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Original Article

Evaluation of The Patients Diagnosed with Urinary Stone Disease in Our Pediatric Nephrology Clinic

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Keywords: Metabolic disorder; pediatric nephrology; urinary stone disease.

ABSTRACT

Objective: Our study aimed to evaluate the demographic characteristics, clinical presentations, metabolic disorders, radiological findings, and treatment outcomes of pediatric patients diagnosed with urinary stone disease in a pediatric nephrology outpatient clinic.

Methods: A retrospective, descriptive study was conducted involving 256 pediatric patients aged between 0-18 years, diagnosed with urinary stone disease from January to December 2016. Clinical data, laboratory results, radiological findings, and treatments were collected from patient files and analyzed statistically.

Results: Among the patients, 52.3% were male, and the median age was 39.5 [15.0-87.0] months. The most common reason for presentation was known urinary stone disease follow-up (21.5%), followed by abdominal pain and restlessness. A positive family history of urinary stone disease was noted in 61.3%, and parental consanguinity was observed in 5.5%. Metabolic abnormalities were detected in 57% of cases, most frequently hypocitraturia (34.4%), hypercalciuria (20.7%), and hyperoxaluria (17.9%). Hypocitraturia was more prevalent in older children, while hypercalciuria was more common in infants. Medical treatment was initiated in 70.7% of patients, primarily with potassium citrate (69.1%). Surgical interventions included extracorporeal shock wave lithotripsy (12.6%), percutaneous nephrolithotomy (2%), and open surgery (3.1%).

Conclusion: Pediatric urinary stone disease commonly presents with metabolic disturbances, particularly hypocitraturia, and frequently involves familial predisposition. Early diagnosis through appropriate imaging and metabolic screening, followed by targeted medical management, is essential for preventing renal complications and reducing the need for surgical interventions.

INTRODUCTION

Pediatric urinary stone disease (USD) is the formation of stones in the urinary tract in children, mainly in the kidneys, ureters, or bladder. This clinical condition is prevalent among the paediatric population. The global prevalence of USD in children has been rising yearly, with the most significant increase among adolescents aged 10 to 14.[1] Over the past 25 years, the annual incidence of pediatric USD has grown globally from 6% to 0%.[1,2] Urinary stone disease has been identified as an endemic disorder in Türkiye, affecting 10% to 20% of the pediatric population.[3]

Socioeconomic, climatic, and dietary factors have been identified as the primary contributors to the prevalence of kidney stones.[4] Genetic and underlying systemic diseases have also been implicated in paediatric populations.[1,3] While USD is observed in children of all ages, the mean age at diagnosis is reported to be between 4.2 and 9.4 years.[5] Hereditary factors should be considered in children diagnosed outside this age range.[6]

While the etiology of USD in children is often not discernible, metabolic causes have been reported with increased frequency in recent years. The underlying metabolic disorder is present in at least 10-70% of children with USD.[5]

The absence of clarity regarding the classical signs and symptoms of USD in children, when compared with adults, results in delayed diagnoses, which can potentially lead to growth retardation, chronic pyelonephritis, or even end-stage renal disease.[3] In contrast, the incidence of chronic renal failure is highest in idiopathic calcium oxalate stones, inherited metabolic disorders such as adenine phosphoribosyltransferase (APRT) deficiency, cystinuria, Dent disease, familial hypomagnesemia with hypercalciuria and

nephrocalcinosis (FHHNC), and primary hyperoxaluria (PH) all frequently cause chronic kidney disease (CKD) and may progress to end-stage renal disease (ESRD).[7] Therefore, early identification, accurate diagnosis, and close clinical monitoring of patients with these inherited metabolic disorders are essential to reduce morbidity and prevent long-term renal complications.

Our study aimed to examine pediatric patients with USD who were followed up in our clinic for one year and to evaluate the frequency and various features.

MATERIALS AND METHODS

Study Design

This is a retrospective, observational, descriptive study conducted at the pediatric nephrology outpatient clinic.

Study Population

Our study included pediatric patients between 0 and 18 years of age diagnosed with USD based on clinical symptoms and confirmed by ultrasonography or other radiological imaging methods, between January 2016 and December 2016. Patients with incomplete data, prior genitourinary surgeries, or systemic diseases affecting calcium or oxalate metabolism were excluded from the analysis.

Ethical Approval and Informed Consent

Our study was conducted according to the Declaration of Helsinki. Ethical approval was obtained from the Kartal Dr. Lütfi Kırdar Training and Research Hospital Ethics Committee (No: 2017/514/104/4, Date: 28/03/2017).

Data Collection and Variables

Demographic characteristics such as age and sex, family history of USD, parental consanguinity, presenting symptoms, laboratory findings, metabolic evaluation results, stone location, imaging modalities, and treatment approaches were extracted from hospital records using a standardized data abstraction form.

Metabolic evaluation included 24-hour urine analyses, including calcium, oxalate, citrate, uric acid, and spot urine tests corrected for creatinine. Hypercalciuria, hyperoxaluria, hyperuricosuria, hypocitraturia, and cystinuria were defined according to standard pediatric reference ranges. The stone location was categorized as unilateral or bilateral.

Statistical Analysis

All statistical analyses were performed using the Jamovi program (Version 2.6) [Computer Software]. The variables were investigated using a visual histogram and Shapiro-Wilk's test to determine whether or not they were normally distributed. Descriptive analyses were presented using median and interquartile range for non-normally distributed data. Categorical variables were analyzed using the chi-square test. The one-way ANOVA test was used to compare multiple groups. P-values were adjusted using the Bonferroni correction method. A p-value <0.05 was considered statistically significant.

RESULTS

Among the 256 patients included in the study, urinary tract anomalies were identified in 22 patients (8.6%). Most patients (66.8%, n=171) had no comorbid conditions. Prematurity (6.3%), congenital heart disease (3.5%), growth retardation (2.3%), undescended testes (2.0%), and hypothyroidism (2.0%) were the most frequently observed associated conditions. Other systemic diseases were reported in 13.7% of the cohort (Table 1).

The most common reasons for admission were follow-up of previously diagnosed USD (21.5%), abdominal pain (10.5%), restlessness (8.6%), and hematuria (5.5%). Other complaints included urinary tract infection (3.9%), red discoloration or visible stones in the diaper (3.5%), urinary symptoms such as dysuria or incontinence (5.9%), and a smaller number of patients presented with fever, flank pain, or constipation. A considerable proportion of patients (27.8%) were diagnosed incidentally through abdominal or urinary tract ultrasonography performed for unrelated complaints. A statistically significant difference was found in symptom distribution across age groups ($p<0.0001$), with restlessness predominating in the 0–2 age group, hematuria in the 2–5 age group, and abdominal pain in children older than 5 years (Table 1).

Renal stones were localized to the left side in 105 patients (41.5%), the right side in 57 (22.5%), and bilaterally in 68 (26.9%) (Table 2). Renal ultrasonography revealed macrocalculi in 162 patients (63.3%), microcalculi in 60 (23.4%), and medullary nephrocalcinosis in 34 (13.3%). There was a statistically significant difference in stone size distribution across age groups ($p<0.001$) (Table 3).

Metabolic evaluation revealed hypocitraturia in 109 patients (50.7%), hyperuricosuria in 56 (28.3%), hyperoxaluria in 59 (28.2%), hypercalciuria in 56 (23.3%), and cystinuria in 2 patients (0.89%). Multiple abnormalities were noted in a subset of patients. Additional laboratory findings included proteinuria in 70 patients (30.7%), elevated spot urine Na/K ratio in 31 (21.2%), pyuria in 34 (13.2%), hematuria in 32 (12.5%), and positive urine cultures in 18 (7.1%). There were no statistically significant differences between age groups regarding the presence of hypercalciuria ($p=0.227$), hypocitraturia, hyperoxaluria, or cystinuria ($p>0.05$) (Table 4). The etiology of USD could not be identified in 47.7% (n=122) of the patients.

Of all patients, 154 (60.1%) received medical therapy, with potassium citrate being the most commonly prescribed agent. Surgical intervention was required in 45 patients (17.6%), including extracorporeal shock wave lithotripsy (12.6%), percutaneous nephrolithotomy (2%), and open surgery (3.1%) (Table 5).

Table 1. Demographic and clinical characteristics of pediatric patients with urinary stone disease

	n=256
Demographics	
Gender, n (%)	
Female	122 (47.7)
Male	134 (52.3)
Age, median [IQR]	39.5 [15.0-87.0]
Mode of delivery, n (%)	
Vaginal	183 (71.8)
Cesarean	72 (28.2)
Neonatal History, n (%)	
Prematurity	16 (6.3)
NICU admission	47 (18.4)
Oxygen therapy	31 (12.1)
Antibiotic therapy	35 (13.7)
Vitamin and Supplement, n (%)	
Usage of vitamin D	255 (99.6)
Duration of vitamin D use	
One year	255 (99.6)
Two years	1 (0.4)
Additional vitamin use	16 (6.3)
Additional medical conditions, n (%)	
None	171 (66.8)
Congenital heart disease	9 (3.5)
Growth retardation	6 (2.3)
Undescended testis	5 (2)
Hypothyroidism	5 (2)
Hemangioma	4 (1.6)
Cerebral palsy	3 (1.2)
Hydrocephalus	3 (1.2)
Autism	3 (1.2)
MTHFR gene mutation	3 (1.2)
Epilepsy	2 (0.8)
Myelomeningocele	2 (0.8)
Retinopathy of prematurity	2 (0.8)
Food allergy	2 (0.8)
Other Nephrology Conditions, n (%)	
None	228 (89)
UPJ obstruction	4 (1.6)
Polycystic kidney disease	3 (1.2)
Neurogenic bladder	3 (1.2)
CKD	2 (0.8)
Hydronephrosis	2 (0.8)
RTA type 2	2 (0.8)
Hypoplastic kidney	2 (0.8)
Renal agenesis	2 (0.8)
AME syndrome	2 (0.8)
Ectopic kidney	1 (0.4)
Megaureter	1 (0.4)
Horseshoe kidney	1 (0.4)
Single kidney	1 (0.4)
AKI	1 (0.4)
RTA Type 1	1 (0.4)
Solitary renal cyst	1 (0.4)
Congenital bladder cyst	1 (0.4)
Family History, n (%)	
Parental consanguinity	
None	242 (94.5)
2°	9 (3.5)
3°	3 (1.2)
4°	2 (0.8)
Family history of USD	
None	99 (38.7)
1°	53 (20.7)
2°	96 (37.5)
3°	8 (3.1)
Reason for Admission, n (%)	
Follow-up for known USD	55 (21.5)
Incidental US for non-nephrology reasons	37 (14.5)
US for other nephrological conditions	34 (13.3)
Abdominal pain	27 (10.5)
Restlessness	22 (8.6)
Hematuria	14 (5.5)
UTI	10 (3.9)
Vomiting	9 (3.5)
Red color or visible stones in the diaper	9 (3.5)
Urinary incontinence	7 (2.7)
Fever	6 (2.3)
Dysuria	5 (2)
Constipation	3 (1.2)
Foul-smelling urine	3 (1.2)
Flank pain	3 (1.2)

IQR: Interquartile range, NICU: Neonatal intensive care unit, MTHFR: Methylene tetrahydrofolate reductase, UPJ: Ureteropelvic junction, CKD: Chronic Kidney Disease, RTA: Renal Tubular Acidosis, AME: Apparent Mineralocorticoid Excess, AKI: Acute Kidney Injury, USD: Urinary Stone Disease, US: Ultrasonography, UTI: Urinary Tract Infection.

Table 2. Radiological findings of pediatric patients with urinary stone disease

Ultrasonography	
Renal stone, n (%)	
Right	57 (22.3)
Left	105 (41.0)
Bilateral	68 (26.6)
Renal stone maximum size, median [IQR]	
Right	3.00 [2.50-4.22]
Left	3.00 [2.37-4.07]
Medullary Nephrocalcinosis, n (%)	11 (4.3)
Ureteral stone, n (%)	11 (4.3)
Pelvicaliectasis, n (%)	25 (9.9)
Calycectasis, n (%)	23 (9.1)
Ureterectasis, n (%)	15 (5.9)
Direct urinary system radiography, n (%)	1 (0.4)

IQR: Interquartile range

Table 3. Age group–based distribution of renal stone size

n (%)	Macrocalculi	Microcalculi	Medullary	p
Total count	162 (63.3)	60 (23.4)	34 (13.3)	<0.012
<2 age	55 (34)	41 (68.4)	4 (11.8)	
2 to 5 age	39 (24)	8 (13.3)	5 (14.7)	
>5 age	68 (42)	11 (18.3)	25 (73.5)	

Table 4. Metabolic abnormalities identified in urinary stone disease according to age

n (%)	Total	<2 age	2 to 5 age	<5 age	p
Hypocitraturia	88 (34,4)	30 (34)	18 (20.5)	40 (45.5)	>0.1
Hypercalciuria	38 (14.8)	14 (36.8)	12 (31.6)	12 (31.6)	>0.1
Hyperoxaluria	16 (6.3)	6 (37.5)	6 (37.5)	4 (25)	>0.1
Cystinuria	2 (0.8)	1 (50)	0 (0)	1 (50)	>0.1
Cystinosis	2 (0.8)	1 (50)	1 (50)	0 (0)	>0.1

Table 5. Treatment modalities applied to pediatric urinary stone disease patients

n (%)	
Medical	181 (70.7)
Potassium citrate	177 (69.1)
Hydrochlorothiazide	2 (0.8)
Tiopronin	1 (0.4)
Cysteamine bitartrate	1 (0.4)
Surgery	45 (17.6)
Extracorporeal shock wave lithotripsy	32 (12.6)
Percutaneous nephrolithotomy	5 (1.9)
Open surgery	8 (3.1)

DISCUSSION

Our study gives pediatric USD by providing a detailed, age-specific analysis of clinical presentation, metabolic risk factors, stone characteristics, and treatment outcomes in a well-defined pediatric population from Türkiye, a known endemic region.

The existing literature shows that USD is more prevalent in males (3,13), while concurrent studies demonstrate a higher incidence of the condition in females.[5,8,9] In our study, the male-to-female ratio was 1.09, and USD was more prevalent in male subjects, consistent with the existing literature. As reported worldwide, urinary tract stones can occur at any age, with the mean age at diagnosis in children ranging from 7.3 to 9.4 years (spanning from 0.2 to 15 years).[7,10,11] Our study comprised 46.09% of subjects aged 0-2 years, 17.19% of subjects aged 2-5 years, and 36.72% of subjects aged over 5 years. The mean age of girls was 4.51±4.24 years, and the mean age of boys was 4.05±4.09.

A key distinguishing feature of our study is the age-related variability in symptomatology. We found that infants under two years old most commonly presented with restlessness or were asymptomatic, while in older children often reported abdominal or flank pain. The statistically significant age-related differences in presenting symptoms ($p<0.0001$) highlight the need for age-specific diagnostic awareness. Additionally, this pattern emphasizes the importance of being vigilant for urinary system disorders in young children who present with vague or behavioral symptoms.[12] Moreover, in 27.8% of patients, USD was diagnosed incidentally through imaging conducted for unrelated reasons. This finding aligns with literature emphasizing the role of ultrasonography in detecting asymptomatic stones due to its accessibility and safety.[13] Routine imaging in high-risk pediatric populations may facilitate early detection and timely intervention.

Metabolic risk factors were identified in 57% of our patients, supporting the idea that pediatric USD is often linked to underlying metabolic issues. Hypocitraturia was the most common abnormality in our cohort, surpassing hypercalciuria, which was traditionally considered the main metabolic etiology cause. This trend matches recent studies indicating a shift toward hypocitraturia as the leading risk factor in pediatric populations.[12] Hypercalciuria ranked second overall but was more common among infants, although this was not statistically significant, whereas hypocitraturia and hyperoxaluria were more common in older children. This age-related variation has also been seen in a national multicenter study by Başтuğ et al.,[3] which reported similar patterns. Additional findings included cases of hyperoxaluria, hyperuricosuria, and cystinuria. These findings highlight the need for comprehensive metabolic screening in all pediatric stone formers to enable timely diagnosis, targeted treatment, and prevention of recurrence.

Our study found that regarding stone size, 75.3% of patients had macrocalculi, while 24.7% had microcalculi. We observed that microcalculi were significantly more common in the 0–2-year age group (p=0.002), likely due to the increased use of high-resolution ultrasonography in young patients. This finding contrasts with the results of Melek et al.,[14] who reported that microcalculi were present in 35.8% of their patients. The lower rate of microcalculi in our cohort may be explained by differences in the definition threshold for stone size, operator dependency of ultrasonographic evaluation, or referral bias toward symptomatic patients with larger calculi. The higher frequency of microcalculi in younger children underscores the importance of early imaging. Therefore, early detection through routine ultrasonography in high-risk infants may allow for timely intervention or close monitoring, potentially avoiding the progression to clinically significant macrocalculi.

A positive family history, present in 61.3% of our patients, and parental consanguinity (5.5%) were notable risk factors. First-degree relatives accounted for 37.5% of familial cases. These findings parallel those of Amancio et al.,[15] who found familial clustering in 85% of cases, with 20.7% involving first-degree relatives. The concordance supports the theory that genetic predisposition, particularly when combined with metabolic abnormalities, plays a crucial role in pediatric USD.

Renal stones were observed in 89.8% of our patients, ureteral stones in 4.3%, and medullary nephrocalcinosis in 4.3%. No bladder or urethral stones were found. These findings differ from those of Ece et al.[5] and Amancio et al.,[15] who reported a higher frequency of lower urinary tract involvement. The predominance of renal localization in our cohort may be attributed to earlier detection through routine ultrasonography.

Our findings also reinforce the importance of metabolic evaluation, particularly in patients with a positive family history or consanguinity. Identification of correctable metabolic abnormalities allows for effective intervention, prevention of recurrence, and preservation of renal function. Regular follow-up and personalized management are essential components of care in pediatric USD.

Medical therapy was started in 70.7% of cases, mainly using potassium citrate (97.8%), reflecting its effectiveness in hypocitraturia, hyperuricosuria, and cystinuria. This is consistent with existing literature recommending potassium citrate for hypocitraturia and its benefits in other metabolic disorders, including hyperuricosuria and cystinuria.[3,16,17] Rare metabolic conditions such as cystinosis and cystinuria were managed with targeted agents like cysteamine and tiopronin. Our individualized treatment strategy-incorporating pharmacologic therapy, dietary counseling, and metabolic monitoring-may have contributed to reducing the need for invasive procedures.

This study has several limitations. First, its retrospective design restricts the ability to establish causality and is subject to potential biases related to data recording and completeness. Second, since it is a single-center study, the findings may not be generalizable to broader pediatric populations. Third, there was no availability of long-term follow-up data regarding stone recurrence, renal function outcomes, and treatment adherence, which prevents a thorough assessment of the effectiveness over time. Future prospective multicenter studies with longer observation periods are needed to address these limitations.

Conclusion

Our study contributes to the understanding of the multifactorial nature of pediatric USD in an endemic setting, with a focus on age-specific clinical features, metabolic risk factors, and family history. The prevalence of hypocitraturia, especially in older children, along with hypercalciuria in infants, may indicate a shifting metabolic profile that requires age-specific evaluation methods. Early imaging, particularly in high-risk or asymptomatic children, can detect microcalculi and help prevent disease progression. While further research is needed, our findings may support the utility of comprehensive metabolic screening and individualized treatment approaches in select patient populations. Although it is a retrospective, single-center study, it may offer valuable insights into the current landscape of pediatric USD and underscores the importance of early diagnosis and customized management to improve long-term kidney health.

Ethics Committee Approval

The study was approved by the Health Sciences University, Kartal Dr. Lütfi Kırdar City Hospital Ethics Committee (Date: 28.03.2017, Decision No: 2017/514/104/4).

Informed Consent

Retrospective study.

Peer-review

Externally peer-reviewed.

Authorship Contributions

Concept: İ.Ü., N.K., N.H.Z., Y.A.; Design: İ.Ü., N.K., N.H.Z., Y.A.; Supervision: İ.Ü., N.K., N.H.Z., Y.A.; Data collection &/or processing: İ.Ü.; Analysis and/or interpretation: İ.Ü., N.K.; Literature search: İ.Ü., N.K.; Writing: İ.Ü., N.K., N.H.Z., Y.A.; Critical review: İ.Ü., N.K., N.H.Z., Y.A.

Conflict of Interest

None declared.

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Pediatrik Nefroloji Polikliniğinde Üriner Sistem Taş Hastalığı Tanısı Alan Hastaların Değerlendirilmesi

Amaç: Bu çalışmada, pediatrik nefroloji polikliniğinde üriner sistem taş hastalığı tanısı alan çocuk hastaların demografik özelliklerinin, klinik başvuru şekillerinin, metabolik bozukluklarının, radyolojik bulgularının ve tedavi sonuçlarının değerlendirilmesi amaçlandı.

Gereç ve Yöntem: Bu retrospektif, tanımlayıcı çalışmaya, Ocak 2016 ve Aralık 2016 tarihleri arasında üriner sistem taş hastalığı tanısı alan 0–18 yaş arası 256 çocuk hasta dahil edildi. Hastaların klinik verileri, laboratuvar sonuçları, radyolojik bulguları ve uygulanan tedavi yöntemleri hasta dosyalarından elde edilerek istatistiksel olarak analiz edildi.

Bulgular: Hastaların %52.3'ü erkekti ve ortanca yaş 39.5 [15.0–87.0] ay olarak belirlendi. En sık başvuru nedeni bilinen taş hastalığı takibi (%21.5) olup bunu abdominal ağrı ve huzursuzluk izledi. Hastaların %61.3'ünde aile öyküsü pozitifti, %5.5'inde ise ebeveynler arasında akraba evliliği mevcuttu. Metabolik anormallikler olguların %57'sinde tespit edildi; en sık saptananlar hipositraturi (%34.4), hiperkalsiüri (%20.7) ve hiperoksalüri (%17.9) idi. Hipositraturi daha çok büyük çocuklarda, hiperkalsiüri ise bebeklerde daha yaygın olarak gözlendi. Hastaların %70.7'sine medikal tedavi başlandı ve bunların %69.1'inde potasyum sitrat kullanıldı. Cerrahi girişimler arasında ekstrakorporeal şok dalga litotripsi (%12.6), perkütan nefrolitotomi (%2) ve açık cerrahi (%3.1) yer aldı.

Sonuç: Pediatrik üriner sistem taş hastalığı sıklıkla metabolik bozukluklarla, özellikle hipositraturi ile ilişkili olup ailevi yatkınlık gösterebilir. Uygun görüntüleme ve metabolik tarama ile erken tanı konulması, ardından hedefe yönelik medikal tedavinin başlanması, renal komplikasyonların önlenmesi ve cerrahi girişim gereksiniminin azaltılması açısından önemlidir.

Anahtar Sözcükler: Metabolik bozukluk; pediatrik nefroloji; üriner sistem taş hastalığı.