CONDITION

# Lamellar ichthyosis

Lamellar ichthyosis is an inherited skin condition that causes widespread thick, dry, plate-like scales.

**WHAT IT IS**
Inherited skin condition causing widespread thick, plate-like scaling

**BODY SYSTEM**
Skin

**MAIN SYMPTOMS**
Large scales, dry skin, overheating, painful cracks

**WHAT CAUSES IT**
Inherited gene changes affecting skin barrier formation

**HOW IT IS DIAGNOSED**
Skin examination, family history, and genetic testing

**MAIN TREATMENT**
Moisturisers, scale-removing treatments, and sometimes retinoids

**CAN IT BE CURED?**
No - inherited changes remain lifelong

**IS IT AN EMERGENCY?**
Sometimes - confusion or collapse during overheating

**LONG-TERM OUTLOOK**
Lifelong, usually manageable with regular skin and eye care

## Overview

Lamellar ichthyosis is a rare, lifelong condition that affects how the outer layer of skin forms and sheds. The skin becomes very dry and develops large, thick scales. These scales may cover most of the body.

The condition is usually present from birth. Many affected babies are born inside a tight, shiny covering called a collodion membrane . This membrane normally peels away during the first few weeks. The skin underneath then develops the features of lamellar ichthyosis.

Lamellar ichthyosis belongs to a group called autosomal recessive congenital ichthyosis , or ARCI. "Congenital" means present from birth. "Autosomal recessive" describes how it is inherited.

The condition is not contagious. It is not caused by poor hygiene, an allergy, or something a parent did during pregnancy. It does not usually affect intelligence.

Lamellar ichthyosis matters because the skin helps control temperature, prevent water loss, and protect against infection. Regular skin care can reduce scaling and discomfort. Some people also need care for their eyes, ears, or ability to cope with heat.

## Symptoms

Symptoms differ in severity, even among people in the same family. The main feature is widespread scaling. The scales are often large, flat, and plate-like. They may be light, brown, or dark and are commonly most noticeable on the trunk, arms, and legs.

Other possible features include:

Ectropion may prevent the eyelids from closing fully. This can dry and irritate the eye's surface. Some people also have lips that turn outward because the surrounding skin is tight.

Newborns with a collodion membrane need careful monitoring. Their skin may lose too much water, interfere with normal temperature control, or become infected. Tight skin around the eyes, mouth, fingers, or toes can also require attention.

Reduced sweating is important throughout life. A person may become overheated during hot weather, exercise, fever, or work in a warm environment. Young children may not recognise or explain early symptoms such as headache, weakness, nausea, or dizziness.

- Very dry, tight, or itchy skin
- Painful cracks, especially around joints, palms, or soles
- Reduced sweating and difficulty staying cool
- Redness beneath or between the scales
- Thickened skin on the palms and soles
- Scalp scaling, hair breakage, or patchy hair loss
- Scale collecting in the ear canals and affecting hearing
- Eyelids turning outward, known as ectropion

## Causes and risk factors

Lamellar ichthyosis is caused by changes in genes involved in making and maintaining the skin

barrier. This barrier is the outer protective layer that limits water loss and helps the skin shed normally.

Changes in the TGM1 gene are a common cause. Changes in several other genes can produce a similar appearance. Because different gene changes can overlap in their effects, the exact genetic cause cannot usually be identified from the skin alone.

Lamellar ichthyosis is most often inherited in an autosomal recessive pattern. This means a child generally has a changed copy of the same gene from each parent. The parents are usually healthy carriers with one changed copy.

When two parents carry changes in the same gene, each pregnancy has a 1 in 4 chance of resulting in an affected child. Each pregnancy is a separate event. The chance may be different in families with another inheritance pattern, so genetic counselling can provide family-specific information.

Having an affected relative or parents who are closely related by blood can increase the chance of inheriting the same rare gene change. However, the condition can also appear in a family with no previous known history.

## Diagnosis

Lamellar ichthyosis is diagnosed from the skin's appearance, the history from birth, and sometimes genetic testing. A dermatologist may ask whether there was a collodion membrane, how the skin changed over time, and whether relatives have similar symptoms.

A blood or saliva test can examine a panel of genes linked to congenital ichthyosis. Finding two relevant gene changes can confirm the genetic cause, guide genetic counselling, and sometimes help distinguish lamellar ichthyosis from related conditions. Testing does not identify a cause in every person.

A small skin sample, called a biopsy, is occasionally examined under a microscope. It can support the diagnosis or help rule out another condition, but it often cannot identify the exact type of inherited ichthyosis.

Assessment may also include the eyes, hearing, scalp, growth, and temperature control. These checks look for effects that may need separate treatment.

## Treatment and management

There is no treatment that removes the underlying inherited gene changes. Management

focuses on softening scale, protecting the skin barrier, easing discomfort, and preventing complications.

Thick moisturising ointments or creams are the foundation of care. They work best when applied regularly, particularly after bathing while the skin is still damp. Ointments usually hold in more water than thin lotions.

Bathing or soaking can hydrate the scales and make them easier to remove. Gentle rubbing may help loosen softened scale. Forceful scraping can damage the skin and lead to pain or infection.

Some creams contain keratolytic ingredients, which help break down and remove thick surface skin. Examples include urea and certain acids. These products can sting on cracked skin. Some ingredients may be absorbed when used over large areas, especially in young children, so product choice should be discussed with the treating dermatology team.

For more severe scaling, a dermatologist may consider an oral retinoid such as acitretin. Retinoids are medicines related to vitamin A that reduce thickening of the outer skin layer. They require specialist supervision and blood monitoring. They can cause serious birth defects if pregnancy occurs during specified risk periods, so strict pregnancy-prevention requirements apply to anyone who could become pregnant.

Eye dryness from ectropion may be managed with lubricating drops or ointment and regular ophthalmology care. Surgery is occasionally considered when the eyelid position threatens the eye or causes persistent symptoms.

Scale in the ear canals may need removal by an ear specialist or another trained professional. Objects or cotton buds should not be pushed into the canal because this can pack the scale more tightly or injure the ear.

Plans for avoiding overheating are also part of treatment. Cooling breaks, water, fans, air conditioning, spray bottles, and suitable clothing may help. School or workplace adjustments can allow access to cooler spaces and reduce heat exposure.

Babies born in a collodion membrane are usually cared for by neonatal and dermatology teams. They may need a humidified environment, careful fluid and temperature monitoring, bland skin ointments, and eye care.

## Complications

Deep cracks can be painful and may become infected. Warning signs include increasing redness, warmth, swelling, pus, worsening pain, or fever.

Reduced sweating can lead to heat exhaustion or heatstroke. Heatstroke is a medical emergency because a very high body temperature can affect the brain and other organs.

Ectropion may cause ongoing dryness, light sensitivity, inflammation, or damage to the clear front surface of the eye. Ear-canal blockage can lead to reduced hearing and may affect speech development if it is not recognised in a young child.

Tight skin can limit movement or make walking uncomfortable. Persistent scalp scale may contribute to hair loss. The visible nature of the condition can also affect confidence, relationships, school, and work. Emotional or psychological support can be an important part of care.

## Prevention

Lamellar ichthyosis cannot be prevented through skin care, diet, or choices during pregnancy because it is inherited. Early treatment can help prevent some complications, but it cannot stop the condition from developing.

If the genetic changes in a family are known, genetic counselling can explain the chance of passing them on. Prenatal testing and testing of embryos during in vitro fertilisation may be available to some families. These are personal reproductive options rather than treatments for an affected person.

## Living with it and what to expect

Lamellar ichthyosis is lifelong. A newborn's collodion membrane usually sheds, but dry, scaly skin remains. The amount of redness may decrease with age, while plate-like scaling often continues. Symptoms can change with climate, illness, hormonal changes, and the amount of time available for skin care.

Daily care may take considerable time. Many people develop a routine involving bathing, moisturising, scale removal, and temperature planning. Treatment needs may change as a child grows or as work, travel, and living conditions change.

Regular follow-up commonly includes skin checks and review of medicines. Eye and hearing assessments may be needed when ectropion or ear-canal scale is present. Children may benefit from school plans that allow moisturiser use, water, cooling breaks, and modified activity during hot weather.

Most people can attend school, work, exercise, have relationships, and raise families. Activities may need adjustment to avoid overheating or painful skin cracking. Swimming is often

possible, although chlorine or salt water may irritate the skin, making rinsing and moisturising afterward helpful.

Lamellar ichthyosis does not usually shorten life when complications are recognised and managed. Individual experiences differ, and severe overheating, infection, or eye problems can have lasting effects if untreated.

## When to get medical help

Arrange routine follow-up if scaling, itching, cracking, or treatment burden is becoming harder to manage. A dermatology review is also appropriate if current products are not helping, side effects develop, or treatment needs change.

Seek prompt medical advice for:

Call emergency services for confusion, collapse, a seizure, severe weakness, or loss of consciousness during heat exposure. Move the person to a cooler place and begin cooling while waiting for help.

A newborn with a tight, shiny membrane needs hospital assessment. Urgent review is especially important if the baby has trouble feeding or breathing, is unusually sleepy, has a temperature problem, or shows signs of infection.

- Increasing skin redness, warmth, swelling, pus, pain, or fever
- New eye redness, persistent eye pain, light sensitivity, or blurred vision
- Reduced hearing or a blocked-ear sensation
- Repeated dizziness, headache, nausea, or weakness in warm conditions
- Deep cracks that bleed, restrict movement, or do not heal

## Common questions

Is lamellar ichthyosis contagious?

No. It is caused by inherited gene changes and cannot spread through touching, sharing towels, swimming, or close contact.

Can lamellar ichthyosis go away?

The condition is lifelong because the underlying genetic changes remain. Scaling and discomfort can often be reduced with regular skin care and, when appropriate, specialist treatment.

Does washing more often remove the scales?

Bathing can soften scales, but lamellar ichthyosis is not caused by dirt. Scrubbing hard or using harsh soap may damage the skin. Gentle soaking, careful scale removal, and moisturising are usually more helpful.

Why do I overheat so easily?

Thick scale can block sweat from reaching the skin's surface. Because sweat helps cool the body, heat may build up during exercise, fever, hot weather, or work in warm surroundings.

Can someone with lamellar ichthyosis have children?

Yes. The chance of passing on the condition depends on the affected gene and whether the other biological parent carries a change in the same gene. Genetic counselling can clarify the chance for a particular family.

Will a child with lamellar ichthyosis develop normally?

The condition does not usually affect intelligence. Skin discomfort, hearing blockage, eye problems, or missed school can affect learning and daily life if they are not addressed. Appropriate medical care and practical support can reduce these barriers.

## Sources

- GeneReviews: Autosomal Recessive Congenital Ichthyosis , University of Washington and National Center for Biotechnology Information
- MedlinePlus Genetics: Lamellar ichthyosis , U.S. National Library of Medicine
- DermNet: Lamellar ichthyosis , DermNet New Zealand
- Mazereeuw-Hautier J, et al. "Management of congenital ichthyoses: European guidelines of care, part one." British Journal of Dermatology . 2019;180(2):272-281
- Mazereeuw-Hautier J, et al. "Management of congenital ichthyoses: European guidelines of care, part two." British Journal of Dermatology . 2019;180(3):484-495
- DailyMed: Soriatane-acitretin capsule prescribing information , U.S. National Library of Medicine