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CASE REPORT

ALPORTS SYNDROME-CASE REPORT

Manish Nagpal[1], Raminder Singh[2], P N Nagpal[1]

INTRODUCTION

Alports syndrome is a rare heritable renal disorder associated with a glomeruler abnormality and commonly associated with anterior lenticonus. This bilateral disease was first definitely described by Alport as an acute haemorrhagic nephropathy with deafness in 1927[1], with the lenticonus recognised later. It is most often transmitted as an X linked recessive trait, severe disease developing in affected males, with some expression in female heterozygotes, autosomal dominant and recessive transmissions are also described. The systemic manifestation in alports syndrome are progressive asymmetric hearing loss and glomerulosclerosis, causing haematuria, proteinuria and renal failure by 20-50 years.

The most typical ophthalmic manifestations in Alports syndrome are anterior lenticonus and anterior polar cataracts. Anterior lenticonus was specifically linked to Alports syndrome by Arnott and coauthors in 1996[2]. Neilson has asserted that anterior lenticonus does not exist apart from Alport's syndrome[3]. Thompson et al stated that anterior lenticonus is a sign of advanced renal disease in this syndrome[4]. Earlier reviews of the literature on anterior lenticonus, notably by Arnott and coauthors in 1966 and by Connor in 1975, indicate that only about 30% to 40% of patients with anterior lenticonus have a concomitant diagnosis of nephritis or Alports syndrome. Slip lamp examination may show calcium crystals in the conjunctiva, juvenile arcus and corneal endothelial pigment. The retinal appearance may mimic Retinitis Pigmentosa or fundus albipunctatus[5], often with abnormal ERG recordings and optic nerve may contain drusen.

[1] Asopolov Eye Hospital
[2] Raghudeep Eye Clinic

Corresponding author: Manish Nagpal Ms, FRCS (Ed), Retina Foundation, Aspalov Eye Hospital, Near Shahibag Underbridge, Ahmedabad-4, gujarat, India.

CASE REPORT

We examined a 17 year old male having Alports syndrome referred to us by a nephrologist for a routine ophthalmic checkup. The patient had undergone Renal transplantation twice in 1993 and 1996 respectively for chronic renal failure. His mother is a diagnosed carrier of this disease and one of his sisters died of renal failure in 1994. Apparently he had normal renal function when he was referred to us. He is completely deaf due to the disease completed with his taking aminoglycosides and frusemide for his renal condition.

On ophthalmic examination he was found to have anterior lenticonus in both eyes. The left eye had a classical lenticonus along with a break in the anterior capsule (Figure 1 & 2) while the right had very minimal changes suggestive of a cone. The fundus showed normal looking discs with some whitish spots at the macula (Figure 3). Fluorescein angiography was not possible due to his condition. His best corrected visual acuity was 6/36p in each eyes.

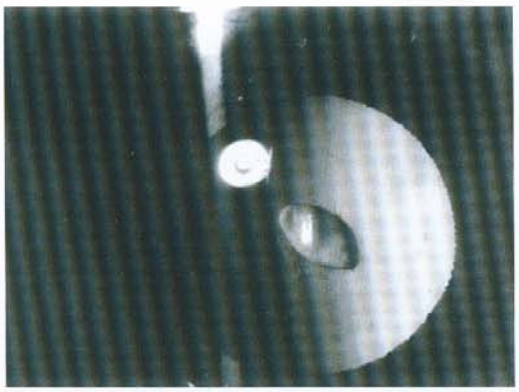

Fig. 1: *Anterior lenticonus with a break in the anterior capsule*

Fig. 2: *Anterior lenticonus on a slit*

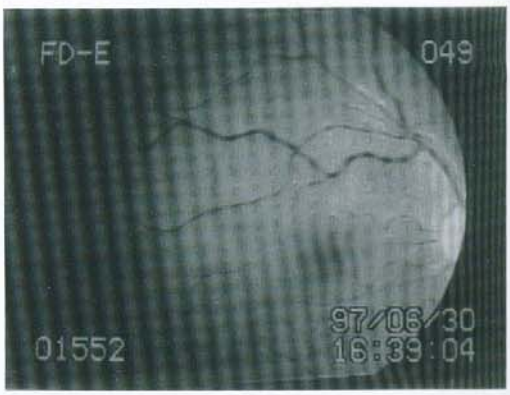

Fig. 3: *Fundus showing macular flecks*

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