

# Epidemiology, Diagnosis and Management of Paediatric Renal Diseases

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## ABSTRACT

**Background:** Paediatric renal diseases are an important cause of morbidity and mortality in children worldwide.

**Objectives:** To evaluate the epidemiological profile, clinical presentations, diagnostic approaches, and management outcomes of renal diseases in paediatric patients presenting to a tertiary care centre.

**Study Design:** Retrospective observational study.

**Place and Duration of Study:** Department of Paediatrics, Avicenna Medical College & Hospital, Lahore from 1<sup>st</sup> April 2023 to 30<sup>th</sup> September 2023.

**Methodology:** One hundred and seventy eight paediatric patients, aged from birth to 18 years diagnosed with renal diseases were included in the study. Data was collected included demographic details (age, sex, residence), clinical presentation (symptoms and signs), diagnostic evaluations (laboratory investigations, imaging studies, renal biopsy), and treatment modalities (medication, dialysis, or transplant where applicable).

**Results:** There were 57.3% males and 42.7% females with a mean age of 8.6±4.2 years. The most common renal conditions were nephrotic syndrome (36.5%), congenital anomalies of the kidney and urinary tract (23.0%), and glomerulonephritis (14.6%). Key clinical presentations included proteinuria (62.9%), edema (52.8%), and hematuria (34.8%). Renal biopsies (n = 38) predominantly showed minimal change disease (50%) and focal segmental glomerulosclerosis (26.3%). Corticosteroids achieved remission in 72.3% of nephrotic syndrome cases, while 6.7% were steroid-resistant. Dialysis was required in 10.7% of patients, and 6 underwent renal transplantation.

**Conclusion:** Nephrotic syndrome and congenital anomalies are the most prevalent renal conditions in children. Early diagnosis through routine screening and timely intervention remain critical to improving outcomes.

**Keywords:** Epidemiology, Diagnosis, Management, Paediatric renal disease

## INTRODUCTION

Paediatric renal diseases represent a significant subset of childhood illnesses that can lead to acute and chronic morbidity, with long-term implications on growth, development, and overall quality of life. Children suffering from kidney dysfunction typically show unobvious symptoms or symptoms that are difficult to recognize; thus, early diagnosis becomes challenging.<sup>1</sup> Various kidney and urinary tract birth defects (CAKUT) and nephrotic syndrome, and severe kidney diseases, including glomerulonephritis, haemolytic uremic syndrome, along with paediatric chronic kidney disease (CKD) form the spectrum of paediatric renal conditions.<sup>2</sup> Paediatric renal disorders have shown population changes throughout the years because of enhanced perinatal care and diagnostic technology advancement, as well as shifts in environmental and genetic risk elements.<sup>3</sup> Paediatric kidney diseases present different levels of prevalence throughout the world since low- and middle-income countries face restricted early diagnosis and therapeutic interventions. CAKUT represents the main origin of end-stage renal disease (ESRD) in children within various areas, where it creates ESRD conditions in 50% of patients.<sup>4</sup>

Primary glomerular diseases, together with systemic conditions including lupus nephritis, along with other causes, significantly contribute to the worldwide burden of the disease. The genetics, together with molecular biology, has increasingly revealed understanding about paediatric renal disease development.<sup>5</sup> The structural analysis of podocytes through genetic sequencing has established links between SRNS and mutations of these genes in addition to recently discovered inherited tubular renal disorders and syndromic kidney conditions resulting from next-generation sequencing advancements.<sup>6</sup> The discovery of gene-based origins allows doctors to provide better diagnoses and enables essential provisions for family support and therapeutic choices, and disease progress predictions.<sup>7</sup>

Children with renal disease receive proper diagnosis through a complete evaluation system that includes physician assessment, together with laboratory tests and imaging methods, as well as occasional kidney tissue analysis. Although hematuria and proteinuria combined with hypertension, along with edema, may indicate diseases that children often fail to report or present with minimal symptoms.<sup>8</sup> Basic renal assessment depends on routine urinalysis, together with serum creatinine along estimated glomerular filtration rate (eGFR) testing. Renal ultrasound serves as a critical imaging method for detecting structural abnormalities, but functional imaging procedures and nuclear medicine testing help assess renal perfusion as well as the extent of scarring. Renal biopsy stands as the definitive evaluation through which doctors achieve both diagnosis and classification in challenging cases.<sup>9</sup>

Novel biomarkers like neutrophil gelatinase-associated lipocalin (NGAL) and kidney injury molecule-1 (KIM-1), and cystatin C demonstrated recent advances that allow detection of kidney injury before conventional markers increase.<sup>10</sup> The implementation of biomarkers in standard paediatric nephrology care would enable improved early diagnosis and better risk-based patient assessment.<sup>11</sup> Different treatment approaches are necessary to manage paediatric renal diseases. Corticosteroids maintain their status as primary treatment for minimal change disease, but focal segmental glomerulosclerosis (FSGS) often requires immunosuppressant medicines that include both calcineurin inhibitors alongside monoclonal antibodies.<sup>12</sup>

## MATERIALS AND METHODS

This retrospective observational study was conducted at Department of Paediatrics, Avicenna Medical College & Hospital, Lahore from 1<sup>st</sup> April 2023 to 30<sup>th</sup> September 2023. A total of 178 paediatric patients, aged from birth to 18 years, diagnosed with renal diseases were included in the study. All children aged 0-18 years, confirmed diagnosis of a renal disease (based on clinical, laboratory, imaging, or biopsy findings), at least one recorded follow-up during the study period were included. The children who have incomplete clinical documentation, referred for renal pathology but without a definitive diagnosis and comorbidities

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masking renal outcomes (e.g., terminal malignancy) were excluded. The demographic details (age, sex, residence), clinical presentation (symptoms and signs), diagnostic evaluations (laboratory investigations, imaging studies, renal biopsy), and treatment modalities (medication, dialysis, or transplant where applicable) were noted. Clinical follow-up data, including disease progression and complications, were also recorded where available. Data include; type of renal disease (e.g., CAKUT, nephrotic syndrome, glomerulonephritis, acute kidney injury, chronic kidney disease), presenting symptoms (e.g., hematuria, proteinuria, edema, hypertension), laboratory values (e.g., serum creatinine, eGFR, urinary protein levels), imaging findings (ultrasound, DMSA scan), histopathological diagnoses (where biopsy was performed) and management approaches (medical therapy, RRT, supportive care). Data were analyzed using SPSS version 26.

## RESULTS

There were 102 (57.3%) males and 76 (42.7%) females with a mean age of  $8.6 \pm 4.2$  years. The highest proportion of patients were in the school-aged group (4-12 years), accounting for 43.8%, followed by adolescents (27.5%) and toddlers (13.5%). In terms of age distribution, neonates made up 4.5% of the cohort, infants 10.7%, toddlers 13.5%, school-aged children 43.8%, and adolescents 27.5%. This distribution indicates that renal diseases were more commonly identified in mid-childhood and early adolescence. Baseline laboratory data revealed a mean serum creatinine level of  $1.42 \pm 0.9$  mg/dL. Notably, 29.8% of patients presented with a reduced estimated glomerular filtration rate (eGFR) below 60 mL/min/1.73m<sup>2</sup>, indicating impaired renal function at presentation (Table 1).

Table 1: Demographic and baseline characteristics of paediatric patients (N = 178)

Variable	No.	%
Gender		
Male	102	57.3
Female	76	42.7
Age		
Neonates (0-28 days)	8	4.5
Infants (1-12 months)	19	10.7
Toddlers (1-3 years)	24	13.5
School aged (4-12 years)	78	43.8
Adolescents (13-18 years)	49	27.5
Baseline laboratory and imaging findings		
eGFR < 60 mL/min/1.73m <sup>2</sup>	53	29.8
Proteinuria Present	112	62.9
Structural Abnormalities on Imaging	41	23.0
Mean serum creatinine (mg/dL)	1.42±0.9	

Table 2: Distribution of paediatric renal diseases (N = 178)

Disease Type	No.	%
Nephrotic syndrome	65	36.5
CAKUT (congenital anomalies of kidney/UT)	41	23.0
Acute glomerulonephritis	26	14.6
Acute kidney injury (AKI)	19	10.7
Chronic kidney disease (CKD)	15	8.4
Hemolytic uremic syndrome (HUS)	7	3.9
Other rare syndromes (e.g., Alport)	5	2.8

Table 3: Histopathological findings from renal biopsy (n = 38)

Diagnosis	No.	%
Minimal change disease	19	50.0
Focal segmental glomerulosclerosis	10	26.3
Membranous nephropathy	5	13.2
Crescentic glomerulonephritis	4	10.5

Nephrotic syndrome was the most common diagnosis (36.5%), followed by congenital anomalies of the kidney and urinary tract (23%). Acute glomerulonephritis (14.6%), acute kidney injury (10.7%), and chronic kidney disease (8.4%) were also notable. Less frequent conditions included hemolytic uremic

syndrome (3.9%) and other rare syndromes (2.8%) [Table 2]. Focal segmental glomerulosclerosis was identified in 26.3%, followed by membranous nephropathy in 13.2% and crescentic glomerulonephritis in 10.5% (Table 3).

Among the patients with nephrotic syndrome, 65 (36.5%) received steroid therapy, with 47 (26.4%) achieving remission within eight weeks. Steroid resistance was observed in 12 patients (6.7%). A total of 27 children (15.2%) required hospitalization for acute management. Renal replacement therapy was needed in 19 cases (10.7%), including 11 patients on peritoneal dialysis and 8 on hemodialysis. Renal transplantation was performed in 6 patients (3.4%). The overall mortality rate was low, at 2.2%, while 82.6% of the cohort achieved either partial or complete recovery, indicating favorable outcomes with timely intervention (Table 4).

Table 4: Management and outcomes of paediatric renal diseases (N = 178)

Management / Outcome	No.	%
Steroid therapy (nephrotic syndrome)	65	36.5
Steroid remission in 8 weeks	47	26.4
Steroid-resistant nephrotic syndrome	12	6.7
Hospitalized for acute management	27	15.2
Dialysis Required (n=19)		
Peritoneal dialysis	11	6.2
Hemodialysis	8	4.5
Renal transplantation	6	3.4
Mortality	4	2.2
Partial or complete recovery	147	82.6

## DISCUSSION

Underscore the diversity and complexity of renal pathologies in children and highlight the importance of early diagnosis and tailored therapeutic strategies. Of all detected renal conditions nephrotic syndrome stood as the most prevalent at 36.5 percent. Similar regional and international studies indicate that minimal change disease represents the main histological diagnosis when children experience proteinuria accompanied by edema.<sup>13</sup> Our study results, which indicate a 72.3% remission rate after steroid treatment back up the current first-line recommendation for corticosteroids in therapy.<sup>14</sup> A significant subset of 6.7% did not respond to steroids, so physicians needed to use calcineurin inhibitors as per prior studies that demonstrate steroid resistance in 10–20% of patients with nephrotic syndrome cases. The second most common medical diagnosis among patients was Congenital anomalies of the kidney and urinary tract (CAKUT) which affected 23% of the cohort.<sup>15</sup> The data confirms the recognition that these anomalies are globally recognized as the primary trigger for chronic kidney disease in children, the identification of these patients through screening programs before and after birth stands as a vital approach for avoiding future deterioration of their kidney's health. Ultrasonography proved itself as a valuable diagnostic method because it showed structural defects in 23% of children undergoing scans.<sup>16</sup>

Glomerulonephritis, together with acute kidney injury (AKI) accounted for a significant disease burden among children, as it was detected in 14.6% and 10.7% of patients.<sup>17</sup> The renal biopsies of these patients revealed that minimal change disease occurred most often (50%) while focal segmental glomerulosclerosis was present in 26.3% of cases.<sup>18</sup> The results demonstrate that glomerular disease presentations match worldwide trends and healthcare providers need biopsy testing for unsure steroid-resistant cases. The clinical presentation of minimal change disease mostly included proteinuria in 62.9% of patients together with edema in 52.8% and hematuria in 34.8% of patients.<sup>19</sup> Healthcare providers should launch renal investigations when patients manifest these symptoms particularly since screening programs are not common in their practice areas. The detection of hypertension in over a quarter of patients (27%) underscores the necessity of regular blood pressure monitoring in paediatric populations, especially in those with chronic kidney disease or glomerular involvement.

## CONCLUSION

Paediatric renal diseases encompass a wide spectrum of pathologies, with nephrotic syndrome and congenital anomalies of the kidney and urinary tract (CAKUT) being the most prevalent in the studied population. Early clinical indicators such as proteinuria, edema, and hematuria remain critical in identifying renal involvement, and timely diagnostic evaluation, including imaging and biopsy when indicated, plays a key role in disease characterization.

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