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Congenital cutis marmorata telangiectatica and syndactyly in a preterm. Case report

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ABSTRACT

Cutis marmorata telangiectatica congenita (CMTC) is a rare, commonly benign, congenital, localized or generalized vascular anomaly of unknown aetiology. It is characterized by persistent cutis marmorata, telangiectasia and phlebectasia. Extracutaneous findings may be associated with CMTC in 18.8-70% of the cases. Diagnosis of the disorder is based on the clinical findings. The prognosis is good and improvement is observed within 2 years after birth. Herein, we report a case of a male neonate with CMTC presented on the skin of all his limbs, trunk and face, and an associated anomaly including syndactyly. We present this case because of its rarity.

Key words: *cutis marmorata telangiectatica congenita, newborn, syndactyly.*

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INTRODUCTION

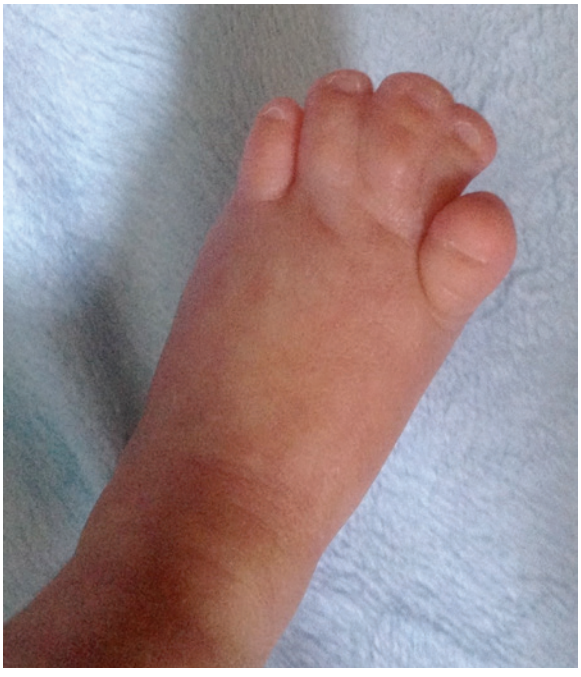
Cutis marmorata telangiectatica congenita (CMTC) is a rare, sporadic congenital vascular malformation characterized by generalized or localized persistent skin changes of cutis marmorata type and telangiectasia.[1] This disorder commonly presents at birth and its aetiology is unknown.[2] Diagnosis of CMTC is mostly made on the basis of clinical features. Associated anomalies such as asymmetry of the body, vascular malformations, skin ulceration or atrophy, glaucoma, psychomotor or mental retardation and limbs anomalies are generally defined.[3] The prognosis is usually good, with minor concomitant anomalies. The skin lesions commonly resolve within the first 2 years of life.[2]

We report a case of CMTC with findings of syndactyly and skin lesions on all the limbs, trunk and face.

CASE REPORT

A male preterm born at 30 gestational weeks was referred to our neonatal intensive care unit shortly after birth because of respiratory distress syndrome requiring surfactant therapy and mechanical ventilation. He was born as second child from consanguineous parents via cesarian section because of the repeat cesarean delivery. The Apgar scores at 1 and 5 minutes were 4 and 6, respectively. His weight was 1080 g (10th percentile), the length was 27 cm (3th percentile) and head circumference was 27 cm (10th-50th percentile). The patient presented respiratory distress and syndactyly in both legs involving second, third and fourth toes (*Figure 1*). Dermatologic examination revealed a fixed,

FIGURE 1. *Syndactyly involving second, third and fourth toes.*



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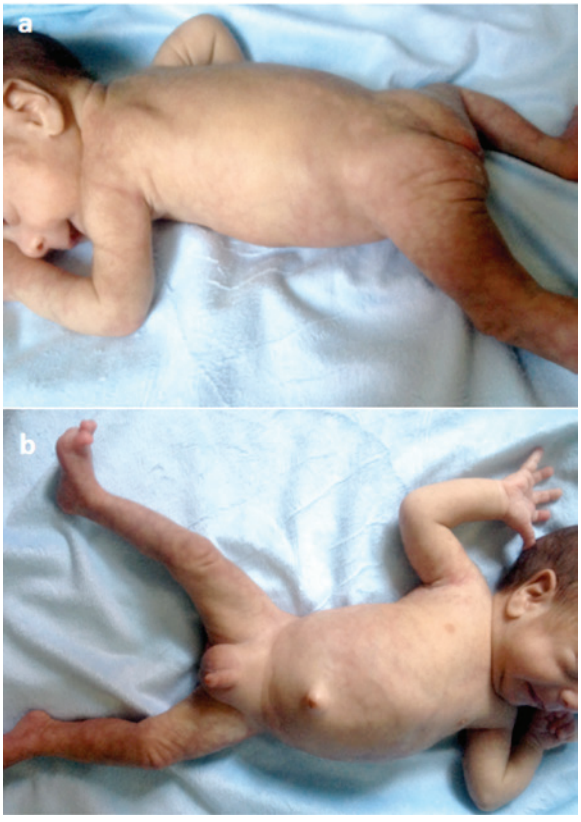
purple, reticulated skin lesion involving upper and lower limbs, trunk and face (left cheek, nose and chin), although scalp, palm, sole or mucous membranes were all normal (*Figure 2*). These lesions were more remarkable over the lower limbs. The body temperature was normal. Skin changes did not disappear with local warming. There was no family history of vascular malformations. No other congenital and neurological anomaly was observed.

A complete blood count, blood gases and chest X-ray were normal. The administration of nasal continuous positive airway pressure was commenced to infant in an incubator. Repeated ultrasound examinations of the head and ophthalmological examinations were normal. Abdomen ultrasound and echocardiographic findings were also unremarkable. Tiroid function tests were all within normal limits. A developmental delay has not been observed during the 1-year follow-up period.

FIGURE 2. *A reticular erythematous patch over the left upper and lower extremities and face (a), lower extremities and abdominal wall (b). There is a symmetrical distribution of dermatological findings and higher intensity of cutis marmorata in the lower extremities.*



Based on these findings and cutaneous pattern, diagnosis of cutis marmorata telangiectatica congenita (CMTC) was made.

DISCUSSION

CMTC is a rare disorder characterized by reticular erythema. It was first described by Van Lohuizen in 1922 and more than 300 cases have been reported to date.[4,5]

CMTC can be either generalized over the whole body or localized to a specific area or limb. In localized form of the CMTC, the lesions are unilateral and if the lesions are in the area of the abdomen, a clear demarcation at the midline is always observed. Trunk, limbs, face and scalp are usually involved in generalized form, but palm, sole or mucous membranes are generally normal. The lower limbs are the most commonly involved areas in the body. The skin lesions mostly present at birth, however, may also be observed up to 3 months to 2 years of age. There is equal representation between sexes.[4,6] The generalized form of CMTC was considered in our patient because of involving large skin surface.

Associated anomalies may be observed in 18.8% to 70% of patients with CMTC. Kienast and Hoeger have reported the presence of associated anomalies in 144 of the 215 patients with CMTC (66.9%). The body asymmetry described as unilateral overgrowth of trunk and/or limbs was the most frequent systemic anomaly (25.1%). Vascular anomalies including port wine stains, angiokeratomas, hemangiomas and Sturge Weber syndrome are the second most common associated anomalies (23.2%). Port wine stains were reported as more common compared with other vascular anomalies.[3,5] Skin atrophy and neurological anomalies have been found in a rate of 5.6% and 5.1%, respectively. Syndactyly may be seen in a rate of 2.8%.[3] Associated skin lesions of CMTC may involve prominent veins, telangiectasias, cutaneous atrophy, ulceration and hyperkeratosis. Cutaneous atrophy and ulceration are the most common skin anomalies in association with CMTC. They can be helpful in the differential diagnosis between congenital and physiologic conditions.[5] Our case had only syndactyly in both legs involving second, third and fourth toes. Although Adams–Oliver syndrome (AOS) is usually associated with CMTC and syndactyly, our infant did not have any other clinical signs for AOS such as aplasia cutis congenita or cardiac malformations.

Macrocephaly has often been reported in

association with CMTC. Because of the high frequency of macrocephaly in these patients, a different subtype was defined named macrocephaly– cutis marmorata telangiectasia congenita (M-CMTC) in 1997. This syndrome includes developmental delay, neonatal hypotonia, segmental overgrowth, syndactyly, asymmetry and connective tissue defects. Unlike CMTC, the prognosis of M-CMTC is significantly worse and the skin lesions are more permanent.[4,7] The head circumference and development of our patient were normal for the age and he had no hypotonia.

Ocular anomalies have been reported in patients with CMCT (3.7%). Glaucoma is the most common concomitant ocular anomaly; is detected frequently in cases with periocular vascular lesions.[2,3] The eye examination for glaucoma was normal in our case.

The diagnosis of the CMTC is mainly based on the clinical features. The skin biopsy is generally not required if the clinical diagnosis is clear. Histopathological examination usually reveals non-specific findings such as dilated capillaries in the deeper dermis, swollen endothelial cells, dilated veins or venous lakes.[8] Many disorders should be excluded before making diagnosis of CMTC.

The main differential diagnosis is physiological cutis marmorata and this condition is resolved by warming the affected area. CMTC resembles cutis marmorata, therefore, in the differential diagnosis physiological cutis marmorata should be considered first. It arises from physiologic dilatation of capillaries and small venules due to low temperatures in healthy infants. Although cutis marmorata resolves with warming of skin, the CMTC persists despite increased temperature.[5] Bockenheimer syndrome is a rare condition presenting as diffuse phlebectasia in early infancy and resulting in progressive venous ectasias that are often large and painful. Adams-Oliver syndrome is an uncommon congenital disorder characterized by aplasia cutis congenita of the scalp and limb defects, and cutis marmorata telangiectatica may be associated with this syndrome.[5,9] Klippel-Trenaunay syndrome is defined with three major features including a wide vascular anomaly, hypertrophy of soft tissue and bone overgrowth, varicose veins and it's more common compared to CMTC.[1,6]

The cutis marmorata may be part of other syndromes as homocystinuria, Down syndrome, Cornelia de Lange syndrome, Divry-Van Bogaert syndrome, but these disorders can be distinguished from children with CMTC by the presence of typical clinical features.[5,6]

In conclusion, CMTC should be suspected in patients with cutis marmoratus in neonatal period. Once diagnosis of CMTC is made, investigations for associated anomalies should be performed. The prognosis of this disease is good. Skin lesions such as cutis marmorata and telangiectatias generally resolve within 3 years of life without any treatment, but the limb asymmetry mostly tends to persist, therefore, the patients should be monitored and seen annually for at least 3 years. ■

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