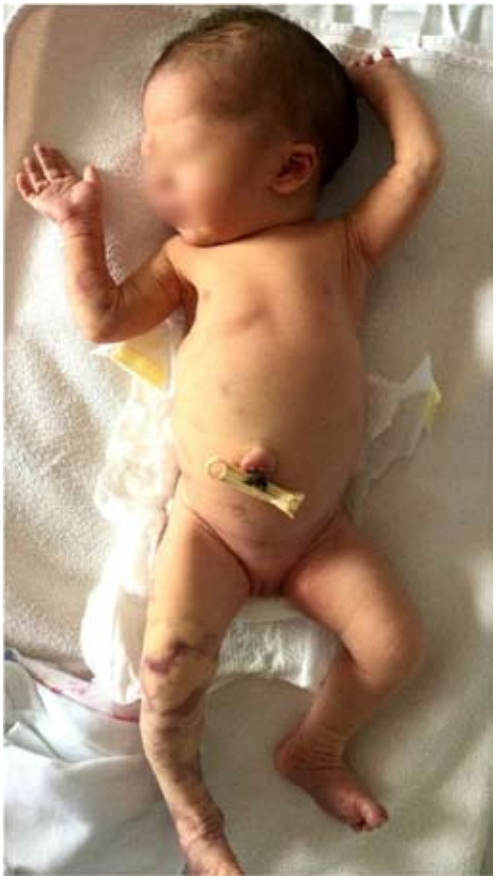
Born in the Purple: An Exceptional Case of Cutis Marmorata Telangiectatica Congenita

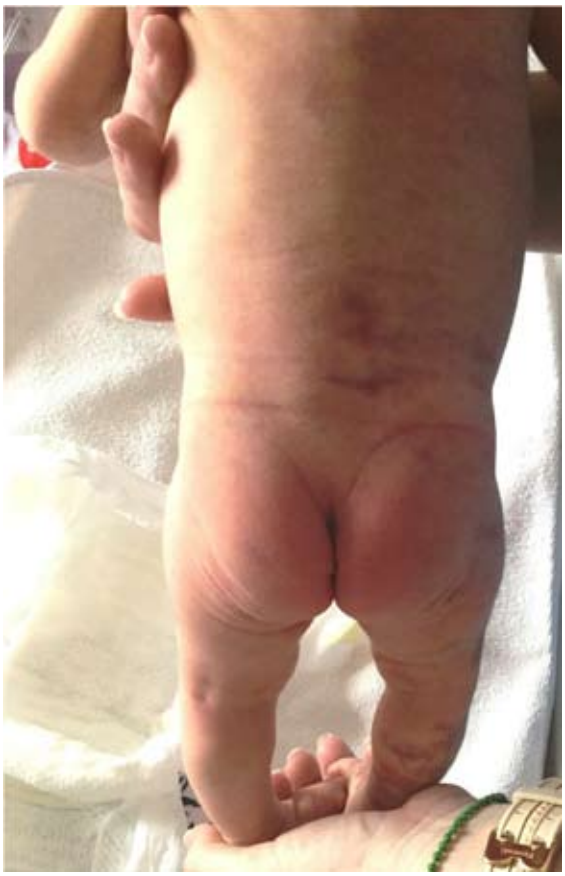
A full-term, 2-day-old female neonate with a congenital non-tender reticular patch that did not disappear with local warming was referred to our department for consultation. The family history as well as the antenatal course and delivery were unremarkable.

On examination, we evidenced a fixed, marbled, bluish to deep purple lesion with a fishnet appearance extending over the right side of her body, face, and scalp. There was presence of atrophy of the involved skin, along with ulceration above the right lateral malleolus. Upon blanching, the lesions could not be emptied completely.

Routine laboratory studies, chest X-rays, and abdominal and cranial ultrasound scan results were nonsignificant. Head and limb circumferences were within normal ranges. The patient was checked by the ophthalmology and neurology department to screen for associated anomalies, which were not detected. Based on the medical history and clinical presentation, the diagnosis of cutis marmorata telangiectatica congenita was established. The infant's parents were reassured about the benign nature of the cutaneous disorder as well as the need for periodic follow-up on an outpatient basis.



Panel A



Panel B

Panel C

Cutis marmorata telangiectatica congenita (CMTC), first described by the Dutch pediatrician Van Lohuizen in 1922, is an exceedingly rare congenital sporadic condition with fewer than 300 cases described in the medical literature to date. Affected infants have discolored vascular patches of skin with a blue-violet marbled appearance. The disorder may present with a segmental or generalized persistent reticular mottling but more frequently has localized distribution over the lower limbs (1,2). Skin lesions are reminiscent of cutis marmorata, a common benign response observed in infants, which resolves with warming of the skin surface. In contrast, CMTC lesions do not disappear with rewarming and occasionally present with ulceration and atrophy of the involved skin (aplasia cutis) (1,3).

Cutis marmorata telangiectatica congenita is described as occuring in association with a plethora of extracutaneous malformations, including undergrowth or overgrowth of the involved extremity, ocular and neurological abnormalities, growth and developmental delays, as well as additional vascular anomalies. The pathogenesis is not fully elucidated, but a lethal gene hypothesis has been suggested by some authors, while others indicate that the disorder may be inherited as an autosomal dominant trait with low penetrance (1,2,4).

Skin biopsy is nonspecific and differential diagnosis is rarely difficult due to the distinctive appearance of cutaneous lesions, and the diagnosis can thus be established on clinical grounds alone. CMTC can be associated with other congenital syndromes including phacomatosis pigmentovascularis and Adams-Oliver syndrome (2,4).

Cutaneous lesions carry a good prognosis and show a tendency for fading or spontaneous resolution over the first years of life in most cases, but the extremity discrepancy tends to persist. A thorough screening for associated anomalies as well as annual controls of skin changes and psychomotor development of the patients should be performed (2-3).

References:

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