



Abstract Type : Oral presentation

Abstract Submission No.: A-0411

Abstract Topic : Pediatric Nephrology + Inherited Kidney Disease

Advancing Pediatric Nephrology Through Genetics - A Retrospective Study From LMIC - A Single Centre Experience

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Objectives : To review the role of clinical genetics and Whole Exome Sequencing in children with Kidney disease. Kidney disease in children presents with an overlapping spectrum of manifestations. Genetic study is indicated basically for confirmation of diagnosis, to look for extra renal manifestations of suspected phenotype, Specific to glomerular disease it helps in avoidance of prolonged immunosuppressive therapy and in tubular disorders to modify and deliver targeted therapies.

Methods : In this Retrospective study we have included 50 children. A detailed family history and relevant clinical details was collected. Children who had clinical diagnose of kidney disease was subjected to Whole Exome sequencing(WES). The main disease categories were glomerular disease, aHUS, Cystic Kidney Disease, Tubular disorders, Nephrocalcinosis/Nephrolithiasis and Resistant Rickets(Table1).

Results : In a cohort of 50 children 24% were females and 76% were males. A positive genetic diagnosis was made in 38 out of 50 with overall yield of 76%. Genetic report was noted to be negative in 12 out of 50(24%)(Figure 1). Consanguinity was noted in 23 out of 50 enrolled children (46%). We also found a strong association between consanguinity and genetic positivity (78%). Out of 38 genetically positive children homozygous and heterozygous mutations were 47% and 53% respectively.

Conclusions : Genetic testing was useful in confirming suspected phenotypic variants, to establish new diagnosis and counselling. In glomerular disease identifying the genetic variants allowed us in forgoing kidney biopsy. Genetic analysis is a crucial step to offer targeted therapy and improve the overall morbidity as observed in our cohort.

Table 1 - Genetic.png



Table 1- Genetic positivity and Disease categories

Category	Diseases	Percentage(n=38)
Glomerular	Nephrotic Syndrome & Alport syndrome	6(15%)
HUS	aHUS	3(8%)
Tubulopathy	Distal Renal tubular acidosis, Proximal Renal tubular acidosis & Gitelman syndrome	13(34%)
Resistant Rickets	VDDR Type 1A	3(8%)
Cystic Kidney Disease	PKD1, MCDK, Nephronophthisis	4(10%)
Nephrocalcinosis/Nephrolithiasis	Renal stone disease without RTA, Hyperoxaluria	5(13%)
Others	White sutton syndrome Lung disease immunodeficiency	4(10%)

Table 1 - Genetic.png

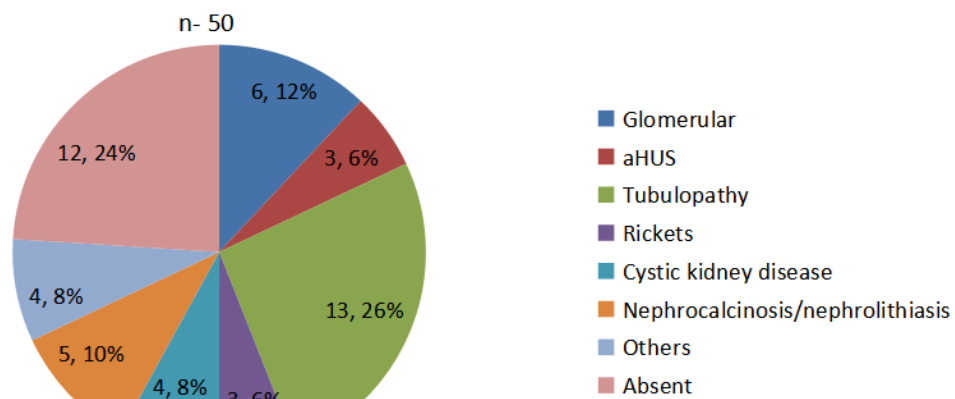


Figure 1 : Disease categories