Cutis Marmorata Telangiectatica Congenita

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A 4-month old female infant was referred to our pediatric department for the evaluation of cutaneous alterations present since birth. A physical examination revealed a purple to red reticulated mottled skin stain affecting the left hemiabdomen and back, left buttock, and left lower limb that did not respond to local warming. No other skin alterations were found, namely atrophy. In the initial study, which included an abdominal ultrasound, lower limbs duplex ultrasound, coagulation profile, and ophthalmic and cardiac evaluations, all was normal. A clinical diagnosis of *cutis marmorata telangiectatica congenita* was made. A multidisciplinary monitoring of the infant was maintained over time. At 6 months old, hypotrophy of the left lower limb was noted, with a difference of 1 cm of the limb girth. At 29 months, a leg length discrepancy was noted, with the left limb being 7 mm shorter. At 6 years old, a magnetic resonance angiography of the limbs was performed, showing normal results. Currently, the patient is 9 years old and the cutaneous alterations are more subtle. Hypotrophy of the left lower limb is slightly worse with a difference of 1.5 cm of the limb girth, without improvement with physiotherapy. Dysmetria remain the same, with the left limb being 7 mm smaller, with no orthopedic indication for intervention.

Cutis marmorata telangiectatica congenita is a rare congenital vascular anomaly of unknown etiology, usually present at birth.[1-3] Recent studies have identified *GNA11* mutations in skin biopsies from *cutis marmorata telangiectatica congenita* affected skin areas, which confirms that the disease is possibly a postzygotic mosaic condition.[1]

Associated abnormalities are common and include port-wine stain, body and limb asymmetry, glaucoma, localized *aplasia cutis* congenital and cleft palate. In more than two thirds of the patients, *cutis marmorata telangiectatica congenita* affects the limbs, which may become discrepant in size and shape in approximately half of the cases,[3] more often with hypotrophy, but occasionally hypertrophy.[4] Glaucoma is the most frequently associated ophthalmic anomaly, mostly in patients with *cutis marmorata telangiectatica congenita* on the face.[1] The differential diagnosis includes physiological *cutis marmorata*, Klippel-Trenaunay, Sturge-Weber and Adams-Oliver syndromes, and reticular hemangioma syndrome, among others.[1,4]

Although the prognosis is generally good, with the improvement of skin lesions in over 50% of the patients, a long-term follow up of associated anomalies is needed.[2]

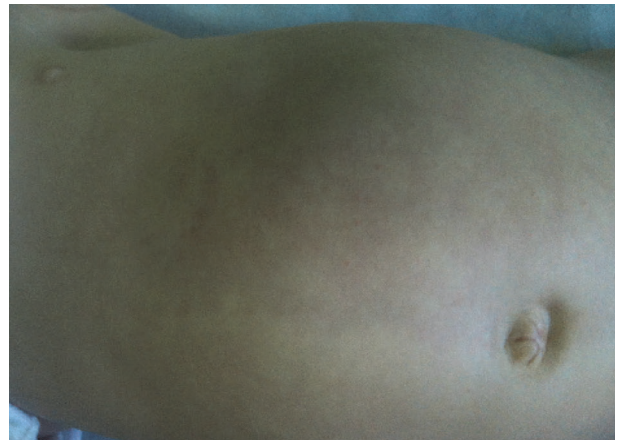

Figure 1. Purple reticulated, mottled skin stain in the left hemiabdomen at 6 months of age.

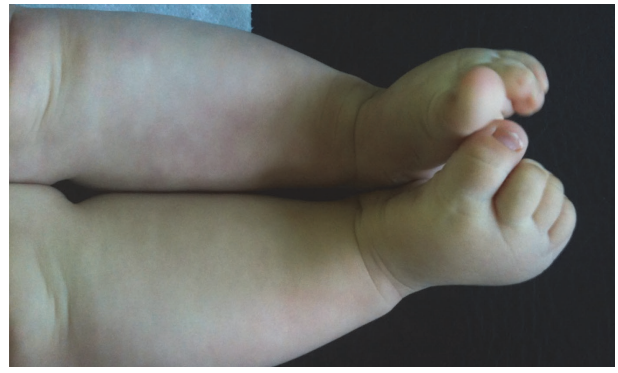

Figure 2. Purple to red reticulated, mottled skin stain in the left lower limb at 6 months of age.

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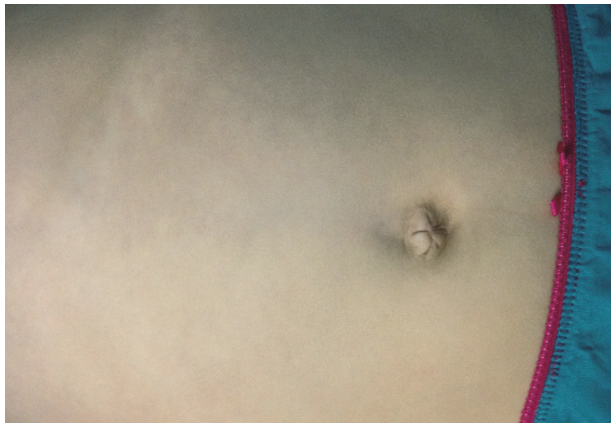

Figure 3. Cutaneous alterations in left hemiabdomen at 33 months of age.

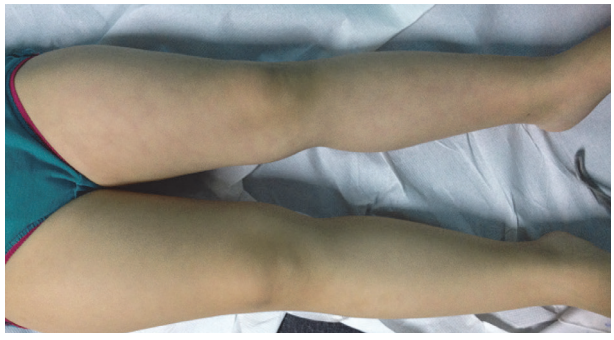

Figure 4. Cutaneous alterations in the left lower limb at 33 months of age.

Keywords: Infant; Leg Length Inequality; Skin Diseases, Vascular/diagnosis; Telangiectasis/complications; Telangiectasis/congenital

WHAT THIS REPORT ADDS

- *Cutis marmorata telangiectatica congenita* is a rare congenital vascular anomaly of unknown etiology, usually present at birth.
- Recent studies have identified *GNA11* mutations in skin biopsies from *cutis marmorata telangiectatica congenita* affected skin areas.
- It is important to consider a vast differential diagnosis, that includes physiological cutis marmorata, Klippel-Trenaunay, Sturge-Weber and Adams-Oliver syndromes, reticular hemangioma syndrome.
- Although the prognosis is generally good, associated abnormalities are common and long-term follow up is needed.

Conflicts of Interest

The authors declare that there were no conflicts of interest in conducting this work.

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Consent for publication was obtained.

Confidentiality of data

The authors declare that they have followed the protocols of their work centre on the publication of patient data.

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