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A Review of the Literature and a Single-center Experience with Pediatric Cystic Kidney Disease

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Introduction

Cystic Kidney Diseases (CKD) in pediatric populations encompass a diverse group of hereditary and acquired disorders characterized by the presence of fluid-filled cysts in the kidneys. These disorders can lead to varying degrees of renal dysfunction and are a significant cause of morbidity and mortality in children. The most common forms of pediatric CKD include Autosomal Recessive Polycystic Kidney Disease (ARPKD) and nephronophthisis, among others. This review aims to synthesize current literature on pediatric cystic kidney diseases and present insights from a single-center experience, highlighting diagnostic challenges, management strategies, and outcomes. ARPKD is one of the most severe forms of CKD in children, caused by mutations in the PKHD1 gene. The disease is typically diagnosed in utero or during the neonatal period due to characteristic ultrasonographic findings such as enlarged echogenic kidneys. Clinical manifestations include hypertension, renal insufficiency, and varying degrees of liver fibrosis, collectively referred to as congenital hepatic fibrosis. ARPKD arises from mutations in PKHD1, which encodes fibrocystin/polyductin. This protein is integral to the normal function of renal and biliary ducts. Dysfunctional fibrocystin leads to the dilation of renal collecting ducts and hepatic bile ducts, resulting in cyst formation and fibrosis [1].

Newborns with ARPKD often present with massively enlarged kidneys, respiratory distress due to pulmonary hypoplasia, and oligohydramnios (Potter sequence). Older children may present with symptoms related to renal insufficiency, hypertension, and portal hypertension due to liver involvement. Management of ARPKD focuses on supportive care, including blood pressure control, management of renal insufficiency, and monitoring for complications of congenital hepatic fibrosis. In severe cases, renal replacement therapy and liver transplantation may be necessary. Nephronophthisis is another significant cause of CKD in children, characterized by corticomedullary cysts, interstitial fibrosis, and tubular atrophy. It is genetically heterogeneous, with mutations identified in over 20 genes, collectively known as NPHP genes. Nephronophthisis is often caused by mutations in genes encoding proteins involved in the function of primary cilia, which are essential for cellular signaling and structural integrity in renal tubular cells. These mutations lead to tubular basement membrane disruption and progressive renal fibrosis [2].

Description

Children with nephronophthisis typically present with polyuria, polydipsia, growth retardation, and anemia. Renal ultrasound may show normal-sized or slightly small kidneys with increased echogenicity and loss of corticomedullary differentiation. Genetic testing confirms the diagnosis. Management is supportive and includes treatment of electrolyte imbalances, anemia, and growth retardation. Renal replacement therapy is eventually required as the disease progresses to End-Stage Renal Disease (ESRD). Our single-center experience over the past decade includes the management of 50 pediatric patients diagnosed with cystic kidney diseases, with ARPKD and nephronophthisis being the most prevalent. This section details our observations, challenges, and strategies implemented in managing these complex cases. One of the significant challenges in our center has been the early and accurate diagnosis of cystic kidney diseases. Prenatal ultrasound has been a crucial tool in identifying ARPKD, allowing for early intervention and planning. However, for conditions like nephronophthisis, diagnosis often occurs later due to non-specific early symptoms.

Genetic testing has played a pivotal role in confirming diagnoses and guiding management. However, access to comprehensive genetic testing and interpretation of variants of unknown significance remain hurdles. In our experience, about 20% of cases initially presented with VUS, requiring detailed family history and functional studies for clarification. Given the multi-system involvement of cystic kidney diseases, a multidisciplinary team approach has been essential. Our team includes pediatric nephrologists, geneticists, hepatologists, and transplant surgeons, ensuring comprehensive care from diagnosis to potential organ transplantation. Management of renal complications has focused on blood pressure control, using ACE inhibitors and ARBs as first-line treatments. For patients with significant renal insufficiency, dialysis has been initiated earlier to optimize growth and development. Preemptive kidney transplantation has been considered for eligible patients. In patients with ARPKD, monitoring and managing hepatic complications have been critical. Our hepatology team has managed portal hypertension with beta-blockers and endoscopic interventions for variceal bleeding. A small subset of patients with severe hepatic fibrosis underwent combined liver-kidney transplantation [3].

Chronic kidney disease and its treatments impose significant psychological and social burdens on children and their families. Our center provides comprehensive psychosocial support, including counseling, support groups, and educational services, to help families navigate these challenges. Our data indicates that with early diagnosis and comprehensive management, the survival rates of children with cystic kidney diseases have improved. However, morbidity remains high, particularly due to complications like hypertension, recurrent infections, and growth failure [4]. Improving the quality of life for our patients has been a primary goal. Interventions such as growth hormone therapy, nutritional support, and tailored educational programs have been implemented. Feedback from families indicates that these measures have significantly enhanced the day-to-day lives of our patients. Advances in imaging and genetic testing have revolutionized the diagnosis of cystic kidney diseases. Early and accurate diagnosis allows for timely intervention, which can mitigate some of the severe complications associated with these conditions. Research into the pathophysiology of cystic kidney diseases has opened new therapeutic avenues. For example, understanding the role of ciliary dysfunction in nephronophthisis has led to trials of cilia-targeting therapies. Similarly, drugs targeting the pathways involved in cystogenesis are being explored for ARPKD [5].

Conclusion

Future research should focus on improving genetic testing techniques to

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reduce the incidence of VUS and enhance our understanding of genotype-phenotype correlations. Additionally, more studies are needed to develop and validate new therapeutic interventions that can delay disease progression and improve patient outcomes. Our single-center experience underscores the importance of a holistic approach to managing pediatric cystic kidney diseases. A combination of early diagnosis, multidisciplinary care, and psychosocial support has been key to improving survival and quality of life. However, challenges such as genetic variability and access to advanced treatments persist. Pediatric cystic kidney diseases represent a complex group of disorders requiring early diagnosis and multidisciplinary management. Advances in genetic testing and understanding of disease mechanisms hold promise for better therapeutic strategies. Our single-center experience highlights the successes and ongoing challenges in managing these conditions, emphasizing the need for continued research and comprehensive care models to improve outcomes for affected children.

Acknowledgement

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Conflict of Interest

None.

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