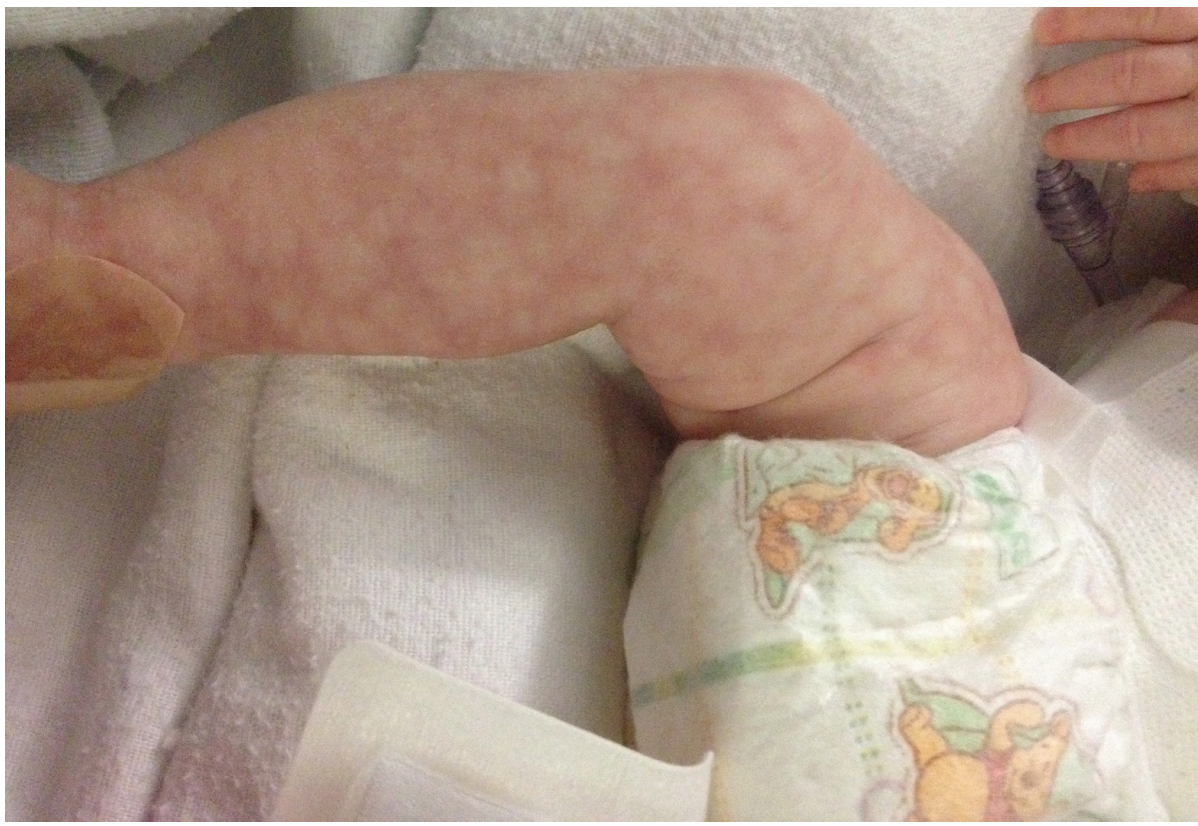
A Photo Quiz to Hone Dermatologic Skills

CARLY GUSS, MD



A 1-month-old full-term girl was admitted to a hospital's pediatrics floor with worsening respiratory distress resulting from bronchiolitis. At the time of evaluation by the rapid response team on the floor, the infant was tachypneic, and the skin of her right lower extremity was noted as having a mottled appearance. She was given a 20-mL/kg bolus of intravenous normal saline, but her leg continued to have the same mottled appearance. A second bolus of saline was administered. Aside from the bronchiolitis and reticulated appearance of the skin, the rest of the examination findings were unremarkable.

Her increased respiratory rate necessitated transfer to the pediatric intensive care unit (PICU) for closer monitoring. Upon examination and history-taking in the PICU, her parents stated that the girl's skin had looked like that since birth, and that her pediatrician had told them that the marbled appearance was normal.

What is the cause of this infant's mottled appearance?

(Answer and discussion on page 298)

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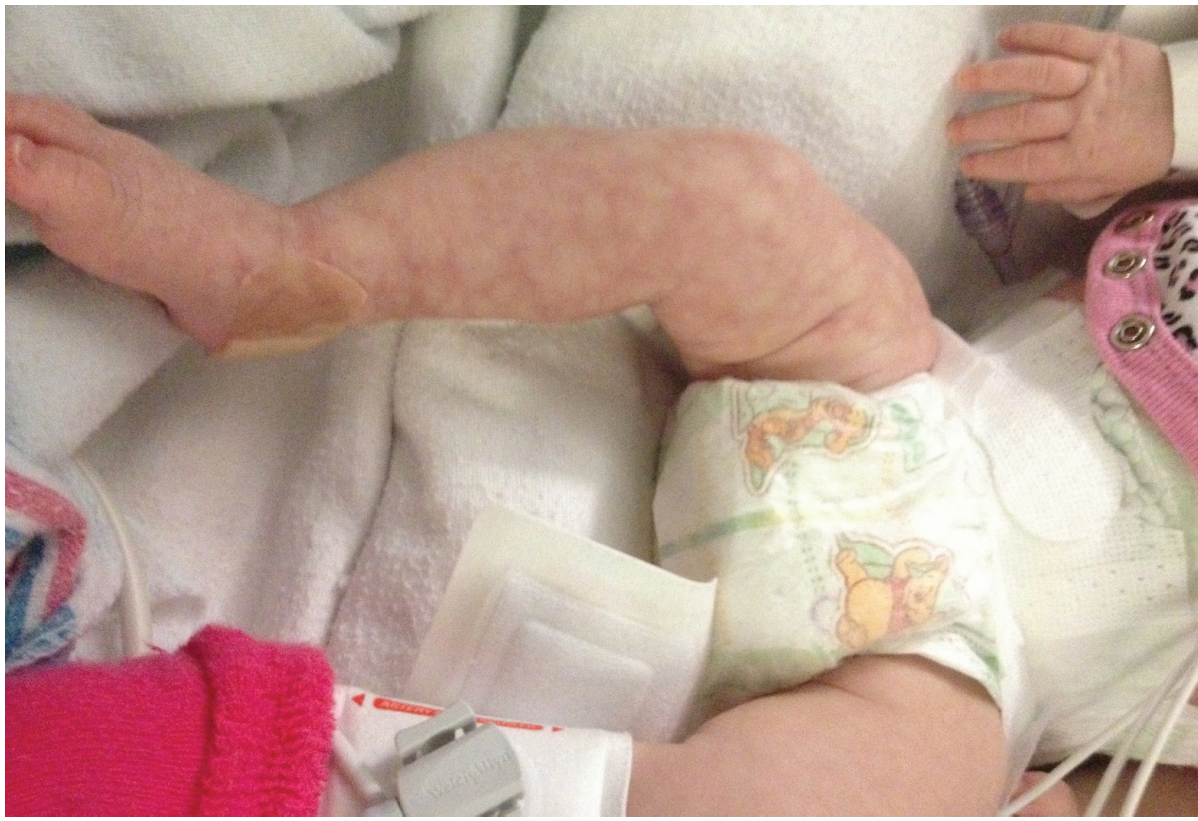

ANSWER: Cutis marmorata telangiectatica congenita

This infant has **cutis marmorata telangiectatica congenita (CMTC)**. This rare congenital condition, known also as Van Lohuizen syndrome, is caused by vascular malformation of capillaries and venous vessels, although the exact pathogenesis is unknown.[1] CMTC is characterized by cutis marmorata (the marbled, lacy appearance of an infant's skin when it is exposed to cold environmental temperatures), telangiectasias, and varicosity.

While CMTC is similar in appearance to physiologic cutis marmorata, the reticulated pattern persists independent of environmental temperature.[2] CMTC typically is localized and affects a single extremity, as with the infant in our case. The appearance of CMTC usually is sporadic, although there have been reports of familial inheritance.[1]

The condition requires no treatment and improves with time. Nevertheless, the presence of other anomalies associated with CMTC have been reported in infants, with estimates ranging from 20% of cases[1] to as many as 80% of cases.[3] These concomitant findings can include other vascular anomalies such as nevi and hemangiomas[2]; ocular anomalies such as glaucoma[1]; neurologic anomalies; musculoskeletal defects such as soft-tissue hypoplasia, limb atrophy and, most commonly, limb asymmetry; and other cutaneous findings such as ulceration and hyperkeratosis. Adams-Oliver syndrome, Klippel-Trénaunay syndrome, and macrocephaly-capillary malformation syndrome also have been reported in association with CMTC.[1]

It is generally recommended that infants who present with CMTC and associated abnormalities be referred to a specialist for further evaluation.

Our patient recovered from her respiratory illness and was discharged with instructions for her parents to follow-up with the girl's pediatrician 2 days later. ■

REFERENCES:
1. Garzon MC, Schweiger E. Cutis marmorata telangiectatica congenita. *Semin Cutan Med Surg.* 2004:23(2):99-106.
2. Levy R, Lam JM. Cutis marmorata telangiectatica congenita: a mimicker of a common disorder. *CMAJ.* 2011:183(4):E249-E251.
3. Kienast AK, Hoeger PH. Cutis marmorata telangiectatica congenita: a prospective study of 27 cases and review of the literature with proposal of diagnostic criteria. *Clin Exp Dermatol.* 2009;34(3):319-323.