A Case Report

A Case of Alport Syndrome in an 18-Year-Old Male: Clinical Presentation, Diagnosis, and Multidisciplinary Management

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ABSTRACT

Alport syndrome is a rare genetic disorder characterized by progressive kidney failure, sensorineural hearing loss, and eye abnormalities. We present the case of an 18-year- old male with shortness of breath and cough with sputum, who was found to have a family history of kidney disease and suffered progressive vision and hearing loss since the age of 10. The patient was diagnosed with Alport syndrome based on clinical criteria. He underwent dialysis and was referred to specialists for management of his vision and hearing loss. The diagnosis and management of Alport syndrome are discussed, highlighting the importance of early diagnosis and timely intervention.

Keywords: *Alport syndrome,Genetic disorder,Kidney failure,Hearing loss,Eye abnormalities*

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INTRODUCTION

AL port syndrome is a hereditary nephropathy. It is caused by mutations in type IV collagen genes COL4A3, COL4A4, and COL4A5 that lead to defective basement membranes in kidneys, ears and eyes(1). It is most commonly inherited as an X-linked disorder. The disease exhibits significant phenotypic variability depending on the pattern of inheritance and type of mutation(2). Here, we present a case of patient with Alport syndrome.

CASE REPORT

An 18-year-old male presented to emergency department with shortness of breath and cough with sputum. His father gave a history of abdominal discomfort, decreased oral intake and vomiting for past few months. Upon evaluation he had bibasilar lung crackles and peripheral edema. Labs showed decreased hemoglobin(7g/dl), creatinine of 10.8mg/dl and blood urea of 208mg/dl. His arterial blood gases revealed pH 7.35, HCO3 12.9 mmol/L and pCO2 23 mmHg. After initial stabilization, patient was admitted in our medical ward.On systemic review, it was found that patient has progressively decreased vision as well as decreased hearing since the age of 10 years. He also had family history of death in maternal uncle at a young age of 25 years due to complications of CKD.

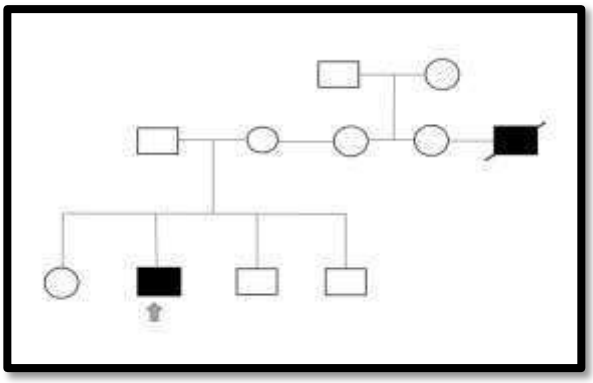
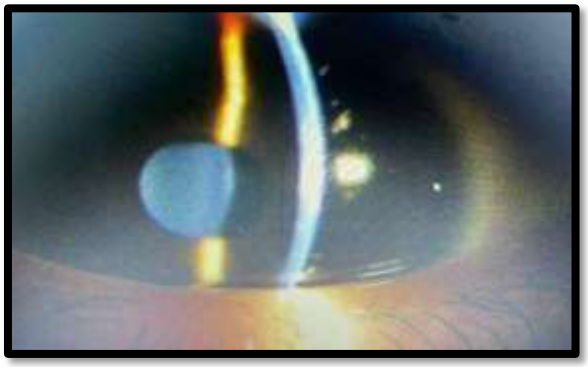


Figure 1. Pedigree chart showing x-linked pattern of inheritance. Arrow=patient On examination, patient had sallow complexion, poor physical growth with thin extremities, protruding ears and half and half nails. Eye examination was remarkable for anterior lenticonus and audiogram showed bilateral sensorineural hearing loss.



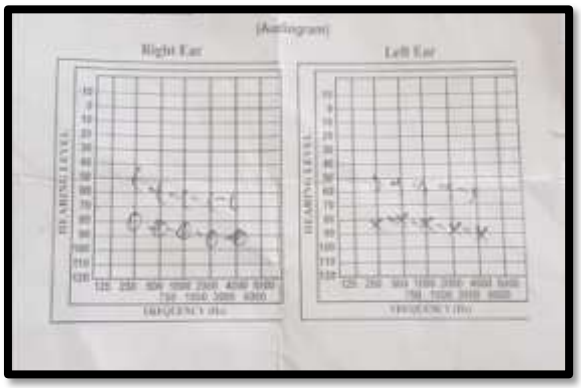


Figure2a.Anterior lenticonus. 2b.Audiogram showing sensorineural hearing loss

Further labs revealed RBCs in urine (10-20/HPF) and proteinuria +++. Ultrasound showed normal sized kidneys with grade III echogenicity. This patient met the clinical criteria for Alport syndrome(3).

MANAGEMENT

Patient was diagnosed with CKD and had few sessions of dialysis that led to significant improvement of his symptoms. Patient was referred to an ophthalmologist and ENT specialist for management of respective problems. A follow-up appointment was arranged with a nephrologist to decide on starting angiotensin converting enzyme inhibitor. Parents were counselled regarding the disease, genetic testing and treatment options as well as about evaluation of mother and siblings and that how early diagnosis and treatment can be beneficial.

DISCUSSION

Alport syndrome is a hereditary nephropathy diagnosed by combination of clinical features, family history and genetic testing. Early diagnosis is crucial because timely intervention can greatly benefit the patients. There is evidence showing the effectiveness of using angiotensin converting enzyme(ACE)

inhibitors to slow down the progression of renal disease and thus delaying the need for replacement therapy and transplant(1,2). The diagnosis should be considered in patients with hematuria detected on urinalysis and family history of kidney disease(3). Treatment at this point is only aimed to slow progression of renal disease. Hearing aids can help with the sensorineural deafness. Emerging therapies for Alport syndrome, such as antisense oligonucleotide(ASO) and others are being investigated and may provide disease-modifying treatment in the future(4).

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Authors Contribution

Concept & Design of Study Ahmad Zeb1

Drafting: Sayyeda Aisha Bahar2 **Data Analysis:**

Sayyeda Aisha Bahar2

Critical Review: Ahmad Zeb1

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