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ALPORT SYNDROME IN A 13 YEAR OLD BOY- A CASE REPORT

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ABSTRACT

Alport syndrome, also referred to as hereditary nephritis, is an inherited progressive form of glomerular disease that is often associated with sensorineural hearing loss and ocular abnormalities**.** Alport syndrome is a primary basement membrane disorder arising from mutations in genes encoding several members of the type IV collagen protein family. The disease accounts for approximately 3% of children and 0.2% of adults with chronic renal failure and for >1% of patients receiving renal replacement therapy. The majority of patients with chronic renal failure are male due to the X-linked inheritance pattern. The symptoms of this disease are renal manifestations, hearing loss, ocular manifestations, leiomyomatosis, arterial disease, phenotype-genotype correlation, heterozygous females with X-linked disease and Other. Here we present a case of 13 year old boy who had symptoms of hearing impairment and chronic kidney disease due to hereditarily acquired alport syndrome. The provision of rescue therapy led to the amelioration of symptoms showing the benefit of early recognition and treatment of Alport syndrome.

KEYWORDS: Alport Syndrome, Hereditary Nephritis, end Stage Renal Failure, Sensorineural Hearing Loss.

INTRODUCTION

Alport syndrome is a congenital and genetic, progressive renal disease that is accompanied by haematuric nephritis, high frequency sensorineural hearing loss (SNHL) and ocular abnormalities. It is the second highest cause of inherited chronic kidney disease (CKD). It is a rare disease that affects 1 in 50,000 live births.[1,2] Patients affected by Alport syndrome experience progressive loss of kidney function, usually resulting in end-stage kidney disease.

Most common symptom of Alport syndrome is the presence of blood in urine (hematuria). 80-99% patients will have glomerulopathy, hematuria, morphological abnormality of the semicircular canal, retinopathy and sensorineural hearing impairment.

Early diagnosis and treatment is essential to reduce mortality and further complications. Diagnosis of the condition is based on family history of the patient, clinical signs and specific testing such as a kidney biopsy. The diagnosis can also be confirmed by genetic testing.

There is no specific treatment for Alport syndrome. Controlling the progression of the disease and treating the symptoms is the main aim of the therapy. Patients with end stage renal failure is treated with peritoneal or hemodialysis and kidney transplantation. High frequency sensorineural hearing loss (SNHL) is treated with the use of a hearing aid.

CASE PRESENTATION

A 13 year old boy presented to our hospital with the complaints of progressive hearing loss for one and a half months. Along with abnormalities in hearing, he also had complicated UTI condition. He also reported the complaints of cold for past two months and dry cough on and off. At the time of presentation, he reported complaints of burning sensation in the abdomen, severe headache, nasal block and ear block. His mother was operated for the complaints of impaired hearing. Similar illness of deafness and chronic kidney disease in the family was reported. The patient had no complaints discharges from the ear, ear pain or sore throat. On systemic examination he was found to be normal. The biochemical workup revealed no abnormal parameters. The patient was treated with cephalosporin antibiotic Tablet cefixime 100mg twice daily, Tablet renosave (Taurine 500mg and N-acetylcysteine 150mg), Tablet B-complex and Tablet sodium bicarbonate500mg. He was temporarily relieved of the present complaints and was asked to report to the hospital if any of the symptoms are further worsened. The patient was asked to visit physician to control the risk factors for renal failure during his entire life. Advised the patient to prevent ototoxic medications and exposure to high noise.

DISCUSSION

Alport syndrome is genetic disorder identified by kidney impairment, hearing loss and eye abnormalities. Cecil A. Alport, in 1927, first described the disorder. Majority of Alport syndrome are inherited in an X linked pattern. The disease is genetically heterogeneous, existing in X-linked, autosomal recessive and autosomal dominant forms.[3] 80% of the

disease is X-linked, 15% autosomal recessive, and 5% autosomal dominant. The most common, X-linked form develops from mutations in COL4A5, the gene encoding the alpha-5 chain of type IV collagen.[4]

Mutation of COL4A3, COL4A4, or COL4A5 causes Alport syndrome. Mutations in these genes result in abnormalities of the type IV collagen in glomeruli that prevent the kidneys from properly filtering the blood. As a consequence, blood and protein pass into the urine which progressively leads to kidney failure. The renal symptoms are generally hematuria, edema and hypertension. Organ of Corti is the inner structure of the ear that is responsible for transforming sound waves into nerve impulses for the brain. Type IV collagen is also an important component of the organ of Corti. Mutations in type IV collagen result in abnormal inner ear functions, which can lead to hearing loss. This may result in high-tone bilateral sensorineural hearing loss.[5] Type IV collagen helps a very important role to maintain the shape of the lens and the cells of the retina. Mutations found in Alport syndrome may affect the retina and the shape of the lens.

Family history and physical examination are used for the diagnosis of alport..Kidney biopsy is done to look for signs of the condition such as abnormalities of the cells of the glomeruli. An ophthalmological examination is also done.

Treatment for Alport syndrome is not definite. However it can be managed with supportive cares such as ACE inhibitors, angiotensin receptor blockers and aldosterone inhibitors.[6,7] In patients with severe proteinuria cyclosporine can be used to prevent disease progression. In those with end stage renal disease,both dialysis and kidney transplantation are the treatment options.[8]

CONCLUSION

It is important to recognize Alport's syndrome early in the course of the disease.

This is facilitated by an integrated approach to diagnosis. Early diagnosis can improve longevity and improve prognosis of Alport's syndrome patients.

Alport syndrome is to be detected at the earliest in order to prevent the severe complications that may be precipitated at the end stages. This can be achieved by strategic and integrated diagnostic workup. Detection of the disease in its earlier stages can lead to increased prognosis in patients affected.

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