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CASE REPORT

Cutis marmorata telangiectasic congenita: a case report

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Abstract

This case report refers to the diagnosis of cutis marmorata congenita in a male newborn, after observing violaceous skin lesions shortly after birth. It is a pathology with few cases described in the literature, which may have associated abnormalities, the main one being limb asymmetry. The condition requires multidisciplinary management and patient follow-up with the aim of tracking possible associated malformations.

INTRODUCTION

Cutis Marmorata Telangiectasic Congenita (CMTC) is a rare, benign vascular malformation that predominantly affects capillaries and venules, initially described by Van Lohuizen in 1922¹. Its main differential diagnosis is physiological cutis marmorata, a very common condition that can contribute to underdiagnoses of this condition². With just over 300 cases described in the literature, it is a pathology of unknown cause, present at birth or shortly after, associated with different anomalies in about 42% of cases, the most frequent being limb asymmetry^{3,4}. Here we present the case of an 18-month-old male patient with vascular lesions present at birth, clinically diagnosed with CMTC and undergoing multidisciplinary follow-up in the city of Lavras, Minas Gerais.

CASE REPORT

Newborn PLSG, male, full-term (38 weeks), born by cesarean section, with Apgar 10/10, weight: 2645g, height: 47cm and Head circumference: 31cm. Mother, 21 years old, 3 pregnancies, 1 vaginal delivery and 1 cesarean section, had prenatal care of late insertion, with only one ultrasound performed, without urinalysis, report of vaginosis and pyoderma during pregnancy. Admission tests: O positive, fasting glucose: 84mg/ dL, post dextrosol glycemia: 95mg/ dL, non-reactive serology. She denied alcoholism and smoking but reported paternal drug addiction.

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During the newborn's (NB) primary examination, violet macules were observed in the right hemi body, in the course of blood vessels, sparing the face, associated with tegumentary hypotrophy in some regions. The case was discussed with the medical team remotely, and the hypothesis of Cutis Marmorata Congenital Telangiectatic Disease (CMTC). The NB evolved well, being discharged after 48 hours of clinical observation. Referred to high-risk follow-up at the State Center for Specialized Care, for *follow-up*, lesion control, ophthalmologic evaluation, observation of neurological development and need for orthopedic control.

During follow-up, he presented alterations related to the diagnosis of CMTC, such as limb asymmetry (right thigh with a circumference 0.5 cm smaller than the contralateral side), weakness in the right side and vicious posture with averted gaze to the left. He presented improvement of these symptoms after physical therapy. The telangiectatic lesions were similar at birth at the beginning of the follow-up and, after 18 months, they evolved to a lighter appearance than initially, associated with an improvement in the signs of skin hypotrophy. There was no ophthalmological and neurological follow-up. Age-appropriate neuropsychomotor development. A new echocardiogram was performed, which showed no changes. Currently, it follows a monthly observation routine.

Table 1 summarizes the main tests performed during patient follow-up. Figure 1 shows the skin lesions exhibited by the NB at birth and after 18 months of life.

DISCUSSION

CMTC is a congenital vascular anomaly, classified as a simple vascular malformation, more specifically a capillary malformation⁵⁻⁹. As it encompasses capillaries and venules, it is a slow-flowing vascular lesion which main cutaneous manifestation is a reticulated marbled erythema very similar to cutis marmorata, however persistent, not disappearing with heating, despite from turning white to digital pressure^{5,10}.

In addition, these children may have skin atrophy in the affected areas, hyperkeratosis, ulceration, and body asymmetry. The condition is benign, but there are some frequently associated anomalies such as congenital glaucoma, central nervous system involvement, presence of other vascular birthmarks and limb asymmetry. Syndactyly, stenosing tendinitis, hip dysplasia, clubfoot, and cleft palate are also described^{5,6,9-11}. In our patient, the presence of prominent veins, telangiectasia, cutaneous atrophy, and asymmetry of limbs were observed.

Despite bearing the term "congenital" in the name, CMTC can manifest up to 2 years after birth. Cutaneous manifestations can be generalized or localized, when they tend to remain unilateral, without crossing the midline^{5,10}. In this form, the most affected limbs are the legs and trunk. The generalized form usually spares mucous membranes, palms of the hands and soles of the feet¹².

It is controversial in the literature whether there is a higher prevalence in females, considering that there is a small number of reported cases^{6,9}. The etiology is still unknown, and most cases occur sporadically, although some rare cases have been described in members of the same family¹³. A genetic theory proposed in 2002 suggested that CMTC may be a lethal autosomal mutation, with patients surviving through mosaicism or incomplete penetrance. There is also the possibility that the occurrence is associated with teratogens^{5,13}. A *GNA11* mutation was identified in skin biopsies from skin areas affected by CMTC, and a study published in 2015 identified the *ARL61P6* gene as a possible gene for a syndromic form of CMTC associated with developmental delay, transient ischemic attacks, and cerebrovascular malformations^{8,11}.

The diagnosis is clinical and, in 2009, diagnostic criteria were proposed by Kienast and Hoeger, which have not been validated. According to the authors, the presence of all three major criteria and two of the five minor criteria would be necessary. The criteria are described below: congenital reticulate erythema (marbled), absence of venectasia in the affected region at 1 year of age or older, and unresponsiveness to local heating. Minor criteria are disappearance of erythema within 2 years, telangiectasia within the CMTC-affected area, port-wine stain outside the CMTC-affected areas, ulceration and skin atrophy^{5,6}. It is noteworthy that many cases have been reported in the literature without proof of the major criterion "absence of venectasia", as is the case of our patient, who presented the other two major criteria associated with the presence of telangiectasia and cutaneous atrophy. Histopathology has no role in the diagnosis of CMTC, as its findings are nonspecific⁵.

The prognosis of the condition is good, and it is closely associated with the presence of extracutaneous alterations, since the dermatological lesions regress with age^{7,10}. Some proposed strategies for the treatment of CMTC include avoidance of cold, vasodilators, aspirin, pentoxifylline, and photodynamic therapies⁶. These patients need annual follow-up for at least 3 years, paying special attention to measurements of circumference and length of the limbs, since asymmetry usually persists¹⁰. Furthermore, these patients may require multidisciplinary follow-up with other specialties such as orthopedics, neurosurgery, ophthalmology, and surgery^{5,6}.

CONCLUSION

CMTC is an essentially benign condition, but it can cause significant morbidity, being associated with other malformations that can impair the quality of life of affected patients. Its diagnosis is essentially clinical, based mainly on a thorough physical examination. The varied involvement of the disease calls for a longitudinal and multidisciplinary approach, with periodic follow-up of patients.

Table 1: Complementary PLSG tests performed during hospitalization and follow-up in the first 18 months of life.

little eye test	Without changes
little heart test	96/96% - normal
US full abdomen	Without changes
Infectious screening (24 hours of life)	Hb 17.9, GL 14100, PLQ 266000, PCR 5.7 (RODWELL HEMATOLOGICAL SCORE 0).
Transfontanellar ultrasound (USTF)	Without changes
Echocardiogram at birth	atrial septal defect
Echocardiogram 18 months	Without changes
little ear test	Normal
Foot test	Normal

Source: Autores, 2022.



Figure 1. A: Comparison between the lesions presented by PLSG at birth; B. After 18 months of life.

Source: Authors, 2022.

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