Hypertensive retinopathy in a young patient! Don't forget Alport syndrome

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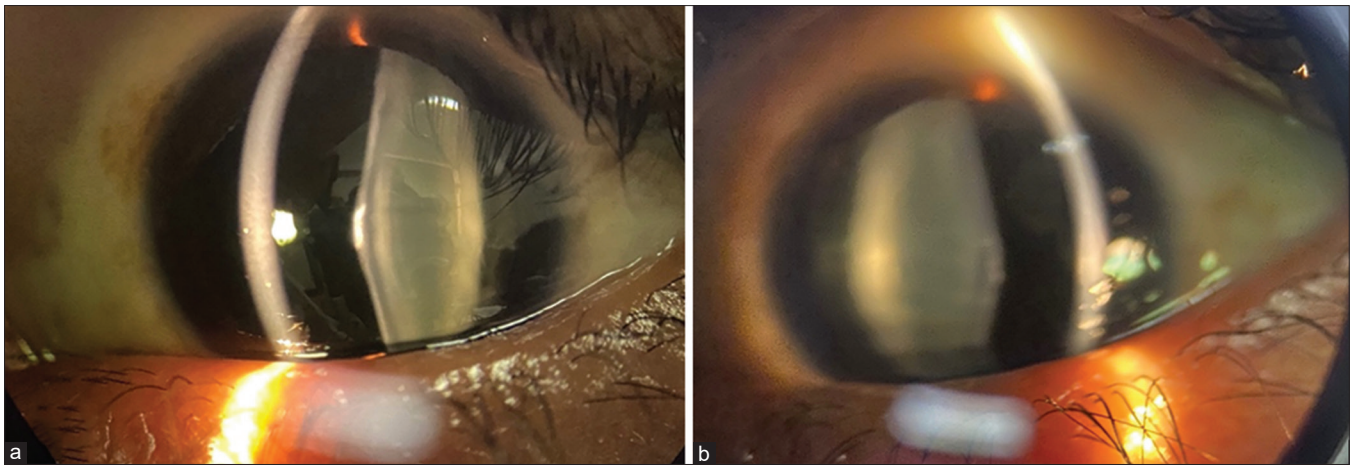

Figure 1: (a and b) Anterior Lenticonus in a patient of suspected Alport's Syndrome

Key words: Alport syndrome, Lenticonus, Nephropathy, Retinopathy

A 24-years-old male diagnosed with chronic kidney disease stage 5 was referred to the ophthalmology department for progressive diminution of vision for the last 1 year. On examination, his best-corrected visual acuity (BCVA) was 20/200 in both eyes. The patient had a clear cornea and a quiet anterior chamber. Dilated examination on a slit lamp showed the presence of anterior lenticonus in both the eyes with few opacities on the anterior as well as posterior capsule [Fig. 1]. Fundus examination revealed arteriolar attenuation, and there were fleck-like opacities in the periphery [Fig. 2]. A provisional diagnosis of Alport syndrome was made, which was confirmed by the presence of sensorineural hearing loss on otological evaluation. Genetic evaluation was advised; however, the patient succumbed to chronic renal failure. Our case is interesting as the diagnosis of Alport syndrome was made after ophthalmic evaluation.

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Furthermore, it highlights the importance of early diagnosis as it may improve patient survival.

Discussion

Alport syndrome is a genetic disorder with multisystem involvement that includes the characteristic clinical triad of chronic renal failure, sensorineural deafness, and ocular signs due to its effect on the alpha chains of collagen type IV.[1] The inheritance is usually X linked (80%); however, it can be autosomal recessive and exceptionally autosomal dominant.[2] Ocular involvement presents in the form of corneal pathology, anterior lenticonus, and retinopathy.[3] Corneal pathology may present as recurrent corneal erosions or posterior polymorphous corneal dystrophy. The lens capsule in these patients is fragile, and thus results in an anterior or, rarely, a posterior lenticonus. The defective collagen in these patients results in a thinned out Bruch's membrane and internal limiting membrane. This combined with the impaired clearance of waste products results in the characteristic dot and fleck retinopathy and "lozenge" sign (dull macular reflex as a result of the demarcation between the perimacular retinopathy and the thinned macula).[4]

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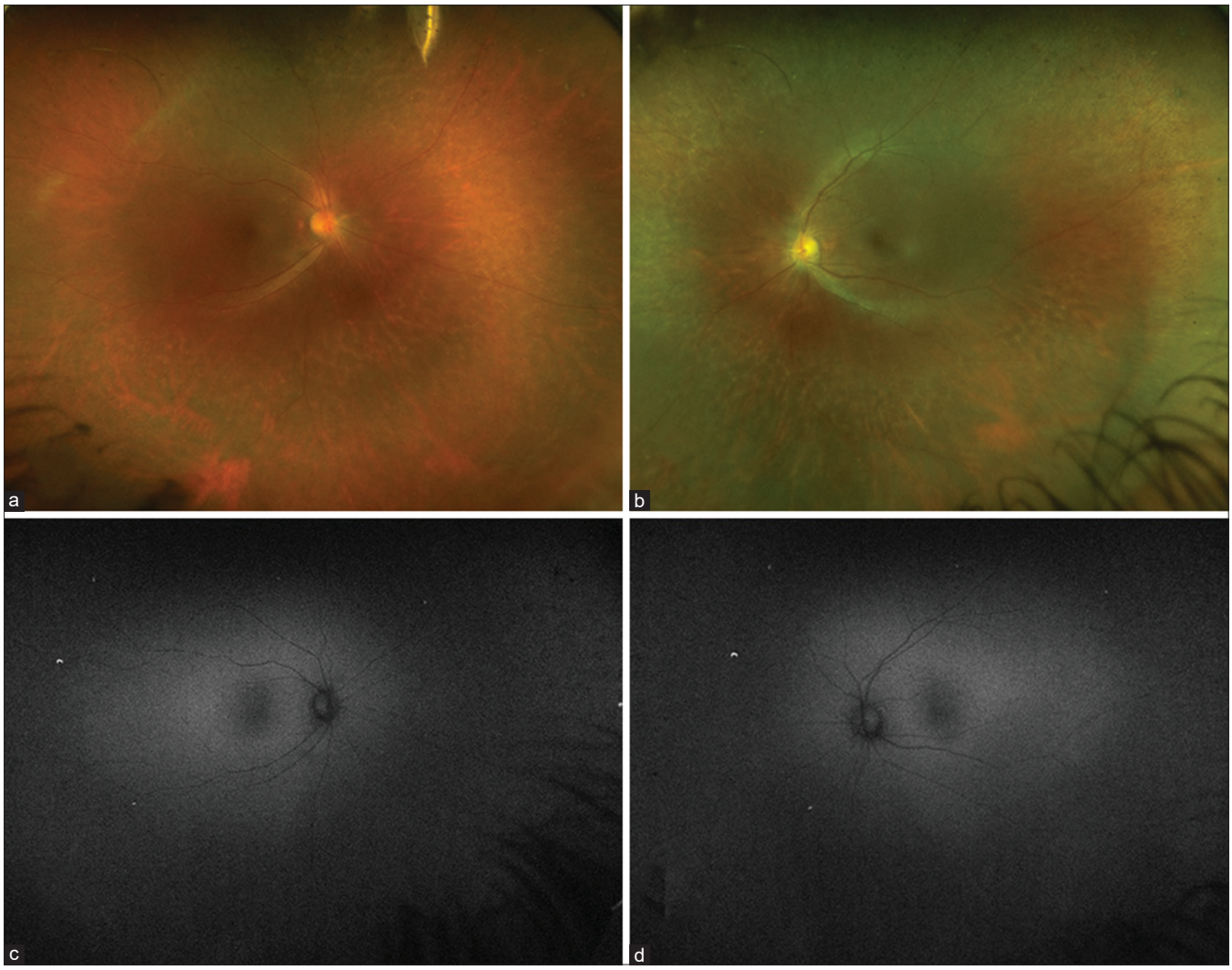

Figure 2: Fundus photographs (a and b) and Fundus Autofluorescence images (c and d) of the patient showing retinal flecks in the peripher

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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