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 marmorata, 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 bazık yaşındaki hipertroidi nedeni ile propiltiyourasil kullanan anneden doğan kız bebeğin her iki alt ekstremitede ve sırtta yerleşimi gösteren renk değişikliğinin eşlik ettiği varıköz lezyonlar nedeniyle yatırıldı. Lezyonların uygunsuz ortam sıcaklığı sağlanmasına rağmen düzelmemiş sahaptı. Kutis marmorata telenjiektatika konjenita tanı konan olgu; eşlik edebilecek ek anomaliler açısından tarama edildi; ek anomali saptanmayan olgu taburcu edildik izleme alındı, postnatal altıncı ayda kutanöz vasküler lezyonların kendiliğinden kaybolup girdi. Yenidoğan döneminde ilk olarak karşımıza çıkan fizyolojik kutis marmoratusu ancak tanısal analizde, ender görülen bir durum olan kutis marmorata telenjiektatika konjenita da akılda tutulmalıdır.

Anahtar sözcükler: Kutis marmorata telenjiektatika konjenita, yeni doğan

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-
coma, hypospadias, and cardiac anomalies are non-vascular cutaneous 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 discoloration (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 /mm³, platelet count 442,000 /mm³, 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-
complete 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 sexes 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.

Informed Consent: Written informed consent was obtained from the patients’ parents.

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References
1. Van Lohuizen CHJ. Über eine seltene angeborne Hautanomalie (cutis marmorata telangiectatica congenita). Acta Derm Venereol 1922; 3: 202−11.
2. Gerritsen MJ, Steijlen PM, Brunner HG, Rieu P. Cutis marmorata telangiectatica congenita: report of 18 cases. Br J Dermatol 2000; 142: 366−9.
3. Amitai DB, Fichman S, Merlob P, Morad Y, Lapidoth M, Metzker A. Cutis marmorata telangiectatica congenita: clinical findings in 85 patients. Pediatr Dermatol 2000;17: 100−4.
4. Fujita M, Darmstadt GL, Dinulos JG. Cutis marmorata telangiectatica congenita with hemangiomatous histopathologic features. J Am Acad Dermatol 2003; 48: 950−4.
5. Baykal C. Genetik hastalıklar. Dermatoloji Atlası. Argos; İstanbul; 2004.p.565.
6. Kurczynski TW. Hereditary cutis marmorata telangiectatica congenita. Pediatrics 1982;70:52−3.
7. Bhargava P, Kuldeep CM, Mathur NK. Cutis marmorata telangiectatica congenita with multiple congenital anomalies. Further clues for a teratogenic cause. Dermatology 1998;196: 368−70.
8. Criado PR, Grizzo Peres Martins AC, Gaviolli CF, Alavi A. Propylthiouracil-Induced Vasculitis With Antineutrophil Cytoplasmic Antibody. Int J Low Extrem Wounds 2015;14:187−91.
9. Garzon MC, Schweiger E. Cutis marmorata telangiectatica congenita. Semin Cutan Med Surg 2004;23:99−106.