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Original Research Paper

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ALPORT'S SYNDROME-A CASE REPORT

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ABSTRACT Alport syndrome is an oculorenal syndrome characterized by a triad of clinical findings consisting of haemorrhagic nephritis, sensorineural hearing loss and characteristic ocular findings which include anterior lenticonus,fleck retinopathy and posterior polymorphous corneal dystrophy We report a case of two siblings who presented to us with similar features of anterior lenticonus and fleck retinopathy.Anterior lenticonus was evident on both ophthalmoscopy and slit lamp examination. Ocular examination is particularly helpful in diagnosis of Alports syndrome when genetic testing is not easily available or results are inconclusive.

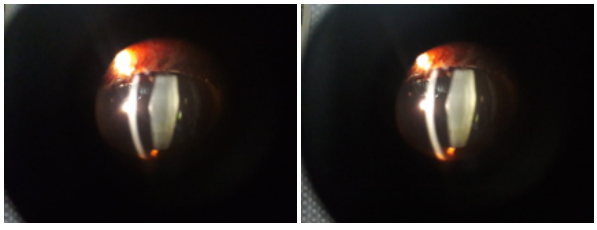
KEYWORDS : Anterior lenticonus, Fleck retinopathy,Hematuria,Haemorrhagic nephritis

CASE REPORT

We are reporting the clinical findings of 2 siblings , 15 and 17 yrs of age respectively, who presented to the Ophthamology Outpatient Department of ACS Medical College in April 2018 ,with complaints of progressively deteriorating vision in both eyes since past 2 years .

There was associated history of hearing loss noticed 5 years ago. There was no history of consanguinity in the parents. Family history revealed that their father had expired due to unknown causes 3 years ago.

Best corrected visual acuity was 6/24 in both their eyes. Anterior segment examination revealed Anterior Lenticonus in both eyes of both patients. Additionally elder brother had Posterior Lenticonus in both eyes .Dilated Fundus examination revealed Retinal Pigment Epithelial mottling in midperipheral retina in both eyes .Keratometry readings showed an average value of 46.00D in both patients.Postdilatation Retinoscopy revealed High Myopia in the range of -10.00 to -18.00 DS



A Probable diagnosis of Alports syndrome was made and the patients were referred to Nephrology clinic and ENT clinic . Audiometry evaluation revealed moderate sensorineural hearing loss in both patients .Laboratory investigations revealed Albuminuria and Haematuria . Nephrologist suggested Oral ACE inhibitors in view of proteinuria. Regular follow up was advised . Nil Ophthalmic intervention planned at present and the patient's family was counseled about need for regular follow up and surgical management in the future .

DISCUSSION

Alport syndrome is characterized by hematuria, progressive renal failure, hearing loss, and ocular abnormalities affecting the cornea, lens, and retina[2,3,4]

Inheritance of Alport syndrome is X-linked in nearly all families (85%), and mutations affect the *COL4A5* gene, which codes for the collagen IV *α*5-chain [8,9].

The etiology of this syndrome is unknown. It has been suggested that there may be a metabolic defect in the biosynthesis of collagen with changes in the glomerular basement membrane, cochlea and capsule of the lens[1]

Alport syndrome affects at least one in 10,000 individuals, and the diagnosis is important because of the risk of disease in other family members; also, early treatment with angiotensin-converting enzyme inhibitors delays the onset of end stage renal failure [6,7].

CONCLUSION: We would like to highlight that the role of the Ophthalmologist is very important in the early detection of this syndrome .

Ocular examination is particularly helpful in diagnosis of Alports syndrome when genetic testing is not easily available or results are inconclusive[5]

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