Cutis marmorata telangiectatica congenita: an unusual cause of lower extremity hypoplasia

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Cutis marmorata telangiectatica congenita is a relatively benign, rare cutaneous disorder with various manifestations. A seven-year-old girl, who presented with extremity hypoplasia, had the characteristic reticular, patchy, blue-pink cutaneous lesions, which became more prominent with exposure to cold temperatures. She had 4.8 cm shortening of her right lower extremity, which was also thinner than on the left side. The patient did not have skin atrophy, ulcers, glaucoma or macrocephaly. She is being followed for a future extremity lengthening procedure.

Key words: telangiectasis, cutis marmorata, skin abnormalities, leg length inequality.

Hypoplasia of the extremities may be classified as congenital or acquired, and there are many causative factors. Cutis marmorata telangiectatica congenita (CMTC) is a rare cutaneous syndrome, and limb asymmetry may be one of its manifestations. Here, we present a case of CMTC with limb hypoplasia and a review of the literature.

Case Report

A seven-year-old girl, second child of a nonconsanguineous marriage, was brought with the complaint of limping. She was born with red stains on most parts of her body that became more prominent when the child was exposed to cold. Soon, the family noticed that her right leg was shorter and thinner than the left, and length discrepancy became more apparent as the child grew older. Skin lesions remained unchanged during this period. Her landmarks of development were normal and she was attending primary school as a successful student. None of the family members had similar cutaneous lesions.

On examination, there was port wine stain on the scalp without any skin lesions on the face. The diffuse port wine stain on the right side of the trunk ended at the midline both posteriorly and anteriorly. All over the right lower extremity there were pale, pink-blue, reticular, macular lesions (cutis marmorata), which became more prominent when the child was exposed to cold (Fig. 1). Similar lesions were found on the anterior part of her left cruris. Skin was not atrophic and there were no ulcers.

Her right lower extremity was 4.5 cm shorter than on the left side by measurement from the superior iliac spine to the medial malleolus (Fig. 2). The right thigh circumference, measured 10 cm proximal to the superior pole of the patella was 5.5 cm thinner than on the left side. The right cruris, measured from the midpoint, was 3 cm atrophic compared to the left side. There were no other deformities and muscle testing was normal.

Blood pressure was measured 95/55 mmHg from both arms. A skeletal survey revealed no abnormalities of the spine or extremities. Leg length discrepancy, measured by computerized tomography (CT) scanogram, was 4.8 cm in total, 2.9 cm from the femur and 1.9 cm from the tibia. Bone age determined from the left wrist was 6 years 10 months. Urinalysis and complete blood count were normal. The genetic karyotype was also reported to be normal. Punch biopsy specimen, obtained from the right arm skin, showed dilated venules in the superficial dermis.

The patient was diagnosed as cutis marmorata telangiectatica congenita. She was prescribed a 3 cm shoe-lift for the right side and is being followed for a future extremity-lengthening procedure.

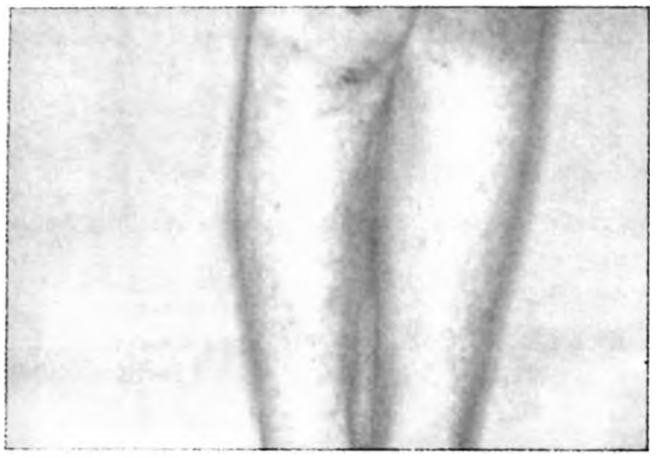

Fig. 1. Closer view of the reticular skin lesions, especially on the right leg.

Fig. 2. Atrophy of the right lower extremity is apparent.

Discussion

Physiological cutis marmorata is frequently observed in newborn babies of Caucasian origin and is accentuated by crying and cold temperatures. Van Lohuizen[1] first described CMTC in 1922, as a localized or generalized, reticulated, blue-violet network, usually present at birth. Various synonyms, such as congenital generalized phlebectasia, nevus vascularis reticularis, and congenital livedo reticularis have also been used in the literature.

Heredity and environmental agents have been proposed in the etiology, but the theory of Happle[4], which suggested a lethal gene surviving by mosaicism for the Klippel-Trenaunay and Sturge-Weber syndromes, better explains the patchy distribution of the lesions and sporadic occurrence of the disease[2-4]. It has also been suggested that Klippel-Trenaunay and Sturge-Weber syndromes and CMTC should be classified as a group of vascular diseases associated with other developmental defects of the mesodermal system during embryonic life[5].

Frequently associated conditions are skin atrophy and ulcerations, nevus flammeus, capillary and cavernous hemangioma, hypertrophy or atrophy of the affected limb, macrocephaly and glaucoma[6,7]. Skin lesions are always patchy and never involve the whole body. When located on the trunk they tend to end at the midline. Skin lesions usually improve, especially during the first two years of life, and this has been attributed to the maturation of the skin, with an increase in the dermis and keratin layers[5,7]. In the current case, the skin lesions were not as dark as observed in photographs of cases presented in the literature, but the family had not noticed any improvement since birth either.

In a review of 35 cases by Devillers et al.[7], 40 percent of the patients had limb asymmetry and in half of these, asymmetry was both in length and circumference. The affected extremity may be atrophic or hypertrophic. In contrast to our patient, who had 4.8 cm shortening, maximum leg length discrepancy in that series was 4 cm. We will follow the patient to observe the changes in discrepancy, and a leg-lengthening procedure will be planned according to the obtained data.

Glaucoma has also been reported, but all of these patients had a skin lesion around the affected eye; routine ophthalmologic evaluation is not recommended[8]. In our case, lesions on the head were restricted to the hairy scalp, and physical examination of the eyes was normal.

To our knowledge, this is the third case of CMTC published from Turkey. One of the previously published cases did not have any anomalies other than the skin lesions[9]. The other case was a newborn with the typical skin lesions and atrial septal defect, but there was no extremity asymmetry[10].

In conclusion, CMTC is a rare and relatively benign condition that should be considered in the differential diagnosis of extremity asymmetry, and associated conditions should be searched.

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