Senior-Loken Syndrome: A Rare Cause of End Stage Kidney Disease in a Child

Dear Editor,

A 14-year-old boy, second-born from a third-degree consanguineous marriage, presented with joint pain in both lower limbs for 2 years. Examination showed pallor, no dysmorphic features, and height and weight <3rd percentile. Oculomotor apraxia was present. Investigations revealed Hb: 5.0 g/dL, serum Ca/PO_4: 6.3/8.7 mg/dL, Alkaline Phosphatase (ALP): 2194U/L, intact parathyroid hormone (iPTH): 1207.2 ng/L, 25(OH)Vit D: 128.64 ng/mL, pH: 7.30, HCO_3: 18 mEq/L, and serum urea/creatinine: 256/6.6 mg/dL. Ultrasound kidney ureter bladder showed bilateral shrunken kidney. Fundus examination showed retinitis pigmentosa [Figure 1]. Senior-Loken syndrome (SLSN) due to chronic kidney disease and retinitis pigmentosa (R) was suspected. Whole exome sequencing showed IQCB1 (ENST00000310864.11): c.488-1G>A, homozygous, pathogenic, autosomal recessive variant (as per Human Genome Variation Society Classification) associated with Senior-Loken syndrome 5 (OMIM#609254), confirming the diagnosis. The child was administered maintenance hemodialysis, antihypertensive drugs, oral sodium bicarbonate, calcium, sevelamer, and calcitriol. Family genetic testing was not done due to financial constraints [Supplementary File].

SLSN is a rare autosomal recessive oculo-renal disease with a prevalence of 1:1,000,000.[1,2] The SLSN5 is caused by mutations in the *NPHP5/ICQB1* gene, first identified by Otto *et al*.[1] in 2005. It was first concurrently described by Senior *et al*.[3] and Loken *et al*.[4] in 1961 as a combination of familial juvenile nephronophthisis and Leber congenital amaurosis. Few cases have emerged from India, of which only one has been genetically proven.[5] Severe infantile, juvenile, and adolescent are the three clinical nephronophthisis types. Severe infantile is the most severe form, and juvenile is the most common. The age of onset is ~5 years, as in this case. This letter intends to raise awareness that ocular involvement is the most common presenting complaint in this syndrome. Our patient suffered from difficulty in vision since 5 years of age and was misdiagnosed with myopia and prescribed spectacles.

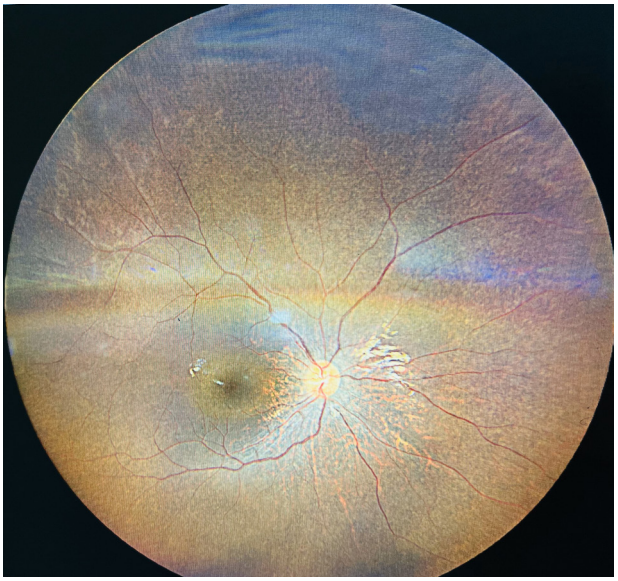

Figure 1: Fundus examination (direct ophthalmoscopy) showing retinitis pigmentosa.

Conflicts of interest: There are no conflicts of interest.

Payal Singh[1], Abhishek Abhinay[1], Ankur Singh[1], Rajniti Prasad[1]

[1]Division of Pediatric Nephrology, Department of Pediatrics, IMS, Banaras Hindu University (BHU), Varanasi, Uttar Pradesh, India

Corresponding author: *Abhishek Abhinay, Division of Pediatric Nephrology, IMS, Banaras Hindu University (BHU), Varanasi, Uttar Pradesh, India. E-mail: abhinay.abhishek@bhu.ac.in*

References

1. Otto EA, Loeys B, Khanna H, Hellemans J, Sudbrak R, Fan S, *et al*. Nephrocystin-5, a ciliary IQ domain protein, is mutated in senior-loken syndrome and interacts with RPGR and calmodulin. Nat Genet 2005;37:282-8.
2. Hildebrandt F, Attanasio M, Otto E. Nephronophthisis: Disease mechanisms of a ciliopathy. J Am Soc Nephrol 2009;20:23-35.
3. Senior B, Friedmann AI, Braudo JL. Juvenile familial nephropathy with tapetoretinal degeneration A new oculorenal dystrophy. Am J Ophthalmol 1961;52:625-33.
4. Loken AC, Hanssen O, Halvorsen S, Jolster NJ. Hereditary renal dysplasia and blindness. Acta Paediatr (Stockh) 1961;50:177-84.
5. Duraiswamy A, C.O. B. Senior-loken syndrome: A case report. KPJ 2020;35:57-60.

How to cite this article: Singh P, Abhinay A, Singh A, Prasad R. Senior-Loken Syndrome: A Rare Cause of End Stage Kidney Disease in a Child. Indian J Nephrol. doi: 10.25259/IJN_168_2025

Received: 17-03-2025; **Accepted:** 28-03-2025; **Online First:** 26-05-2025; **Published:** ***

DOI: 10.25259/IJN_168_2025

Supplementary available at: https://dx.doi.org/10.25259/IJN_168_2025