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Endocrinology & Metabolic Syndrome

Letter to Editor

Letter on Alport Syndrome

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Introduction

Alport syndrome is a rare genetic disorder. People with this syndrome experience Kidney malfunction, Hearing loss, Abnormalities in Eyes [1].

Individuals who are experiencing Alport syndrome will have Hematuria (Blood in the Urine) and also proteinuria (high levels of protein). With these abnormalities, kidneys are not able to function normally results end-stage renal disease (ESRD).

Individuals experiencing Alport syndrome also develop hearing loss. Abnormalities cause in inner ear. They also develop abnormalities in eyes (vision loss).

This Syndrome is mostly seen in Men than in females. Severity of the disease can be seen equally in males and females.

Causes of Syndrome

Alport syndrome is an inherited disease, which caused by the mutations in COL4A5 gene and is inherited in an X-linked pattern. A characteristic of X-linked inheritance is father cannot pass X-linked traits to their sons. It is an autoimmune disorder [2].

The estimated effect of this rare disorder is approximately 1 in 5,000-10,000 people and account 3% of children with chronic kidney disease and 0.2% of adults with end-stage renal disease in the United States.

Few disorders are related to Alport Syndrome based upon the similar symptoms.

Diagnosis

This disorder can suspect on symptoms. Various specialized tests need to perform to confirm.

Clinical testing, Genetic testing, Skin biopsy, Kidney biopsy, Urine analysis, Hearing test, Eye test, Speech test should perform [3].

Treatment

There is no cure and no treatment completely.

A team of specialists like paediatricians, nephrologists, audiologists, ophthalmologists, genetic counsellor, and other healthcare professionals requires in treating this rare genetic disorder [4].

Psychosocial support for the entire family is necessary as well.

Angiotensin-converting enzyme (ACE) inhibitors have been used to treat individuals with Alport syndrome [5-10].

Three clinical trials have demonstrated the benefit of SGLT2 inhibitors in treating the Alport syndrome [11,12].

Dialysis is one of the procedures to treat syndrome. A kidney transplant is also preferred for individuals. Alport syndrome does not recur in kidney transplants. However about 3% or less of transplanted Alport patients make antibodies to normal collagen proteins in the transplanted kidney, causing severe inflammation of the transplant.

Hearing aids can be used to treat hearing loss.

Surgeries will perform to remove cataracts when it is necessary.

Registries

Alport syndrome registries have been established in several countries. Two of the largest Alport syndrome registries are in USA. Registries have also been established in the Europe, United Kingdom, France, Italy, Australia, China and other countries. A registry is a special database that contains information about individuals with a specific disorder or group of conditions. The collection of data about rare disorders may enable researchers to increase the understanding of disorders, treatments, and accelerate clinical trials in specific treatment. Medical practitioners are encouraged to submit data to treat patients with Alport syndrome.

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Received: July 23, 2020; **Accepted:** July 27, 2020; **Published:** July 31, 2020

Citation: Himabindhu G (2020) Letter on Alport Syndrome. Endocrinol Metab Syndr. 9:309. doi: 10.4172/ 2161-1017.20.9.309

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