CONDITION

Van lohuizen syndrome

A birth-present capillary malformation that causes a fixed, net-like blue-purple skin pattern and sometimes uneven limb growth.

WHAT IT IS
Congenital net-like capillary malformation of the skin

BODY SYSTEM
Skin and small blood vessels

MAIN SYMPTOMS
Marbled skin, visible veins, skin thinning, limb asymmetry

WHAT CAUSES IT
Usually a mosaic GNA11 gene change

HOW IT IS DIAGNOSED
Clinical examination, sometimes affected-skin genetic testing

MAIN TREATMENT
Monitoring, skin care, and treatment of associated problems

CAN IT BE CURED?
No - markings often fade naturally

IS IT AN EMERGENCY?
Sometimes - sudden eye pain or vision loss

LONG-TERM OUTLOOK
Often stable or fading, with growth monitoring

Overview

Van Lohuizen syndrome is the older name for cutis marmorata telangiectatica congenita, often shortened to CMTC. It is a capillary malformation, meaning that some of the smallest blood vessels in the skin developed differently before birth.

CMTC causes a blue, purple, or red net-like pattern on the skin. The pattern can look like marble. Unlike ordinary mottling caused by cold, it does not fully disappear when the skin becomes warm.

The markings are usually present at birth or become clear soon afterward. They may cover one area, such as part of a leg, or several areas. One side of the body is often more affected than the other.

CMTC mainly affects the skin. In some people, the tissues beneath the affected area grow differently. This can lead to a thinner or, less often, larger limb. Rarely, there are eye problems or other associated physical differences. Recognising these features allows growth, skin health, and vision to be monitored when needed.

Symptoms

The main feature is a fixed, branching or net-like skin pattern. Its colour and visibility can change with temperature, crying, exercise, or circulation, but the pattern does not completely vanish with warming.

Other skin features may include:

The pattern usually affects a limited part of the body. A leg is a common location, although the arms, trunk, face, or several regions can be involved.

An affected arm or leg may be smaller in width or length. This is called undergrowth or hypotrophy. Less often, the affected area is larger. Differences may be subtle in infancy and become easier to see as a child grows.

CMTC itself is not usually painful or itchy. Pain may come from an open ulcer, an infection, joint strain caused by unequal leg length, or another condition affecting the same area.

When the face is involved, especially near an eye, there may be a higher risk of glaucoma. Glaucoma is damage related to pressure inside the eye. In babies and children, possible signs include a cloudy or enlarged eye, excessive tearing, discomfort in bright light, or frequent eye rubbing.

- Prominent small veins or widened surface blood vessels
- Thin or slightly sunken skin
- Affected skin that feels cooler than nearby skin
- Small ulcers, particularly in a severely affected limb
- Scarring where an ulcer has healed

Causes and risk factors

CMTC is usually linked to a genetic change that occurs after conception while an embryo is developing. This is called a mosaic change. It means that only some cells carry the change, while the rest do not.

Changes in a gene called GNA11 have been found in affected tissue in many people with CMTC. This gene helps control signals involved in blood-vessel development. A genetic change is not identified in every person who is tested, so a negative test does not rule out the condition.

Because the change happens by chance after conception, CMTC is not usually inherited from a parent. It is also not caused by anything a parent did during pregnancy. There are no established dietary, environmental, or lifestyle risk factors.

The chance that parents will have another child with CMTC is thought to be low. A genetics professional can explain recurrence risk when there are unusual features or a relevant family history.

Diagnosis

CMTC is usually diagnosed by examining the skin and reviewing when the markings first appeared. There is no routine blood test that confirms it.

The clinician will look at whether the pattern remains after warming. Ordinary newborn mottling, called physiological cutis marmorata, usually fades as the skin warms. They will also check for skin thinning, ulcers, visible veins, and differences in limb size or length.

Measurements of the arms or legs may be recorded and repeated during growth. Photographs, taken with consent, can help show whether the markings or body proportions are changing.

Most people do not need a skin biopsy or scan. Ultrasound, magnetic resonance imaging, or other tests may be considered if there is significant swelling, unusual overgrowth, pain, or concern about a deeper vascular malformation.

Genetic testing may be offered when the diagnosis is uncertain or the findings are extensive. Because the genetic change is mosaic, testing affected skin may be more useful than testing blood. Even affected-skin testing can be negative.

Other conditions can resemble CMTC. These include ordinary cold-related mottling, port-wine birthmarks, Klippel-Trénaunay syndrome, Adams-Oliver syndrome, and conditions involving both vascular and pigment birthmarks. A specialist assessment may be useful when the pattern is widespread or accompanied by head enlargement, major overgrowth, missing skin at birth, or unusual hands and feet.

Treatment and management

Many people need monitoring rather than active treatment. There is no treatment that

changes the underlying mosaic genetic change, and skin markings often become lighter during childhood.

Management is based on the features present:

A vascular-anomalies or dermatology service may coordinate care when several body areas are involved. Orthopaedic assessment can help if unequal leg length affects posture or walking.

Vascular lasers, such as pulsed-dye lasers, have been used to reduce visible colour. Results are inconsistent because CMTC vessels can lie at different depths. Laser treatment may also cause pain, pigment changes, or scarring. A specialist can explain the likely benefit and limitations for a particular area, but complete removal cannot be guaranteed.

Surgery is not usually needed for the skin marking itself. It may occasionally be considered for a major limb-length difference or another structural problem.

- Gentle skin care helps protect thin or fragile areas
- Dressings and wound care are used for ulcers
- Infection, pain, or bleeding is treated if it occurs
- Limb measurements help identify a growing length difference
- Physiotherapy, shoe lifts, or orthopaedic care may help with significant asymmetry
- Eye examinations may be arranged, particularly when the face is affected

Complications

Most complications are local to the affected area rather than problems throughout the body.

Thin skin can break down and form an ulcer. An ulcer may be painful, heal slowly, or leave a scar. Infection is possible if bacteria enter through broken skin.

Unequal growth may cause one limb to become shorter, narrower, or occasionally larger than the other. A substantial difference in leg length can affect walking, hip position, or the back. Because this may change during childhood and adolescence, repeated measurements can be more helpful than a single assessment.

Glaucoma is an uncommon but important associated problem. The risk is of particular concern when CMTC affects the face or skin around an eye. Early eye assessment can identify pressure changes before they cause lasting vision problems.

Visible markings or body asymmetry can also affect confidence, especially during adolescence. Camouflage products, practical clothing choices, and psychological support may

help if appearance causes distress or unwanted attention.

Not every medical finding in someone with CMTC is caused by the condition. New or unexpected symptoms still need their own assessment.

Prevention

CMTC cannot currently be prevented. The mosaic genetic change usually happens by chance during early development, and there is no known action before or during pregnancy that prevents it.

Skin protection cannot remove the condition, but it may reduce avoidable problems. Moisturising dry skin, protecting fragile areas from rubbing, and treating cuts promptly may lower the chance of skin breakdown.

Regular growth and eye checks do not prevent CMTC either. Their purpose is to find associated problems early, when support or treatment may be more effective.

Living with it and what to expect

The skin pattern often becomes less noticeable during the first years of life. Some markings fade considerably, while others remain visible into adulthood. Cold conditions may continue to make the pattern look darker even after it has faded.

Skin thinning, scarring, or differences in limb size may not improve when the colour fades. Limb-length differences can become more apparent during growth spurts, so children may need routine measurements until growth is complete.

Follow-up is usually more frequent when there is facial involvement, an ulcer, or unequal limb growth. A person with a small, stable marking and no associated findings may eventually need little or no specialist follow-up.

Most people with isolated CMTC can take part in usual school, work, exercise, and family activities. Activities may need adjustment if fragile skin, pain, or a meaningful leg-length difference causes difficulty. CMTC is not contagious and does not turn into skin cancer.

The condition is usually not passed to children because the genetic change is present only in a group of the affected person's cells. Genetic counselling may be helpful for anyone who wants a personal discussion about family planning.

When to get medical help

Arrange a routine appointment if a birthmark has a persistent net-like pattern that does not disappear with warming. A child with known CMTC should also be reviewed if one limb appears to be growing differently, walking becomes uneven, or the skin pattern changes unexpectedly.

Seek prompt medical advice for:

Get emergency help for sudden loss of vision or severe eye pain. Uncontrolled bleeding from an open area also needs emergency care. These events are not typical of uncomplicated CMTC, but they require immediate assessment.

- A new ulcer or a wound that is not healing
- Increasing pain, warmth, swelling, or redness
- Pus, fever, or other signs of infection
- New difficulty walking or using an affected limb
- A cloudy or enlarged eye, excessive tearing, or sensitivity to light

Common questions

Is Van Lohuizen syndrome the same as CMTC?

Yes. Van Lohuizen syndrome is an older name for cutis marmorata telangiectatica congenita. CMTC is the term now used more often in medical care.

Will the marbled skin go away?

It often fades during childhood, especially in the first few years. It may not disappear completely. Thin skin, scars, and differences in limb growth can remain even when the colour becomes lighter.

Is CMTC inherited?

Usually not. It is generally caused by a mosaic genetic change that happens after conception and affects only some cells. Parents did not cause it, and the chance of passing it to a child is thought to be low.

Does CMTC hurt?

The skin marking itself is usually painless. Pain may indicate an ulcer, infection, joint strain, or another problem and should be assessed rather than assumed to be part of CMTC.

Can laser treatment remove the birthmark?

Laser treatment may reduce colour in some cases, but results are unpredictable and complete

removal is unlikely. A vascular laser specialist can assess the depth and location of the affected vessels and explain possible adverse effects.

Will an affected leg always be shorter?

No. Many people have no meaningful leg-length difference. When undergrowth is present, the difference may stay small or become more noticeable during growth. Regular measurements help determine whether treatment is needed.

Sources

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