Cutis Marmorata Telangiectatica Congenita with Limb Asymmetry: A Case Report

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ABSTRACT

Cutis marmorata telangiectatica congenita (CMTC) is a congenital capillary malformation having a characteristic appearance. Although benign, CMTC tends to be associated with many local and systemic anomalies. Body asymmetry and vascular anomalies are the commonest associations. To the authors' knowledge, there have been about 300 cases of CMTC reported in the world literature to date. The authors report a case of a female neonate who presented with persistent reticulate bluish erythema over the right lumbosacral area extending to the right lower limb and gradually developed limb asymmetry. She also had an ulcerated haemangioma on the sole of her right foot. Diagnosis was made clinically, and the proposed diagnostic criteria were fulfilled. The patient was followed prospectively through the early years to monitor skin lesions, associated anomalies, and psychomotor development.

Key Words: *Cutis marmorata telangiectatica congenita, Limb asymmetry, Capillary malformation*.

How to cite this article: Arooba Z, Aman S. Cutis Marmorata Telangiectatica Congenita with Limb Asymmetry: A Case Report. *JCPSP Case Rep* 2025; **3**:358-360.

INTRODUCTION

Cutis marmorata telangiectatica congenita (CMTC) is an exceedingly rare congenital vascular malformation with only a few hundred cases reported globally to date.[1,2] It is characterised by mesh-like, marbled erythema present at or shortly after birth that does not disappear with warmth. It may be segmental or generalised, but most frequently, it has unilateral distribution localised over the lower limbs. In some cases, epidermal atrophy and even ulceration of the lesion can be seen.[1-4] CMTC has been reported to be associated with local and systemic anomalies in 18-80% of cases. The commonest association is with body asymmetry that affects limbs, trunk, or face.[1-3] Vascular lesions frequently accompany, either within or far from the area of CTMC and include port-wine stain, haemangiomas, nevus anaemicus, and angiokeratomas.[2,4] Other reported associations include glaucoma, macrocephaly, seizures, developmental delay, Mongolian spot, genitourinary anomalies, and cardiovascular defects.[1-3] Diagnosis of CMTC is clinical, and a set of diagnostic criteria has been designed. The major criteria include presence of a reticular erythema since birth, unresponsiveness to local heating, and no venectasia in affected sites at one year of age, while minor criteria include erythema fading within two years, presence of a port-wine stain outside the CMTC area, telangiectasia, ulceration, and skin atrophy within a CMTC-affected site.[2]

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Received: March 07, 2025; Revised: May 15, 2025; Accepted: July 08, 2025
DOI: https://doi.org/10.29271/jcpspcr.2025.358

CASE REPORT

A 3-month female child presented with a fixed network of reddish blue discolouration over her back and her right lower limb since birth. Parents of the patient reported darkening of the discoloured area on forceful crying or in a hot environment. There was an ulcerated nodule over the sole of the right foot that was initially a small patch at birth and increased in size progressively, became ulcerated, and used to bleed profusely on minor trauma. She was the second child born to a non-consanguineous marriage and was delivered vaginally at full term after an uneventful pregnancy.

Examination revealed a marbled, reddish-blue reticular erythema over the right half of the back of the trunk and right lower limb, circumferentially extending from the inguinal area till the sole of the foot (Figure 1). The erythema did not improve on local warming and was only partially blanchable. Few telangiectasias were appreciated within this network; however, there was no skin atrophy. The nodule on the sole of the foot was located within the discoloured network and was ulcerated with yellowish exudate (Figure 2). A large Mongolian spot could be seen around the midline over the sacral area. There were no varicosities or lymphangiomas on the affected limb. Her ophthalmological, neurological, and cardiac evaluations were normal. Colour Doppler of the reticulate area revealed small dilated capillaries, but no varicosities were seen. The nodule over the sole of the foot was detected as a highly vascular hamartoma (haemangioma).

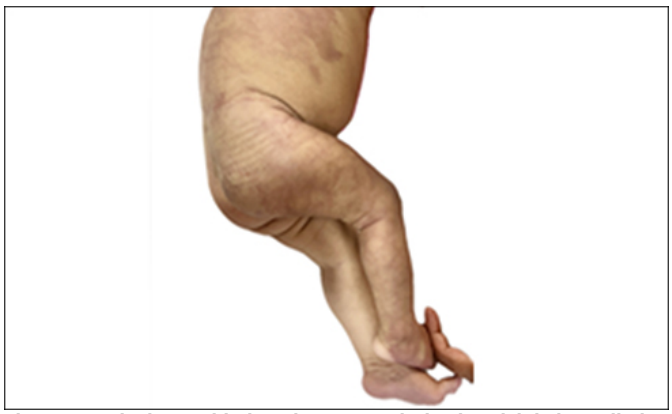

Figure 1: Reticular marbled erythema over the back and right lower limb.

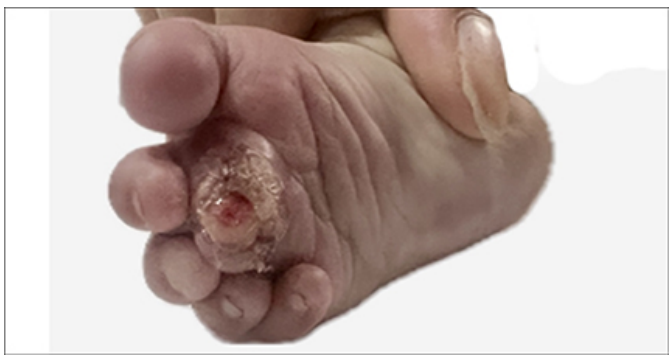

Figure 2: Ulcerated haemangioma over the sole of the right foot.

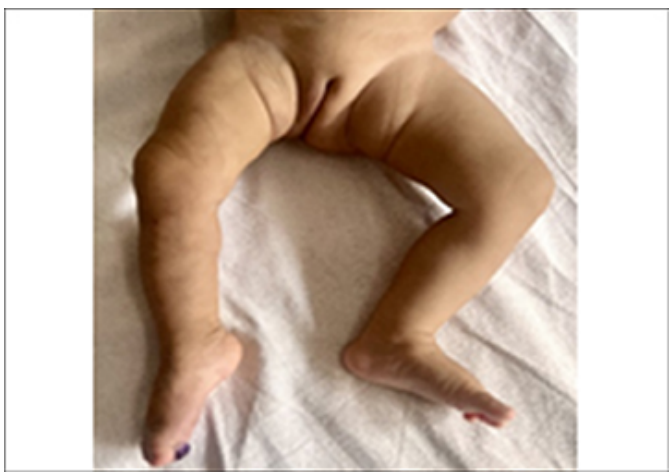

Figure 3: The affected limb (right) showing reduced length and girth compared to the healthy one at two years of age.

Based on the clinical presentation consistent with diagnostic criteria by Kienast *et al*., diagnosis of the CMTC was established. Haemangioma on the foot was initially treated with antibiotics, and then a topical beta-blocker, timolol, was prescribed. The lesion remained unchanged in size. At 9 months of age, oral propranolol was started at a dose of 0.5 mg/kg/day in two divided doses and resulted in a remarkable reduction in size within two months. The vascular network over the leg gradually faded out, with total disappearance in some areas. The limbs developed gradual asymmetry, which parents noticed when the child was around one year, with the affected limb showing reduced limb girth and length compared to the other limb. At two years of the age, the right lower limb was 1.8 cm shorter in length as compared to the left one (Figure 3). The child's psychomotor development was monitored, and no delay or disturbance was found.

DISCUSSION

The authors report an infant with CMTC, which was followed prospectively till 2 years of age. CMTC is diagnosed by its distinctive cutaneous manifestations. Histopathology of the cutaneous lesion is non-specific and does not give any additional information.[4,5] It is distinguished from physiological cutis marmorata, which is a transient response to cold in neonates and vanishes after rewarming.[5,6] Klippel-Trenauny syndrome was easy to rule out because of the absence of venectasia, lymphatic malformation, and soft tissue hypertrophy.[2] Another differential is reticular haemangioma, which is characterised by the presence of a vascular tumour coexisting with ulcerations, and urogenital and sacral anomalies.[2] Port-wine stain can also rarely present in a reticulate pattern, making it difficult to differentiate from CMTC, as both may be unilateral with respect to the midline. However, contrary to port-wine stains, CMTC tends to fade down often within the first two years, which helps in an optimal retrospective diagnosis. Besides, port-wine stain has well-defined, geographical borders and is not associated with atrophy.[6] Adams-Oliver syndrome may present as generalised CMTC accompanied by aplasia cutis, limb defects, cardiac malformations, and anomalies of the scalp and cranium.[2,6]

A review of existing literature reveals several other case reports of the CMTC with similar presentations of unilateral limb involvement and associated vascular anomalies, further supporting the findings in this patient.[4-6]

Once the clinical diagnosis of the CMTC is made, the patient must be carefully evaluated for associated anomalies, ideally in a multidisciplinary setup with a dermatologist, paediatrician, ophthalmologist, and eventually, an orthopaedic surgeon for limb length discrepancy.[1,5] The cutaneous lesion generally fades in the early years of life, but limb discrepancy tends to persist. Regular follow-up should be continued during early childhood to monitor the outcomes of associated anomalies.[2,4,5]

In conclusion, this case highlights the characteristic presentation of the CMTC, associated with significant limb asymmetry and a haemangioma. It underscores the importance of long-term follow-up to monitor associated anomalies despite the potential fading of the cutaneous findings.

PATIENT'S CONSENT:

Informed consent was taken from the parents of the patient for publication of this case.

COMPETING INTEREST:

The authors declared no conflict of interest.

AUTHORS' CONTRIBUTION:

ZA: Conception, design of the study, case identification, writing of the manuscript, and accountability for the work's accuracy and integrity.

SA: Analysis, proofreading, revision of the manuscript, and providing specialised knowledge related to the case.
Both authors approved the final version of the manuscript to be published.

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