Cutis Marmorata Telangiectatica Congenita: A Case Report and Review of the Literature

KUTİS MARMORATA TELANJİEKTATİKA KONJENİTA: OLGU SUNUMU VE LİTERATÜRÜN İNCELENMESİ

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Abstract

Cutis marmorata telangiectatica congenita is a rare, benign, sporadic skin lesion that presents itself as a localized or generalized, reticulated, blue-violet, cutaneous vascular network at birth. The prognosis is usually good and the lesions mostly improve within 2 years after birth. There is actually no specific treatment for the lesions but the laser therapy is under investigation. We here present you a case of cutis marmorata telangiectatica congenita diagnosed clinically at birth, with no associated anomalies. The lesion involved mainly the right lower extremity and resolved completely and spontaneously in a 5-month period.

Key Words: Skin; abnormalities; congenital

Özet

Kutis marmorata telanjiektatika konjenita, kendini doğumda lokalize veya jeneralize, retiküle, mavi-mor renkli kutanöz vasküler bir ağ olarak gösteren nadir, benign ve sporadik bir cilt lezyonudur. Prognozu genellikle iyi olup lezyonlar doğumdan sonraki 2 yıl içinde gerilerler. Günümüzde spesifik bir tedavisi olmamakla birlikte lazer tedavisi araştırma altındadır. Biz burada, doğumda tanı konmuş ve eşlik eden herhangi bir anomalinin bulunmadığı bir kutis marmorata telanjiektatika konjenita vakasını sunuyoruz. Cilt lezyonu doğumda esas olarak sağ alt ekstremitede izlenmekteyken 5 aylık süre içinde spontan olarak tamamen kayboldu.

Anahtar Kelimeler: Cilt; anomali; konjenital

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First described by the Dutch pediatrician Van Lohuizen in 1922, cutis marmorata telangiectatica congenita (CMTC) is a rare, benign, sporadic skin lesion that presents itself as a localized or generalized, reticulated, blue-violet, cutaneous vascular network at birth.[1] Lesions commonly occur on the legs, arms and trunk and rarely involve the face and the scalp and are usually associated with skin atrophy and ulcerations.[2] Associated abnormalities such as macrocephaly, body asymmetry, vascular and neurological anomalies, glaucoma and psychomotor retardation occur in most of the patients.

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The prognosis is usually good and the lesions mostly improve within 2 years after birth. There is actually no specific treatment for the lesions but the laser therapy is under investigation. Long term follow-up is indicated in the presence of associated anomalies.

We here present a case of cutis marmorata telangiectatica congenita diagnosed clinically at birth, with no associated anomalies. The lesions involved mainly the right lower extremity and resolved completely and spontaneously in a 5-month period.

Case Report

Antenatal surveillance of the 35-year-old mother, gravida 3, para 1 was performed beginning from the early gestational weeks, in our clinic. They were not consanguineous with her husband. Her first pregnancy in 2001 ended up with a spon-

taneous miscarriage at about seventh gestational week. Her second pregnancy was in 2004, and she delivered a 3200 gram boy at term. This baby had no signs of any important diseases up to now, including cutis marmorata telangiectatica congenita.

Detailed ultrasonographic examination performed at 19th gestational week revealed no anomalies. Lateral ventricle was measured as 0.8 mm and the posterior fossa as 0.52 mm. Both lower and upper extremities were within the normal range and no asymmetry was encountered. The amniocentesis was performed due to advanced maternal age, and revealed normal karyotype.

An elective cesarean section was performed at 38 weeks and 3 days, and a female baby was born with an apgar score of 9/10. Weight and height at birth were at the 50th centile. Just after birth, a reticulated, vascular, blue-violet cutaneous lesion localized to right lower extremity was observed (Figures 1 and 2). There was no size discrepancy between both lower extremities. The remainder of the examination, in particular neurologic and ophtalmologic findings, was normal. Cranial and abdominal ultrasonographic examinations were within the normal range.

The psychomotor development of the baby was within the normal range in the following 5 months, and the lesion resolved completely and spontaneously, without any treatment, within this period.

Discussion

The diagnosis of cutis marmorata telangiectatica congenita is made upon clinical manifestations. Histologic examination is usually not diagnostic and usually demonstrates an increase in the size and number of capillaries, veins and lymphatics.[3] We did not perform any histologic examination based on this knowledge in our case.

Although several hypotheses such as environmental factors, a multifactorial cause or an autosomal dominant inheritance with low or variable penetrance have been proposed, the pathogenesis of CMTC is obscure and most cases are sporadic.

Additional abnormalities are common in the presence of CMTC. They are present in about 50% of the more than 300 cases reported so far. The mostly encountered ones are macrocephaly, body asymmetry, vascular and neurological anomalies, glaucoma and psychomotor retardation.[4-6] In 1997, Moore et al.[7] and Clayton-Smith et al.[8] independently reported 13 and 9 children respectively with a malformation complex consisting of macrocephaly and cutis marmorata telangiectatica congenita. This common association was then described as a unique disorder. In our case, no sign of macro-

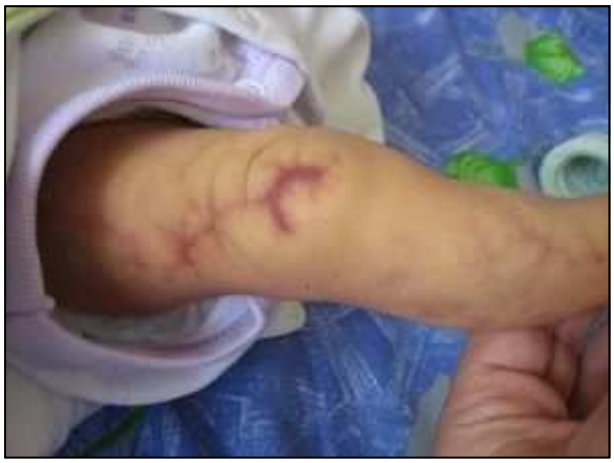

Figure 1.

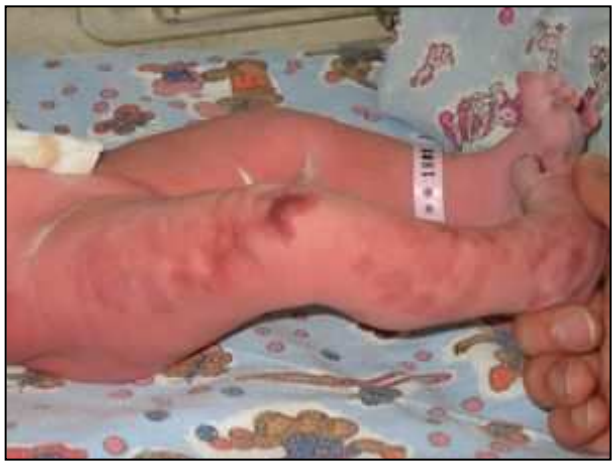

Figure 2.

cephaly was detected either in prenatal ultrasonographic examination or in postpartum cranial ultrasonographic examination.

The lesions are usually associated with a rapid improvement and hardly distinguishable residual spots. This rapid improvement is usually due to the normal accelerated maturation process in infants, resulting in rapid thickening of the epidermis and dermis. Rarely, the lesions remain the same as years pass, a condition which may lead to disturbed psychomotor development.

CMTC is usually a mild condition with good prognosis and therapy is rarely indicated. Laser is the most commonly preferred treatment modality but the reported success rates are not brilliant, probably due to the dilated and deep capillaries.[9]

In conclusion, parents of the newborn with CMTC should be counseled that the lesions will most probably resolve but may rarely remain unchanged, that the severity of the condition depends on the presence of other anomalies and that the treatment is rarely indicated and successful.

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