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Review Paper

Multicystic Dysplastic Kidney Disease: A Review

Mohsen Akhavan Sepahi[1, 2*], Mohammad Hoseinn Akhavan Sepahi[3], Ali Akhavan Sepahi[4]

1. Department of Pediatric Nephrology, School of Medicine, Qom University of Medical Sciences, Qom, Iran.
2. Clinical Research Development Unit, Hazrat-E Fatemeh Masoumeh Hospital, Qom University of Medical Sciences, Qom, Iran.
3. Department of Dentistry, School of Medicine, A.I. Yevdokimov Moscow State University of Medical and Dentistry Sciences, Moscow, Russia.
4. Federal State Budgetary Educational Institution of Higher Education, A.I. Yevdokimov Moscow State University of Medicine and Dentistry, Moscow, Russia.

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Corresponding Author:
Mohsen Akhavan Sepahi, MD.
Address: Department of Pediatric Nephrology, School of Medicine, Qom University of Medical Sciences, Qom, Iran.
E-mail: akhavansepahimm@gmail.com

ABSTRACT

The cystic kidney is one of the most common abnormalities detectable by ultrasound before and after birth and is also a cause of hydronephrosis; therefore, ultrasound is necessary for accurate diagnosis. Multicystic dysplastic kidney (MCDK) is a congenital disease in which the kidney is filled with multiple cysts and does not function properly. This article describes an article review of MCDK and new attention. This study aimed to increase awareness of the importance of diseases that cause kidney cysts, especially dysplastic multicystic kidney disease, to promote understanding of diagnostic methods before and after birth, and to investigate its cause.

Keywords: Multicystic dysplastic kidney (MCDK), Hematuria, Vesicoureteral reflux (VUR), Imaging

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Introduction

Multicystic dysplastic kidney (MCDK) is a condition that develops during fetal development and leads to kidney abnormalities [1, 2]. MCDK is a disease that causes kidney cysts and abdominal masses in infants and children; however, most kidneys are insensitive at birth [1-5]. The prevalence of MCDK across studies ranges from 1 in 1000 to 1 in 4300 live births, with a higher prevalence in males, and predominantly affecting the left side [2-4]. In a 20-week fetal ultrasound, prenatal diagnosis of MCDK is possible by observing multiple cysts, which are rarely detectable using other diagnostic methods [3]. MCDK is a rare congenital disease in which the normal kidney contralateral to an MCDK exhibits a unique growth pattern in intrauterine and postnatal life. In prenatal diagnosis of dysplastic changes and cyst formation in the kidney during the fetal period, the developmental mechanism is currently unclear, unknown, and confusing [6, 7]. Renal changes include a collection of thin-walled, multiple non-communicating, irregular cysts of various sizes with little or no functioning renal parenchyma [1, 4, 7-9]. The classic features of Potter syndrome in infants are bilateral and fatal. In most cases, one kidney fails to develop properly and is affected unilaterally, with the left kidney being preferred [3, 4]. The majority of MCDK cases resolve over time [10]. A total of 94.1% of MCDK cases are diagnosed using prenatal ultrasound [11]. MCDK is a common cause of childhood endstage renal failure [12]. Up to 30% of MCDK cases have other genitourinary tract abnormalities, such as contralateral renal agenesis, vesicoureteral reflux (VUR), ureteral ectopy, ureterocoele, renal hypoplasia, ureteral-pelvic or ureteral-vesical junction obstruction, and VUR [8, 10, 13-17]. MCDK may present with symptoms, such as flank pain, recurrent urinary tract infections (UTI) (secondary to VUR), hypertension, and a palpable mass [13]. In adults, unilateral MCDK typically presents asymptomatically [15]. Over time, as patients with MCDK progress, the affected kidney often regresses and becomes smaller, while the contralateral kidney is compensated by hypertrophy [14]. With the increase in routine prenatal imaging, increased accuracy of colleagues, and greater collaboration between physicians in different disciplines, the number of renal abnormalities identified during pregnancy has increased significantly. However, the underlying causes of these abnormalities and the clinical significance of these findings remain unclear [7, 17].

Etiology

The etiology of MCDK is unclear, but some studies have described the etiology of MCDK as sporadic, non-familial, and genetic [8, 9]. Several studies have shown that patients with MCDK have an increased risk of other congenital malformations [2, 9]. MCDK is caused by abnormal inductive interactions between the metanephric mesenchyme and the ureteral bud during embryonic development, and due to intrauterine obstruction of the fetal urinary tract. However, the ureter is absent at later stages of kidney development [1, 2, 18]. Genetic studies of individuals with MCDK and their genomic landscape in humans suggest that both genetic causes and mutations in the affected individual's gene pool are involved [18]. Bilateral MCDK usually causes Potter syndrome (widely separated eyes and nasal bridge, incomplete limbs, underdeveloped ears, hypoplastic lungs), although the disease is predominantly unilateral [1, 11].

Diagnosis

MCDK can be diagnosed prenatally postnatally; however, in some cases, the condition is not detected until after birth. Accurate evaluation of the growth status of the affected kidney and the contralateral kidney with serial ultrasound, voiding cystoureterography (VCUG), dimercaptosuccinic acid (DMSA) nuclear scan, computed tomography, and magnetic resonance imaging (MRI) plays a crucial role in confirming the diagnosis and evaluating the disease [1, 15, 19, 20]. MCDK is diagnosed through prenatal ultrasound, histological examination of associated pathological findings, and clinical status, with an emphasis on characterizing MCDK compared to other forms of cystic kidney disease [2, 7]. Given prenatal information, genetic counseling helps diagnose [2]. Histological examination of MCDK reveals immature epithelium and connective tissue [18]. In all patients, postpartum evaluation is necessary to confirm the diagnosis of suspected prenatal cases [3]. Prenatal diagnosis of a fetus with MCDK is usually performed by ultrasound [2]. The patient should be monitored by ultrasound for a long time after MCDK is diagnosed prenatally. In defining the development of MCDK, ultrasound screening is important and improves the quality of life of affected children by preventing complications such as infection or malignancy [3, 9].

Ultrasound

Ultrasound is the first-line screening tool and basic imaging technique for the follow-up and evaluation of patients with cystic renal anomalies [3]. One of the

most common renal abnormalities that can be detected by ultrasound examination during pregnancy (94.1%) is MCDK [4, 11, 13]. On ultrasound, MCDK is diagnosed by observing complete replacement of the kidney with irregular and disorganized cysts of various sizes, without surrounding normal tissue, and the presence of an atretic ureter [14]. The differential diagnosis of MCDK includes severe hydronephrosis and other abdominal masses [2].

VCUG

A second-line investigation, such as VCUG, is suggested for the diagnosis of VUR [3]. However, several studies have shown that routine renal scanning and VCUG are unnecessary in children with MCDK without UTI and hydroureteronephrosis [8, 19]. Some studies suggested that VCUG is necessary in all patients, given the reported incidence of VUR in MCDK for screening [5]. However, in our study, we recommended performing VCUG only in patients with a history of UTI or ultrasound findings suggestive of VUR, such as urinary tract dilatation [8].

Scintigraphy

Renal cortical tissue function, scar, hypodysplasia, and contralateral kidney involvement have been demonstrated on renal scintigraphy in patients with MCDK. Therefore, a Tc-99m DMSA scan is recommended at least once during the follow-up period in MCDK patients [17]. In cases of severe urethral dilatation, a MAG-3 scan is recommended to detect obstruction [3].

MRI

Detailed anatomical and functional information is provided on the MRI of the kidney and urinary tract. If gadolinium is used, functional and differential information of the kidney can be obtained. The high costs and the need to lie down for a long time are disadvantages of MRI; however, the indications for MRU may expand in the future [3].

Anomalies associated with MCDK

Associated congenital genitourinary anomalies are common (21-75%). VUR is the most common abnormality on the contralateral side in MCDK [2]. Screening for VUR in the contralateral kidney is essential in genital tract abnormalities [5, 21-24]. Contralateral renal abnormalities were reported in 21-75% and extrarenal abnormalities associated with MCDK in 35-5% of cases [2, 25]. Associated abnormalities include chromosomal abnormalities, cardiac D-George syndrome, and non-perforated anus. Prenatal information about extrarenal abnormalities is important, but careful postnatal examination is critical [2, 8].

Complication

Other complications include high blood pressure, UTI, mild proteinuria (increased protein in the urine due to kidney damage), and malignancy [13]. In MCDK, chromosomal abnormalities and syndromes (7%) and abnormalities in other organ systems (23.4%) were observed [4]. About 1.5% to 3% of children with unilateral MCKD develop hypertension [1, 9]. In some cases, hypertension resolves after removal of the dysplastic tissue [1, 16]. Aberrant renin expression has been documented in macrophage-like interstitial cells [1]. Hypertension is usually underdiagnosed in children with unilateral MCDK because its prevalence may be underestimated. However, blood pressure in MCDK patients may not be higher than in the population [2, 8, 25]. One study showed that hypertension in affected children is lower than in the population [9]. Multiple cases of Wilms tumor and renal carcinoma with the origin of cystic tissue have been reported [1]. On the other hand, due to the rapid growth of Wilms tumors, effective ultrasound screening should be performed every 3 months for 8 years [1]. However, some studies showed that the rate of long-term cancer in MCDK patients is similar to that of the general population [2, 25].

Prognosis

If, in unilateral MCDK, the contralateral kidney has normal function, patients will have an excellent prognosis [26, 27]. In 60% of MCDK cases, complete and precise spontaneous recovery is observed [10, 13], and other studies have shown that spontaneous regression occurs in 24% of cases by age 5 [28]. Those at increased risk of death or need for dialysis include infants with prenatal diagnosis of bilateral MCDK, extrarenal abnormalities, contralateral renal abnormalities, or anhydramnios [2]. Conservative nonsurgical management is an appropriate treatment strategy for MCDK, given the low risk of hypertension, tumor formation, and UTI [9, 29]. Due to the hidden nature of MCKD, some physicians recommend monitoring for complications with frequent assessment of kidney function markers, blood pressure, proteinuria, ultrasound evaluation, and VCUG [5, 10-12], although some do not recommend the latter [30-32]. Surgery is indicated only in cases of abdominal pain, recurrent UTI, and a sudden or progressive increase in kidney size [13]. Recently, the nonsurgical approach has become more popular as the benign natural history of MCDK is better understood [9].

Conclusion

Conservative nonsurgical management is an appropriate treatment strategy for MCDK, given the low risk of hypertension, tumor formation, and UTI. Recently, the nonsurgical approach has become more popular as the benign natural history of MCDK is better understood

Ethical Considerations

Compliance with ethical guidelines

There were no ethical considerations to be considered in this research.

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Authors' contributions

Conceptualization, Design, and Revision of the manuscript and critical evaluation of the intellectual content: All authors; Final draft and Critical appraisal of the manuscript: Mohsen Akhavan Sepahi.

Conflict of interest

The authors declared no conflict of interests.

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