Cutis marmorata telangiectatica congenita

Babies commonly present with skin anomalies at or shortly after birth. While physiological cutis marmorata is a frequently encountered condition, we report a rare entity that appears similar, but should not be confused with it. Cutis marmorata telangiectatica congenita (CMTC) is a rare, sporadic condition that presents at birth as a localised or generalised reticulated, blue-violet vascular network in the skin. The prognosis is usually good, with minor associated anomalies. Clinical improvement occurs within the first 2 years in most cases but the lesions occasionally persist.

A full-term baby of non-consanguineous Chinese parents, whose family history, antenatal course and delivery were unremarkable, had purplish reticular marks over the right abdominal wall, lower limbs, and buttocks at birth (Figs 1 and 2). Upon blanching, these non-tender reticular marks could not be emptied completely. There were no periocular vascular lesions, dysmorphism, hepatosplenomegaly, limb atrophy or hypertrophy. His growth was appropriate and body temperature was normal. A complete blood picture, blood gases, and chest X-ray were normal and blood cultures were negative.

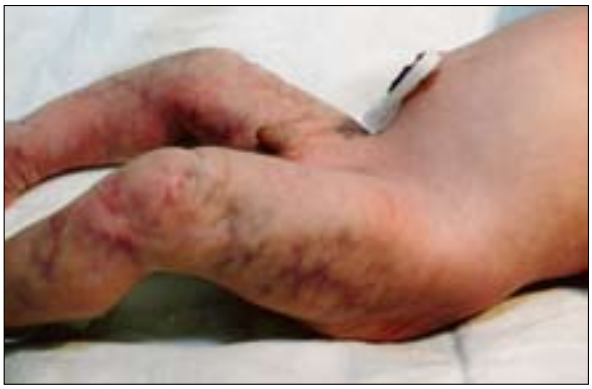

FIG 1. Lateral view of the patient's lower limbs, showing the characteristic purplish reticular pattern on the skin

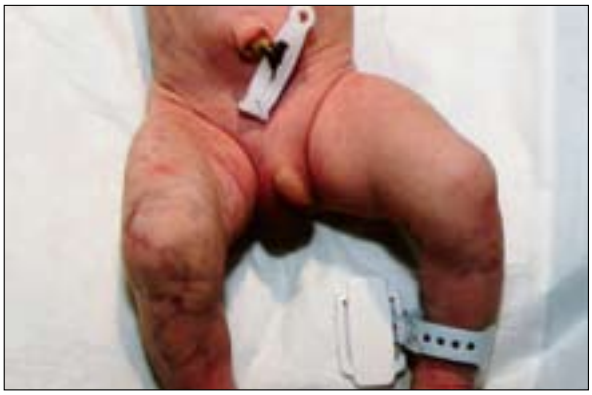

FIG 2. A front view of the same patient, showing the involvement of the lower limbs as well as the abdominal wall

The clinical diagnosis was CMTC, a skin condition first described by Dutch paediatrician Van Lohuizen in 1922. More than 200 cases have been published worldwide under various synonyms, including congenital generalised phlebectasia, naevus vascularis reticularis, livedo telangiectatica, congenital livedo reticularis and Van Lohuizen syndrome. It is an uncommon, sporadic, congenital cutaneous disorder characterised by a localised, segmental, or generalised persistent reticular vascular skin pattern with a marbled bluish to deep purple appearance, which is present at, or shortly after, birth. Major features include persistent cutis marmorata, phlebectasia, telangiectasia and, occasionally, ulceration and atrophy of the involved skin. The reticulate mottling frequently becomes more prominent in a cold environment but tends not to disappear with rewarming. The extremities, especially the lower limbs, are most commonly involved, followed by the trunk and face.

Different hypotheses have been proposed to explain this phenomenon, such as failure of development of the mesodermal vessels in the early embryonic stage, peripheral neural dysfunction, lethal gene theory of Happle or external factors such as a teratogenic agent. Pehr and Moroz[1] reviewed 126 cases and reported associated abnormalities in 68%, the most common ones being body asymmetry (limb hypertrophy or hypoplasia), other vascular anomalies (mostly capillary malformations), glaucoma, cutaneous atrophy and ulcerations, psychomotor and mental retardation.

The diagnosis of CMTC is made on clinical grounds. Skin biopsy is not required if the clinical diagnosis is definite. The histological findings are often non-specific, such as dilated capillaries in the deeper dermis, swollen endothelial cells, dilated veins or venous lakes.[2] This disease must be differentiated from other reticulated vascular lesions. A rare type of diffuse capillary malformation, generalised reticulate capillary malformation, can be confused with CMTC although the former lacks the patchy, linear atrophy and telangiectasia seen in CMTC. Physiological cutis marmorata, a normal response to chilling that occurs during the first few weeks of life, has a much finer,

symmetrical pattern over the trunk and extremities, and disappears in warm temperatures. Livedo reticularis and telangiectasia may also be initial signs of neonatal lupus erythematosus (LE). They can occur without obvious inflammatory lesions and CMTC-like lesions have been described in neonatal LE.[3] A thorough clinical history and maternal blood testing can confirm whether this clinical suspicion is correct as mothers of infants with neonatal LE have anti-Ro autoantibodies in 95% of cases. Diffuse phlebectasia, or Brockenheimer's disease, is a rare progressive hamartomatous malformation involving the deeper veins, characterised by gradual onset during childhood with irregular, painful venous dilatation, usually affecting one limb. Naevus flammeus, also easily confused with CMTC, presents with a light-coloured, reticular pattern, and does not undergo spontaneous resolution.

It is important to perform a careful clinical examination on all patients with CMTC to exclude possible associated anomalies. An ophthalmological assessment is particularly important in order to rule out glaucoma if periocular vascular lesions are present. Patients with CMTC require long-term follow-up for their skin lesions, associated abnormalities and psychomotor development. Associated developmental defects are usually more common in children with widespread generalised CMTC than with more localised or regional CMTC. Fortunately, our case is of the latter type. The prognosis of the skin condition is usually good. Most improve with time, especially within the first 2 years,[4] which is probably related to the normal thickening and maturation of skin. Treatment is not required. Few cases of persistent CMTC have been reported, and laser therapy has been tried with variable outcomes.[1]

In conclusion, CMTC is a sporadic congenital condition with a typical reticular skin pattern at birth and has a number of associated abnormalities. It carries a good prognosis and usually resolves within the first 2 years.

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