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ORIGINAL ARTICLE

Prevalence and Pattern of Congenital Anomalies of the Kidney and Urinary Tract in a Tertiary Hospital in Bangladesh

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ABSTRACT

Background & objective: Congenital anomalies of the kidney and urinary tract (CAKUT) are a leading cause of chronic kidney disease, but data on their prevalence and patterns in Bangladesh is limited. This study aimed to address this gap by characterizing CAKUT in a local population.

Methods: This was a hospital-based cross-sectional study conducted over one year at a tertiary hospital. A total of 100 patients (infants, children, and adults) with confirmed CAKUT were included. Data on demographics, maternal history, and specific types of anomalies were collected and analyzed.

Results: The study found a prevalence of 11.3% for CAKUT among all urology cases attended. Demographically, the cohort was predominantly male (65%), with a notable finding that 66% of patients received their first diagnosis after the age of 20 years. Maternal history revealed a high rate of oligohydramnios (35%) in index pregnancies. The most frequent anomalies were hydronephrosis (77%) and ureteropelvic junction (UPJ) obstruction (66%), with some patients presenting with multiple anomalies concurrently.

Conclusion: The findings suggest that CAKUT constitutes a significant disease burden in the study population. The high proportion of patients diagnosed in adulthood underscores a critical need for enhanced prenatal and postnatal screening programs. Timely detection and intervention are crucial to mitigate the progression of these conditions and improve long-term renal outcomes in Bangladesh.

Key words: Prevalence, Pattern, Congenital Anomalies, Kidney, Urinary Tract etc.

INTRODUCTION:

Congenital anomalies (CAs) are a significant cause of morbidity and mortality in early life, affecting an estimated 1-6% of viable pregnancies worldwide.[1-4] These conditions are responsible for approximately 3.3 million deaths annually in children under five years of age[3] and account for a quarter of all infant deaths in high-income countries.[5-7] For long-term survivors, the prognosis varies, but many experience significant physical and psychological impairments, leading to sustained health and social care needs with considerable economic cost.[8]

Among these conditions, congenital anomalies of the kidney and urinary tract (CAKUT) represent a diverse group of structural malformations involving the kidneys, renal collecting system, bladder, or urethra. These anomalies occur with a frequency of approximately 1 in 500 live births.[8,9] CAKUT is a leading cause of chronic kidney disease (CKD), accounting for 30-60% of pediatric end-stage renal disease (ESRD) cases that may

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require renal transplantation in childhood.[10,11] Furthermore, renal transplantation for ESRD occurs significantly earlier in patients with CAKUT (mean age 31 years) compared to those without these abnormalities (mean age 61 years). The association between CAKUT and other congenital anomalies, such as scoliosis, is also notable, with incidence rates ranging from 11-37%.[12-15]

While many patients with CAKUT may remain asymptomatic, long-term studies have demonstrated a high risk of progressive renal function impairment.[10,11,16,17] Most cases are not detected until clinical symptoms emerge, or they are diagnosed incidentally in patients who have already progressed to ESRD, requiring dialysis or transplantation. CAKUTs are responsible for 40-50% of pediatric and 7% of adult ESRD cases globally. The etiology of the majority of CAKUT cases remains unknown, although a strong genetic component is suggested by familial aggregation studies, which indicate that 10-50% of affected children have a family history of kidney abnormalities or urinary tract disease.[18,19] Pathogenic variants in genes causing CAKUT are linked to monogenic diseases, such as polycystic kidney disease and ciliopathies, as well as syndromes with other associated abnormalities. Prenatal diagnosis, primarily through ultrasonography,[20] is an important first step for early prenatal counseling.

Despite the significant burden of CAKUT, most previously published studies have focused on the morphology, epidemiology, or pathogenesis in children and have been conducted outside of Bangladesh. There is a pressing need to determine the prevalence and patterns of CAKUT in the local context of Bangladesh, particularly across infants, children, and adults. Such data will facilitate early counseling and intervention, potentially mitigating the progression to more severe disease. The present study aims to address this gap by determining the prevalence and pattern of CAKUT in a tertiary hospital in Bangladesh.

METHODS:

This was a hospital-based cross-sectional study conducted from June 2023 to May 2024 over a period of one year. The study was approved by the Ethical Review Committee of Chattagram International Medical College, Chattagram. The study was conducted in the Department of Urology and the Department of Ultrasound & Imaging of the same medical college. The study population consisted of infants, children, and adults who were suspected of having CAKUT. A total of 400 infants, children, and adults presenting with signs and symptoms of CAKUT or incidentally diagnosed with CAKUT during urological or other abdominal surgeries were consecutively enrolled after obtaining informed consent from them. For minor patients, their parents or legal guardians provided informed consent to participate. However, individuals suspected of having CAKUT but were unwilling to undergo an ultrasound examination were excluded.

Data were collected through a combination of clinical examination & ultrasound imaging, as per standard diagnostic procedures and were processed and analyzed using the Statistical Package for Social Sciences (SPSS), version 23.0. Descriptive statistics were used to summarize the data. While quantitative data were analyzed using Unpaired t-Test, qualitative data were analyzed using Chi-square (χ^2) or Fisher's Exact (a variant of Chi-square) Probability Test. The level of significance was set at $p < 0.05$.

RESULTS:

Prevalence of CAKUT:

The total number of cases attended/admitted during the period of data collection was 997; of them, 113 cases had one or more forms of CAKUT, although 100 cases, who gave consent to participate in the study, were included as a sample. Therefore, the proportion of CAKUT cases attended/admitted in the Urology Department at any given time is (113/997)×100=11.3%.

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Demographic Characteristics:

The study population's demographic characteristics revealed several key findings. The majority of patients were male (65%), resulting in a male to female ratio of approximately 2:1. The median age at the time of their first confirmatory diagnosis was over 20 years, with 66% of patients falling into this age group. Geographically, 60% of patients were from rural areas, while the remaining 40% were from urban settings. The median monthly family income was BDT 20,000, with nearly half of the cohort (47%) earning between BDT 10,000 and BDT 20,000 per month. The vast majority of participants (92%) were from Muslim families.

Maternal and Birth Characteristics

Maternal characteristics during the index pregnancy were also documented. The mean maternal age at the time of birth was 25.6 ± 5.8 years, with two-thirds of mothers being between 20 and 30 years old. The birth order for two-thirds of the patients was either first or second. Pre-existing maternal health conditions and complications during pregnancy were noted in a small percentage of cases. Oligohydramnios was reported in 35% of pregnancies, followed by gestational diabetes mellitus (GDM) in 13% and pregnancy-induced hypertension (PIH) in 5%. Other less common conditions included severe maternal anemia (6%), pregestational diabetes (8%), and pregestational hypertension (4%).

Pattern of Congenital Anomalies

The study identified a diverse spectrum of CAKUT abnormalities. Hydronephrosis was the most prevalent anomaly, affecting 77% of patients. This was followed by ureteropelvic junction (UPJ) obstruction in 66% of cases. Other anomalies, in descending order of frequency, included: ureterovesical junction obstruction (11%), posterior urethral valve defects (13%), vesicoureteral reflux (4%), duplicate collecting system (3%), renal agenesis (1%). It's important to note that some patients presented with multiple concurrent congenital anomalies. Rarer malformations, such as horseshoe kidney, were found in only one case.

Table I. Distribution of patients by their demographic characteristics (n = 100)

Demographic characteristics	Frequency	Percentage
Age (years)		
≤ 5	4	4.0
6 – 10	7	7.0
11 – 15	15	15.0
16 – 20	8	8.0
> 20	66	66.0
Sex		
Male	65	65.0
Female	35	35.0
Residence		
Urban	42	42.0
Rural	58	58.0
Monthly family income* (Taka)		
< 10000	11	11.0
10000 – 20000	47	47.0
> 20000	42	42.0
Religion		
Islam	92	92.0
Hinduism	6	6.0
Buddhism	2	2.0

***Median ± SEM** = 30 ± 2.1 years; **range** = (2 – 95) years.
***Median income** = 20000 ± 611 Taka.

Table II. Distribution of patients by their maternal age group (n=100)

Maternal age at birth (years)	Frequency	Percentage
< 20	16	16.0
20 – 30	67	67.0
> 30	17	17.0

***Mean maternal age** = 25.6 ± 5.8 years; **range** = (14 – 40) years.

Table III. Distribution of patients by obstetric history of the index pregnancy (n=100)

Obstetric history	Frequency	Percentage
Birth order		
1 – 2	67	67.0
3 – 5	33	33.0
Gestational diabetes mellitus (GDM)	13	13.0
Oligohydramnios	35	35.0
Pregnancy-induced hypertension (PIH)	5	5.0

Table IV. Patients stratified by medical problems during index pregnancy (n=100)

Maternal medical problem	Frequency	Percentage
Severe maternal anemia	6	6.0
Pre-gestational hypertension	4	4.0
Pre-gestational diabetes	8	8.0

Table V. Distribution of patients by their pattern of CACUT (n=100*)

Congenital anomalies of kidney and urinary tract (CAKUT)	Frequency	Percentage
Vesicoureteral reflex (VUR) defect	4	4.0
Ureterovesical junction (UVJ) obstruction	11	11.0
Ureteropelvic junction (UPJ) obstruction	66	66.0
Duplicate collecting system	3	3.0
Renal Agenesis	1	1.0
Hydronephrosis	77	77.0

*Total will not correspond to 100%, for multiple response

Table VI. Patients stratified by defects in fusion bladder/ urethral development

Defects in fusion or bladder/ urethral development	Frequency	Percentage
Horseshoe kidney	1	1.0
Posterior urethral valves	13	13.0

*Total will not correspond to 100%, for multiple response

DISCUSSION:

This study provides valuable insights into the prevalence and patterns of congenital anomalies of the kidney and urinary tract (CAKUT) in a tertiary hospital setting in Bangladesh, a topic that has been under-researched in the local context. The findings from this study are discussed in comparison to existing literature, highlighting both similarities and disparities.

Prevalence and Demographics

The observed prevalence of CAKUT, at 11.3% of all urology cases, underscores its significant burden on healthcare services in Bangladesh. While a direct comparison with population-based prevalence studies (e.g., 1 in 500 live births globally) is difficult due to the hospital-based nature of our data, our findings suggest that CAKUT constitutes a substantial proportion of urological presentations. This aligns with the global literature that identifies CAKUT as a leading cause of pediatric and adult kidney disease.

The study's finding that the majority of patients (66%) were diagnosed after the age of 20 is particularly noteworthy. This is in stark contrast to studies from high-income countries where widespread prenatal and postnatal screening often leads to an earlier diagnosis.[21] This delayed presentation in our cohort suggests a lack of systematic screening, leading to diagnoses only when symptoms of advanced disease-such as renal failure or urinary tract infections-manifest. This highlights a critical need for enhanced public health initiatives and screening programs to enable earlier detection and intervention.

The observed male predominance (male-to-female ratio of 2:1) is consistent with many international studies on CAKUT, particularly those focusing on obstructive uropathies like posterior urethral valve (PUV), which are more common in males.[22] Our findings of a higher number of patients from rural areas (60%) may reflect the demographic distribution of the region or, alternatively, could indicate disparities in access to specialized healthcare, where individuals from rural areas may only seek care at a tertiary center when their condition is advanced. The family income distribution in our cohort, with the majority falling into the middle-income range, suggests that while poverty may be a barrier to healthcare access, it's not the only factor, & individuals from various socioeconomic backgrounds are affected.

Maternal and Birth Characteristics

The mean maternal age at birth (25.6 years) is within the typical reproductive age range for the region. However, the high prevalence of oligohydramnios (35%) is a significant finding. Oligohydramnios, a known complication of CAKUT, can be both a cause and a consequence of urinary tract malformations, particularly those leading to reduced fetal urine output, such as renal agenesis or severe obstructive uropathies. Our findings are consistent with studies that report a strong association between oligohydramnios and CAKUT.[23] The presence of other maternal health issues like gestational diabetes, pregestational

hypertension, and anemia, while not a direct cause, can be co-morbidities that may complicate the clinical management of both the mother and the infant with CAKUT.

Pattern of Anomalies

The pattern of CAKUT identified in this study aligns with global trends where hydronephrosis and ureteropelvic junction (UPJ) obstruction are the most common diagnoses. Hydronephrosis, which is often a consequence of obstructive anomalies like UPJ obstruction, was the most prevalent finding (77%). The high incidence of UPJ obstruction (66%) and posterior urethral valve (PUV) defects (13%) in our cohort is particularly concerning as these conditions are known to cause progressive kidney damage if left untreated.[24] The co-occurrence of multiple anomalies in some patients further complicates management and underscores the need for a comprehensive diagnostic approach.

The low prevalence of other anomalies like vesicoureteral reflux (VUR) and renal agenesis in our adult-heavy cohort may be a result of the study's design. VUR, for instance, is often detected in infancy or childhood due to recurrent urinary tract infections, and its prevalence might be underrepresented in an adult population. Similarly, bilateral renal agenesis is a severe condition often incompatible with life, which might explain its low representation. The single case of horseshoe kidney reflects the rarity of this anomaly.

Limitations and Future Directions

This study, being hospital-based and cross-sectional, has limitations. The results may not be generalizable to the entire Bangladeshi population, & the sample size of 100 is relatively small. Furthermore, the lack of long-term follow-up data prevents us from assessing the progression of the disease in our cohort.

CONCLUSION:

The study confirms that congenital anomalies of the kidney and urinary tract (CAKUT) are a significant health issue in Bangladesh, as evidenced by their 11.3% prevalence in the urology department. A critical finding is the delayed diagnosis, with two-thirds of patients presenting after age 20, likely due to a lack of systematic screening. The most common anomalies were hydronephrosis and ureteropelvic junction obstruction. The high rate of oligohydramnios (35%) in maternal histories also points to early-stage developmental issues. These findings highlight a pressing need for a structured public health approach, including enhanced prenatal and postnatal screening, to enable earlier detection and improve long-term patient outcomes in the country. The implementation of enhanced prenatal and postnatal ultrasound screening programs is essential for early diagnosis, which in turn can facilitate timely interventions and mitigate the progression to chronic and end-stage renal disease. Future, large-scale, population-based studies are required to more accurately determine the true prevalence of CAKUT in the country and to assess long-term patient outcomes.

REFERENCES:

1. British Isles Network of Congenital Anomaly Registers (BINOCAR). Congenital anomaly statistics 2012 England and Wales. http://www.binocar.org/content/Annual%20report%202012_FINAL_nologo.pdf. Accessed 1 Apr 2015.
2. Centers for Disease Control and Prevention (CDC) Update on overall prevalence of major birth defects - Atlanta, Georgia, 1978-2005. MMWR Morb Mortal Wkly Rep 2008;57:1–5.
3. Christianson A, Howson CP, Modell B. March of Dimes global report on birth defects: the hidden toll of dying and disabled children. White Plains: March of Dimes Birth Defects Foundation; 2006.
4. Dolk H, Loane M, Garne E. The prevalence of congenital anomalies in Europe. *Adv Exp Med Biol* 2010; 686:349–64. doi: 10.1007/978-90-481-9485-8_20.
5. Murphy SL, Xu J, Kochanek KD. Deaths: final data for 2010. *Natl Vital Stat Rep* 2013;61:1–117.
6. Word Health Organization. Birth defects. Report by the Secretariat for Sixty-Third World Health Assembly. 2010. http://apps.who.int/gb/ebwha/pdf_files/WHA63/A63_10-en.pdf. Accessed 8 May 2015.

7. CDC's National Center on Birth Defects & Developmental Disabilities. Strategic plan 2011-2015. 2011. http: //www.cdc.gov/NCBDDD/AboutUs/documents /NCBDDD _StrategicPlan_2-10-11.pdf. Accessed 2 June 2015.

8. Nakanishi K, Yoshikawa N. Genetic disorders of human congenital anomalies of the kidney and urinary tract (CAKUT) *Pediatrics Int* 2003;5:610–16.

9. Nicolaou N, Renkema KY, Bongers EM, et al. Genetic, environmental, and epigenetic factors involved in cakut. *Nature Rev Nephrol* 2015;12:720–31.

10. Japanese Society of Nephrology. Evidence-based clinical practice guideline for CKD 2013. *Clin Exp Nephrol* 2014;3:346–423.

11. Harambat J, van Stralen KJ, Kim JJ, et al. Epidemiology of chronic kidney disease in children. *Pediatric Nephrol* 2012;3:363–73.

12. MacEwen GD, Winter RB, Hardy JH. Evaluation of kidney anomalies in congenital scoliosis. *J Bone Joint Surg* 1972;7:1451–54.

13. Drvaric DM, Ruderman RJ, Conrad RW, et al. Congenital scoliosis and urinary tract abnormalities: Are intravenous pyelograms necessary? *J Pediatr Orthop* 1987;4:441–43.

14. Rai AS, Taylor TK, Smith GH, et al. Congenital abnormalities of the urogenital tract in association with congenital vertebral malformations. *J Bone Joint Surg Br* 2002;6:891–95.

15. Shen J, Wang Z, Liu J, et al. Abnormalities associated with congenital scoliosis: A retrospective study of 226 Chinese surgical cases. *Spine* 2013;10:814–18.

16. Tain YL, Luh H, Lin CY, et al. Incidence and risks of congenital anomalies of kidney and urinary tract in newborns: A population-based case-control study in Taiwan. *Medicine* 2016;5:e2659.

17. Melo BF, Aguiar MB, Bouzada MC, Aguiar RL, Pereira AK, Paixão GM, et al. Early risk factors for neonatal mortality in CAKUT: analysis of 524 affected newborns. *Pediatr Nephrol* 2012;27(6):965-72.doi:10.1007/s00 467-012-2107-y.

18. Weber S. Novel genetic aspects of congenital anomalies of kidney and urinary tract. *Curr Opin Pediatr* 2012; 24(2):212–218.

19. Bulum B, Ozçakar ZB, Ustüner E, Düşünceli E, Kavaz A, Duman D, et al. High frequency of kidney and urinary tract anomalies in asymptomatic first-degree relatives of patients with CAKUT. *Pediatr Nephrol* 2013;28(11) :2143-7. doi: 10.1007/s00467-013-2530-8.

20. Asha N. Talati, Carolyn M. Webster, Neeta L. Prenatal genetic considerations of congenital anomalies of the kidney and urinary tract (CAKUT). *Vora Prenat Diagn* 2019;39(9): 679–692. doi: 10.1002/pd.5536

21. Brockwell M, Hergenrother S, Satariano M, Shah R, Raina R. Pathophysiology of Congenital Anomalies of the Kidney and Urinary Tract: A Comprehensive Review. *Cells* 2024;13(22):1866.https://doi.org/10. 3390/cells13221866

22. Urology-Textbook. Ureteropelvic Junction or UPJ-Obstruction: Diagnosis and Treatment. Available from:https://www.urology-textbook.com/upj-obstructi on.html. UK Kidney Association. Congenital Anomalies of the Kidneys and Urinary Tracts. Available from: https://www.ukkidney.org/rare-renal/clinician-zinformation/ congenital-anomalies-kidneys-and-urinary-tracts.

23. Caiulo VA, Caiulo S, Gargasole C, Mele G, Massafra R. Congenital anomalies of the kidney and urinary tract (CAKUT) in critically ill infants: a multicenter cohort study. *Pediatric Nephrology* 2022;38(Suppl 1):1–12.

24. Sepulveda M. Giant Hydronephrosis - A Late Diagnosis of Ureteropelvic Junction Obstruction. World Journal of Nephrology and Urology. Gavin Publishers. Prevalence of Congenital Anomalies of the Kidney and Urinary Tract (CAKUT) in the Greater Buffalo Region: A Retrospective Review. Available from:https://www.gavinpublishers.com /article/view/prevalence-of-congenital-anomalies-of-th e-kidney-and-urinary-tract-cakut-in-the-greater-buffal o-region-a-retrospective-review).