CONDITION

CHILD naevus

Learn how CHILD naevus affects the skin and sometimes the limbs or internal organs, including its genetic cause, diagnosis, treatment, and follow-up.

WHAT IT IS
Rare, usually lifelong genetic skin condition

BODY SYSTEM
Skin

MAIN SYMPTOMS
One-sided scaly patches, itching, cracking, limb differences

WHAT CAUSES IT
Genetic, usually an NSDHL change

HOW IT IS DIAGNOSED
Clinical pattern, skin biopsy, genetic testing

MAIN TREATMENT
Supportive skin care and symptom management

CAN IT BE CURED?
No, underlying genetic change remains

IS IT AN EMERGENCY?
Sometimes, severe breathing difficulty or loss of consciousness

LONG-TERM OUTLOOK
Usually lifelong with widely variable severity

Overview

CHILD naevus is a rare, usually lifelong skin condition caused by a change in how the body makes cholesterol. It is the characteristic skin finding of CHILD syndrome. CHILD stands for congenital hemidysplasia with ichthyosiform naevus and limb defects .

"Congenital" means present at birth. "Hemidysplasia" means that structures on one side of the body have not developed in the usual way. "Ichthyosiform" describes thick, dry, or scaly skin. A naevus is a localized area of skin that developed differently from the surrounding skin.

The affected skin is usually limited mainly to one side of the body and may stop sharply at the midline. The same genetic change can also affect a limb, the skeleton, or, less often, internal organs on that side. Some people have mostly skin findings, while others have the broader features of CHILD syndrome.

CHILD naevus matters because the skin can become thick, itchy, cracked, or prone to infection. Limb differences may affect movement and daily activities. Identifying the condition can also guide genetic counseling and help clinicians check for related developmental differences.

Care is usually coordinated by a dermatologist. Depending on the person's features, the team may also include genetics, orthopedics, rehabilitation, pediatrics, and other specialists.

Symptoms

The skin changes are often visible at birth or appear during early infancy. Their extent and severity vary widely.

Typical skin features include:

The face is often less affected than the trunk and limbs, although patterns differ. The skin changes may follow developmental lines in the skin rather than the shape of a nerve or blood vessel.

Changes outside the skin may occur on the same side as the naevus. These can include:

Less commonly, the condition may involve the brain, heart, lungs, kidneys, or other organs. The type of evaluation needed depends on the person's examination and symptoms. Not everyone needs every possible test.

- Red, pink, orange, or yellowish patches covered by waxy or thick scale
- A sharply defined pattern affecting mainly one side of the body
- A border that may stop near the middle of the chest, abdomen, or back
- Greater involvement of skin folds, such as the groin, armpit, or crease between the buttocks
- Thick, rough, or wart-like areas
- Itching, tenderness, cracking, or occasional bleeding
- Areas that become moist, sore, or irritated from friction
- A shortened, underdeveloped, or missing part of a limb
- Differences in fingers or toes
- Joint stiffness or reduced range of movement
- Curvature or uneven development of the spine

- Differences in the muscles or bones of the pelvis or chest

Causes and risk factors

CHILD naevus is most often caused by a disease-causing change in the NSDHL gene. This gene provides instructions for an enzyme involved in the final stages of cholesterol production.

Cholesterol is not only related to blood cholesterol levels. The body also needs cholesterol to build cell membranes, make certain hormones, and support normal development. In CHILD syndrome, affected skin cells cannot complete part of the cholesterol-making pathway. They may have too little cholesterol and a buildup of other sterol substances. This disrupts skin growth and causes inflammation and scaling.

The NSDHL gene is on the X chromosome. The condition is therefore described as X-linked dominant . Most recognized cases occur in girls and women.

In females, each cell usually turns off one of its two X chromosomes. This process is called X-chromosome inactivation. If different cells keep different X chromosomes active, the body contains a mixture of affected and unaffected cells. This biological mosaic helps explain the one-sided or patchy pattern.

The condition is much less common in males. A genetic change affecting most cells may not be compatible with development in a male fetus. Rare surviving males may have the change in only some cells, known as mosaicism, or may have another unusual genetic situation.

Many cases result from a new genetic change rather than one inherited from a parent. The chance of the condition occurring again in a family depends on the exact genetic finding and whether the change is present in a parent's blood or reproductive cells. A genetics professional can explain this after testing.

CHILD naevus is not caused by an infection, an allergy, or anything a parent did during pregnancy. Diet and ordinary cholesterol levels do not cause it.

Diagnosis

Diagnosis begins with the appearance and distribution of the skin changes. A clinician will ask when they first appeared, how they have changed, and whether there are concerns about growth, movement, or development.

A full examination may include the skin, limbs, joints, spine, and body symmetry. The clinician may also check the heart, lungs, nervous system, and abdomen when the medical history or

examination suggests broader involvement.

A small skin biopsy can help support the diagnosis. During a biopsy, a clinician removes a small sample after numbing the area. Under a microscope, CHILD naevus may show thickened surface skin and clusters of lipid-filled cells called foam cells. These findings are helpful but are not always unique to this condition.

Genetic testing can look for a disease-causing change in NSDHL. Blood testing may confirm the diagnosis, but it can sometimes miss a change that is present mainly in the affected skin. In that situation, testing a skin sample may be considered by a genetics or dermatology team.

Other conditions can resemble CHILD naevus. These include other types of epidermal naevus, inflammatory linear verrucous epidermal naevus, naevus sebaceous, and certain disorders of cholesterol production. Diagnosis should therefore combine the clinical pattern, biopsy findings, and genetic results rather than rely on one feature alone.

Imaging or other tests are selected according to individual findings. For example, limb or spine differences may lead to X-rays or other imaging. Heart, kidney, or neurological testing is not automatically required for every person.

Treatment and management

There is no single treatment plan that fits everyone. Management focuses on improving skin comfort, limiting complications, supporting mobility, and addressing any associated developmental differences.

Daily skin care may include bland moisturizers or ointments to soften scale and reduce cracking. Gentle washing and avoidance of harsh soaps can help protect the skin barrier. Dressings may be useful over areas exposed to rubbing.

Clinicians sometimes use medicines that loosen thick scale, called keratolytics, or topical retinoids that regulate skin-cell growth. Topical corticosteroids may briefly reduce inflammation in selected areas. These treatments do not correct the underlying cholesterol pathway, and irritation can limit their use.

A more targeted approach combines topical cholesterol with a topical statin, such as lovastatin or simvastatin. The cholesterol replaces a needed end product, while the statin like atorvastatin or simvastatin is intended to reduce the buildup of potentially harmful precursor substances. Published reports describe meaningful improvement in some patients, but the evidence comes mainly from case reports and small series rather than large clinical trials [4,5].

These preparations often need to be specially compounded by a pharmacy. They are not the same as taking a statin by mouth for high blood cholesterol. The concentration, area treated,

monitoring, and potential for absorption require specialist oversight, especially for infants or when large areas of skin are involved.

Procedures may be considered when a localized area remains painful, very thick, or difficult to manage. Options described in specialist care include surgical removal, skin grafting, or laser treatment. Results vary, and skin changes can return after some procedures. The likely benefit must be weighed against scarring, healing problems, anesthesia, and the size and location of the lesion.

Treatment for limb or skeletal differences may involve:

Psychological support can be valuable, particularly when visible skin changes, repeated procedures, or physical differences affect confidence, school, work, or social activities.

- Physical or occupational therapy to preserve movement and independence
- Splints, braces, mobility aids, or adapted footwear
- Orthopedic procedures when they are expected to improve function
- Pain management tailored to the cause of discomfort

Complications

Thick or fragile skin may split and create painful fissures. Open areas can allow bacterial or fungal infection. Ongoing moisture and friction in skin folds may also cause irritation and odor.

Extensive scale can make temperature control and skin care more difficult, particularly in young children. Persistent itching or discomfort may disturb sleep and concentration.

Limb and skeletal differences can contribute to joint stiffness, uneven walking, back discomfort, or reduced ability to carry out everyday tasks. Early rehabilitation may help maintain function and prevent avoidable contractures, which are joints that become fixed in a bent or limited position.

The emotional effect should not be overlooked. A visible naevus or limb difference may lead to unwanted attention, social stress, or concerns about body image. These experiences are valid reasons to seek support.

Persistent ulcers, new lumps, or unexplained bleeding within a long-standing lesion should be assessed. Serious skin changes appear to be uncommon, but any new or clearly changing area deserves examination rather than being assumed to be part of the naevus.

Follow up

Routine follow-up is based on age, symptoms, and the parts of the body involved. Children may need periodic review as they grow because limb length, spine alignment, joint movement, and support needs can change over time.

A routine appointment is appropriate for increasing scale, itching, repeated cracking, treatment irritation, new mobility problems, or concerns about growth and development. Medication and compounded-treatment plans should be reviewed rather than changed without professional advice.

Seek prompt medical advice for increasing warmth, swelling, pain, pus, rapidly spreading redness, or fever, as these may suggest a skin infection. A painful joint that suddenly loses movement or a limb that becomes unusually cold, pale, or swollen also needs urgent assessment.

Call emergency services for severe breathing difficulty, loss of consciousness, a prolonged seizure, or other immediately life-threatening symptoms. These are not routine features of CHILD naevus, but they require emergency care regardless of the underlying condition.

Genetic counseling may be useful during adolescence or adulthood, before pregnancy, or when another family member has related skin or developmental findings.

Sources

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