CONDITION

Ohio type amyloidosis

Ohio type amyloidosis is a rare inherited condition in which abnormal transthyretin protein builds up in nerves and other organs.

WHAT IT IS
Rare inherited transthyretin amyloidosis

BODY SYSTEM
Nervous system, heart, circulation, and digestive system

MAIN SYMPTOMS
Numbness, weakness, standing dizziness, heart problems

WHAT CAUSES IT
Inherited TTR gene change

HOW IT IS DIAGNOSED
TTR genetic testing and amyloid protein typing

MAIN TREATMENT
TTR-targeted medicines and organ-specific supportive care

CAN IT BE CURED?
No - treatment may slow progression

IS IT AN EMERGENCY?
Sometimes - severe breathlessness, chest pain, or fainting

LONG-TERM OUTLOOK
Often progressive, but treatment can slow organ damage

Overview

Ohio type amyloidosis, also called Type VII, is a historical name for a very rare inherited form of transthyretin amyloidosis. It belongs to the group now called hereditary transthyretin amyloidosis, or ATTRv amyloidosis.

Transthyretin, often shortened to TTR, is a protein mainly made by the liver. It normally carries thyroid hormone and vitamin A through the blood. In this condition, an inherited change in the TTR gene makes the protein less stable.

Unstable TTR can separate into pieces that form amyloid. Amyloid is abnormal protein material that collects between cells. Over time, these deposits can interfere with nerves, the heart, the digestive system, and other tissues.

The name "Ohio type" came from the family in which this pattern was historically described. Doctors now usually classify the condition by the affected gene, the exact gene change, and the organs involved rather than by the older numbered type.

Ohio type amyloidosis is not the same as AL amyloidosis, which is caused by abnormal antibody-producing cells. Correctly identifying the amyloid protein matters because the treatments are different.

Symptoms

Symptoms usually develop gradually. The age when they begin and the organs affected can differ, even among relatives with the same TTR gene change. Some people carry a gene change for years without symptoms.

Peripheral neuropathy is common. This means damage to nerves outside the brain and spinal cord. It often begins in the feet and may slowly move upward.

Possible symptoms include:

TTR amyloid can also collect in the heart. This may make the heart muscle thick and stiff, a problem called amyloid cardiomyopathy. The heart may then have difficulty filling and pumping normally.

Heart involvement can cause breathlessness, reduced exercise tolerance, ankle swelling, a racing or irregular heartbeat, or fainting. Some people have electrical conduction problems, meaning the signal that controls the heartbeat moves too slowly or irregularly.

Not every symptom is caused by amyloidosis. Numbness, bowel changes, dizziness, and heart symptoms have many more common causes. Testing is needed to find the reason.

- Tingling, burning pain, numbness, or reduced temperature sensation
- Weakness, poor balance, or difficulty walking
- Dizziness or faintness after standing
- Diarrhoea, constipation, nausea, or feeling full quickly
- Reduced sweating or unusual sweating
- Bladder or sexual difficulties
- Carpal tunnel symptoms, such as numb or painful hands

Causes and risk factors

Ohio type amyloidosis is caused by a disease-causing change in the TTR gene. The condition is usually inherited in an autosomal dominant pattern. This means a person with the gene change has a 1 in 2 chance of passing it to each child.

Inheriting the gene change does not always mean that symptoms will develop. This is called reduced penetrance. The chance of becoming unwell and the age when symptoms begin can be influenced by the exact TTR change, age, family history, and other factors that are not fully understood.

People of any sex can inherit and pass on the gene change. It is not caused by diet, infection, alcohol use, or anything a parent did during pregnancy.

A known TTR gene change in a biological relative raises the chance that another family member carries it. A family history may not always be obvious. Relatives may have been diagnosed with heart failure, unexplained neuropathy, or another type of amyloidosis before genetic testing was available.

Diagnosis

Diagnosis can take time because the early symptoms resemble diabetes-related nerve damage, common heart conditions, spinal problems, and other causes of neuropathy.

An assessment usually begins with your symptoms, family history, and a physical examination. Neurological testing may check strength, reflexes, balance, and the ability to feel touch, pain, or temperature.

Nerve conduction studies measure electrical signals in larger nerves. These tests can support a diagnosis of neuropathy, although early damage to very small nerve fibres may not appear on routine studies. Tests of blood pressure, sweating, digestion, or bladder function may be used to look for autonomic nerve damage.

Heart assessment may include an electrocardiogram, or ECG, an ultrasound scan called an echocardiogram, blood tests, and sometimes cardiac MRI. A nuclear medicine bone-tracer scan can provide strong evidence of ATTR amyloid in the heart under specific circumstances.

Blood and urine tests are also used to look for an abnormal antibody protein. This step helps doctors distinguish ATTR amyloidosis from AL amyloidosis. A positive heart scan by itself should not be assumed to identify the amyloid type when an abnormal antibody protein is present.

A tissue biopsy may be needed. A small sample of fat, nerve, heart, or another affected tissue is examined for amyloid. If amyloid is found, the protein should be typed, often using mass spectrometry. This laboratory method identifies which protein formed the deposit.

Genetic testing can identify a disease-causing TTR change and establish that the ATTR amyloidosis is inherited. Genetic counselling can help you understand the result, including uncertain findings and what a confirmed result could mean for relatives.

Treatment and management

Treatment aims to slow the production or formation of TTR amyloid, manage organ problems, and preserve independence. It cannot reliably remove damage that is already established, so timely recognition matters.

TTR-targeted medicines work in two main ways. TTR stabilisers help keep the protein intact so it is less likely to form amyloid. Gene-silencing medicines reduce the amount of TTR made by the liver. Which treatment may be appropriate is influenced by whether the nerves, heart, or both are affected, as well as local approval and access.

Treatment is usually combined with care for specific symptoms. This may include support for neuropathic pain, walking difficulties, blood pressure drops on standing, bowel problems, or bladder symptoms. Physical or occupational therapy can help with balance, mobility, hand function, and practical adaptations.

Heart care may include treatment for fluid build-up and monitoring for abnormal rhythms. Some people need a pacemaker or another implanted heart device because of electrical conduction problems. Medicines commonly used for other types of heart failure are not always tolerated in cardiac amyloidosis, so treatment needs specialist oversight.

Liver transplantation was historically used because most circulating TTR is produced in the liver. It is now considered only in selected circumstances because TTR-targeted medicines are available and amyloid-related problems can continue after transplantation. Heart or combined-organ transplantation may be considered for a small number of people with advanced organ damage.

Regular follow-up may include nerve examinations, walking or strength assessments, heart tests, kidney and liver blood tests, weight checks, and questions about digestion or blood pressure symptoms. The schedule is based on the organs involved and the treatment being used.

Complications

Progressive nerve damage can lead to muscle weakness, falls, foot injuries, and increasing difficulty walking. Reduced pain or temperature sensation can allow cuts, pressure sores, or

burns to go unnoticed.

Autonomic nerve damage can cause severe blood pressure drops, dehydration from ongoing diarrhoea, poor nutrition, bladder retention, or sexual difficulties. Changes in digestion and unintended weight loss can affect strength and quality of life.

Heart complications include heart failure, abnormal heart rhythms, and conduction block. Heart failure means the heart cannot meet the body's needs without pressure or fluid building up; it does not mean that the heart has stopped.

Complications are not inevitable, and their timing cannot be predicted from the historical "Ohio type" label alone. The exact gene change, age, organs affected, and response to treatment are more useful for planning care.

Prevention

The inherited TTR gene change cannot be prevented through lifestyle choices. Someone who has inherited it did not cause the condition.

If a familial TTR change is known, genetic counselling and voluntary testing can help adult relatives understand whether they carry it. Testing a relative is most informative after the specific family gene change has been identified.

People who carry the gene change but have no symptoms may be offered regular monitoring. This does not prevent inheritance, but it may identify early nerve or heart changes when treatment and follow-up can be considered.

People planning a family can discuss reproductive options with a genetics service. These are personal choices, and counselling should include the medical limits and emotional implications of testing.

Living with it and what to expect

Ohio type amyloidosis is usually a long-term, progressive condition. Changes tend to occur over years rather than days, although the pace differs between people. Symptoms that come on suddenly should not automatically be attributed to amyloidosis.

Numbness may slowly spread, and weakness or balance problems may become more noticeable. Autonomic symptoms can change over time. Heart involvement may appear before, alongside, or after nerve symptoms.

Treatment may slow further decline, but it may not restore sensation, strength, or heart

function that has already been lost. Some symptoms can still improve when blood pressure, fluid retention, pain, nutrition, or mobility problems are treated directly.

Many people continue with work, family activities, travel, and exercise adapted to their abilities. Others eventually need walking aids, help with daily tasks, or more intensive heart care. Rehabilitation, foot care, nutrition support, and practical home changes can help maintain independence.

Follow-up remains important even when symptoms seem stable. Reviews are used to detect meaningful changes, manage treatment effects, and address new organ involvement. A confirmed genetic result may also give relatives the option of counselling and testing, but sharing that information can be emotionally difficult. A genetics team or patient support organisation may help with family conversations.

When to get medical help

Arrange a routine medical appointment if you develop persistent numbness, burning pain, unexplained weakness, dizziness when standing, ongoing bowel changes, ankle swelling, or declining exercise tolerance. These symptoms have several possible causes and need assessment rather than self-diagnosis.

You should also seek routine advice if a biological relative is diagnosed with hereditary ATTR amyloidosis or a disease-causing TTR gene change. Genetic testing is best arranged with counselling so that you understand what the result can and cannot show.

Seek urgent medical advice for rapidly worsening swelling, new repeated fainting, a sustained racing or irregular heartbeat, or a major reduction in urine output.

Call emergency services for:

These signs can have causes other than amyloidosis, but they require prompt assessment.

- Chest pain, severe breathlessness, or collapse
- Fainting with injury or failure to recover promptly
- Sudden facial drooping, arm weakness, or speech difficulty
- Severe weakness that appears suddenly rather than gradually

Common questions

Is Ohio type amyloidosis the same as hereditary ATTR amyloidosis?

Ohio type, or Type VII, is an older name within the hereditary transthyretin amyloidosis group. Modern diagnosis uses the TTR gene result and the organs affected because these details are more informative than the historical type number.

If I have the gene change, will I definitely become ill?

Not necessarily. Some people with a disease-causing TTR change develop symptoms, while others remain well into later life. The exact gene change and family pattern can offer clues, but they cannot predict an individual course with certainty.

Can my children inherit it?

A person with an autosomal dominant TTR gene change has a 1 in 2 chance of passing it on in each pregnancy. The chance is the same for sons and daughters. Inheritance of the change does not predict when or whether symptoms will begin.

Is Ohio type amyloidosis contagious?

No. It cannot spread through contact, food, blood, sex, or shared living space. It results from an inherited gene change.

Can amyloid deposits be removed?

Current treatment mainly aims to prevent or slow the formation of new deposits. The body may clear some material over time, but established nerve or heart damage does not always reverse. Supportive treatment remains important even after TTR-targeted therapy begins.

Why is it called the Ohio type?

The name reflects the family and location connected with its original description. It does not mean the condition is limited to Ohio or caused by living there.

Sources

- Sekijima Y, Ueda M, Koike H, Misawa S, Ishii T, Ando Y. Hereditary Transthyretin Amyloidosis . GeneReviews , University of Washington, Seattle.
- Adams D, Ando Y, Beirão JM, et al. "Expert consensus recommendations to improve diagnosis of ATTR amyloidosis with polyneuropathy." Journal of Neurology . 2021;268:2109-2122.
- Maurer MS, Bokhari S, Damy T, et al. "Expert Consensus Recommendations for the Suspicion and Diagnosis of Transthyretin Cardiac Amyloidosis." Circulation: Heart Failure . 2019;12:e006075.



- Benson MD, Buxbaum JN, Eisenberg DS, et al. "Amyloid nomenclature 2020: update and recommendations by the International Society of Amyloidosis nomenclature committee." Amyloid . 2020;27(4):217-222.
- Hawkins PN, Ando Y, Dispenzeri A, Gonzalez-Duarte A, Adams D, Suhr OB. "Evolving landscape in the management of transthyretin amyloidosis." Annals of Medicine . 2015;47(8):625-638.