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Case Report / Olgu Sunumu

A rare vascular lesion of newborn: cutis marmorata telangiectatica congenita

Yenidoğanın nadir vasküler lezyonu: kutis marmorata telenjiektatika konjenita

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Abstract

Cutis marmorata telangiectatica congenita is a rare, benign, sporadic and cutaneous vascular disease. A newborn female baby whose mother was aged 29 years and used propylthiouracil during pregnancy was hospitalized because of varicose lesions on the skin of the lower extremity and on the back, which were present at birth. It was observed that the lesions did not disappear, although appropriate room temperature was provided. The patient was diagnosed as having cutis marmorata telangiectatica congenita and screened for additional anomalies. She had no additional anomalies, and she was discharged and monitored. At the postnatal sixth month, the cutaneous vascular lesions disappeared spontaneously. Cutis marmorata telangiectatica congenita, which is a rare condition, should be kept in mind in the differential diagnosis of physiologic cutis marmoratus, which occurs frequently in the neonatal period.

Keywords: Cutis marmorata telangiectatica congenita, newborn

Öz

Kutis marmorata telenjiektatika konjenita; nadir görülen, selim, sporadik, deriyi tutan vasküler bir hastalıktır. Yirmi dokuz yaşında hipertroidi nedeni ile propiltiyourasil kullanan anneden doğan kız bebek, her iki alt ekstremitede ve sırtta yerleşim gösteren renk değişikliğinin eşlik ettiği variköz lezyonlar nedeniyle yatırıldı, lezyonların uygun ortam sıcaklığı sağlanmasına rağmen düzelmediği saptandı. Kutis marmorata telenjiektatika konjenita tanısı konan olgu; eşlik edebilecek ek anomaliler açısından tarandı; ek anomali saptanmayan olgu taburcu edilerek izleme alındı, postnatal altıncı ayda kutanöz vasküler lezyonların kendiliğinden kaybolduğu görüldü. Yenidoğan döneminde sık olarak karşımıza çıkan fizyolojik kutis marmoratus ayırıcı tanısında, ender görülen bir durum olan kutis marmorata telenjiektatika konjenita da akılda tutulmalıdır.

Anahtar sözcükler: Kutis marmorata telenjiektatika konjenita, yenidoğan

Introduction

The skin's diffuse, transient, and maculated appearance caused by the effect of cold in babies and many adults, is a well-known condition. The excessive vasomotor response of the capillary vessels, which gives the skin a reticular-marmoral appearance, is named cutis marmorata (CM). The congenital form of cutis marmorata was defined by van Lohuizen for the first time in 1922 and named as cutis marmorata telangiectatica congenita (CMTC) (1). Similar to cutis marmorata, CMTC may become prominent with exposure to cold. However, skin abnormalities may not return to normal with warming and may be associated with telangiectasia, phlebectasia, skin atrophy, and ulcers in contrast to CM. These lesions may be limited or diffuse (2).

More than 300 cases have been reported in the world literature up to the present time (3). Some congenital anomalies associated with cutis marmorata telangiectatica congenita occur with rates ranging between 19% and 70% in different series (2, 3). Hemihypertrophy, extremity defects, various ophthalmic anomalies including mainly glau-

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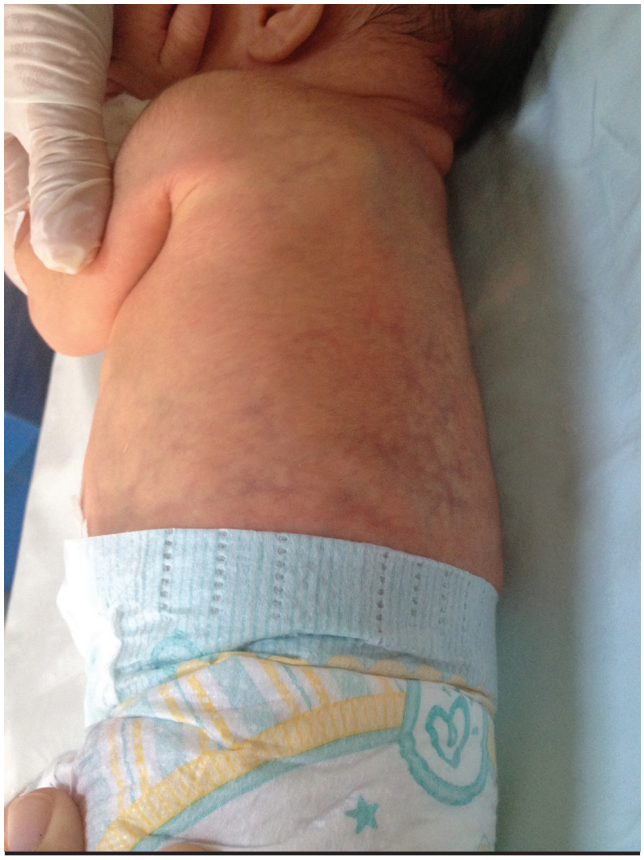

Figure 1. Cutis marmorata telangiectatica congenita localized on the back

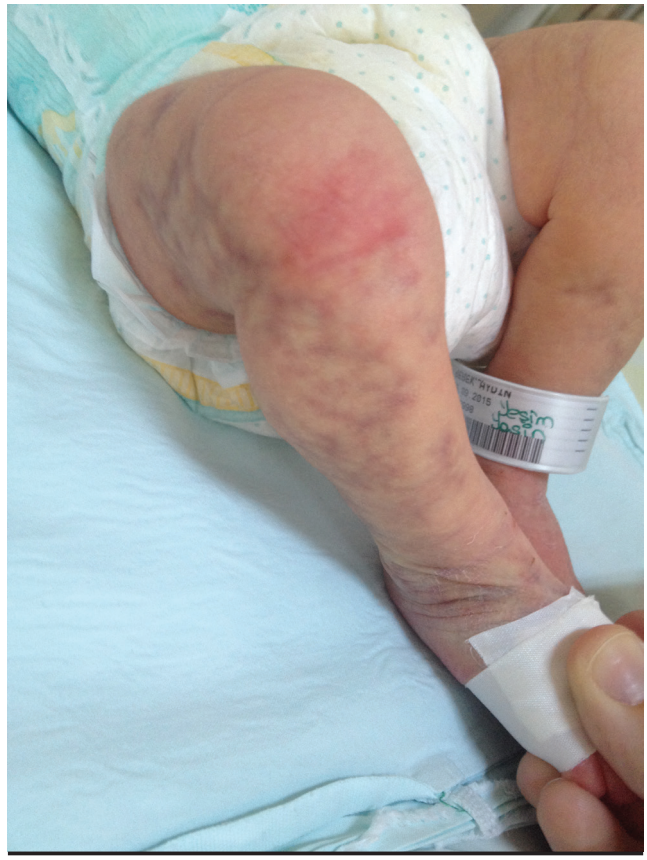

Figure 2. Non-blanchable varicose lesions more diffuse on the right lower extremity accompanied by discoloration

coma, hypospadias, and cardiac anomalies are non-vasculocutaneous abnormalities that are observed frequently in these patients. The neurologic findings described in the literature include seizure and psychomotor retardation.

The etiopathogenesis is not known exactly. This picture, which may affect both sexes equally, is considered to occur sporadically. The diagnosis is made with clinical findings. In most cases, marked improvement is observed in CM and telangiectasias in the first two years. Although the prognosis is good in most patients, long-term follow up is needed in terms of accompanying anomalies (3).

Case

It was learned that the female baby who was born by cesarean section because of cephalopelvic disproportion in the 39th gestational week according to an ultrasonographic examination (in the 39–40th gestational week according to the last menstruation period). The birth weight was 2920 g, and the APGAR score in the first minute was 8, and 10 in the 5th minute. The mother, who had hyperthyroidism, was aged 29 years (gravida 1, parity 0, abortus 0, curettage 0). There was no parental consanguinity. In the antenatal period, the ultrasonographic follow-up examinations were found to be normal. The mother had used propylthiouracil (PTU) throughout pregnancy and antibiotics in the final three months of pregnancy because of a urinary tract infection.

The baby had a body weight of 2920 g (10–25%), a height of 50 cm (50%) and a head circumference of 35 cm (50%). Her vital signs were found to be stable and she did not have oxygen requirement. A physical examination revealed no abnormal findings except for diffuse non-blanchable varicose lesions on both lower extremities, on the back, and on the front side of the trunk (Fig. 1), which were more diffuse especially on the right lower extremity accompanied by discolorations (Fig. 2).

The arterial pulses were palpable in both lower extremities and no temperature difference was found between the lower extremities. The laboratory tests were found to be normal (hemoglobin 16.7 g/dL, hematocrit 49.2 %, white blood cells (WBC) 23,650 /mm^3, platelet count 442,000 /mm^3, prothrombin time 12.6 s, INR 1.11, partial thromboplastin time 35.8 s, and fibrinogen 232 mg/dL). In the peripheral blood smear, platelets were observed to be abundant and in clusters in accordance with the com-

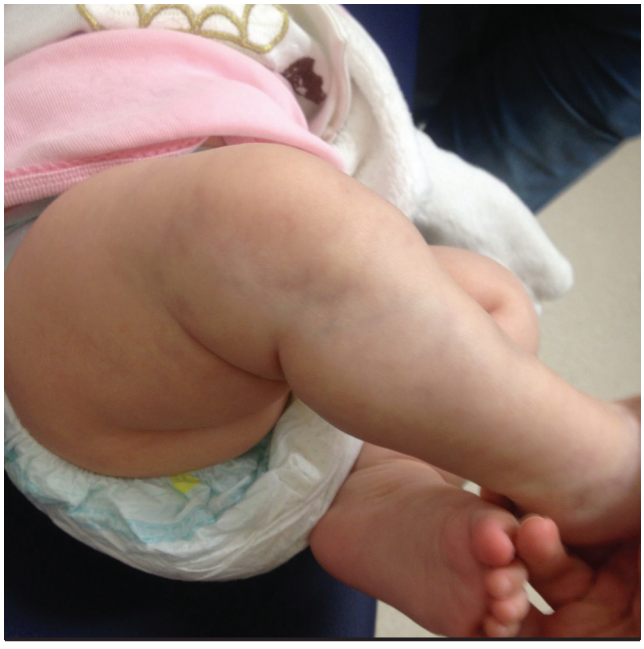

Figure 3. Appearance of the right leg at the sixth month

plete blood count. Thyroid-stimulating hormone (TSH) (1.14 uIU/mL) and thyroxine (fT4) (1.6 ng/dL) were found to be normal in the blood sample obtained on the postnatal 5th day in the infant, whose mother had a medical history of hyperthyroidism. Cranial and abdominal ultrasonographic examinations and extremity Doppler examinations were found to be normal.

In the follow-up, no increase was observed in the lesions, but there was also no improvement, even though an appropriate environmental temperature was provided. A diagnosis of CMTC was made with the present clinical findings. The patient was examined in terms of potential accompanying anomalies; Doppler imaging of the portal vein and renal arteries and veins was performed in terms of vascular pathologies and no pathology was observed. Cranial magnetic resonance imaging and electroencephalography were performed in terms of neurologic involvement and found to be compatible with the age. Whole-body bone radiography was obtained in terms of body asymmetry and evaluated by a radiologist; no pathology was found. Echocardiography was performed in terms of cardiac involvement and was found to be normal. Eye examination was performed in terms of ophthalmic pathology; glaucoma was not found. The patient who was considered to have isolated CMTC, was discharged to be followed up in the outpatient clinic.

The patient was regularly followed up in our neonatology outpatient clinic. Her growth and development was compatible with her age. At the age of six months, the lesions on the back and front side of the trunk improved completely, whereas the lesions on the lower extremities and around the knees continued, though they regressed (Fig. 3). Written informed consent was obtained from the patient's parents.

Discussion

Cutis marmorata, which is defined as diffuse reticular-marmoral appearance with the effect of cold, is a physiologic condition that may be observed in many newborn babies, whereas CMTC is a capillary and/or venous, vascular congenital malformation that does not improve even if an appropriate temperature is provided.

Biopsy was not performed in our patient because biopsy findings in the literature are controversial and not diagnostic. The clinical findings of CMTC include discoloration in the skin, prominence in the capillaries and veins, telangiectasia and skin atrophy and ulceration; the diagnosis is made with clinical findings (4, 5).

Cutis marmorata telangiectatica congenita affect both s equally. One study reported that the lesions were more frequent and showed a limited location in male subjects (5). The lesions showed diffuse localization in our female patient.

The etiopathogenesis is not clear and the cause may be multifactorial. Although most cases are sporadic, autosomal dominant inheritance was reported in some families (6). Teratogens and autosomal dominant inheritance have been considered to be involved in the etiology (7). In the literature, there are no data related to the association of the action of fetal PTU with CMTC; as known, PTU is the preferred as anti-thyroid drug in the treatment of hyperthyroidism in pregnancy, because methimazole (MMI) has high teratogenic activity. However, recently, the number of publications reporting an association between PTU use and vasculitis in adults has gradually increased (8). The CMTC in this patient was considered sporadic because a positive familial history was absent, but it might be the result of fetal PTU use. Although use of propylthiouracil is considered safer compared with MMI in pregnancy, further studies should be conducted to assess the use of PTU in pregnancy and its teratogenic activity.

Cutis marmorata telangiectatica congenita is a self-limiting condition which shows improvement with growth until the age of two years. Treatment is not needed unless an anomaly accompanies. Most frequently, body asymmetry, vascular anomalies, neurologic disorders, eye problems, and syndactyly may accompany (3). Psychomotor retardation has been reported in different publications with a rate raging between 0% and 22% (9). Our assessments

showed that this was an isolated case of CMTC with no accompanying anomalies.

In conclusion, CMTC, which is a rare condition, should also be considered if lesions do not improve despite appropriate environmental temperature in the differential diagnosis of physiological cutis marmoratus, which is observed frequently in the neonatal period, and patients should be evaluated in terms of potential comorbid anomalies. The fetal PTU effect observed in our case may shed light on future cases of CMTC, the etiology of which has not been fully elucidated.

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