Familial Renal Disease with Autosomal Dominant Inheritance

Dear Editor,

A 22-year-old female was admitted with 5-month amenorrhea (G2P1L1), edema over feet and reduced urine output for 20 days. Urine protein-to-creatinine ratio was 2.0 and serum creatinine was 6.0 mg/dL. Ultrasonography showed right kidney 7.9 × 2.8 and left kidney 7.2 × 2.3, both with lost corticomedullary differentiation. Due to worsening renal function, dialysis was initiated, and pregnancy was terminated.

At 40 years, her grandfather had kidney failure, managed medically until his death at 60. At 38, her father developed kidney failure, undergoing dialysis until his death at 55. Her son showed leg edema, with 2 + urine albumin on routine analysis, leading to further investigations.

A genetic analysis revealed a mutation in exon 2 of the INF2 gene., with a cytogenetic location at 14q32.33.[1] Its corresponding phenotypic MIM number is 613237.[2] This is suggestive of focal segmental glomerulosclerosis type 5, inherited in an autosomal dominant manner. A limited number of studies have documented similar genetic mutation.[3-5]

Identifying the mother's genetic pathology aids in treating her children. When treating her child, we can consider steroid-resistant nephrotic syndrome, avoiding unnecessary steroid use. Genetic analysis avoids unnecessary biopsy, and the genetic form of FSGS does not recur after kidney transplantation.[6] However, FSGS due to a circulating factor has a good response to steroids, but recurrence after transplant is high. The INF2 mutation is rare among Indians.[7] To date, only 38 cases of autosomal dominant FSGS have been identified globally.[8] Studying it involves genetic analysis in renal failure families, aiding in treatment decisions, early screening of family members, and guiding post-transplant outcomes.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

Conflicts of interest

There are no conflicts of interest.

Gurusamy Karthikeyan[1], Ramasubramanian Viswanathan[1], P. Shankar[1], Kannan Bhaba Velu[1]

[1]Department of Nephrology, Tirunelveli Medical College Hospital, Tirunelveli, Tamil Nadu, India

Corresponding author:
Gurusamy Karthikeyan, Department of Nephrology, Tirunelveli Medical College Hospital, Tirunelveli, Tamil Nadu, India.
E-mail: drkarthikeyan111@gmail.com

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