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Central Flecks and Temporal Macular Thinning in Alport Syndrome

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Abstract | **Case Report**

A 43-year-old man was referred to our department for ophthalmic examination as a candidate for simultaneous liver and kidney transplantation. The underlying cause of liver and kidney failure had not been identified. The examination of the anterior segment revealed posterior polymorphous corneal dystrophy and corneal guttata. Fundus examination confirmed bilateral foveal sparing perimacular fleck retinopathy. Optical coherence tomography demonstrated symmetrical temporal retinal thinning. Based on these findings, Alport syndrome was strongly suspected. Clinician should be aware of the importance of evaluating ophthalmic examinations in patients with unexplained renal dysfunction.

Keywords: Alport syndrome, fleck retinopathy, temporal macular thinning.

INTRODUCTION

Alport syndrome is an inherited disease characterized by progressive renal failure, hearing loss, and ocular abnormalities [1, 2]. The characteristic ocular features of Alport syndrome are corneal opacities and posterior polymorphous corneal dystrophy, anterior lenticonus and cataract, central perimacular and peripheral coalescing fleck retinopathies, and temporal retinal thinning [1-4]. These findings are highly suggestive of Alport syndrome in the diagnostic process. Herein, we report a case of suspected Alport syndrome in a patient with unexplained renal dysfunction.

CASE REPORT

A 43-year-old man was referred to our department for ophthalmic examination as a candidate for simultaneous liver and kidney transplantation. The underlying cause of liver and kidney failure had not been identified. The patient had a history of cataract surgery at the age of 4. He is undergoing hemodialysis. His best-corrected visual acuity was 20/20 in both eyes. Slit lamp examination of the anterior segment revealed posterior polymorphous corneal dystrophy (Figure 1).

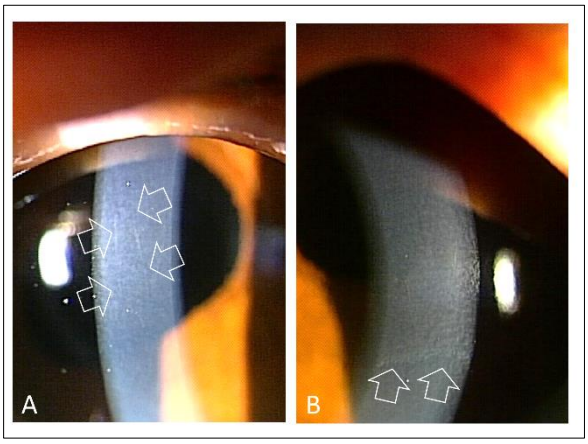

Figure 1: Slit lamp photograph of the right (A) and left (B) eyes Note posterior polymorphous corneal dystrophy (arrows). Specular microscopy of the corneal endothelium demonstrated corneal guttata (Figure 2).

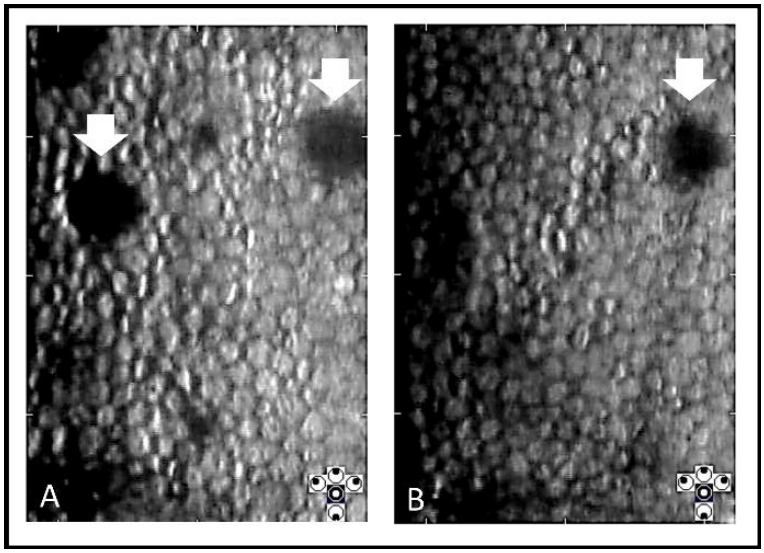

Figure 2: Specular microscopic finding of the right (A) and left (B) eyes Note corneal guttata (arrows). Fundus photography confirmed bilateral foveal sparing perimacular fleck retinopathy (Figure 3).

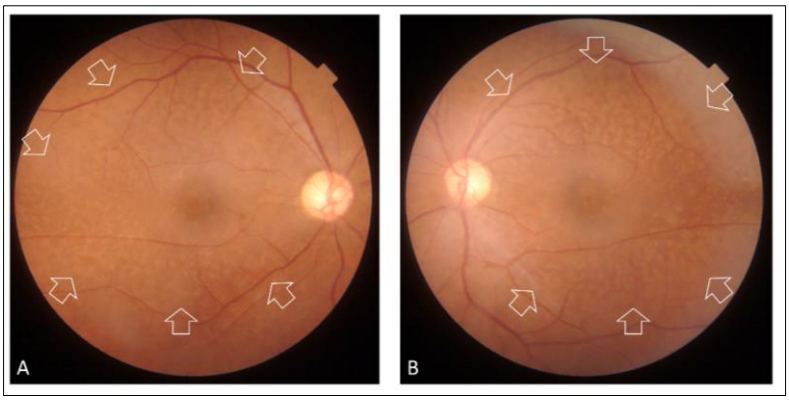

Figure 3: Fundus photograph of the right (A) and left (B) eyes Note bilateral foveal sparing perimacular flecks (arrows).

Optical coherence tomography demonstrated the accumulation of hyperreflective materials on the internal limiting membrane surface with symmetrical temporal retinal thinning (Figure 4). Based on these findings and his history, Alport syndrome was strongly suspected.

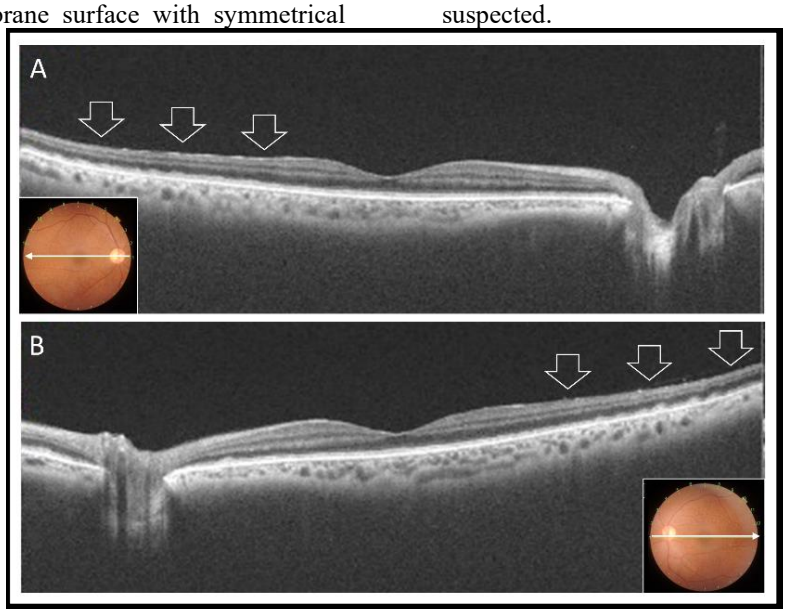

Figure 4. Optical coherence tomography of the right (A) and left (B) eyes Note the accumulation of hyperreflective materials on the internal limiting membrane surface with temporal retinal thinning (arrows).

Discussion

Alport syndrome is an inherited disease characterized by progressive renal failure, hearing loss, and ocular abnormalities. Mutations in the *COL4A5* (X-linked), or *COL4A3* and *COL4A4* (autosomal recessive) genes result in absence of the collagen IV α3α4α5 network from the basement membranes of the cornea, lens capsule, and retina and are associated with corneal opacities, anterior lenticonus, fleck retinopathy, and temporal retinal thinning [1].

Central fleck retinopathy is present in 60% of men and at least 15% of women with X-linked Alport syndrome and 50% of individuals with recessive disease [1, 2]. It is more common with early-onset renal failure and lenticonus. The central retinopathy varies from scattered whitish-yellow dots and flecks to a dense, almost confluent annulus around the region of temporal retinal thinning. The clear demarcation of the central retinopathy from the foveola is consistent with involvement of the inner limiting membrane and retinal nerve fiber layer. This is not a true membrane but rather, results from fusion of the Muller cell end plates and incorporates the collagen IV α3α4α5 network. Temporal retinal thinning is very common in men and women with X-linked Alport syndrome, and with recessive disease [1, 2]. Therefore, the investigation for ocular abnormalities has an important clinical role in Alport syndrome.

Conclusion

This case highlights the importance for clinicians to be aware of evaluating ophthalmic examinations in patients with unexplained renal dysfunction.

References

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