Pierson Syndrome: Case Report

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Abstract:

Background: Pierson syndrome comprises of congenital nephrotic syndrome (CNS) and peculiar ocular changes. LAMB2 gene mutation has been reported to be causative reason for this rare autosomal recessive disorder.

Methods: An observational case series reports comprising of two children presenting with symptoms like heavy proteinuria, haematuria are being discussed. Physical examination along with ophthalmological assessment, hearing assessment, varied blood investigations, urinalysis, renal biopsy and gene testing were carried out to diagnose the condition.

Results: Pearson syndrome was detected with mutations of LAMB2 gene detected by Whole Exom Sequencing test in one of the case study. The Ocular abnormality in both patients comprised of squint hypertropia, a new variant ocular finding related to Pearson syndrome.

Conclusions: The clinical finding of squint hypertropia is a novel finding associated with Pearson syndrome, reported here for the first time.

Keywords:

Autosomal recessive; Pearson syndrome; Congenital Nephrotic Syndrome, Children.