CONDITION

Familial cutaneous amyloidosis

Familial cutaneous amyloidosis is an inherited condition that causes itchy, discoloured amyloid deposits in the skin.

WHAT IT IS
Inherited skin amyloid deposits causing itchy, discoloured patches

BODY SYSTEM
Skin

MAIN SYMPTOMS
Itching, rippled dark patches, thickened bumps, scratch marks

WHAT CAUSES IT
Inherited, sometimes involving OSMR or IL31RA gene changes

HOW IT IS DIAGNOSED
Skin examination, biopsy, and sometimes genetic testing

MAIN TREATMENT
Itch control and skin-directed treatments

CAN IT BE CURED?
No - inherited skin tendency remains

IS IT AN EMERGENCY?
No - routine dermatology assessment is appropriate

LONG-TERM OUTLOOK
Chronic symptoms that often wax and wane

Overview

Familial cutaneous amyloidosis is an inherited condition in which a protein material called amyloid collects in the skin. Amyloid forms when proteins fold abnormally and build up in tissues.

In this condition, the deposits usually come from damaged skin cells called keratinocytes. They are limited mainly to the upper layer of the skin rather than being produced by an abnormal protein circulating throughout the body.

The two main patterns are:

Some people have features of both patterns. This is sometimes called biphasic amyloidosis.

The word "familial" means the tendency runs in a family. Symptoms may begin during

childhood, adolescence, or adulthood. Their severity can differ even among relatives with the same inherited change.

Familial cutaneous amyloidosis is usually a skin-limited condition. This makes it different from systemic amyloidosis, in which amyloid deposits affect internal organs. It is also different from nodular cutaneous amyloidosis, a less common form that may require additional assessment.

- Macular amyloidosis , which causes flat, brown or grey-brown patches, often with a rippled appearance
- Lichen amyloidosis , which causes clusters of small, firm, itchy bumps

Symptoms

Itching is often the main symptom. It may be mild or persistent and can interfere with sleep, concentration, or daily activities. Heat, sweating, dry skin, friction, and repeated scratching may make it feel worse.

Macular amyloidosis usually causes brown, grey-brown, or darker patches. Fine, wavy lines can create a rippled or net-like pattern. The patches often develop on the upper back, especially between the shoulder blades, or on the arms and legs.

Lichen amyloidosis causes many closely grouped bumps. They are usually firm, rough, and similar in colour to the surrounding skin or slightly darker. The shins are a common location, although other areas can be affected.

Repeated scratching may lead to:

The condition does not usually cause weakness, numbness, shortness of breath, kidney problems, or unexplained swelling. Symptoms such as these need a separate medical assessment rather than being assumed to come from cutaneous amyloidosis.

- Thickened or leathery skin
- Scratch marks, crusts, or small areas of bleeding
- Lighter or darker skin after inflammation
- Tenderness if the skin becomes broken

Causes and risk factors

Familial cutaneous amyloidosis is caused by an inherited tendency that affects how skin cells respond to irritation and how itch signals are processed.

Changes in the OSMR or IL31RA genes have been found in some families. These genes help form receptors, which are structures that allow cells to receive chemical signals. Changes affecting these receptors may make itching and skin-cell damage more likely. In many affected families, however, a specific gene change is not identified.

The condition is often inherited in an autosomal dominant pattern. This means one altered copy of a gene can be enough to cause the condition. A parent may pass that change to a child, but relatives who inherit it may not have symptoms of the same type or severity.

Friction and scratching do not create the inherited tendency. They can contribute to skin damage and may make deposits, itching, and discolouration more noticeable. Rough clothing, vigorous rubbing, and abrasive washing tools can aggravate affected areas.

A family history of similar itchy, rippled patches or persistent bumps supports the possibility of a familial condition. Similar-looking skin changes can also occur without a known family history.

Diagnosis

Diagnosis usually begins with a review of the skin changes, when they started, where they occur, and whether relatives have similar symptoms. A clinician may also ask about medicines, allergies, other skin conditions, and symptoms that could suggest a different form of amyloidosis.

A skin biopsy is the main test used to confirm cutaneous amyloidosis. After numbing the area, a small sample of skin is removed and examined under a microscope. Special stains, including Congo red, can help identify amyloid deposits. Under polarised light, Congo red-stained amyloid has a characteristic appearance.

The biopsy also helps distinguish macular or lichen amyloidosis from eczema, lichen planus, prurigo, and other conditions that can cause itchy or darkened skin.

Genetic testing may be offered when several relatives are affected, symptoms begin unusually early, or the result could help with family planning. Testing may look for changes in genes such as OSMR and IL31RA. A negative result does not rule out familial cutaneous amyloidosis because not every responsible genetic change is known.

Blood or urine testing is not routinely needed for a typical skin-limited macular or lichen pattern. Additional tests may be considered if the biopsy shows nodular amyloidosis or if symptoms raise concern about internal organ involvement.

Treatment and management

There is no treatment that removes the inherited tendency. Management aims to reduce itching, protect the skin, and improve the appearance or thickness of affected areas.

Regular moisturising can reduce dryness and support the skin barrier. Fragrance-free creams or ointments are often less irritating than perfumed products. Gentle washing and avoiding abrasive scrubbing may help limit further damage.

Depending on the pattern and severity, treatment may include:

Some treatments can thin or irritate the skin if used incorrectly. A treatment plan should account for the location involved, the thickness of the lesions, skin colour, previous treatment, and possible side effects.

Evidence for many treatments is limited because familial cutaneous amyloidosis is uncommon. A treatment may help itching without clearing the colour change, or improve bumps without preventing their return. Several approaches may need to be tried over time.

Breaking the itch-scratch cycle is an important part of management. Keeping nails short, wearing soft clothing, using cool compresses, and covering frequently scratched areas at night may reduce unintentional damage. These measures manage symptoms but do not cure the condition.

- Anti-inflammatory creams, such as topical corticosteroids or calcineurin inhibitors
- Medicines or preparations aimed at reducing itch
- Keratolytic products that soften thick or scaly skin
- Light treatment, also called phototherapy
- Laser treatment, dermabrasion, or other procedures for selected areas

Complications

The main complications come from long-term itching and scratching rather than from damage to internal organs.

Repeated scratching can cause thickened skin, persistent pigment changes, open sores, or scarring. Broken skin can occasionally become infected. Signs include increasing pain, warmth, swelling, spreading redness, or pus.

Ongoing itch may disturb sleep and affect mood, work, exercise, or social activities. Visible lesions can also affect confidence, especially when they occur on exposed skin.

Typical familial macular and lichen amyloidosis generally remain limited to the skin. They are not considered a usual route to systemic amyloidosis.

Nodular cutaneous amyloidosis is different because its deposits come from immune cells called plasma cells. If a biopsy identifies the nodular form, follow-up may include tests for amyloid or related protein abnormalities elsewhere in the body.

Prevention

Familial cutaneous amyloidosis cannot currently be prevented because the underlying tendency is inherited.

Reducing friction and scratching may help prevent lesions from becoming thicker, more inflamed, or more discoloured. This does not prevent inheritance or guarantee that new areas will not develop.

Genetic counselling can help families understand the likely inheritance pattern, the limits of genetic testing, and reproductive options. It does not tell exactly when symptoms will begin or how severe they will be.

Living with it and what to expect

Familial cutaneous amyloidosis is usually long-lasting. Symptoms may stay in the same areas for years, slowly extend, or become more or less noticeable over time. Periods of increased itching can alternate with quieter periods.

Treatment often controls symptoms rather than clearing every mark. Darkening may remain after the itch has improved because pigment changes can fade slowly. Thickened bumps may flatten with treatment but can return, especially if scratching continues.

The condition does not spread through contact and does not usually limit physical activity, work, relationships, or life expectancy. Many people manage it with skin care, itch control, and occasional dermatology follow-up.

Routine follow-up is useful when symptoms are affecting sleep or daily life, treatment is irritating the skin, or lesions are changing despite management. Follow-up may also be appropriate after a nodular biopsy result or when genetic test results need to be explained to relatives.

When to get medical help

Arrange a routine appointment if you develop persistent itchy patches or bumps, particularly

when relatives have similar skin changes. An examination is important because eczema, lichen planus, and other skin conditions can look similar.

Seek earlier medical advice if:

Prompt assessment is appropriate for increasing redness, warmth, swelling, pain, pus, or fever, as these may indicate a skin infection.

Familial cutaneous amyloidosis itself is not usually an emergency. Call emergency services for severe breathing difficulty or swelling of the lips, tongue, or throat after using a medicine or skin product, as these can be signs of a serious allergic reaction.

- Itching is disrupting sleep or daily activities
- A lesion develops a new lump, ulcer, or repeated bleeding
- Treatment causes persistent burning, thinning, or worsening irritation
- The diagnosis is uncertain or the skin changes are spreading quickly

Common questions

Is familial cutaneous amyloidosis cancer?

No. The usual macular and lichen forms are not cancer. They involve deposits made from damaged skin-cell proteins. A new or changing lump should still be examined because other conditions can occur separately.

Can skin amyloidosis spread to my organs?

Typical familial macular and lichen amyloidosis generally stay in the skin and are different from systemic amyloidosis. Nodular cutaneous amyloidosis has different follow-up needs, so the exact biopsy diagnosis matters.

Is it contagious?

No. You cannot catch it through touching, sharing towels, sexual contact, or living with someone who has it. It results from an inherited tendency, not an infection.

Will my children inherit it?

Many families have an autosomal dominant inheritance pattern. If a disease-causing variant is confirmed in a parent, each child has a one-in-two chance of inheriting that variant. Inheriting it does not predict the exact age of onset or severity. A genetics professional can explain results for a particular family.

Why does scratching make it worse?

Scratching damages keratinocytes, the cells that make up much of the outer skin. Their

damaged proteins can contribute to local amyloid deposits. Scratching also thickens the skin and strengthens the itch-scratch cycle.

Can the marks be removed permanently?

Treatments may flatten bumps, reduce itching, or make discolouration less noticeable. Complete and permanent removal is difficult, and lesions can return because the underlying inherited tendency remains.

Sources

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