CASE REPORT

Cutis marmorata telangiectatica congenita and major lower limb asymmetry

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Accepted 27 November 2017

SUMMARY

A 39-week-old male newborn presented at birth with atrophic erythematous and purpuric skin lesions, in a typical right-sided segmental distribution. Lesions were persistent and unaffected by rewarming in the postpartum period. Postnatal echocardiogram showed a predominance of the right cavities and an upper atrial septal defect. Cerebral and abdominal ultrasound were normal along with ophthalmological examination. On follow-up, lower limbs asymmetry was noted. The right lower limb was shorter in length and had a smaller diameter. At 6 months, the right lower limb was 1.5 cm shorter than the left, most likely related to nutritive vessels malformations. The discrepancy was even more pronounced at the age of 9 months. This leg-length asymmetry can lead to severe functional limitations in the future.

BACKGROUND

Cutis marmorata telangiectatica congenita (CMTC) is a rare congenital vascular anomaly, that presents with localised or generalised persistent telangiectasia in a reticulate erythema pattern, associated with skin atrophy and ulceration.¹⁻³ Even though the aetiology is unknown, the typical segmental distribution with midline separation suggests that CMTC may be caused by genetic mosaicism.³ Nevertheless, other studies advocate an autosomal dominant inheritance pattern with incomplete penetrance and also a teratogenic cause.^{1,4}

The existence of gender differences remains controversial, not only due to the reduced number of patients but also due to the inexistence of statistically significant differences.⁵ Although described as rare, the incidence of CMTC is probably underestimated, as it is established based on clinical features.⁶ The vascular malformation, that primarily involves

the subcutaneous vascular system, is manifested by purplish erythematous macules located on extremities, upper body or face. Facial involvement might occur, but mucous membranes are usually spared.³ Body asymmetry is the most common anomaly associated with CMTC, mainly trunk and lower limbs, and the histopathological examination typically reveals multiple dilated capillaries and veins within the dermis.^{2,6}

The authors present the case of a neonate with CMTC and the associated features of this syndrome.

CASE PRESENTATION

A 39-week-old male newborn presented at birth generalised linear purple reticulate lesions of the limbs, gluteal region and upper body. He was the first-born child of non-consanguineous parents. Prenatal echocardiogram revealed a predominance of the right cavities and a tortuous aortic cross. Apgar scores were 9 and 10 at 1 and 5 min, respectively. The newborn was admitted to a Neonatal Intensive Care Unit shortly after birth, for clinical surveillance. On physical examination, he had fixed and confluent erythematous purpuric lesions with marked atrophy, predominantly on the right knee and dorsum. The largest skin area affected had up to 15 mm in diameter, and a characteristic segmental distribution could be seen on the right side of the body (figures 1 and 2). Even though there was a well-delimited area in the midline of the anterior face of the upper body, the newborn also presented purpuric lesions in the left hand (figure 3) and left malleolus.

Lesions were persistent and unaffected by rewarming in the postpartum period. These findings lead us to the diagnosis of CMTC. There was no family history of vascular malformations. Postnatal echocardiogram revealed a predominance of

Figure 1 (A) Purpura reticulate lesions and (B) typical segmental distribution on the right half body.

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To cite: Amaral J, Peixoto S, Mimoso G, et al. *BMJ Case Rep* Published Online First: [please include Day Month Year]. doi:10.1136/bcr-2017-222269

Figure 2 Atrophic lesion on the right knee.

the right cavities and an atrial septal defect in the upper third of the septum. Both cerebral and abdominal ultrasound were normal. There were no findings on ophthalmological examination. He was discharged on the sixth day of life with a scheduled follow-up appointment.

OUTCOME AND FOLLOW-UP

Regular follow-up was arranged to allow early detection of other features of the disease. There was gradual improvement of the purpuric reticulate lesions, with a reduction in extension and pigmentation. At 6 months, lesions were mainly confined to the lower limbs and less evident in the upper limbs and hands (figures 4 and 5). The cephalic perimeter remained stable at the 50th percentile. Follow-up echocardiogram at 6 months revealed normalisation of the previous alterations and no impairment of the cardiac function.

Despite the rather benign course of CMTC, the lower limbs were particularly affected in our case. For this reason, orthopaedic outpatient visits were arranged. At 6 months, the

Figure 3 Atrophic lesion on the left hand.

Figure 4 Atrophic scar in the right limb at 6 months of age.

asymmetry was particularly significant with an expressive difference of 1.5 cm in extension from the right to the left lower limb, being the right lower limb shorter than the left. Both thigh and leg diameters were also substantially asymmetric, with a 3 cm difference in thigh's diameter and 2.5 cm comparing between legs (figure 5). The hips were clinically and ultrasonographically normal. At 9 months, the difference between both members, both in size and circumference, was even more significant (figure 6).

Even though there was no facial involvement, glaucoma and retinal disease were excluded by ophthalmological observation.

Neurodevelopmental assessment was adequate throughout the entire follow-up.

DISCUSSION

CMTC was first described by Van Lohuizen in 1922, and since then, more than 300 cases have been reported.^{2 3 7} Associated

Figure 5 Asymmetric lesions in lower limbs at 6 months of age, with a difference of 3 cm in thigh's diameter and 2.5 cm comparing between legs.

Figure 6 Leg-length discrepancy at 9 months of age: right leg length 23.5 cm, left leg length 25.2 cm.

anomalies are present in 18.8% to 70% of patients diagnosed with CMTC, generally predominant on one half-body.²

In accordance with other reported cases, the diagnosis of our patient was reinforced by his right-sided cutaneous lesions and atrophy, with a clear demarcation at the midline. These signs, associated with the purpura's persistence even with rewarming, were essential to the differential diagnosis between CMTC and other conditions.^{4–6,8} The main differential diagnosis is the reticulated plane angioma, in which atrophy and necrosis are absent.⁵

The cephalic perimeter was regularly evaluated and remained stable at the 50th percentile. This finding leads us to exclude macrocephaly CMTC, a recently recognised syndrome.¹² The recognition of this syndrome is of utmost importance due to the risk of severe neurological abnormalities and life-threatening complications, such as heart anomalies and developmental delay.^{2,6} Neurodevelopmental evaluation was also normal which, along with the absence of cutis aplasia, allows the exclusion of conditions, such as homocystinuria, trisomy 21 and Cornelia de Lange syndrome.^{5,9}

At 6-month follow-up, the reticulate pattern had significantly improved, but lower members had a clear asymmetry, most likely related to the nutritive vessels malformations.³ At 9 months, the asymmetry in size and circumference was even more pronounced.

Some previous findings suggested that Sturge-Weber syndrome, Klippel-Trenaunay syndrome and CMTC form a group of vascular diseases, resulting from defects of the mesodermal system during embryonic life.³ Therefore, they are considered to be differential diagnosis. As stated before, the infant does not have any other abnormalities, such as neurological impairment nor port-wine stain like in Sturge-Weber syndrome,⁸ nor bone and soft tissue hypertrophy in the affected side, as seen in Klippel-Trenaunay syndrome,^{6,8,10} nor diffuse superficial phlebotonic characteristic of Bockenheimer syndrome, nor aplasia cutis nor syndactyly associated with D'Adams-Oliver syndrome.^{3,11} Ocular anomalies have been reported in patients with CMTC (3.7%).¹¹ Glaucoma is the most common concomitant ocular anomaly detected.¹¹ Ocular involvement was not found in this case.

The prognosis is usually good, with minor concomitant anomalies. In 50% of the babies, the lesions disappear during the first 2 years of life.⁵ However, the residual scarring lesions will remain unchanged throughout the patient's life. In cases of significant aesthetic impact, the pulsed laser treatment may be considered, even though its efficacy may be limited.⁵

The lower limbs asymmetry is the main concern in our case, since the subsequent growth is affected and it will be proportional to the initial degree of limb asymmetry.¹⁰ This patient will probably have severe consequences of his body asymmetry in the future. He currently has a right lower limb 1.5 cm shorter than the left.

There are studies that describe limb-length discrepancy in half of the patients with CMTC.⁷ Limb-length discrepancies greater than 1 cm can cause not only alterations in biomechanics but also severe functional limitation as a consequence of scoliosis, back and lower extremity joint pain, pelvic tilt and abnormal gait.¹² These comorbidities will secondarily affect the patient's neurodevelopment. The limb-length discrepancies usually require lengthening procedures. Radiographs should be performed at the age of 10 years (girls) or 12 years (boys) or if the leg-length discrepancy is ≥ 2 cm.⁷ Therefore, follow-up by orthopaedic surgery is recommended.

Learning points

- ▶ Cutis marmorata telangiectatica congenita (CMTC) presents with persistent erythematous purpuric lesions that are unaffected by rewarming in the postpartum period.
- ▶ Lesions of CMTC typically have a segmental distribution.
- ▶ Despite its benign course, it can be associated with marked cutaneous atrophy and lower limbs asymmetry.
- ▶ Body asymmetry is the most common associated anomaly, and most likely results from malformations regarding the nutritive vessels.
- ▶ Leg-length discrepancy greater than 1 cm can lead to severe functional limitation in the future.

Contributors JA and SP were responsible for the bibliographic research, drafted the initial manuscript and approved the final manuscript to be submitted. GM and DP reviewed and revised the manuscript and approved the final manuscript to be submitted. All authors approved the final manuscript to be submitted and agreed to be accountable for all aspects of the work.

Competing interests None declared.

Patient consent Guardian consent obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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