CONDITION

Recessive x-linked ichthyosis

An inherited condition in which steroid sulfatase deficiency causes dry, dark, firmly attached skin scales.

WHAT IT IS
Inherited skin scaling from steroid sulfatase deficiency

BODY SYSTEM
Skin, with possible eye, testicular, and developmental effects

MAIN SYMPTOMS
Dark scales, dry skin, itching, reduced sweating

WHAT CAUSES IT
Inherited STS gene change or deletion

HOW IT IS DIAGNOSED
STS genetic testing, sometimes steroid sulfatase enzyme testing

MAIN TREATMENT
Moisturisers, scale-removing creams, and specialist retinoid treatment

CAN IT BE CURED?
No - inherited enzyme deficiency remains lifelong

IS IT AN EMERGENCY?
Sometimes - severe overheating or rapidly spreading skin infection

LONG-TERM OUTLOOK
Usually normal lifespan with lifelong, manageable scaling

Overview

Recessive X-linked ichthyosis is an inherited condition that affects the skin's ability to shed dead cells normally. These cells collect on the surface and form dry, firmly attached scales.

The condition is caused by reduced or absent activity of an enzyme called steroid sulfatase. An enzyme is a protein that helps the body carry out a chemical process. Without enough steroid sulfatase, certain fats build up in the outer skin layer and interfere with normal shedding.

The skin changes usually begin in infancy or early childhood and continue throughout life. They mainly affect males because the altered gene is on the X chromosome. Females can carry the altered gene and occasionally have mild or, rarely, more noticeable symptoms.

The scaling can usually be managed with regular skin care. However, the genetic change may

sometimes be associated with undescended testicles, cloudy areas in the cornea of the eye, or developmental and attention-related differences.

Symptoms

A baby's skin may look normal at birth. Dryness and scaling often become noticeable during the first weeks or months of life, although mild cases may not be recognised until later.

Typical skin changes include:

The folds of the elbows and knees, palms, soles, and central face are often less affected. The pattern can differ from one person to another.

Some people sweat less effectively because thick scale blocks the sweat ducts. This can make it harder to cool down during hot weather or exercise.

The condition does not usually cause the intense skin redness seen in some other inherited ichthyoses. "Ichthyosis" is the name for a group of conditions that cause dry, scaly skin.

- Large, flat scales that may be grey, brown, or darker than nearby skin
- Scaling on the neck, trunk, scalp, and outer surfaces of the limbs
- Dryness, tightness, or mild itching
- Symptoms that worsen in cold, dry weather

Causes and risk factors

Recessive X-linked ichthyosis results from a change affecting the STS gene. This gene provides instructions for making steroid sulfatase. Most affected people have a deletion, meaning that some genetic material containing the gene is missing. Others have a smaller change within the gene.

The STS gene is on the X chromosome. Males usually have one X chromosome and one Y chromosome. If their X chromosome carries a non-working STS gene, they will usually develop the condition.

Females usually have two X chromosomes. If one carries the altered gene, the other often provides enough enzyme activity to prevent obvious symptoms. Such a female is called a carrier. Some carriers have dry or mildly scaly skin. More substantial symptoms are rare but possible.

A carrier has a chance of passing the altered X chromosome to each child. A son who inherits it

will usually be affected, while a daughter who inherits it will usually be a carrier. An affected father passes his altered X chromosome to all his daughters and none of his sons.

Sometimes the deletion includes genes next to STS . These larger deletions may cause additional features, such as developmental delay or other physical differences. Genetic testing can show whether only STS , or a wider chromosome region, is affected.

During a pregnancy involving an affected fetus, steroid like prednisone or hydrocortisone sulfatase deficiency in the placenta may cause a low estriol result on prenatal screening. It may also be associated with labour starting late or progressing slowly. These findings can have several causes and do not diagnose the condition by themselves.

Diagnosis

A clinician may suspect recessive X-linked ichthyosis from the scale's appearance, its distribution, when it began, and the family history. Other forms of ichthyosis can look similar, so examination alone may not establish the type.

Genetic testing can confirm a deletion or another relevant change involving the STS gene. Chromosomal microarray testing may be used to identify a deletion and show whether nearby genes are also missing.

An enzyme test can measure steroid sulfatase activity, although it is not available everywhere. A skin biopsy, which removes a small sample for examination, is rarely needed and does not usually confirm the genetic cause.

Relatives may be offered carrier testing after the family's genetic change has been identified. Genetic counselling can help explain what a result means for the person tested and other family members.

Treatment and management

There is no treatment that replaces the missing gene or permanently restores steroid sulfatase activity. Care focuses on reducing scale, keeping the skin comfortable, and managing associated concerns.

Regular use of an emollient is central to treatment. An emollient is a moisturising product that softens the skin and reduces water loss. Ointments and thick creams generally hold moisture longer than lotions. Applying them after bathing can help trap water in the skin.

Scale-removing products may contain urea, lactic acid, or another alpha-hydroxy acid. These

are called keratolytics because they loosen thickened dead skin. They can sting cracked or inflamed areas, and not every product is suitable for young children.

Salicylic acid can be absorbed through the skin. Using strong products over large areas, particularly in infants and young children, can cause toxicity. It should only be used in this setting with appropriate medical guidance.

Helpful daily measures may include:

A dermatologist may consider a topical retinoid for limited troublesome areas. Oral retinoids, medicines related to vitamin A, are reserved for more severe symptoms because they can cause significant side effects and require monitoring. They also carry serious pregnancy risks.

Associated concerns are treated separately. For example, an undescended testicle needs assessment, while learning or attention difficulties may lead to developmental or educational support.

- Short baths or showers in warm rather than very hot water
- Gentle, fragrance-free cleansers instead of harsh soaps
- Moisturiser applied while the skin remains slightly damp
- Soft clothing and attention to overheating during activity

Complications

Thick scale can crack, become painful, or occasionally allow a skin infection to develop. Reduced sweating may increase the risk of overheating, especially in hot, humid conditions or during strenuous exercise.

Some affected males are born with one or both testicles outside the scrotum. This is called cryptorchidism, or undescended testicles. It should be assessed because treatment may be needed for reasons not limited to the skin condition.

Small cloudy areas can develop in the cornea, the clear front surface of the eye. These usually do not affect vision, but new visual symptoms should not be assumed to come from ichthyosis.

Attention-deficit hyperactivity disorder, autism-related traits, and some mood or developmental differences have been reported more often among people with X-linked ichthyosis. Most affected people do not have all these concerns. A larger deletion involving nearby genes can increase the likelihood of additional features.

Prevention

Recessive X-linked ichthyosis cannot be prevented through diet, skin care, or choices made during pregnancy. It is caused by an inherited genetic change.

Genetic counselling can help people understand carrier testing and the chance of passing the altered gene to a child. Depending on personal circumstances and local availability, reproductive options may include prenatal testing or testing embryos created through in vitro fertilisation. These are personal choices rather than required steps.

Moisturising and avoiding overheating do not prevent the condition, but they can reduce symptoms and complications.

Living with it and what to expect

The scaling is lifelong, although its visibility and severity can change. It often becomes more obvious in winter and improves during warm, humid weather. Skin care usually needs to continue even when symptoms are mild.

Finding a manageable routine can take time. Some people need only regular moisturiser, while others use scale-removing treatments or periodic dermatology care. Visible scaling may affect confidence, clothing choices, or comfort in social situations, even when it causes little physical discomfort.

Most people with an isolated STS gene change have a normal lifespan and can take part in school, work, exercise, relationships, and family life. Adjustments may be needed in hot conditions if sweating is reduced.

Follow-up is guided by individual features. Routine appointments may review skin comfort, treatment side effects, heat tolerance, testicular health, vision, and any learning or attention concerns.

When to get medical help

Arrange a routine appointment if persistent scaling has not been assessed, especially when it began in infancy or occurs in male relatives. A confirmed diagnosis can guide skin care and clarify inheritance.

Routine medical review is also appropriate for:

Seek urgent medical help for spreading redness, increasing warmth, swelling, pus, or fever, as these may suggest a skin infection.

Move to a cool place and seek urgent help if someone becomes unusually weak, dizzy, nauseated, or develops a severe headache in the heat. Confusion, fainting, a seizure, or inability to cool down may indicate severe heat illness and requires emergency care.

- Painful cracks, persistent itching, or treatment irritation
- Difficulty sweating or repeated overheating
- An undescended testicle
- New problems with vision
- Concerns about learning, attention, mood, or development
- Questions about carrier testing, pregnancy, or inheritance

Common questions

Is recessive X-linked ichthyosis contagious?

No. It is caused by an inherited genetic change. It cannot spread through touch, shared towels, swimming, sex, or close contact.

Will a child grow out of it?

Usually not. The underlying enzyme deficiency remains throughout life. The scale may become less noticeable at certain times, particularly in warm, humid weather, but ongoing skin care is commonly needed.

Can females have X-linked ichthyosis?

Females are more often carriers because they usually have a second working copy of the STS gene. Some carriers have mild dryness or scaling. More pronounced symptoms can occur in unusual genetic circumstances but are uncommon.

Why does it get worse in winter?

Cold outdoor air and heated indoor air contain less moisture. This increases water loss from the skin, making scale drier and more visible. Regular use of a thicker emollient may help during these months.

Can a father pass it to his son?

No. A father gives his Y chromosome, not his X chromosome, to a son. An affected father passes the altered X chromosome to all his daughters, who will usually be carriers if their other X chromosome is unaffected.

Does it affect life expectancy?

Isolated recessive X-linked ichthyosis does not usually shorten life. The outlook may be different when a larger chromosome deletion causes additional medical or developmental conditions.

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