CONDITION

Finnish type amyloidosis

An inherited condition in which abnormal gelsolin protein deposits damage the eyes, nerves, skin, and sometimes internal organs.

WHAT IT IS
Inherited condition causing gelsolin protein deposits in tissues

BODY SYSTEM
Eyes, skin, nerves, kidneys, and heart

MAIN SYMPTOMS
Corneal clouding, facial weakness, loose skin, numbness

WHAT CAUSES IT
Inherited GSN gene change

HOW IT IS DIAGNOSED
GSN genetic testing, sometimes biopsy with amyloid typing

MAIN TREATMENT
Eye care, symptom relief, and complication monitoring

CAN IT BE CURED?
No - treatments cannot remove the gene change

IS IT AN EMERGENCY?
Sometimes - with sudden vision loss or stroke-like symptoms

LONG-TERM OUTLOOK
Usually slow progression; severe organ damage is uncommon

Overview

Finnish type amyloidosis is a rare inherited condition caused by a change in the GSN gene. It is also called gelsolin amyloidosis , AGel amyloidosis , Meretoja syndrome, or Type V amyloidosis.

The GSN gene provides instructions for making gelsolin, a protein involved in cell structure and movement. In this condition, abnormal gelsolin fragments accumulate between cells as amyloid deposits .

Deposits most often affect the cornea, the clear front surface of the eye. They can also damage facial and peripheral nerves and affect the skin. Kidney or heart involvement can occur, but serious internal-organ damage is uncommon in most people with one altered gene copy.

Symptoms usually begin gradually in adulthood. Eye changes often appear first, followed over time by facial weakness, loose skin, or nerve symptoms.

The condition was first described in Finland, where it is more common because of a founder gene change. It can occur in people of any ethnic or geographic background.

Symptoms

Symptoms vary, even among relatives with the same gene change, and usually progress slowly.

Eye symptoms often begin between the 20s and 40s. Amyloid forms branching lines in the cornea, causing lattice corneal dystrophy . Symptoms may include:

Reduced tear production and difficulty closing the eyelids can worsen dryness and corneal injury.

Damage to cranial nerves may cause weakness on both sides of the face. This can reduce facial expression, prevent full eye closure, cause drooping around the mouth, or make it harder to keep food and liquids inside the mouth.

Skin may become loose, thin, or sagging, particularly around the forehead, scalp, eyelids, and face. This is called cutis laxa . Fragile blood vessels and supporting tissue may also cause easy bruising.

Peripheral neuropathy can cause numbness, tingling, burning pain, or reduced temperature awareness, often beginning in the feet. Carpal tunnel syndrome may cause hand numbness or weakness.

Less common features include swallowing or speech problems, reduced balance, abnormal heart rhythms, protein in the urine, and reduced kidney function. These require assessment because they may also have unrelated causes.

- Dry, gritty, or irritated eyes
- Sensitivity to light
- Blurred or reduced vision
- Repeated painful corneal scratches or erosions

Causes and risk factors

Finnish type amyloidosis results from a harmful change in the GSN gene. The altered gelsolin

protein is cut into unstable fragments that form deposits and gradually build up in tissues.

The condition usually has an autosomal dominant inheritance pattern. One altered gene copy is enough to cause it. An affected person has a 1 in 2 chance of passing the change to each biological child. The outcome of one pregnancy does not affect another.

Family history is the main risk factor, but the condition may not have been recognised in previous generations. Relatives may have mild symptoms, a different diagnosis, or signs that developed late in life.

Very rarely, someone inherits altered copies from both parents. This homozygous form can begin earlier and be more severe, with a greater risk of kidney damage.

Finnish ancestry may increase the chance of carrying the Finnish founder variant, but ancestry cannot confirm or exclude the condition. Other GSN variants occur outside Finland.

Diagnosis

A combination of corneal changes, facial weakness, loose skin, neuropathy, and family history may suggest Finnish type amyloidosis.

An ophthalmologist can examine the cornea with a slit lamp , which may reveal the branching lines of lattice corneal dystrophy. Similar changes occur in other inherited disorders, so an eye examination alone may not identify the exact cause.

A blood or saliva test showing a harmful GSN variant can confirm the diagnosis. Genetic counselling can explain what the result means for you and your biological relatives. Testing relatives is most informative after the family's specific variant has been identified.

A biopsy is not always needed if genetic testing provides a clear result. If tissue is sampled, Congo red staining can show amyloid. The amyloid should then be typed using specialised analysis because several unrelated forms of amyloidosis exist.

Other tests look for complications rather than confirm the diagnosis. Depending on symptoms, they may include nerve studies, kidney blood and urine tests, an electrocardiogram, or heart imaging.

Treatment and management

There is no established treatment that removes the GSN change or clears all gelsolin amyloid. Care focuses on protecting the eyes, relieving symptoms, maintaining function, and detecting complications.

Regular ophthalmology care is important. Lubricating drops or ointments can reduce dryness and protect the cornea. People who cannot close an eyelid fully may need additional protection for the exposed eye surface.

Laser treatment or corneal transplantation may be considered for repeated erosions, scarring, or clouding that significantly affects comfort or vision. Amyloid can return in treated or transplanted tissue, so long-term follow-up remains necessary.

Procedures to improve eyelid closure or support loose facial skin may help with eye protection, vision, eating, or daily activities. Benefits should be balanced against possible healing and bleeding concerns.

Medicines intended for nerve pain may help neuropathy more than ordinary painkillers. Physiotherapy can support strength, balance, and mobility, while occupational therapy can help with hand weakness and daily tasks. Carpal tunnel syndrome is treated with standard measures, including surgery when appropriate.

Kidney disease, heart rhythm problems, and other complications are treated according to the specific finding. Care may involve ophthalmology, neurology, clinical genetics, kidney or heart specialists, and rehabilitation services.

Complications

Repeated corneal erosions may cause pain, infection, scarring, and vision loss. Poor eyelid closure raises the risk of exposure keratopathy , in which the uncovered cornea dries out and becomes damaged.

Facial weakness may affect eye closure, speech, chewing, or control of food and liquids. Loose eyelid skin can interfere with sight. Peripheral nerve damage may impair balance or make foot injuries difficult to notice.

Kidney involvement can cause protein in the urine, swelling, high blood pressure, or reduced kidney function. Severe kidney failure is unusual with one altered GSN copy but is a greater concern in the rare homozygous form.

Amyloid may affect the heart's electrical system, causing a slow or irregular heartbeat. Significant heart involvement is less common than in several other hereditary amyloidoses.

Eye or facial procedures may be followed by bleeding, delayed healing, infection, or recurrence of amyloid-related changes. Procedures are not always unsuitable, but the surgical team should know about the condition.

Prevention

Finnish type amyloidosis cannot be prevented in someone who has inherited the GSN change. Lifestyle choices do not cause it or stop the altered protein from being produced.

Genetic counselling can explain inheritance and reproductive options, including testing during pregnancy or testing embryos created through in vitro fertilisation. These are personal decisions, and counselling provides information rather than directing a choice.

Corneal protection, planned eye follow-up, and monitoring based on symptoms can reduce or identify secondary problems. Prompt assessment of changes in vision, eyelid closure, nerve function, urine, or heart rhythm may prevent avoidable harm.

Living with it and what to expect

Finnish type amyloidosis usually progresses over years or decades. Corneal findings often come first, while facial weakness, loose skin, and peripheral neuropathy may become more noticeable later.

Progression differs between people. Dryness, erosions, and corneal clouding can impair vision, but treatment may improve comfort and function. Facial weakness and skin looseness are generally long-lasting because treatment does not remove the underlying amyloid process.

Most people with one altered gene copy do not develop the severe heart, kidney, or nerve failure seen in some other amyloidoses. Finnish long-term studies suggest overall survival may remain close to that of the wider population, although eye, facial, and nerve symptoms can create a substantial burden. People with two altered copies may have a more serious course.

The condition does not usually affect intelligence or memory. Many people continue to work, maintain relationships, and live independently, although adjustments may be needed for vision, facial movement, hand sensation, or balance.

Follow-up is usually lifelong and should reflect the body systems affected rather than follow the same schedule for everyone.

When to get medical help

Arrange routine follow-up for persistent dry eyes, gradual vision changes, facial weakness, numbness, balance problems, or skin changes. Seek assessment for new hand weakness, difficulty swallowing, foamy urine, ankle swelling, or repeated palpitations.

Contact an eye care service promptly if an eye becomes painful, red, unusually sensitive to light, or difficult to open. Prompt assessment is also important if you cannot fully close an eyelid or develop a new defect on the eye surface.

Seek urgent or emergency care for:

These symptoms may not be caused by Finnish type amyloidosis, but they should not wait for a routine appointment.

- Sudden loss of vision
- New one-sided weakness, facial drooping, or difficulty speaking
- Severe chest pain or major difficulty breathing
- Fainting with ongoing symptoms or an irregular heartbeat
- A severely painful red eye with nausea or rapidly worsening vision

Common questions

Is Finnish type amyloidosis the same as other types of amyloidosis?

No. Amyloidosis includes conditions caused by different proteins. Finnish type amyloidosis is caused by altered gelsolin and differs from light-chain and transthyretin amyloidosis in its inheritance, usual symptoms, organ risks, and treatment.

Will I go blind?

Not everyone develops severe sight loss. Corneal deposits, dryness, erosions, and poor eyelid closure can reduce vision. Regular eye care and treatment of surface damage can protect comfort and sight, although corneal changes may progress.

Does everyone with the gene change develop symptoms?

Many people with a harmful GSN variant eventually develop features, but age at onset and severity vary. A mildly affected parent does not guarantee that a child will have the same experience.

Can Finnish type amyloidosis affect my children?

Yes. Each biological child of an affected parent usually has a 1 in 2 chance of inheriting the altered gene. Genetic testing can show whether an adult relative carries the family variant.

Is there a special diet for gelsolin amyloidosis?

No diet has been shown to prevent gelsolin amyloid. Dietary changes may be advised for a separate problem, such as impaired kidney function, but they do not treat the gene change.

Can surgery remove the condition?

Surgery can treat specific problems such as corneal clouding, poor eyelid closure, loose facial skin, or carpal tunnel syndrome. It does not remove amyloid throughout the body, and local changes can return.

Sources

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