CONDITION

CMTC

Learn about cutis marmorata telangiectatica congenita (CMTC), a rare vascular birthmark, including symptoms, diagnosis, monitoring, and treatment.

WHAT IT IS
Rare congenital capillary malformation

BODY SYSTEM
Skin and blood vessels

MAIN SYMPTOMS
Net-like skin pattern, thin skin, limb-size differences, skin ulcers

WHAT CAUSES IT
Unknown, sometimes mosaic genetic changes

HOW IT IS DIAGNOSED
Skin appearance and physical examination

MAIN TREATMENT
Observation and symptom-based care

CAN IT BE CURED?
No, color may fade but persist

IS IT AN EMERGENCY?
Sometimes, sudden vision loss, severe infection, or uncontrolled bleeding

LONG-TERM OUTLOOK
Many remain healthy, though skin changes may persist

Overview

CMTC stands for cutis marmorata telangiectatica congenita . It is a rare type of vascular birthmark caused by blood vessels that developed differently before birth. The affected skin has a fixed, net-like pattern that may look red, purple, or blue.

The name describes the condition:

CMTC is usually visible at birth, although it may become clearer during the first weeks of life. It most often affects one arm or leg. It can also involve the trunk, face, or more than one area. The pattern may look darker when the skin is cold, but unlike normal temporary skin mottling, it does not disappear completely after warming.

CMTC is considered a capillary malformation , meaning a structural difference in small blood

vessels rather than a tumor or infection. It is not contagious and is not caused by anything a parent did during pregnancy.

In many children, the color becomes lighter during early childhood. Some skin changes may remain into adulthood. CMTC matters because a minority of affected children have differences in limb growth, fragile or ulcerated skin, or another medical concern that needs monitoring. A careful examination helps determine whether the birthmark is isolated or part of a broader pattern of findings.

CMTC is not the same as macrocephaly-capillary malformation syndrome, now generally called PIK3CA-related overgrowth spectrum or megalencephaly-capillary malformation-polymicrogyria syndrome . These are separate conditions, although older medical literature sometimes used overlapping names.

- Cutis marmorata means marbled-looking skin.
- Telangiectatica refers to widened small blood vessels.
- Congenita means present at birth.

Symptoms

The main sign of CMTC is a persistent, branching or net-like skin pattern. Its color may range from pink-red to deep purple or blue. The pattern often follows part of a limb rather than crossing the entire body evenly.

The affected area may partially lighten when pressed because pressure briefly pushes blood out of the small vessels. Cold temperatures can make the pattern more noticeable. Warming may soften its appearance, but the birthmark usually remains visible.

CMTC commonly causes no pain or itching. Other possible features include:

Limb differences most often involve undergrowth, with the affected arm or leg being thinner or shorter. Overgrowth can occur, but it should prompt clinicians to consider whether another vascular or overgrowth condition may better explain the findings.

CMTC on the face, especially near an eye, deserves particular attention. Some people with facial involvement have glaucoma, a condition in which pressure damages the optic nerve. Glaucoma may not cause obvious symptoms at first, so an eye examination may be recommended even when vision seems normal.

Symptoms and associated findings vary widely. Many people have only the skin pattern and remain otherwise healthy.

- Skin that looks thin, indented, or slightly wasted in places
- More visible veins within or next to the birthmark
- A difference in the length or width of an affected limb
- Small areas of skin breakdown or ulceration, especially in infancy
- Scarring after an ulcer heals
- Pale patches called nevus anemicus , caused by locally narrowed blood vessels

Causes and risk factors

CMTC develops when small blood vessels form in an unusual pattern before birth. The altered vessels are mainly close to the skin surface, which creates the visible marbled appearance.

The exact cause is not known in every person. Research has identified changes in a gene called GNA11 in some affected tissue. These changes may be mosaic , meaning they occur in only some cells rather than every cell in the body. A mosaic change may arise early in fetal development and affect one limited body area.

Because mosaic genetic changes may not be present in blood, a routine blood-based genetic test can be normal even when a genetic cause exists in the affected skin. Testing is not needed for every person with typical, isolated CMTC.

Most cases are sporadic, meaning they occur in a person with no family history of the condition. CMTC is generally not considered an inherited disorder, and the chance of it happening again in another child appears to be low. A genetics professional can provide more specific counseling when CMTC occurs with unusual growth, developmental differences, or several congenital findings.

There are no well-established actions, foods, medicines, or pregnancy exposures known to cause typical CMTC. Cold weather can make the birthmark look darker, but it does not cause the condition.

Diagnosis

CMTC is usually diagnosed from the skin's appearance and a physical examination. There is no single laboratory test that confirms every case.

A pediatrician, dermatologist, or vascular-anomalies specialist may examine whether the pattern remains after warming. This helps distinguish CMTC from normal physiologic cutis

marmorata , a temporary mottled appearance that commonly occurs when infants are cold and disappears with warming.

The examination may include:

Photographs and measurements can provide a baseline for future comparison.

Imaging is not routinely required for a typical superficial birthmark. Ultrasound or magnetic resonance imaging may be considered if the clinician suspects deeper blood-vessel involvement, a significant limb difference, or another diagnosis. A skin biopsy is rarely necessary.

An ophthalmology examination may be recommended when CMTC affects the face or skin around an eye. The eye specialist can check pressure inside the eye and examine the optic nerve.

Genetic evaluation may be useful if the person has marked overgrowth or undergrowth, an unusually large head, developmental concerns, several vascular birthmarks, or abnormalities involving other organs. These findings do not automatically mean a genetic syndrome is present, but they may change which tests or monitoring are appropriate.

Conditions that may resemble CMTC include temporary newborn mottling, other capillary malformations, venous malformations, and vascular-overgrowth disorders. Specialist review can help when the diagnosis is uncertain.

- Mapping which areas of skin are involved
- Checking for thin skin, ulcers, scars, or visible veins
- Measuring the length and circumference of both arms or legs
- Looking for differences in head or body growth
- Assessing movement, strength, and walking when age appropriate
- Reviewing development and any other birth differences

Treatment and management

Many people with CMTC do not need treatment for the birthmark itself. Observation is often appropriate when the skin is healthy, limb growth is balanced, and there are no associated concerns.

The color frequently fades during childhood, although it may not disappear completely. Treatment decisions are usually based on symptoms, function, skin health, and the person's preferences rather than appearance alone.

Gentle skin care can help protect affected areas. A plain moisturizer may reduce dryness and cracking. Clothing and footwear should avoid repeated rubbing over thin or fragile skin. Keeping the area comfortably warm may make the color less prominent, but excessive heat should be avoided.

Any ulcer needs medical assessment. Care may include protective dressings, wound-care guidance, and treatment of infection if one develops. Because management depends on the ulcer's depth, location, and cause, families should not apply medicated products unless advised by a clinician.

If one limb grows differently, management may involve a pediatric orthopedist, physiotherapist, or rehabilitation specialist. Small differences may only need measurement over time. A more noticeable leg-length difference may affect walking and could require a shoe adjustment or another orthopedic approach. These decisions depend on growth and function.

Vascular laser treatment, often with a pulsed-dye laser, may be considered for persistent color or visible small vessels. Results are variable, and several sessions may be needed. Laser treatment may not correct thin skin or limb-size differences. A dermatologist or vascular-anomalies team can explain likely benefits, discomfort, scarring risk, and whether treatment is suitable for the specific skin area.

Glaucoma requires treatment by an ophthalmologist. Its management is separate from treatment of the skin birthmark.

Visible birthmarks can also affect confidence or attract unwanted questions. Age-appropriate explanations, school support, counseling, or connection with a vascular-birthmark support organization may be helpful if appearance causes distress.

Complications

Most people with CMTC do not develop serious complications. Possible problems depend on the location and extent of the affected skin.

Skin ulceration is one of the more important early complications. An ulcer can be painful and may leave a scar. Open skin also creates a path for infection, although infection is not an expected feature of uncomplicated CMTC.

A difference in limb length or circumference may become clearer as a child grows. Mild differences may have little effect. Larger differences can influence posture, footwear, walking, or use of an arm and may need orthopedic monitoring.

Thin skin and reduced soft tissue can persist even after the purple color fades. Some people

also report greater sensitivity to cold in the affected area.

Glaucoma is an uncommon but important concern when the face or area around an eye is involved. Without treatment, glaucoma can harm vision. This is why preventive eye assessment may be advised before symptoms appear.

The visible birthmark may have a social or emotional effect, particularly during school years or adolescence. This impact is real even when the condition does not cause physical symptoms.

Follow up

Follow-up is tailored to the person's age, the location of CMTC, and whether growth or skin problems are present. A child with isolated, stable skin changes may need only routine review. Children with limb differences, facial involvement, ulcers, or other findings may need more frequent specialist visits.

During childhood, clinicians may repeat limb measurements and assess walking, movement, skin integrity, and overall growth. Families can take occasional photographs under similar lighting, but daily checking is usually unnecessary.

Contact the usual clinician for a routine appointment if the birthmark changes gradually, a limb-size difference becomes more noticeable, or the condition begins to affect movement, footwear, comfort, or emotional well-being.

Seek prompt medical advice for:

Seek emergency care for sudden loss of vision, severe illness with signs of infection, or bleeding that does not stop after continuous firm pressure. These events are not typical of uncomplicated CMTC, but they require immediate assessment.

- A new ulcer, increasing pain, or skin that is becoming warm and swollen
- Pus, spreading redness, or fever with an open area
- New eye redness, eye pain, light sensitivity, or blurred vision
- A rapid or unexpected change in limb size

Sources

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