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LETTER TO THE EDITORS

Outcome of females with Alport syndrome

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Dear Editors,

We read with great interest the paper by Goka et al. [1]. We want to present the clinical characteristics of our female patients with Alport syndrome. In the last 10 years, we had 8 cases. The presenting feature was persistent microscopic hematuria in six patients, recurrent macroscopic hematuria in one patient, and the last patient had kidney failure at admission. Mean ages at the time of presentation and at diagnosis were 5.87 ± 3.08 (1.5–10.0) and 8.43 ± 2.91 (4.5–12.0) years, respectively. Five patients had a family history of chronic kidney disease. All patients had microscopic hematuria and three had proteinuria at admission. Only the patient with kidney failure had hypertension. An 11.5-year-old girl had unilateral mild sensorineural hearing loss; the other patients had normal audiology exams. Seven patients were diagnosed by genetic analysis and one by kidney biopsy. The patient with kidney failure had homozygous *COL4A3* mutation (p.Val1476fsX (c.4425_4426insT)). One patient had homozygous *COL4A4* mutation (c.4684 delT (Tyr1562fs)) and one had compound heterozygous *COL4A5* mutation (p.Gly953Val (c.2858G > T) and p.Ile444Ser (c.1331 T > G)). The other 4 cases had heterozygous *COL4A3* mutations (p.Asp263Asn (c.787G > A), p.Gly392Glu (c.1175 G > A), p.Arg408His (c.1223 G > A) and c.547–9 A > C). Mean follow-up duration was 5.28 ± 1.87 (4.0–9.0) years, and mean age at last follow-up visit was 13.71 ± 3.30 (8.5–18.5) years. The patient with kidney failure underwent kidney transplantation. At the end of the follow-up period, all cases had an estimated glomerular filtration rate > 90 ml/min/1.73 m^2. Only one of the 8 patients had macroscopic hematuria as a new sign. None of the patients developed new-onset proteinuria. The patient with unilateral mild sensorineural hearing loss had bilateral significant hearing loss when she was 14 years old.

In our series, 5 of 7 patients had *COL4A3* mutations, different from the report by Goka et al. [1], in which most cases had *COL4A5* mutations. The clinical signs of Alport syndrome in females may be variable. Although the only sign was persistent microscopic hematuria in many of the patients, some may develop kidney failure at early age. Hearing loss may not be present at childhood.

Declarations

Conflict of interest The authors declare no competing interests.

Reference

1. Goka S, Copelovitch L, Levy Erez D (2021) Long-term outcome among females with Alport syndrome from a single pediatric center. Pediatr Nephrol 36:945–995

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