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Original Article

Morphological Spectrum of Congenital Anomalies of Kidney and Urinary Tract in Children: A Single-center Prospective Observational Study

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ABSTRACT

Background and Aim: The range of congenital anomalies affecting the kidneys and urinary tract (CAKUT) is wide, extending from asymptomatic ectopic kidneys to severe bilateral kidney agenesis. This study unveils the spectrum of morphological anomalies in the kidneys and urinary tract among pediatric patients.

Methods: Over 2 years, 116 patients underwent comprehensive evaluation, including lab tests, imaging, and radionuclide scans.

Results: Among the 650 patients, 116 cases were identified as CAKUT, yielding a prevalence rate of 17.8%. Among these, 67 of 116(57.7%) were male, with the majority (85%) aged 0 to 5 years. Abnormal ultrasonographic findings drove hospital visits in 46 of 116(39.7%) cases, followed by recurrent fevers (19.8%), and urinary dribbling (12%). Anemia and renal dysfunction were the main lab abnormalities in 90 of 116(77.5%) and 40 of 116(34.5%) subjects, respectively. Meanwhile, CAKUT cases were categorized as non-obstructive in 78 of 116(68.1%) and obstructive in 37 of 116(31.9%), Primary vesicoureteral reflux (VUR) in 25/116 (21.6%) and ureteropelvic junction obstruction (UPJO) in 18/116 (15.6%) were most common non-obstructive and obstructive anomalies respectively. Among VUR cases, grade II and grade III VUR accounted for 9 of 25(36%) and 7 of 25(28%) subjects, respectively; also, aspects associated with hypertension were in 18 of 25(72%) and renal scarring in 4 of 25 (16%). UPJO was present in 18 of 116(15.5%) and typically mild in 14 of 18(61%) on ultrasonography (USG). Posterior urethral valves (PUV) accounted for 16 in 116(13.7%) of cases, presenting with renal dysfunction in 13 of 16(81%) and hydronephrosis in 12 of 16 (75%).

Conclusion: This study provides valuable insights into the prevalence, clinical patterns, and morphological diversity of CAKUT. These findings underscore the importance of early diagnosis and multidisciplinary management for optimizing outcomes.

Keywords: CAKUT, Vesicoureteral reflux (VUR), Ureteropelvic junction obstruction, Posterior urethral valve, Multi-cystic kidney disease

Introduction

The investigation of human kidney and urinary tract abnormalities reveals substantial phenotypic variability, leading to challenges in clinical classification and diagnosis. In response to this intricate overlap, contemporary researchers have categorized kidney and urologic malformations under the encompassing term congenital anomalies of the kidney and urinary tract (CAKUT) [1]. The CAKUT spectrum includes a diverse array of conditions, such as renal agenesis, horseshoe kidney, ectopic kidney, primary vesicoureteral reflux (VUR), ureteropelvic junction obstruction (UPJO), diverticula of urethras or bladder, duplicated ureters, megaureters, posterior urethral valves (PUV), vesicoureteral junction obstruction, multi-cystic dysplastic kidney (MCDK), and renal hypo/dysplasia. These anomalies significantly contribute to renal impairment among children, potentially progressing to end-stage renal disease [2]. Birth defect registries from California, USA, report a prevalence of three to six cases of CAKUT per 1000 births, underscoring the potentially serious consequences these abnormalities pose to future life [3]. Accordingly, this study explores the morphological spectrum of CAKUT, seeking to gain firsthand insights into this condition within our union territory.

Materials and Methods

This cross-sectional study was conducted at the Department of Pediatrics, Government Medical College Srinagar, Jammu and Kashmir, India, following the Ethics Committee approval. Over two years, from September 2019 to September 2021, a total of 116 patients up to 18 years of age, exhibiting clinical, laboratory, or imaging evidence suggestive of CAKUT, were enrolled. Patients with neurogenic bladder were excluded. The study encompassed a wide range of units, including nephrology, neonatal intensive care units, and pediatric intensive care units. Comprehensive information, including prenatal, natal, and family history, was gathered from caregivers. Urinalysis, complete blood counts, serum electrolytes, and kidney function tests were conducted for all patients. Additionally, imaging studies such as ultrasonography (USG) focusing on the genitourinary system, X-ray abdomen, micturating cystourethrogram, computed tomography abdomen, computed tomography/magnetic resonance imaging urography, and radionuclide studies (DTPA and DMSA) were performed based on clinical indications. Expert opinions were sought from specialists in pediatric radiology, pediatric surgery, and nuclear medicine when necessary.

Statistical analysis

Statistical analysis was carried out using the SPSS software, version 23. Descriptive statistics, including Mean±SD, median, and range, were used for numerical data, while categorical data was presented as percentages.

Results

Among the 650 patients who visited the pediatric nephrology unit during the study period, 116 cases were diagnosed with CAKUT, resulting in a prevalence rate of 17.18% (116 out of 650×100=17.18%). Among these cases, males accounted for 57.7% (67 out of 116), with the majority (85%) falling within the 0-5 years age group. Notably, abnormal ultrasonographic findings (39.7%) were the most common reason for hospital presentations, followed by recurrent fever (19.8%), urinary dribbling, and abdominal distension (12% each). Anemia was the most frequent laboratory abnormality, affecting 77.5% of cases, followed by elevated C-reactive protein, microhematuria, and renal dysfunction (57%, 43%, and 35%, respectively). CAKUT cases were categorized as non-obstructive in 78 out of 116(68.1%) and obstructive in 37 out of 116(31.9%); meanwhile, primary VUR in 25 out of 116(21.6%) and UPJO in 18 out of 116(15.6%) were most common non-obstructive and obstructive anomalies, respectively. Among VUR cases, grade II and grade III VUR accounted for 9 in 25 (36%) and 7 in 25(28%) subjects respectively; also, aspects associated with hypertension were in 18 out of 25(72%) and renal scarring in 4 out of 25(16%). UPJO was present in 18 out of 116(15.5%), typically mild in 14/18 (61%) subjects on USG. PUV accounted for 16 in 116(13.7%) of cases, presenting with renal dysfunction in 13 in 16(81%) and hydronephrosis in 12 in 16(75%). MCDK constituted 6% (7 in 116) of cases, primarily unilateral, with a notable right-kidney predilection in 5 in 7(71%) subjects. The results are summarized in Tables 1, 2, 3 and 4.

Discussion

This study investigated the prevalence and varied presentation of CAKUT among pediatric patients from a single center in northern India. The observed prevalence of 17.8% within the total pediatric nephrology cohort highlights the substantial impact of CAKUT on this demographic. The prevalence of 20% to 30% was reported by Queisser-Luft et al. in a study from Germany [4] which is similar to our results. This finding underscores the significance of early detection and management strategies in pediatric nephrology.

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Table 1. Morphological pattern of congenital anomalies affecting the kidneys and urinary tract

Non-obstructive	No. (%)
	Cases
Primary VUR	25(21.6)
Non-obstructive hydronephrosis	21(18.1)
MCDK	7(6)
Hypoplastic kidney	6(5)
Hypospadias	6(5)
Ectopic kidney	5(4)
Renal agenesis	3(2.6)
Polycystic kidney disease	3(2.6)
Horse shoe kidney	2(2)
Supernumery kidney	1(0.8)
Total	79(68.1)

Obstructive	No. (%)
	Cases
Uretero-pelvic junction obstruction	18(15.6)
Posterior urethral valve	16(13.8)
Vesico-ureteric junction obstruction	2(2)
Complete duplex collecting system	1(0.8)
Total	37(31.9)

Table 2. Profile of children with UPJO (n=18)

Characteristics		No. (%)
Gender (male/female)		16/2
Time of diagnosis (prenatal/postnatal)		8/10
Unilateral/Bilateral		13/5
Severity for each renal unit	Mild	14(61)
	Mild to moderate	4(17.4)
	Moderate	3(13)
	Moderate to severe	1(4.3)
	Severe	1(4.3)

Table 3. Profile of patients with PUV (n=16)

Characteristics	No. (%)
Male/Female	16/0
Age at diagnosis in months (Mean±SD)	16.0±2.8
Hypertension	12(75)
Renal dysfunction	13(81)
Hydronephrosis on ultrasound sonography test (unilateral/bilateral)	3/9
Significant post-void residual urine on ultrasound sonography test	8(50)
VUR on voiding cystourethrogram (unilateral/bilateral)	3/9
Dilated posterior urethra on voiding cystourethrogram	15(94)
Detrusor instability on urodynamic study	3(18.8)

Table 4. Profile of patients with MCDK (n=7)

Characteristics	No. (%)
Right/Left	5(71)/2(29)
Bilateral	0
Antenatal/postnatal diagnosis	5(71)/2(29)
Associated secondary VUR	2(29)
Associated ureterocele and ectopic ureter	1(14)
Contralateral renal hypertrophy on ultrasound sonography test	4(58)

The predominance of males within the CAKUT cases (57.7%) echoes previously documented gender trends in renal anomalies. The higher incidence in males could be attributed to anatomical differences and developmental factors in the urinary tract. A similar trend was observed by Saha et al. [5]; however, in another study by Bulum et al. [6], females outnumbered males. The reason for this discrepancy is difficult to explain but could be due to the complex interplay of genetic, environmental, and possible pregnancy-related adverse events.

The age distribution of CAKUT cases, with 85% occurring in the 0-5 years age group, aligns with the notion that many of these anomalies are recognized early in life due to associated clinical manifestations or incidental findings. The prevalence of abnormal ultrasonographic findings (39.7%) as the primary reason for hospital visits underscores the vital role of imaging techniques in diagnosis, enabling prompt intervention.

Renal dysfunction was present in 35% of patients in our cohort. CAKUT is known to be responsible for 30% to 50% of cases of chronic kidney disease requiring kidney replacement therapy in children [2]. The notable presence of anemia (77.5%) and renal dysfunction highlights the systemic impact of CAKUT, emphasizing the need for comprehensive evaluations beyond imaging alone.

The prevalence of both non-obstructive (68.1%) and obstructive (31.9%) CAKUT variants indicates the diverse nature of these anomalies. Primary VUR (21.6%) and UPJO (13.8%) were prominent among these anomalies (Table 1). Primary VUR is the most common urologic finding in children ranging from 15% [7, 8] in children with antenatal hydronephrosis to 30% to 45% [9, 10] in children with recurrent febrile urinary tract infections. The observation of grade II and III VUR as the most common variants reiterates the importance of stratifying the severity of these conditions for clinical decision-making.

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The findings related to UPJO highlight its predominance in males, unilateral presentation, and varied degrees of ultrasonographic severity (Table 2). Male predominance has been shown by Morin et al. [9] and Duong et al. [10]. Bilateral UPJO was slightly more prevalent in our study (28%) as against the reported prevalence of around 10% [10, 11]. The reason for this discrepancy is unclear. The normal voiding cystourethrogram results rule out significant VUR as a cause of hydronephronephrosis.

PUV accounted for 13.7% of cases, exclusively affecting males. This reaffirms the well-established gender predilection for PUV. The presence of renal dysfunction and hypertension in PUV cases underscores the potential long-term consequences of this anomaly. Renal dysfunction was present in 81% of patients with PUV in our study (Table 3). PUV is reported as the most common cause of obstructive CKD in children [12]. Approximately 15% to 20% of patients with PUV progress to end-stage renal disease [13, 14].

The incidence of MCDK varies according to the country and the study, ranging from 1 per 3600 to 1 per 4300 live births [15, 16]. The presence of MCDK in 6% of cases indicates its relatively rare occurrence (Table 4). MCKD is the most severe form of cystic kidney disease. The unilateral prevalence and association with VUR and ectopic ureter highlight the complex interplay of multiple anomalies in some cases. Male predominance and unilateral involvement have been reported by other studies as well [17].

Additionally, the identification of ectopic kidneys, hypoplastic/dysplastic kidneys, and unilateral renal agenesis demonstrates the wide morphological spectrum of CAKUT, underlining the need for individualized management approaches.

Conclusion

This study provides valuable insights into the prevalence, clinical patterns, and morphological diversity of CAKUT among pediatric patients. These findings underscore the importance of early diagnosis, multidisciplinary management, and long-term follow-up strategies for optimizing outcomes in this vulnerable population.

Limitations

This study faced certain limitations. Firstly, it is a single-center study of short duration catering to a limited demographic and ethnic group. Low levels of awareness among the population in general and among treating physicians about the importance and occurrence of CAKUT, could have resulted in low referrals and consequent underestimation of this problem.

Ethical Considerations

Compliance with ethical guidelines

This study is approved by the Ethics Committee of Government Medical College Srinagar, Srinagar, India (Code: ETH/GMC/50). Written consent has been taken from the guardians of patients.

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Authors' contributions

Study design and conceptualization: Nucksheeba Aziz Bhat, Mohd Ashraf, Naseer Ul Hassan Khan, Muzafar Jan, Rayees Yousuf Sheikh and Sheikh Quyoom Hussain; Data collection: Nucksheeba Aziz Bhat, Mohd Ashraf, Naseer Ul Hassan Khan, and Rayees Yousuf Sheikh; Data analysis and interpretation: All Authors; Writing the initial draft: Nucksheeba Aziz Bhat; Review and editing: Mohd Ashraf, Naseer Ul Hassan Khan, Muzafar Jan, Rayees Yousuf Sheikh, Sheikh Quyoom Hussain, and Waseem Shafi Sheikh; Final approval: Rayees Yousuf Sheikh, Sheikh Quyoom Hussain1, and Waseem Shafi Sheikh.

Conflict of interest

The authors declared no conflict of interests.

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