

Original Article:**Multicystic dysplastic kidney: a retrospective study with clinicopathological correlation**

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ABSTRACT

Background: Multicystic dysplastic kidney (MCDK) is one of the most frequently observed congenital urinary tract abnormalities.

Methods: Retrospective study of MCDK in nephrectomy specimens seen over a 20 year period at a tertiary care teaching hospital in South India.

Results: MCDK was evident in 6 of the 230 nephrectomy specimens (2.6%) seen during the 20 year period of study. Their median age [interquartile range (IQR)] was 2.25 (1.075-7.750) years; there were 3 males. MCDK more frequently involved the right kidney (5/6); 4 cases presented with megaureter. Salient histopathological findings included cystically dilated spaces lined by flattened to cuboidal lining epithelium with intervening stroma showing entrapped, dilated, narrowed immature tubules with surrounding smooth muscle cuffing.

Conclusions: Histopathological examination remains the mainstay in the diagnosis and helps in differentiating MCDK from other cystic lesions.

Key words: *Multicystic dysplastic kidney, Clinicopathological correlation*

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INTRODUCTION

Dysplasia is a developmental disorder and may affect several organs in embryonic life.¹ Multicystic dysplastic kidney (MCDK) is one of the most frequently observed congenital urinary tract abnormalities.² It is also referred to as renal dysplasia, renal dysgenesis, multicystic kidney and potters type II renal cystic disease. It is found in 2% of paediatric autopsies and the incidence is one in 3640-4300 live births.² It is seen in 10% of refluxing kidneys. MCDK occurs most often sporadically, but familial cases sometimes occur but no significant recurrence risk has been observed for future pregnancies.³ The inheritance pattern is autosomal dominant with incomplete penetrance. Majority (77% - 88%) of MCDK cases are diagnosed by prenatal ultrasonography usually during the third trimester, but can be detected in second trimester also.⁴ MCDK is most commonly seen in boys usually with a left-sided unilateral involvement. Less commonly, it pre-

sents in older children/adults (age range 12-70 years). Bilateral presentation and focal/segmental involvement are occasionally observed.^{5,6} MCDK is commonly associated with abnormalities like ureteral atresia, pyelocaliceal obstruction, vesico-ureteral reflux, uretero pelvic junction obstruction and uretero-vesical junction obstruction.^{4,6,7} We present a retrospective analysis of 6 cases of MCDK seen at our tertiary care teaching hospital.

MATERIAL AND METHODS

We retrospectively reviewed the medical records of patients who had undergone nephrectomy for various indications during the 20-year period from March 1992 to March 2012 at Sri Venkateswara Institute of Medical Sciences (SVIMS), Tirupati a tertiary care teaching hospital in South India. Of these, we identified cases that were diagnosed to have MCDK on histopathological examination of the nephre-

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ctomy specimens. The ultrasonographic findings of these patients were obtained from the records of the Department of Radiodiagnosis.

RESULTS

MCDK was evident in 6 of the 230 (2.6%) nephrectomy specimens that were evaluated during the study period.

Their median age [interquartile range (IQR)] was 2.25 (1.075-7.750) years. The youngest patient was a one-year-old female child and the oldest was a ten year old girl. There were 3 males. Out of the six cases, only one case presented with recurrent urinary tract infection (UTI) characterized by fever, chills, pyuria and terminal haematuria. Four cases presented with abdominal mass and one case was detected antenatally as persisting hydronephrosis. All the cases had undergone ultrasonography (Table 1). On ultrasonography, right side involvement was seen in 4 patients while left side involvement and bilateral involvement were observed in one patient each. Gross pathological examination revealed that 3 nephrectomy specimens were hypoplastic and one was rudimentary and the other two were multicystic with a nodular outer surface (Figure 1) and the cut surface revealed multiple cystic spaces of varying sizes (Figure 2) filled with clear fluid. Salient histopathological findings include cystically dilated



Figure 1: Gross pathology photograph of multicystic right nephrectomy specimen measuring 4.5 x 3 x 1 cm with attached dilated ureter measuring 1.5 cm



Figure 2: Cut-section of nephrectomy specimen reveals multiple cysts of varying sizes with intervening grey white stromal tissue

spaces lined by flattened to cuboidal lining epithelium with intervening stroma showing entrapped, dilated, narrowed immature tubules with surrounding smooth muscle cuffing. Focal cystic glomerular collections, nerve bundles, cartilage and congested blood vessels were also present (Figures 3 and 4).

Table 1 : Ultrasonography findings of the six MCDK cases

Case No.	Age (years)	Sex	Kidney	USG findings	Follow-up
1	1	Female	Right	Right dysplastic kidney with associated megaureter and left hydroureteronephrosis	Lost
2	7	Female	Right	Right dysplastic kidney with associated megaureter and left hydroureteronephrosis	8 months; no complaints
3	1.5	Male	Left	Left dysplastic kidney and right hydroureteronephrosis	Lost
4	3	Male	Right	Right non-functioning kidney with reflux megaureter and left primary obstructing megaureter	Lost
5	1.1	Male	Right	Right hydroureteronephrosis and left normal kidney	5 years; no complaints
6	10	Female	Right	Bilateral megaureters with right hydroureteronephrosis	Lost

MCDK=multicystic dysplastic kidney; USG=ultrasonography

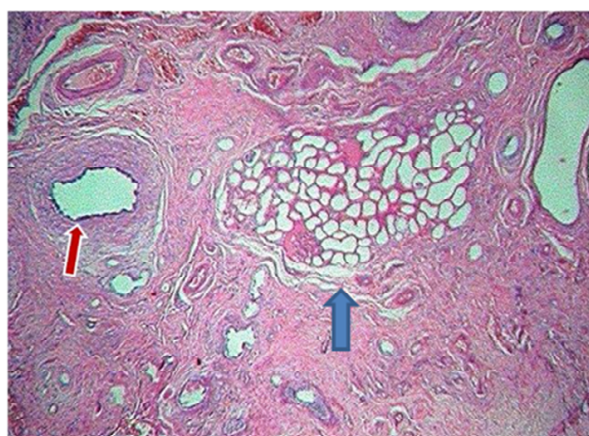


Figure 3: Photomicrograph showing dilated cysts (thin arrow) lined by cuboidal to flattened lining epithelium with surrounding smooth muscle cuffing and cystic glomerular collections (thick arrow) (Haematoxylin and eosin, $\times 50$)

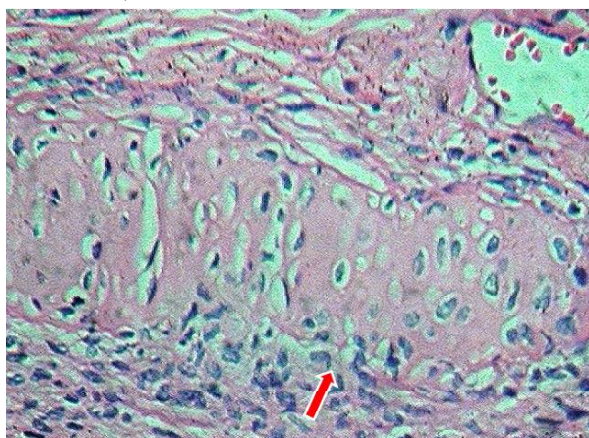


Figure 4: Photomicrograph showing cartilage (arrow) and congested thin walled blood vessel (Haematoxylin and eosin, $\times 400$)

DISCUSSION

In 1936 Schwartz first described MCDK in a seven month old child.⁸ The characteristics of the disease was first described by Spence in 1955.⁹ In MCDK the kidney has the appearance of a bunch of grapes with little stroma between the cysts. Morphologically it is characterized by replacement of normal architecture of the kidney with numerous cysts, undifferentiated epithelium and primitive ducts with surrounding fibromuscular connective tissue.¹⁰

MCDK is the most common type of renal cystic disease and the second most common cause of an abdominal mass in a new born after hydronephrosis. It results from abnormal differentiation of metanephros, possibly as a result

of disturbed connection of ureteric bud with renal blastema and abnormal division at the stage of metanephros.^{10,11} Various aetiological factors have been said to be associated with MCDK, including teratogens like intrauterine viral infections and medications. In one study it was reported that 1% - 3% of children with congenital kidney malformations had amniotic fluid culture positive for enterovirus, cytomegalovirus and adenovirus.¹² In another study four infants with MCDK were born to epileptic mothers treated with antiepileptics during pregnancy.¹³

In our study the age of the patients ranged from 1-10 years and both genders were equally affected. In two other studies^{2,14} male preponderance was observed. While left kidney is more commonly involved^{2,14}, we found right kidney was more frequently involved in five out of the six cases. In our study we found unilateral involvement more frequently (5 out of 6). Similar observations were found in another study.¹⁴

Many coexistent urinary tract abnormalities have been described in patients with MCDK, the most common being vesico-ureteral reflux (VUR) to the contralateral kidney. In a study (n=143) of patients who underwent vesicocystourethrogram VUR was observed to occur in 19% to the contralateral kidney and 16% to the kidney affected with MCDK.¹⁵ In our study four cases had associated megaureters and five cases had hydronephrosis. MCDK associated with contralateral ureteric stenosis with hydronephrosis has been described.¹⁶ MCDK is also associated with multiple congenital abnormalities in other organ systems. The anomalies of gastrointestinal tract like transoesophageal fistula, jejunal stenosis and anal atresia, cardiovascular anomalies like ventricular septal defect, patent ductus arteriosus, truncus arteriosus and venous haemangiomas. One study¹⁶ (n=10) reported multiple congenital anomalies predominantly affecting musculoskeletal system, three cases

had pulmonary hypoplasia, three had meningoencephalocele and one each with phocomelia and syringomyelia. In our study, no such congenital anomalies were found. Urinary tract abnormalities like contralateral uretero pelvic junction obstruction has been observed in 7%-15% cases.¹⁷ MCDK tend to involute with involution rates varying from 19% -74% over nine months to 10 years. The involution is so severe that the affected kidneys show compensatory hypertrophy.¹⁸

The clinical presentation may vary widely with most patients presenting with non-tender abdominal mass, infection or hypertension. Rarely it may present with haematuria. In our study one case presented with recurrent UTI with pyuria and terminal haematuria, four cases with abdominal masses and one case with persistent hydronephrosis and dysuria. In a study of 887 patients with MCDK only six cases (0.7%) had hypertension.¹⁹ In other studies four of the 15 cases¹⁴ and three of 22 cases¹⁶ were found to have hypertension. In contrast to these studies, none of the cases in the present study had hypertension. Several studies^{8,20} including the report from the multicystic kidney registry in 1993⁸ have demonstrated the incidence of hypertension to be less than 1%. A much higher incidence of hypertension was reported in another study.²¹ MCDK is most often identified by prenatal ultrasonography and in newborn ultrasonography is usually repeated within a few days or a month later to confirm it and also to evaluate the contralateral kidney and bladder. In a few cases it is difficult to differentiate MCDK from severe hydronephrosis.^{22,23} MCDK has a haphazard distribution of cysts of varying sizes without a large or central medial cyst and without visible communication and presence of small cysts in between large cysts. In ureteropelvic junction obstruction the cysts are seen at the periphery of kidney and interconnected with the central/medial cyst and

absence of small cysts in between large cysts. In the difficult cases radioisotope studies may be helpful. Hydronephrotic kidneys show some function on dimercaptosuccinic acid (DMSA) or Technetium^{99m} mercaptoacetyl triglycine, whereas the renal uptake is seldom seen in MCDK. In our study ultrasonography findings revealed dysplastic kidneys (n=3), hydro-ureteronephrosis (n=2) and nonfunctioning kidney (n=1). Four were associated with megaureters.

Grossly the kidney in MCDK may be either enlarged, multicystic, losing its reniform shape or it may be hypoplastic/small or rudimentary. In our study three cases were hypoplastic, two were multicystic and one was rudimentary. Histological features considered specific for MCDK are collections of primitive ducts with surrounding smooth muscle condensation, foci of cartilage, fetal/primitive glomeruli and primitive tubules and it needs to be differentiated from cystic nephroma, cystic partially differentiated nephroblastoma and polycystic kidney disease. Cystic nephroma and cystic partially differentiated nephroblastoma are sharply encapsulated neoplasms with no normal renal elements inside the capsule, whereas in MCDK grossly there is loss of reniform shape and microscopically there is presence of primitive ducts, mal-developed nephrons and cartilage in one third of the cases. In polycystic kidney disease bilateral involvement and grossly markedly enlarged kidneys retaining reniform configuration is seen. Microscopically cysts are dilated tubular structures with intervening uninvolved nephrons. No primitive tubules with smooth muscle cuffing, primitive ducts or cartilage could be seen. Histopathologically in our study all cases revealed primitive ducts with smooth muscle cuffing, primitive glomerular collections and thickened blood vessels and only three cases showed cartilage and nerve bundles.

Studies show that MCDK cases may undergo malignant transformation. Case reports of Wilms' tumour, renal cell carcinoma and urothelial carcinoma arising from MCDK have been published.^{24,25} In our study no such malignant transformation was observed. Of the six cases, four were lost for follow-up. Of the two cases who could be followed-up, one is a male child coming for follow-up for the past five years with ultrasonography showing normal left kidney and the other is a female child coming for follow-up for the past eight months with ultrasonography showing moderate dilatation of left ureter.

Though MCDK is a frequently encountered congenital abnormality, only a few case studies were reported in Indian literature. The clinical presentation may range from being asymptomatic to neonatal death, abdominal lump, recurrent infections or hypertension. It may be associated with multiple congenital anomalies. Histopathological examination remains the mainstay in the diagnosis and helps in differentiating MCDK from other cystic lesions.

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