Indian Journal of Pediatrics (April 2022) 89(4):411
https://doi.org/10.1007/s12098-021-04053-4

CORRESPONDENCE

# Homozygous *ARHGDIA* Gene Mutation in an 11-Month-Old Infant with Steroid-Resistant Nephrotic Syndrome

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Received: 26 July 2021 / Accepted: 12 November 2021 / Published online: 21 January 2022


*To the Editor:* An 11-mo-old developmentally normal girl was referred for evaluation of anasarca for the preceding 3 mo. At presentation, she had periorbital, pedal edema, severe ascites, and vulval edema. The weight was 9.3 kg (–1.30 *z*), length 79 cm (–0.61 *z*), and head circumference 44 cm (–0.66 *z*). She had stage 2 hypertension. Investigations showed hemoglobin 10.5 g/dL, leucocyte count $6.9\times10^9$/L, platelet count $285\times10^9$/L, blood urea 92 mg/dL, and serum creatinine 3.25 mg/dL. Hypoalbuminemia (2.1 g/dL), hypercholesterolemia (270 mg/dL), and nephrotic-range proteinuria (Up:Uc 5.5) without microscopic hematuria were noted. Ophthalmological evaluation and other systemic examination were unremarkable. She was born at 38 wk gestation as the firstborn to Telugu-speaking third-degree consanguineous parents (birth-weight 2.5 kg). Despite 8 wk of daily prednisolone, she had not attained remission.

We initiated peritoneal dialysis, but she remained anuric and eventually died 18 d later due to multiorgan failure. Renal biopsy could not be performed. Clinical exome sequencing revealed homozygous missense likely-pathogenic variation in exon 6 of *ARHGDIA* gene (chr17:g.79826865C>G) resulting in amino acid substitution of arginine for glycine (p.Gly168Arg). Prenatal counseling was provided.

Infantile nephrotic syndrome (INS) from India has been characterized by *NPHS1*, *NPHS2*, *LAMB2*, *WT1*, or *PLCE1* gene mutations [1, 2]. *ARHGDIA* mutations have not been reported in two large Indian series [1, 2] on INS. *ARHGDIA* mutations in INS have been reported so far in Pakistani [3], Moroccan, and Ashkenazi Jewish communities [4]. These cases had infantile-onset; showed diffuse mesangial sclerosis on renal biopsy; and rapidly progressed to end-stage renal disease (ESRD) or died [3, 4]. *ARHGDIA* mutations cause Rho-GTPase activation, causing podocyte damage [3, 4]. Unlike some previous cases [4], our patient did not have intellectual disability. Our report of INS due to *ARHGDIA* gene mutation adds to existing spectrum of genetic mutations in INS from India.

## Declarations

**Informed Consent** Written informed consent for publication of the child's clinical details was obtained from the parents.

**Conflict of Interest** None.

## References

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