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Averting Complications in Alport's Syndrome through Early Diagnosis and Treatment

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ABSTRACT

Alport syndrome is a rare genetic disorder of specialized basement membranes in the kidney, ear, and eye, characterized by haematuria with progressive hereditary nephritis, high-frequency sensorineural hearing loss (SNHL) and pathognomonic ocular lesions. It is one of the spectra of diseases representing hereditary nephritis, which inevitably leads to end-stage renal disease (ESRD). Microscopic or frank haematuria persistent from childhood constitutes the clinical clue for its early recognition. It occurs as a result of genetically inherited or de novo mutations in type IV collagen genes. The most common mode of inheritance is X-linked and men are more severely affected. We report a case of a middle aged man, in his fourth decade of life presenting with persistent haematuria, thrombocytopenia associated with SNHL and anterior lenticonus, diagnosed as a previously undetected case of Alport syndrome.