary hypoplasia.

This mother presented in preterm labour with breech presentation at POA of 36 weeks. Emergency lower segment caesarean section was done. The baby had poor respiratory effort and expired a few hours after birth. A pathological autopsy was carried out.

The body was that of a non-dysmorphic, female neonate. Foetal weight and measurements were as follows: body weight: 2025g (between 50th and 10th centile); occipito-frontal circumference: 300mm; crown rump length: 280mm (standard for the stated gestation 318+/-39mm); crown heal length: 410mm (standard for the stated gestation 434+/-59mm); foot length: 62mm (standard for the stated gestation 69+/-11mm).The head was normal in size and shape. The eyes, nose, lips, palate, mouth and ears were normally formed. There are no features of Potter facies such as recessed chin, far apart eyes, flat nasal bridge and low set ears. The neck was of normal length and shape. The chest, abdomen and spine were normally formed. The abdomen was distended. The anus was patent, and the external genitalia were of a normal female. All four limbs were symmetrical, having normal length and have five normal sized separate digits (Figure 1).

Figure 1: External appearance of the baby with distended abdomen

On internal examination, both lungs were very small (Figure 2), and lung hypoplasia was present. The lungs showed normal lobation. The heart had normal shape and size. It showed normal atrial situs and normal ventricles with normal interrelationship of aorta and pulmonary trunk. The atria and ventricles were normal in size and in wall thickness. There was a large ostium secondum atrial septal defect. The interventricular septae was normally formed and the ductus arteriosus was closed. The kidneys were enlarged and distorted by variably sized cysts (Figure 2B).

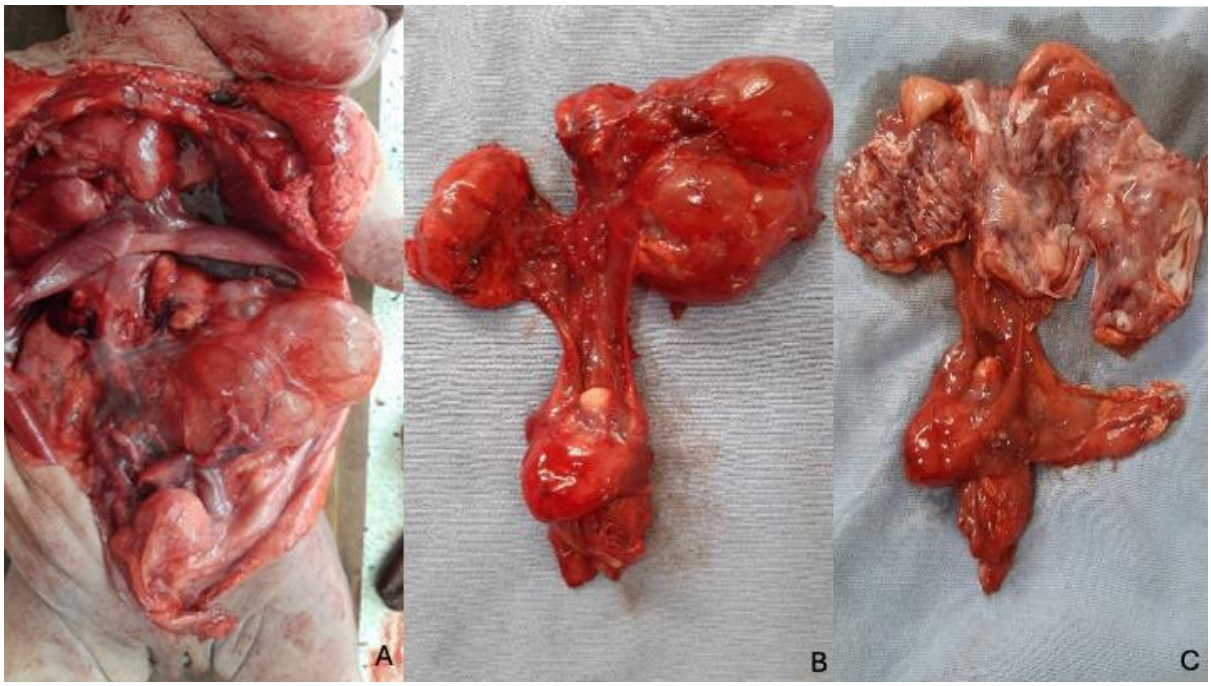

Figure 2: A. Small lungs and bilateral cystic kidneys, B. Bilateral cystic kidneys, C. Bilateral cystic kidneys – cut surface

The left kidney was larger than the right kidney (Figure 2B). The left kidney measured 5x4x3.5cm and the right kidney measured 4x3.5x3cm. The cysts were more prominent in the left kidney. The cut surfaces of both kidneys showed variably sized cysts with indistinct corticomedullary junction (Figure 2C). The adrenals were present in their normal position and were normal in size. The left ureter was atretic and the right ureter was normally formed. The bladder was normal and there was no hypertrophy of bladder wall. The placenta was not received for pathological examination.

On microscopic examination both kidneys showed variably sized cysts lined by cuboidal epithelial cells (Figure 3 A & B), surrounded by immature stromal elements. Scattered primitive tubules and glomerular structures were present. Islands of immature cartilage were present (Figure 3 C and D). Thymus, myocardium, lungs, liver, spleen and brain demonstrated tissue with appropriate development for stated gestation.

Discussion

Multicystic dysplastic kidneys (MCDK) are usually non-functional and contain multiple variable sized non-communicating cysts. Macroscopically the affected kidney has the appearance similar to that of a bunch of grapes (4). The cysts are separated by dysplastic renal parenchyma and there is an abnormal pelvicalyceal system. Histologically, cysts, primitive ducts and the metaplastic cartilage are the main characteristic features. If metaplastic cartilage is not seen, it is very important to take multiple sections to find a focus of metaplastic cartilage (4). In some cases, undifferentiated mesenchyme can be seen. Most cases are associated with ureteropelvic obstruction, ureteral agenesis, ureteral atresia or reflux and horseshoe kidney (4). MCDK can also be associated with several malformations including cardiac malformations, intestinal abnormalities, metabolic abnormalities and genetic syndromes. Bilateral renal dysplasia is not compatible with life. It usually results in stillbirth and is associated with lung hypoplasia and oligohydramnios as in this case (4,5). Most of the time these are diagnosed antenatally. Ultrasonically the cysts involve the whole kidney, and the larger cysts are usually located in the periphery. In this case, the baby was non-syndromic. Postmortem examination revealed bilateral enlarged kidneys with a

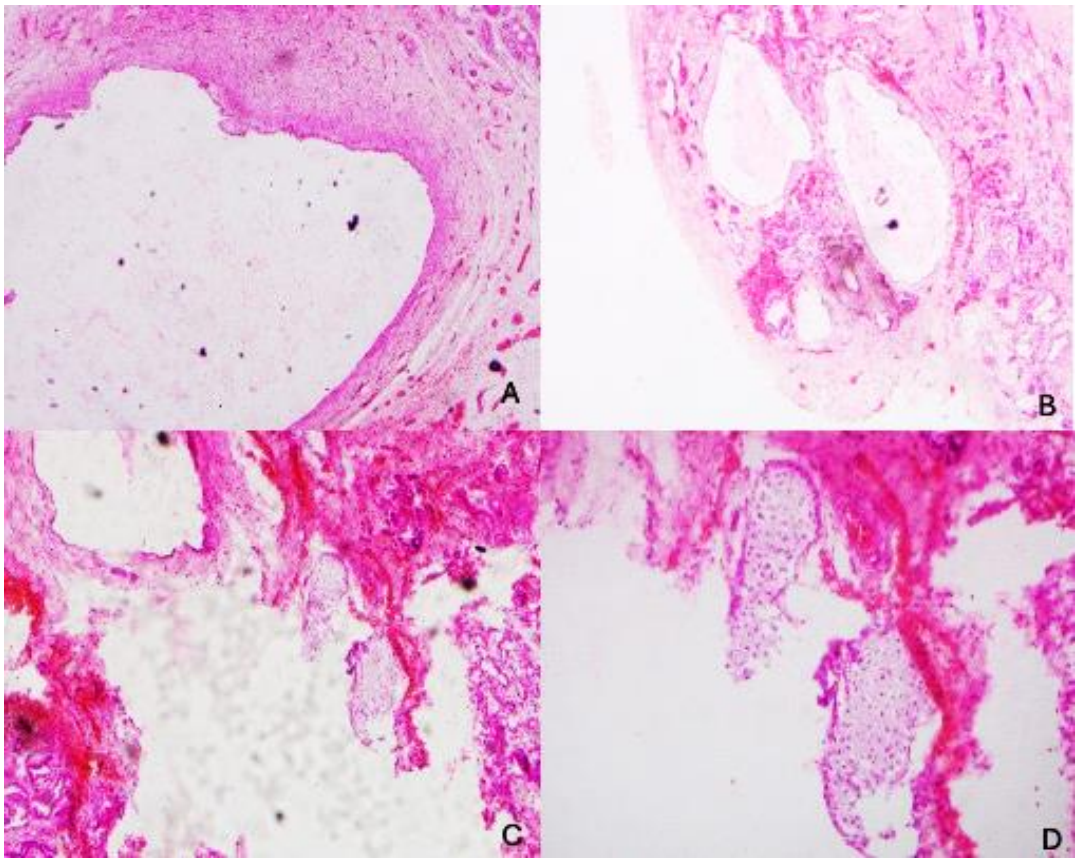

Figure 3: A. Left kidney showing large renal cysts (Haematoxylin and eosin (H&E x40), B. Right kidney showing small cysts (H&E x40), C. Islands of immature cartilage (H&E x100), D. Islands of immature cartilage (H &W x200)

prominent enlargement of left kidney, with multiple bulging large cysts. The cysts were filled with serous fluid. Left ureteric atresia was also present and there was bilateral lung hypoplasia and a large ostium secondum ASD. The above mentioned characteristic histological features were also found. Therefore, the final diagnosis of multicystic renal dysplasia was made.

The differential diagnosis considered were paediatric nephroma and ciliopathies including autosomal recessive polycystic kidney disease and autosomal dominant polycystic kidney disease (4).

When considering the paediatric cystic nephroma, it is an exclusively cystic neoplasm of young children of less than 24 months of age. The lesion shows a pseudo-capsule, and the cysts are varying in size and are lined by flattened, cuboidal, hobnailed or denuded epithelium. The septa between the cysts contain well differentiated tubules. Immature nephroblastic elements are not seen. These tumours can show *DICER1* mutations (6,7). The presence of metaplastic cartilage and immature mesenchymal elements and the characteristic macroscopy excludes the diagnosis of cystic nephroma.

Autosomal recessive polycystic kidney disease (ARPKD) is associated with the mutations in polycystic kidney hepatic disease gene (*PKHD1*) and is characterized by bilateral markedly enlarged kidneys without altering the reniform configuration. The cut surfaces show multiple linear cysts, radiating from medulla to outer cortex. The cysts appear as dilated tubular structures lined by cuboidal or flattened epithelium. The intervening tissue shows uninvolved nephrons depending on the severity. This can be associated with hepatic disease, mainly hepatic fibrosis. In this case

there were no cysts with a linear arrangement and the liver histology was normal. Therefore, autosomal recessive polycystic kidney disease was excluded.

Autosomal dominant polycystic kidney disease (ADPKD) is less common in foetuses and newborns when compared to the adults and it is characterized by expanding cysts progressively destroying the renal parenchyma. Mutations are associated with two genes, *PKD1* and *PKD2*. The cysts develop in all segments of renal tubule and glomerular capsule. As they enlarge, they get disconnected from the tubular origin. The intervening renal parenchyma shows interstitial scarring and tubular atrophy microscopically. The other abnormalities that can be associated with ADPKD are hepatic and pancreatic cysts, coronary artery aneurysms and abnormalities in cardiac valves. (6,7). The above-mentioned features were not observed in this baby.

MCDK is a birth defect that happens during intrauterine development. It is not an inherited condition. Various teratogens and in-utero exposure to various viral infections including cytomegalovirus, enterovirus and adenovirus are thought to be associated with the disease origin (4). However, still a definite aetiology is not understood.

Since this condition is not a hereditary cystic renal disease, the risk of getting MCDK in subsequent pregnancies is very low. This information is very important in counselling the parents after the still birth and planning a subsequent pregnancy and emphasizes the value of performing foetal autopsies.

Conclusion

Multicystic renal dysplasia is a rare, non-hereditary cystic renal malformation that is usually detected antenatally through foetal ultrasound. The diagnosis is predominantly established macroscopically during foetal or perinatal autopsy, with microscopic findings providing supportive evidence. A thorough post-mortem examination is essential not only to confirm the diagnosis but also to exclude associated anomalies and to provide valuable information for obstetric counselling and management of future pregnancies.

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