

Cutis marmorata telangiectatica congenita

Vinoth N. Ponnurangam, Venkataraman Paramasivam

Department of Pediatrics,
Sri Ramachandra Medical
College, Chennai,
Tamil Nadu, India

ABSTRACT

Cutis marmorata telangiectatica congenita (CMTC) is an uncommon, congenital, vascular malformation, characterised by a fixed, generalised or localised, reticulate erythematous rash. We report an eighty day old male child who presented with the above lesion involving the left upper and lower limbs with underlying atrophic changes.

Key words: Atrophy, cutis marmorata telangiectatica congenita, vascular malformation

INTRODUCTION

Cutis marmorata telangiectatica congenita is an uncommon, congenital, vascular malformation composed of capillary and venous sized vessels. It presents with a fixed reticulate erythema that is similar to physiologic cutis marmorata, however skin lesions do not resolve with warming of the skin surface. It may have a localized or generalized pattern on the skin. When localized, the lesion tends to remain unilateral and do not cross the midline, and it may be sharply demarcated. Many associated anomalies have been reported to occur in individuals with cutis marmorata telangiectatica congenita. Most commonly reported are limb asymmetry and the coexistence of other vascular birthmarks. We report here an eighty day old infant with CMTC involving left upper and lower limbs with underlying atrophic changes seen over the left extremities.

CASE REPORT

An eighty-day-old female child, first born to nonconsanguineous parents, presented with reduced growth of left upper and lower limbs. Birth and developmental history were normal. There was no family history of vascular malformations. Physical examination revealed a fixed, purple, reticulated skin lesion involving left upper and lower limbs [Figure 1] with underlying atrophic changes seen over the left extremities [Figure 2]. Anthropometric measurements were within the normal limits except for the reduced left upper and lower limb girths. Systemic examination

did not reveal any abnormality. With the above findings, clinically we made the diagnosis as cutis marmorata telangiectatica congenita. Parents were reassured about the benign nature of the skin lesion and the need for periodic follow up on outpatient basis.

DISCUSSION

Cutis marmorata telangiectatica congenita (CMTC) is an uncommon, congenital, vascular malformation first described by Van Lohuizen in 1922. The presence of a reticular erythema is pathognomic of CMTC. This erythema may be either generalized or localized to a specific area or limb. Localized lesions remain unilateral and sharply demarcated, and do not cross the midline. The color of the lesion varies from deep violet to red. The skin lesion usually manifests at birth but may also appear up to 3 months to 2 years of age. There is no sexual predilection. However, many studies reported that this condition affects girls more than boys, but their number is minute and the difference is not statistically significant.

The pathogenesis of this condition is not very clear and the etiology may be multifactorial. A genetic etiology has been suggested; however it is generally considered a sporadic condition.^[1] Lethal gene hypothesis has been suggested by some authors for the patchy distribution of the lesion and its sporadic occurrence.

CMTC closely resembles physiological cutis marmorata. Dermatological findings in CMTC include prominent veins, telangiectasias,

Access this article online

Website: www.idoj.in

DOI: 10.4103/2229-5178.126042

Quick Response Code:



Address for correspondence:

Dr. Vinoth N.
Ponnurangam,
Department
of Pediatrics,
Sri Ramachandra
Medical College, Porur,
Chennai, Tamil Nadu,
India.
E-mail: vindoc1977@gmail.com



Figure 1: Fixed, purple, reticulated skin lesion seen in the left upper and lower limbs

cutaneous atrophy, ulceration, and hyperkeratosis. The presence of atrophy, skin ulceration, and sharp demarcation of a localized lesion that does not disappear after warming all distinguish CMTC from physiological cutis marmorata.^[1,2]

Congenital anomalies have been reported in 20-80% of patients with CMTC.^[1,3] There is some controversy regarding these figures as some studies have included patients with definite genetic disorders, including macrocephaly and capillary malformation and there may be overestimation of these rates due to coincidental associated findings.^[4] However, no data exists on the association between the size and severity of the lesion in CMTC and its relationship with dermatological and systemic anomalies.^[1]

Asymmetry, especially of the limbs, is the most common systemic abnormality seen in CMTC. The affected limb may be either atrophied or hypertrophied, and overlying cutaneous atrophy is frequently present. Syndactyly, tendinitis stenans, hip dysplasia, club foot, and cleft palate are other skeletal defects reported to be associated with CMTC.^[1] A detailed musculoskeletal examination is necessary to identify any of the above-mentioned skeletal defects. Our patient had reduced left upper limb and lower limb girth due to underlying atrophy.

The most common ocular association of CMTC is glaucoma, which has been detected in children with facial lesions.^[1] Vascular anomalies like port wine stains, angiokeratomas, hemangiomas, and Sturge Weber syndrome have been reported to be associated with CMTC. Of these anomalies, port wine stains were reported frequently.^[5] Our patient did not have any of the above-mentioned abnormalities.

Macrocephaly has been reported frequently in association with CMTC. This has led to the designation of a distinct subtype called macrocephaly-CMTC in 1997.^[6] Other features of this

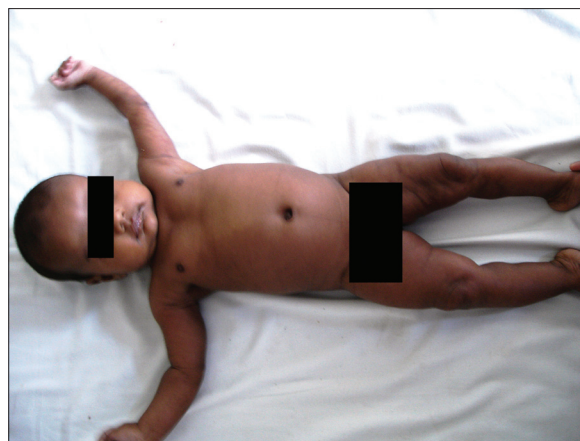


Figure 2: Atrophic changes seen over the left upper and lower limbs

syndrome are developmental delay, neonatal hypotonia, segmental overgrowth, syndactyly, asymmetry, and connective tissue defects. More recently, this entity has been renamed as macrocephaly-capillary malformation, because the skin findings are more consistent with capillary malformation rather than true CMTC.^[6] Physicians should assess the development of the child and also measure the head circumference to rule out macrocephaly-capillary malformation. Our patient had normal development and head circumference for the age.

CMTC is a clinical diagnosis and histopathological examination of skin is usually not diagnostic. Several disorders should be ruled out before making a diagnosis of CMTC. This can generally be accomplished by a thorough screening for associated anomalies. The most common lesion presenting with reticulate erythema is cutis marmorata, which may be physiological and it disappears during early childhood. Persistent reticulate skin lesions are seen in children with Down's syndrome, de-Lange syndrome, homocystinuria, and Divry-van Bogaert syndrome.^[1] Bockenheimer's syndrome is characterized by diffuse phlebectasia presenting in early infancy and progressing to large painful venous ectasias. Adams Oliver syndrome presents as generalized CMTC with associated cardiac, limb and scalp abnormalities. Klippel-Trenaunay syndrome consists of vascular malformation, venectasia, and soft tissue hypertrophy. Capillary malformations such as reticular port wine stains, remain a very close differential diagnosis to CMTC. Both remain unilateral and localized without crossing midline. CMTC tends to fade in color as age progresses, often within the first 2 years of life. Port wine stains have more distinct borders that do not fade and are not associated with underlying atrophy. Neonatal lupus erythematosus and other vasculitides may also enter the differential diagnosis of reticulate telangiectatic atrophy, but congenital lupus is bilateral and symmetrical, with a greater frequency of facial lesions.^[2] In our case, antinuclear antibodies and anti-Ro/SSA antibodies were negative in both mother and child.

Kienast and Hoeger have recently proposed diagnostic criteria for CMTC. However, the validity of these criteria has not been established.^[3] Once diagnosed as CMTC and other associated anomalies ruled out, these children should be seen annually for a minimum period of 3 years. On follow-up, limb girth and length measurements are to be monitored along with a thorough physical examination to rule out associated anomalies.^[4] Although the vascular lesion often lightens with time, the limb asymmetry tends to persist.^[1] CMTC has a good prognosis with improvement of skin lesions occurring during the first 2 years of life. This has been attributed to the maturation of the skin along with the growth of the child.^[7]

REFERENCES

1. Garzon MC, Schweiger E. Cutis marmorata telangiectatica congenita. *Semin Cutan Med Surg* 2004;23:99-106.
2. Moss C, Shahidullah H: Naevi and other Developmental Defects. In: Rook other Devk of Dermatology. 8th ed. Wiley: Blackwell; 2010. p. 18.1-7.
3. Kienast AK, Hoeger PH. Cutis marmorata telangiectatica congenita: A prospective study of 27 cases and review of the literature with proposal of diagnostic criteria. *Cli Exp Dermatol* 2009;34:319-23.
4. Levy R, Lam JM. Cutis marmorata telangiectatica congenita: A mimicker of a common disorder. *CMAJ* 2011;183:E249-51.
5. 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.
6. Gonzalez ME, Burk CJ, Barbouth DS, Connelly EA. Macrocephaly-capillary malformation: A report of three cases and review of literature. *Pediatr Dermatol* 2009;26:342-6.
7. Chatterjee R, Dey S. Cutis marmorata telangiectatica congenita with skin ulcerations in a newborn. *Indian J Dermatol* 2009;54:375-7.

Cite this article as: Ponnurangam VN, Paramasivam V. Cutis marmorata telangiectatica congenita. *Indian Dermatol Online J* 2014;5:80-2.

Source of Support: Nil, **Conflict of Interest:** None declared.