CONDITION

Alstrom syndrome

Learn how Alstrom syndrome affects vision, hearing, the heart, metabolism, liver, and kidneys, and how lifelong monitoring and supportive care can help.

WHAT IT IS
Rare genetic ciliopathy affecting multiple organs

BODY SYSTEM
Multiple body systems

MAIN SYMPTOMS
Progressive vision loss, hearing loss, childhood obesity, heart muscle disease

WHAT CAUSES IT
Inherited ALMS1 gene variants

HOW IT IS DIAGNOSED
ALMS1 genetic testing

MAIN TREATMENT
Supportive multidisciplinary care

CAN IT BE CURED?
No, underlying genetic change remains uncorrected

IS IT AN EMERGENCY?
Sometimes, severe breathing trouble, fainting, or chest pain

LONG-TERM OUTLOOK
Variable, with progressive multisystem organ damage

Overview

Alstrom syndrome is a rare genetic condition that can affect many parts of the body. Common features include progressive vision and hearing loss, obesity beginning in childhood, insulin resistance, type 2 diabetes, heart muscle disease, and problems involving the liver or kidneys.

The condition is caused by changes in a gene called `ALMS1`. This gene helps tiny structures called cilia work properly. Cilia are found on many cells and help cells sense signals and carry out normal functions. Alstrom syndrome is therefore described as a ciliopathy , meaning a disorder related to cilia.

Symptoms and severity vary widely. A person may not develop every possible feature, and problems may appear at different ages. Some infants develop serious heart disease, while others first come to medical attention because of unusual eye movements, light sensitivity,

hearing loss, or rapid weight gain.

There is currently no cure that corrects the underlying genetic change. However, early recognition and coordinated care can help protect organ function, support development, and treat complications. Because several organs may be involved even before symptoms are obvious, lifelong monitoring is an important part of care.

Symptoms

Eye symptoms often begin during the first year of life. Alstrom syndrome causes cone-rod dystrophy , a disorder of the retina at the back of the eye. The retina changes light into signals that the brain interprets as vision.

Early signs may include rapid, uncontrolled eye movements called nystagmus and strong sensitivity to light. Vision usually becomes progressively reduced. Color vision and the ability to see fine details are often affected early. Peripheral and night vision may also decline over time. The pace of vision loss differs from person to person.

Hearing loss usually affects both ears and often develops during childhood. It is typically sensorineural hearing loss , which means it results from changes in the inner ear or the nerve pathways that carry sound signals. Hearing may continue to decline, making regular testing important even when a child appears to respond normally to sound.

Many children develop rapid weight gain or obesity, especially around the trunk. This is related to the condition's biology and is not simply a matter of personal choices. Some people develop increased hunger, but appetite patterns vary.

The body may also become less responsive to insulin, the hormone that controls blood sugar. This is called insulin resistance. It can eventually lead to type 2 diabetes. Possible symptoms of high blood sugar include increased thirst, frequent urination, tiredness, and blurred vision, although diabetes may initially cause no noticeable symptoms.

Alstrom syndrome can affect the heart muscle. Cardiomyopathy means that the heart muscle is weakened or otherwise abnormal. Some infants develop dilated cardiomyopathy, in which the heart becomes enlarged and has difficulty pumping blood. Symptoms may include poor feeding, rapid breathing, sweating with feeds, unusual tiredness, or swelling. Heart disease may improve after infancy but can return later. Other people first develop cardiomyopathy during adolescence or adulthood.

Additional possible features include:

Intelligence is usually not directly affected. However, combined vision and hearing loss can influence communication, learning, mobility, and social development. Accessible education and

early communication support can make a major difference.

- High cholesterol or triglyceride levels
- Fat buildup, inflammation, or scarring in the liver
- Gradual loss of kidney function
- High blood pressure
- Short stature or unusual growth patterns
- Delayed puberty or other hormone-related problems
- Bladder or urinary symptoms
- Breathing problems, including sleep apnea
- Digestive symptoms such as reflux
- Joint, spine, or foot problems
- Dark, thickened skin patches related to insulin resistance

Causes and risk factors

Alstrom syndrome is caused by disease-causing variants in both copies of the `ALMS1` gene. A variant is a change in genetic instructions. The condition follows an autosomal recessive inheritance pattern.

A person usually inherits one altered copy of the gene from each parent. The parents are called carriers and generally do not have Alstrom syndrome themselves. When both parents carry an `ALMS1` variant, each pregnancy has a 25% chance of resulting in a child with the condition. Each pregnancy also has a 50% chance of resulting in a carrier and a 25% chance of resulting in a child who inherited neither familial variant.

These probabilities apply separately to each pregnancy. They do not mean that one out of every four children in a family will necessarily have the condition.

Having a sibling with Alstrom syndrome or known carrier parents is the main risk factor. The condition occurs in people of many ethnic backgrounds. It may be more common in some closely related families or isolated populations because relatives are more likely to carry the same rare gene variant.

Alstrom syndrome is not caused by parenting, diet, infection, or something a person did during pregnancy. Lifestyle can influence some complications, but it does not cause the genetic condition.

Diagnosis

Diagnosis is based on the pattern of symptoms, medical examinations, and genetic testing. Because features develop gradually, diagnosis may be difficult in a young child who has only eye findings or cardiomyopathy.

An evaluation may include an eye examination and retinal testing. An electroretinogram measures the retina's electrical response to light and can help identify cone-rod dystrophy. Hearing tests assess the type and degree of hearing loss.

Heart evaluation may include an electrocardiogram, which records the heart's electrical activity, and an echocardiogram, which uses ultrasound to examine heart structure and pumping function. Blood and urine tests may check glucose, cholesterol, triglycerides, liver function, and kidney function. Blood pressure, growth, puberty, and thyroid function may also be assessed.

Genetic testing can look for disease-causing variants in `ALMS1`. Finding a pathogenic or likely pathogenic variant in both gene copies can confirm the diagnosis. Sometimes testing finds a variant whose meaning is uncertain. This result does not confirm or exclude Alstrom syndrome by itself and should be interpreted by genetics professionals.

Other genetic conditions can have overlapping features. These include Bardet-Biedl syndrome, Usher syndrome, some mitochondrial disorders, and other inherited retinal or metabolic conditions. The combination of clinical findings and genetic results helps the care team distinguish among them.

Genetic counseling can explain test results, inheritance, testing options for relatives, and reproductive choices.

Treatment and management

Care usually involves a coordinated team because no single specialist can address every part of Alstrom syndrome. The team may include genetics, cardiology, ophthalmology, audiology, endocrinology, liver and kidney specialists, dietitians, therapists, and educational professionals.

There is no established treatment that restores retinal function. Care focuses on using remaining vision and reducing barriers. Tinted lenses may help with light sensitivity. Magnifiers, high-contrast materials, screen readers, mobility instruction, and other low-vision services can support independence. Educational planning should account for both visual and hearing needs.

Hearing aids may improve access to speech and environmental sounds. Some people with severe hearing loss may be evaluated for cochlear implants, which send sound signals directly

to the hearing nerve. Communication support should begin early and may include spoken-language services, sign language, or a combination based on the person's needs.

Cardiomyopathy is treated according to the type and severity of heart dysfunction. Treatment may include medicines that reduce the heart's workload, control blood pressure, or manage excess fluid. Some people need implanted heart devices or advanced heart-failure care. These decisions require individualized assessment by a cardiology team.

Nutrition and physical activity plans should be realistic, supportive, and adapted for sensory loss, heart disease, and mobility needs. The goal is to support health rather than assign blame for weight. Dietitians familiar with insulin resistance and high triglycerides can help families plan meals.

Type 2 diabetes may be managed with nutrition changes, activity, glucose monitoring, and diabetes medicines. Treatment depends on age, blood sugar levels, kidney and liver health, and other medical factors. High cholesterol, high triglycerides, and high blood pressure may also require medication.

Liver and kidney problems are managed according to their stage. Care may include avoiding substances that could further damage these organs, adjusting medicines when organ function changes, and treating related complications. Advanced disease may require specialized therapies, including consideration of organ transplantation in selected cases.

Physical, occupational, speech, and behavioral health support can help with mobility, communication, daily activities, and the emotional effects of living with a progressive condition.

Complications

Alstrom syndrome can lead to progressive damage in several organs. Possible complications include heart failure, abnormal heart rhythms, advanced liver scarring, chronic kidney disease, and complications of diabetes.

Very high triglyceride levels can increase the risk of pancreatitis, which is inflammation of the pancreas. This may cause severe upper abdominal pain, vomiting, and tenderness. Diabetes can also affect the eyes, nerves, kidneys, and blood vessels, although careful management may reduce these risks.

Combined hearing and vision loss can limit access to communication if accommodations are delayed. It may also increase the risk of falls, isolation, anxiety, or depression. These effects are not inevitable, and accessible technology, rehabilitation, counseling, and community support can help.

Some medical problems may be present without obvious symptoms. This is why routine monitoring remains important even when a person feels well.

Follow up

Follow-up should be coordinated through a clinician familiar with Alstrom syndrome when possible. The timing of visits and tests depends on age, existing organ involvement, previous results, and current symptoms.

Routine care commonly includes monitoring of:

Keep an updated list of medicines and specialists. Clinicians should consider kidney, liver, and heart function when selecting or adjusting treatments. Families may also benefit from an emergency care summary explaining the diagnosis and major medical concerns.

Contact the usual care team promptly about gradually increasing shortness of breath, swelling, reduced exercise ability, persistent vomiting, major changes in thirst or urination, new hearing changes, or worsening sleep symptoms. Sudden vision or hearing loss is not an expected part of routine progression and needs urgent medical assessment.

Seek emergency care for severe trouble breathing, blue or gray lips, fainting, new chest pain, severe weakness, confusion, or difficulty waking. A baby with poor feeding, rapid breathing, sweating during feeds, or unusual sleepiness also needs urgent assessment.

- Vision, hearing, communication, and access needs
- Heart structure, heart rhythm, and blood pressure
- Growth, weight, puberty, and other hormone functions
- Blood sugar, cholesterol, and triglycerides
- Liver and kidney function
- Breathing during sleep and daytime respiratory symptoms
- Mobility, joint health, emotional well-being, and school or workplace support

Sources

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