Joshi Shivani et al., J Nephrol Ther 2014, 4:4
http://dx.doi.org/10.4172/2161-0959.S1.013

OMICS Group Conferences
Accelerating Scientific Discovery

3rd International Conference on
Nephrology & Therapeutics
June 26-27, 2014 Valencia Conference Centre, Valencia, Spain

Mutation spectrum in Danish patients with steroid resistant nephrotic syndrome

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Variations in genes encoding glomerular proteins like *NPHS1*, *NPHS2* and *INF2* have shown to be associated with steroid resistant nephrotic syndrome (SRNS). Patients with genetic forms of SRNS are usually unresponsive to immunosuppressive therapy and are at risk of developing end stage renal disease. Thus, knowing the underlying genetic etiology has important implications in therapeutic decisions. The study aimed to establish a rapid and highly sensitive method for scanning variations in SRNS genes and to determine their spectrum in the Danish patients. Variations in coding regions of *NPHS1* and *NPHS2* as well as hot spot exons of INF2 were scanned using high-resolution melting analysis and confirmed by bi-directional sequencing in 40 Danish patients diagnosed with sporadic SRNS. This analysis identified some previously described pathogenic variations like c.550G>A in INF2, c.686G>A and c.855_856del in *NPHS2* as well as c.349G>A, c.1223G>A, c.3047G>A and c.3230A>G in *NPHS1*. In both patients and controls we also identified known polymorphisms in *NPHS2* and *NPHS1*. In addition, it was identified that some novel exonic and intronic variants of unkown significance that are predicted to affect splicing of *NPHS1* and INF2. Interestingly, while most patients are compound heterozygous for potential pathogenic variations in *NPHS1*, some have single heterozygous variations in both *NPHS1* and *NPHS2*, respectively. Our results show that 1) high-resolution melting analysis is a rapid and sensitive technique for mutation screening of SRNS genes and 2) the identified variations do explain the occurrence of SRNS in approximately 50% of the patients suggesting further loci heterogeneity in Danish patients with SRNS.

Biography

Joshi Shivani was awarded an international mobility PhD scholarship from Aarhus University in 2010 and is currently pursuing a PhD in Translational Molecular Medicine at Aarhus. Her research is focused on studying the mutation spectrum in Danish patients with steroid resistant nephrotic syndrome; identifying new genes in steroid sensitive nephrotic syndrome and using animal models to understand the pathophysiology of nephrotic syndrome. She has presented her work at various national and international conferences. She has won several research grants in connection to her PhD, published a review on genetics of Nephrotic syndrome, is a member of Danish Society of Nephrology and is a Youth Goodwill Ambassador for Denmark.

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