Published on *My46* (https://www.my46.org) 01/23/2022

My46 Trait Profile

Alstrom syndrome

Alstrom syndrome is a rare genetic condition that affects many organs of the body. It can cause vision, hearing, heart, kidney, lung, and liver problems. It is caused by mutations in the ALMS1 gene that makes a protein called alstrom syndrome protein 1.

Characteristics of Alstrom syndrome

Alstrom syndrome is a rare genetic condition that often causes blindness, cardiomyopathy (condition affecting the heart muscle), sensorineural hearing loss (damage to the inner ear or the nerve pathway from the inner ear to the brain), and childhood obesity (overweight). Diabetes and obesity are also seen in many affected individuals. Other features commonly seen in Alstrom syndrome include seizures, respiratory problems (such as bronchitis, asthma), and kidney and liver problems. Intelligence is usually normal in those affected with Alstrom syndrome though some can have delayed development. Affected individuals usually die before the age 50 due to organ failure. People affected with this condition may show variation of the features, even within the same family.

Diagnosis/Testing

Individuals with Alstrom syndrome have a change or mutation in a gene called ALMS1. This gene makes a protein called the Alstrom syndrome protein 1. Although the exact function of this protein is not known, it is present in many areas or tissues of the body. This may explain why many different organ systems are affected in Alstrom syndrome. This protein is thought to be involved in the movement of other proteins within the body (protein trafficking) and cilia function (cilia are hair-like structures on cells that help to move various substances in the body). Changes or mutations in the ALMS1 gene are thought to interfere with this “moving system.” Genetic testing is available and can detect the mutations in some, but not all, affected individuals. Therefore, the diagnosis is made primarily by physical examination and other laboratory tests.

Management/Surveillance

Management of Alstrom syndrome includes treatment for vision (e.g. prescription glasses) and hearing impairment (e.g. hearing aids). Nutritional intervention (healthy diet), exercise and treatment for the heart disease and diabetes are also needed. Other interventions maybe required depending on other symptoms the affected individual has. In general, regular evaluations are recommended for the areas of the body that are commonly affected in Alstrom syndrome (eyes, ears, heart, kidney, liver, etc).

Individuals with Alstrom syndrome should avoid any substance(s) that are not recommended for persons with kidney or heart failure. Careful attention is needed to ensure treatment for one system/symptom does not affect another system in a negative way – e.g. certain medications used to treat diabetes may not be recommended if the person also has heart failure.

Mode of inheritance

Alstrom syndrome is inherited in an autosomal recessive pattern. This means that an individual has to inherit two ALMS1 mutations (i.e., one from each parent) to be affected with Alstrom syndrome. If both parents are carriers of an ALMS1 mutation, they have a 1 in 4 (25%) chance with each pregnancy of having a child with Alstrom syndrome.

Risk to family members

Parents of a child with Alstrom syndrome are carriers of Alstrom syndrome. If a sibling of a child with Alstrom syndrome is unaffected, he/she has a 2 in 3 (66%) chance of being a carrier of Alstrom syndrome.

Special considerations

None

Resources

Alstrom Syndrome International
http://www.alstrom.org/
Genetics Home Reference: Alstrom syndrome
http://ghr.nlm.nih.gov/condition/alstrom-syndrome

References

Joy, T. et al. (2007).Alstrom syndrome (OMIM 203800): a case report and literature review." Orphanet Journal of Rare Diseases (2): 49.

Marshall JD, Paisey RB, Carey C, Macdermott S. (Updated 31 May 2012). Alstrom Syndrome. In: GeneReviews at GeneTests Medical Genetics Information Resource (database online). Copyright, University of Washington, Seattle. 1997-2013. Available at http://www.ncbi.nlm.nih.gov/books/NBK1116/. Accessed [04/17/2014].

Marshall, J. et al. (2013).Clinical utility gene card for: Alstrom syndrome – update 2013." European Journal of Human Genetics 21(11).

Marshall, J. et al. (2011).Alstrom Syndrome: Genetics and Clinical Overview." Current Genomics 12(3): 225-235.

Created: 04/2014 **Created by:** Marcia Jodah, MS, CGC
Updated: mm/yyyy **Edited by:** Seema Jamal, MSc, LCGC