A Case of Cutis Marmorata Telangiectatica Congenita

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Cutis marmorata telangiectatica congenita(CMTC) is a rare congenital cutaneous vascular anomaly. The major skin findings are persistent, fixed cutis marmorata, telangiectasia, and phlebectasia. In approximately 50 % of patients, the disorder tends to be associated with various congenital anomalies. We describe in this report a 4-month-old male infant with CMTC. The patient had red or violet marbled patches, telangiectases, and atrophy on the right arm present at birth. During the follow up period of 12 months, there was no change in the cutis marmorata pattern and telangiectases, whereas the atrophy has been less prominent than at the initial visit.(Ann Dermatol 8:(1)43~46, 1996).

Key Words : Cutis marmorata telangiectatica congenita, Atrophy

Cutis marmorata telangiectatica congenita (CMTC) is a rare congenital cutaneous condition first described by Van Lohuizen[1] in 1922. By this time more than one hundred cases of CMTC were reported in the literatures under various names, including congenital generalized phlebectasia[2,3], congenital generalized telangiectasia, nevus vascularis reticularis[4], and congenital livedo reticularis[5,6]. The major skin findings are persistent, fixed cutis marmorata, telangiectasia, and phlebectasia[7]. Approximately 50 % of reported cases have been associated with various congenital anomalies including the atrophy or hypertrophy of an involved extremity, the most frequently associated anomaly[8]. We describe in this report a 4-month-old male infant with CMTC.

REPORT OF A CASE

A 4-month-old male had red or violet marbled patches, telangiectases, and atrophy on the right upper arm. The lesions had been present since birth. The patient was born by cesarean section at full term. His birth weight was 4.0 kg. The mother was 27-year-old primiparity. There was no history of medications taken during pregnancy or of maternal illness. The family history was unremarkable.

Physical examination revealed reddish to violet marbled or mottled patches, which blanched with pressure, on the right upper extremity. The pattern appeared to be reticulated. The telangiectasia and subcutaneous atrophy were present in some areas(Fig. 1). The right arm was not smaller in circumference than the left. The blood pressure of the right arm was lower than the left(right 105/70 mmHg; left 120/77 mmHg).

Routine laboratory findings including complete blood cell count, urine analysis, liver function test, glucose, blood urea nitrogen, creatinine, VDRL, hepatitis B antigen and antibody, antinuclear antibody, cryoglobulin, and cold agglutinin were within normal limits or negative. Two-dimensional echocardiography and electrocardiography showed normal findings. The X-ray findings of the chest, skull, abdomen, pelvis, both upper and lower extremities were normal. The MRI of both upper extremities revealed a slight decrease in thickness of subcutaneous fat in the right side(Fig. 2). There

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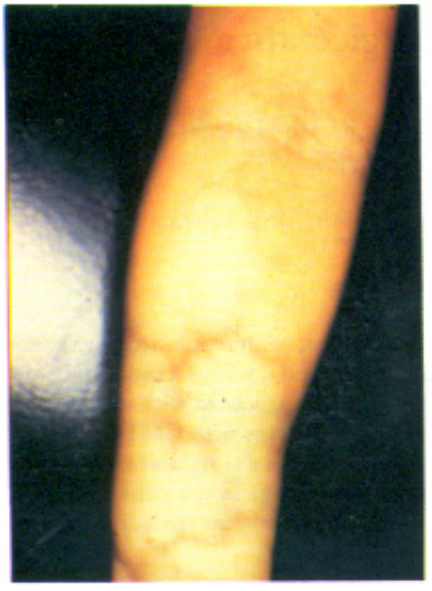

Fig. 1. Reddish to violet reticulated patches with telangiectasia and atrophy on the right upper extremity.

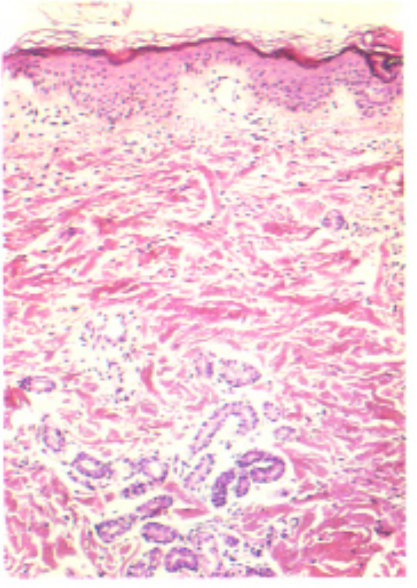

Fig. 3. Skin biopsy shows vascular ectasis in the upper and mid dermis (H & E stain, ×40).

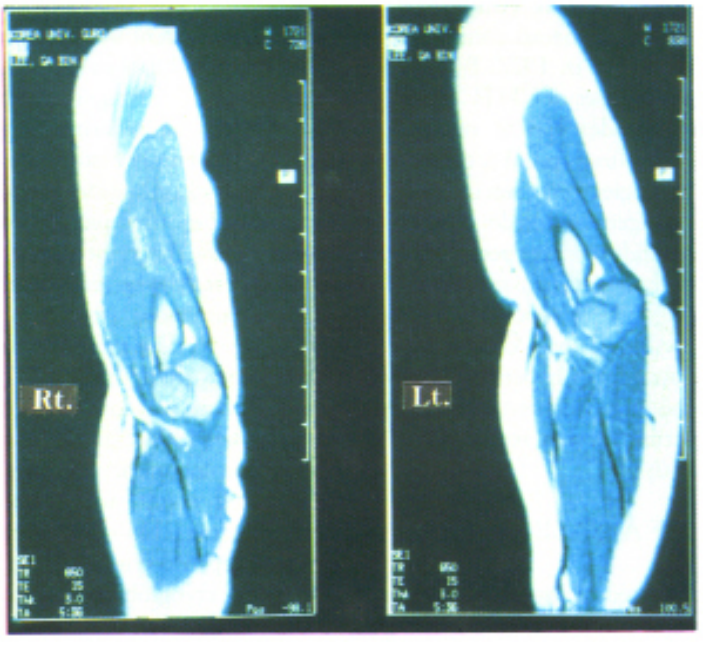

Fig. 2. MRI showing a slight decrease in thickness of subcutaneous fat in the right upper extremity.

was no difference in blood flow measured by color doppler flower metry(Acouson) between the right and left arm.

An excision biopsy was performed. The biopsy specimen showed the vascular ectasis in the upper and mid dermis(Fig. 3).

At the age of 12 months, the patient's skin was re-examined and no changes had occurred in the

reticulated(cutis marmorata) pattern and telangiectases. The atrophy was less prominent than at the initial visit. No ulcer or erosion were present.

DISCUSSION

Cutis marmorata telangiectatica congenita (CMTC) is a rare vascular anomaly. Van Lohuizen[1] used the term "CMTC" to describe the persistent cutis marmorata pattern, spider-like telangiectasia, phlebectasia, and ulceration in an infant girl.

It is characterized by persistent cutis marmorata, telangiectasia, and phlebectasia[7,8]. The reticulated cutaneous vascular network of blue-violet color can be localized or generalized and usually is present at birth. The area of skin enclosed by the reticulated pattern may be normal or erythematous. The red-purple hue of the lesions may be enhanced by a decrease in ambient temperature, by vigorous movement, or by crying. However the marbled pattern is always readily discernible even when the infant is warm and at rest, unlike the physiologic cutis marmorata. Telangiectases are often noted in associated with the reticulated bands[7,8]. Atrophy of the skin and subcutis in the reticulated bands of CMTC has been reported by many authors. Ulceration also can be a prominent finding and usually heal without incident, but scarring is not unexpected[3,4,9-13]. Although Way et al.[14] and Powell et al.[8] noted a predominance of female children and South et al.[3] reported a preponderance of male patients in their series, CMTC appears to affect both sexes equally[15]. CMTC is usually present at birth or shortly thereafter, but may not develope until two years after birth[8,16]. Our patient was born with an cutis marmorata pattern, comprising a reticulated network of dilated veins.

The histopathologic findings of CMTC are nondiagnostic. Van Lohuizen[1] described dilated capillaries in all layers of the dermis and subcutaneous tissue, as well as dilated veins and venous lakes. Andreev and Pramatarov[12] observed swelling of endothelial cells in addition to capillary dilation. A sparse perivascular lymphocytic infiltration also has been reported[13,17,18]. Ultrastructural studies performed by Lynch and Zelickson[17] revealed increased numbers of perithelial cells. Way et al.[14] described an atypical vascular endothelium with a discontinuous basal lamina.

The cause of CMTC is unknown. Although the disorder appears to be sporadic in most instances, a genetic basis has been proposed by some authors. Andreev and Pramatarov[12] described the cases of two sisters and suggested that the condition might be inherited as a dominant trait with low penetrance. Kurczynski[15] reported a case with CMTC whose father and paternal grandfather had similar lesions. Way et al.[14] proposed a multifactorial cause for CMTC. Rogers and Poyzer[19] suggested the dysmorphogenic environmental agent as the cause of CMTC reported in four cases. No teratogenic factor has been postulated. There was no apparent hereditary pattern associated with the disorder in our case.

Petrozzi et al.[20] reported the case of CMTC combined with Sturge-Weber syndrome and patent ductus arteriosus in 1970. Subsequently, Way et al.[14] reviewed the literature on CMTC and noted associated abnormalities in more than 50% of the 41 reported cases. Hypoplasia or hyperplasia of an affected or unaffected limb was the most common defect. Other associated findings included capillary hemangioma, asymmetric skull, micrognathia, triangular face, scaphoid scapular, dystrophic teeth, high-arched palate, syndactyly of toes, short stature, short fingers, and acral cyanosis[3,9,14]. Lee et al.[18] reported a case of CMTC with mental retardation, cleft palate, simian lines on the palm, diffuse demineralization of the bones, and weakness of the long extensor muscles of both thumbs. Additional cases have been described in association with aplasia cutis congenita[3], spina bifida[8], neonatal ascites[10], and macrocephaly[9]. In our case, atrophy of the skin and subcutis was clinically obvious, being most notable over the areas of maximal vascular dilatation, This finding was more clarified in MRI.

CMTC must be differentiated from several other vascular disorders, including Bockenheimer's syndrome(diffuse genuine phlebectasia) and Klippel-Trenaunay-Weber syndrome. Bockenheimer's syndrome has its onset in childhood and is characterized by the gradual development of multiple large and often painful venous ectasis, usually affecting a single limb[3,13]. The patients with Klippel-Trenaunay-Weber syndrome have a vascular lesion(most often a port wine stain), venous varicosities, and hypertrophy of soft tissue and sometimes bones of involved parts[21]. Even more confusing are the cases of patients with light-colored port wine

stains that have a reticulated appearance. In these patients it is often difficult to be sure of the correct diagnosis, and long-term follow-up may offer the only avenue to certainty. Homocystinuria, Down syndrome, and de Lange's syndrome are congenital disorders associated with a pronounced physiologic cutis marmorata that must be considered in differential diagnosis of CMTC, but usually other clinical features will differentiate between them[7,8]. Although neonatal lupus erythematosus may also present a livedo reticularis, appropriate serologic tests will clarify the diagnosis[22].

There is no treatment but the argon or dye laser therapy may be helpful in decreasing the vascular lesions. Many reports have indicated that the lesions tend to improve with time[7,15,16]. However some patients may continue to have lesions throughout their lives. This should be considered when counseling parents of children who have this disorder[8].

In conclusion, a 4-month-old male was seen with characteristic features of CMTC associated with an atrophy of involved extremity. We could not determine familial, environmental, or in utero factors responsible for his disorder, which has improved slightly during the follow up period of 12 months.

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