Case report

Alport Syndrome: case report and review of ocular manifestations

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Abstract

Background: Alport Syndrome is an uncommon disease.

Case: We report a case of a young Indian male who presented with the characteristic ocular findings and systemic features of Alport Syndrome.

Conclusion: Any young patient with a chronic renal disease should have a careful ophthalmologic examination for Alport Syndrome.

Key words: Alport Syndrome, hereditary nephritis, lenticonus, deafness, retinopathy

Introduction

Alport Syndrome is an oculorenal syndrome characterized by a clinical triad of hemorrhagic nephritis, progressive sensorineural deafness and characteristic ocular abnormalities (Govan 1983, Colville et al 1997). The common ocular findings include dot and fleck retinopathy, anterior lenticonus and posterior polymorphous corneal dystrophy. However, posterior lenticonus, cataract, recurrent corneal erosion and macular degeneration have also been reported (Jayaprasad B et al 1994).

Case report

A 21-year-old male patient attended our out-patient department with bilateral progressive diminution of vision. The patient was a diagnosed case of chronic hemorrhagic nephritis and was on dialysis. He had one an elder brother who had died of chronic renal disease at the age of twenty three. Besides visual complaints and dysuria, the patient also complained of progressive hearing loss and swelling all over the body.

On systemic examination the patient was found to be anaemic (Hb 4.3 mg/dl) and hypertensive. His biochemical examination showed increased blood urea (232 mg/dl) and serum creatinine (13.1 mg/dl).

There was a borderline hyponatremia (131 mg/dl) and hypokalemia (6.6 mg/L). Urine examination showed proteinuria (albumin 4+) and microscopic haematuria. Renal ultrasound showed normal-size kidneys with increased echogenicity consistent with medical renal disease. Audiometry revealed bilateral moderate to severe sensorineural hearing loss.

On ocular examination, uncorrected visual acuity was 5/60 in both eyes which had improved to 6/36 with – 1.75 DS. Slit-lamp examination revealed anterior lenticonus in both eyes. A characteristic old droplet appearance was seen on retro-illumination. The rest of the anterior segment was normal. Fundus examination revealed no specific pathology in the retina.

A clinical diagnosis of Alport Syndrome was made on the basis of the classical triad and the family history.

Received on: 25.01.2011 Accepted on: 12.05.2011
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Right Eye

Left Eye

Slit-examination showing anterior lenticonus

Discussion

Alport Syndrome, or hereditary nephritis, is a rare, progressive form of glomerular disease with an approximate incidence of 1 in 50,000 live births (Mukerji et al 2003). 85 % of patients show X-linked dominant type of inheritance though autosomal recessive and autosomal dominant inheritance patterns are also reported (Mahajan et al 2003).

Alport Syndrome is a primary basement membrane disorder arising from mutations in type IV collagen biosynthesis genes (Lemmink et al 1997, Srinivasana et al 2009) .The impaired integrity of type IV collagen in the glomerular basement membrane, cochlea and anterior lens capsule leads to the clinical manifestations seen in Alport Syndrome.

Alport Syndrome often affects young adult males who develop recurrent or persistent hematuria and proteinuria often ending in renal failure. Hearing loss and ocular abnormalities are never present at birth and usually become apparent by late childhood or early adolescence.

Anterior lenticonus is the pathognomic feature of Alport Syndrome and its presence in any individual is highly suggestive of Alport Syndrome (Mahajan et al 2003, Arnolt et al 1966; Singh et al 1977). It is a valuable marker of the disease severity and is almost entirely restricted to patients with progression to end stage renal disease and deafness before age 30.

Anterior lenticonus presents at an average age of 26 years and is bilateral in 75 % of patients. It causes slowly progressive axial myopia leading to the necessity of having to frequently change glasses. The diagnosis is confirmed when the central part of lens projects anteriorly by 3 - 4 mm in an axial projection on biomicroscopic examination. This condition rarely progresses to spontaneous rupture of lens capsule (Sathish et al 2001). Attenuation and fracturing of the anterior lens capsule have been demonstrated by electron microscopy in patients with lenticonus (Citrik et al 2007).

Dot and fleck retinopathy is the most common ocular manifestation of Alport Syndrome which usually becomes apparent at the onset of renal failure (Gehrs et al 1995). The rare ocular manifestations of Alport Syndrome are posterior polymorphous corneal dystrophy, posterior lenticonus, cataract, recurrent corneal erosion and macular degeneration.

The diagnosis of Alport Syndrome can be made clinically when the disease presents with classical features of hereditary hemorrhagic nephritis, sensorineural deafness and anterior lenticonus. Electron microscopic examination of renal biopsy specimen is diagnostic in cases without classic finding.

Treatment involves medial management of progressive renal insufficiency with dialysis or renal transplantation for renal failure. Sensorineural hearing loss can be managed with hearing aids. Anterior lenticonus, when visually significant, may be managed by lens extraction (John et al 1995). The ophthalmologist plays an important role in the early detection of this syndrome (Zhang et al 2008).

Conclusion: Any young patient with a chronic renal disease should have a careful ophthalmologic examination.

References

Alport AC (1927). Hereditary familial congenital hemorrhagic nephritis. Br. Med J; 1: 504-6.

Arnolt, Crawfurd, Toghill (1966). Anterior lenticonus and Alport's Syndrome. Br J Ophthalmol; 50: 390.

Citrik M, Batmen C, Men G(2007). Electron microscopic examination of anterior lens capsule in case of Alport's syndrome. Clin Exp Optom; 90(5): 367 -70.

Colville DJ, Savige J (1997). Alport syndrome. A review of the ocular manifestations. Ophthalmic Genet; 18 (4): 161-73.

Gehrs KM, Pollock SC, Zikha G (1995). Clinical features and pathogenesis of Alport retinopathy. Retina; 25: 305.

Govan AA (1983). Ocular manifestations of Alport's syndrome: a hereditary disorder of basement membrane. Br J Ophthalmol; 67: 493-503.

Jayaprasad B, Sathish KR, Chandrasekhar N, Upadhyaya NS, Mehta S (1994). Alport's syndrome: A case report. Indian J Ophthalmol; 42: 211-2.

John ME, Noblitt RL, Coots SD, Ballew C, Kenyon J (1995). Clear lens extraction and intraocular lens implantation in a patient with bilateral anterior lenticonus secondary to Alport's Syndrome. J Cataract Refract Surg; 21:363-4.

Lemmink HH, Schroder CH, Monnens LA (1997). The clinical spectrum of type IV collagen mutations Hum Mutat; 9 (6):477-99.

Mahajan SK, Sud S, Sood BR, Patial RK, Prashar N, Prashar BS (2003). Alport syndrome: Case report. JIACM; 4:337-9.

Mukerji N & Dodson K (2003). X-linked Alport Syndrome: A Case Report. The Internet J of Nephrology; 1: 42.

Prasad RA, Bhatnagar RK, Ratnakar C,Veliath AJ (1996). Alport's syndrome: A case report. Indian J Pathol Microbiol; 39: 225-7.

Sathish KR, Chandrasekhar N, Pai V, Bhatia P, Kakkar G (2001). Spontaneous capsular ruptures in Alport syndrome. Annals of Ophthalmology; 33: 131-5.

Singh DS, Bisht DB, Kapoor S, Sharma RN, Sankaran K, Majumdar NK (1977). Lenticonus in Alport's syndrome. Acta Ophthalmologica; 55: 164-70.

Srinivasana M, Uzel SGM, Gautieri A, Keten S, Buchler MJ (2009). Alport Syndrome mutations in type IV tropocollagen alter molecular structure and nanomechanical properties. J. Structural Biology; 168(3): 503-10.

Zhang KW, Colville D, Tan R (2008). The use of ocular abnormalities to diagnose X-linked Alport Syndrome in children. Pediatr. Nephrol; 23(8):1245-50.

Source of support: nil. Conflict of interest: none