







Pathophysiology

The pathogenesis of cutis marmorata telangiectatica congenita (CMTC) remains unclear, and the cause may be multifactorial. Most cases occur sporadically, although rare cases occur in families. Cases of cutis marmorata telangiectatica congenita are reported in association with fetal ascites⁴ and an elevated maternal beta-human chorionic gonadotropin (beta-hCG) level, although a direct relationship has not been established.

Some authors suggest that the Happle lethal gene hypothesis (ie, the lethal dominant gene survives by means of mosaicism) best explains the patchy distribution of the lesions and sporadic occurrence of the disease. Other authors suggest that a possible teratogen is the cause, and yet others consider cutis marmorata telangiectatica congenita to be an autosomal dominant genetic disorder with incomplete penetrance.

Cutis marmorata telangiectatica congenita is described to occur in association with other discrete syndromes such as Sturge-Weber syndrome and Klippel-Trenaunay syndrome. Some have suggested that Sturge-Weber syndrome, Klippel-Trenaunay syndrome, and cutis marmorata telangiectatica congenita may be included in a group of vascular diseases that are associated with other developmental defects of the mesodermal system during embryonic life.

History

Cutis marmorata telangiectatica congenita (CMTC) is generally present at birth or shortly thereafter. The reticulated mottling frequently becomes more prominent in a cold environment (eg, physiologic cutis marmorata), but it tends not to disappear with rewarming

Physical

Cutis marmorata telangiectatica congenita (CMTC) principally affects the skin. Cutis marmorata telangiectatica congenita tends to occur more frequently on the lower limbs, although the upper extremities, trunk, and face may also be involved. When located on the trunk, cutis marmorata telangiectatica congenita tends to have a midline distribution.

The primary lesion is characterized by pinkish blue, reticular, and patchy skin changes. Lesions may be localized or generalized. Localized lesions were observed in 60% of the patients in one series, but this percentage varies. Persistent cutis marmorata, telangiectasia, and phlebectasia may occasionally be associated with cutaneous atrophy and ulceration of the involved skin.⁶

The incidence of abnormalities associated with cutis marmorata telangiectatica congenita is high, varying from 18.8-89%, as follows:

- Way et al, 1974 - 50%⁷
- South and Jacobs, 1978 - 89%⁸
- Picascia and Esterly, 1989 - 27%⁹
- Pehr and Moroz, 1993 - 68%¹⁰
- Devillers et al, 1999 - 80%¹¹
- Amitai et al, 2000 - 18.8%¹²

Skin atrophy and ulcerations, capillary malformations (ie, nevus flammeus), capillary and cavernous hemangioma, atrophy or hypertrophy of the affected extremity, macrocephaly (macrocephaly cutis marmorata telangiectatica congenita syndrome), and glaucoma are frequently associated with cutis marmorata telangiectatica congenita.

Other conditions may be associated with cutis marmorata telangiectatica congenita.

Common associations include the following:

- Body asymmetry (hypoplasia and hypertrophy of the affected limbs)
- Vascular anomalies (capillary and cavernous hemangiomas, nevus flammeus, Sturge-Weber syndrome, Klippel-Trenaunay syndrome, Adams Oliver syndrome) 13
- Glaucoma and retinal detachment¹⁴
- Cutaneous atrophy
- Neurologic anomalies^{15,16}

Uncommon associations include the following:

- Mental retardation
- Psychomotor retardation
- Aplasia cutis congenita
- Cleft palate

Rare associations include the following:

- Patent ductus arteriosus and double aortic arc
- Congenital hypothyroidism
- Distal limb defects and scoliosis
- Mild growth deficiency
- Stenosis of a deep femoral artery
- Congenital generalized fibromatosis
- Disseminated blue nevi
- Syndactyly
- High arched palate
- Micrognathia
- Nevus anemicus
- Neonatal ascites
- Hypoplasia of the right iliac and femoral veins¹⁷
- Café au lait spots
- Mongolian spots¹⁸
- Hypospadias
- Multicystic renal disease
- Elevated maternal hCG level
- Hemophagocytic lymphohistiocytosis¹⁹
- Iliac artery stenosis²⁰
- Airway obstruction: Airway obstruction due to unilateral hypertrophy of vocal cords, in addition to brainstem compromise, may produce apnea in patients with signs and symptoms of cervicomedullary cord compression. 21

Causes

The risk factors and prognostic factors of cutis marmorata telangiectatica congenita (CMTC) are still unknown. The cause may be multifactorial.

Although the disorder most commonly has a sporadic occurrence, some authors suggest that cutis marmorata telangiectatica congenita may be inherited as an autosomal dominant trait with low penetrance. The role of external factors, including viral infections, is suggested because several cases of cutis marmorata telangiectatica congenita occurred in the same geographic area. In theory, some factors can influence vascular development during intrauterine growth.

Laboratory Studies

Physical examination helps in diagnosing cutis marmorata telangiectatica congenita (CMTC).

Imaging Studies

Imaging studies are indicated only for the evaluation of other suspected congenital anomalies. Such studies are specifically performed according to the clinician's suspicion.

Histologic Findings

A skin biopsy is not necessary in cutis marmorata telangiectatica congenita (CMTC) because the histologic findings are nonspecific and nondiagnostic. Microscopic findings include dilated capillaries in the deeper dermis, swollen endothelial cells, and sometimes dilated veins or venous lakes.

Medical Care

No treatment is needed for cutis marmorata telangiectatica congenita (CMTC) unless

associated anomalies (eg, glaucoma, hypospadias, syndactyly, multicystic renal disease, cardiac malformation, limb asymmetry) require treatment.

Consultations

Consultation with an orthopedist and/or neurosurgeon may be necessary for evaluation of associated anomalies (eg, limb or cranial defects).

Consultation with an ophthalmologist may be necessary because glaucoma has been reported in association with cutis marmorata telangiectatica congenita. However, all of the patients with glaucoma had periocular skin changes around the affected eye. Therefore, ophthalmologic evaluation is probably only indicated in this setting.