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COMMENTARY

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Causes, Symptoms and Preventive Measures of Alstrom Hallgren Syndrome

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Description

A relatively uncommon autosomal recessive genetic illness called Alström syndrome (AS), also known as Alström Hallgren syndrome, is distinguished by childhood obesity and various organ failure. Early-onset type 2 diabetes cone-rod dystrophy which causes blindness, sensorineural hearing loss, and dilated cardiomyopathy are among the symptoms. Typically, endocrine conditions including hypergonadotrophic hypogonadism, hypothyroidism, and acanthosis nigricans brought on by hyperinsulinemia also manifest. Nearly half of those with Alström syndrome experience developmental delay. Alström syndrome is a ciliopathy because it results from mutations in the gene *ALMS1*, which is important in the development of cellular cilia. As of 2015, at least 239 *ALMS1* mutations that cause disease have been identified. Bardet-Biedl syndrome, another ciliopathy with comparable symptoms, is occasionally mistaken for Alström syndrome, but Bardet-Biedl syndrome is more common involves polydactyly and is brought on by mutations in the Bardet–Biedl syndrome genes.

Causes

The *ALMS1* gene, which is situated on the short arm of chromosome 2 (2p13.2), is the source of Alström syndrome. The gene mutation is an autosomal recessive trait that is inherited. This means that even if the parents may not exhibit any symptoms or signs of the disorder, a faulty copy of the *ALMS1* gene must be passed from one parent to the other for their child to have the syndrome. *ALMS1* is a particular protein that can be produced by the *ALMS1* gene. The protein is then implicated in intracellular transport, cell cycle regulation, and ciliary function. The protein is also present in all of the body's organ tissues. It plays a part in the development, maintenance, and appropriate operation of cilia, which are present in all body cell types. As of 2015, at least 239 *ALMS1* mutations that cause disease have been identified. The majority of these mutations have produced low quantities of defective *ALMS1* proteins, which are present in tissues.

Symptoms

The age at which Alström syndrome symptoms first manifest is quite variable. Heart failure (dilated cardiomyopathy) occurs in over 60% of instances typically in the first few weeks after birth however it can also manifest in adolescence or maturity.

- Cone-rod dystrophy and light sensitivity in all instances, often before 15 months of birth and getting worse until around 20 years of age.
- Early developmental delays, milestone delays in 50% of cases, and learning difficulties in about 30% of cases.
- Obesity is present in all cases by the age of five, however it is frequently present from birth (Alström newborns typically have normal birth weights, and by adolescence, weights tend to be in the high-normal to normal range).
- Nystagmus, which typically affects children, is one of the first symptoms to appear and results in uncontrollable rapid eye movement.
- Bilateral sensorineural hearing loss mild to moderate.
- Type 2 diabetes typically manifests in childhood.
- Hyperinsulinemia/insulin resistance, which is the rise in blood levels of insulin.
- Hypertriglyceridemia.
- Cirrhosis and liver failure can develop in some patients with steatosis, or fatty liver, and increased

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transaminases (liver enzymes), which frequently start in childhood.

- Slowly progressing kidney failure can happen in the second to fourth decade of life and may be caused by endocrine dysfunctions, such as an under- or overactive thyroid gland, weak growth hormone, excessive androgen in females, and low testosterone in males.

Prevention

Because Alström syndrome is a hereditary illness, prevention is thought to be more difficult than for other diseases or syndromes. For parents, however, who have an Alström syndrome family history, there are still other choices. People can meet with a genetic counsellor to explore the risks of having children with the disease. Genetic testing and counselling are also offered. Prior to conception, the genetic counsellor may also assist in identifying whether a person carries the *ALMS1* gene deficiency. Chorionic villus sampling (CVS), preimplantation genetic diagnosis (PGD), and amniocentesis are a few of the tests the genetic counsellors run. Only the embryos that are unaffected by the *ALMS1* gene may be chosen for implantation by *in vitro* fertilisation in PGD.