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Congenital nephrotic syndrome and the heart: Lest we forget!

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Congenital nephrotic syndrome is defined by the presence of nephrotic range proteinuria, hypoalbuminemia and edema, with onset in the first three months of life. It is usually secondary to genetic mutations of the components of the glomerular filtration barrier, although infective causes must be ruled out. Congenital heart disease is extremely rare in congenital nephrotic syndrome, accounting for less than 20% of cases and is mostly associated with podocin mutation. We report a two month girl, presenting with anasarca in the first two months of life and was diagnosed to have congenital nephrotic syndrome. Infectious causes including malaria, cytomegalovirus, toxoplasmosis, syphilis, human immunodeficiency virus and rubella were ruled out. In view of a systolic murmur, echocardiography was done which revealed ostium secundum atrial septal defect and branch pulmonary artery stenosis. Genetic analysis showed homozygous single base pair duplication in exon 20 of the *NPHS1* gene (chr19:36332624dupG; Depth: 216x) resulting in a frameshift and premature truncation of the protein 6 amino acids downstream to codon 937 (p.Ser937GlnfsTer6; ENST00000378910.5). This is the first case of *NPHS1* (nephrin) mutation associated with congenital cardiac disease along with congenital nephrotic syndrome.

Biography

Deepanjan Bhattacharya has completed his MBBS from West Bengal University of Health Sciences and is currently a Postgraduate Resident Doctor in PGIMER, Chandigarh. He has published five papers in reputed journals.

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