Cutis Marmorata Telangiectatica Congenita in a Preterm Female Newborn: Case Report and Review of the Literature

Cutis Marmorata Telangiectatica Congenita in una neonata pretermine: Descrizione del caso e revisione della letteratura

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Key words: Cutis marmorata telangiectatica congenita, congenital vascular skin disorder, benign vascular anomaly, congenital glaucoma

Riassunto

La Cutis Marmorata Telangiectatica Congenita (CMTC) è una condizione rara, di solito presente alla nascita, caratterizzata da cutis marmorata persistente, localizzata o generalizzata, telangectasia e flebectasia.
Riportiamo il caso di una neonata pretermine, terzogenita di genitori caucasici non consanguinei, con CMTC alla nascita che mostrava le tipiche lesioni cutanee e glaucoma congenito monolaterale.
La patogenesi di questo disordine non è nota e la causa è probabilmente multifattoriale.
Come fattori etiologici sono stati ipotizzati l'esposizione ad agenti teratogeni ed un meccanismo di trasmissione autosomico dominante a penetranza incompleta.
La prognosi, nei casi senza complicazioni, è buona.

Abstract

Cutis Marmorata Telangiectatica Congenita (CMTC) is a rare, sporadic condition usually present at birth characterized by localized or generalized persistent cutis marmorata, telangiectasia and phlebectasia.
We report a preterm female newborn, the third child of non-related caucasian parents, with CMTC at birth who showed typical cutaneous features and monolateral congenital glaucoma.
The pathogenesis of this disorder is unknown and the cause is probably multifactorial.
Teratogens and autosomal dominant mode of inheritance with incomplete penetrance have been considered as etiological factors.
Prognosis, in uncomplicated cases, is good.

Case report

M.R. is a female newborn spontaneously delivered at 33 weeks and 5 days of gestational age (GA) for maternal preeclampsia. The baby needed no resuscitation at birth. Apgar score was 9-9 at 1' and 5' minutes of life respectively. Family medical history was noncontributory. During first hours of life, for incoming of respiratory distress syndrome, exogenous porcine surfactant was administered and mechanical ventilation performed for about 24 hours. Hypoglycemia was corrected by infusion of 10% glucose solution through umbilical venous catheter. Since birth marbled bluish and deep purple reticulated skin lesions involving the whole body were noted. The lesions were prominent over the trunk and face but more pronounced over the limbs and became more visible with crying and exposure to room temperature (Fig.1). Clinical characteristics and persistence of cutis marmorata prompted the diagnosis of Cutis Marmorata Telangiectatica Congenita (CMTC). The baby had normal face, head circumference 32.5 cm (90° percentile), birth weight 2,688 Kg (>97° percentile), length 48 cm (>97° percentile), without other vascular anomalies or asymmetry of limbs growth.
Neurological examination showed mild hypotonia of the trunk, poor spontaneous motor activity, autonomic instability and immaturity of the organization of behavioral states, while ophthalmological evaluation detected unilateral congenital glaucoma in the left eye.
Cardiac examination, audiologic screening, Magnetic Resonance Imaging (MRI) and abdominal ultrasound were normal.
At four month of life, under general anesthesia, she underwent surgical trabeculotomy ab externo in the left eye with uneventful postoperative course.
In the meantime, asymmetry in growth of the lower limbs became increasingly evident with circumference of right thigh and leg grea-

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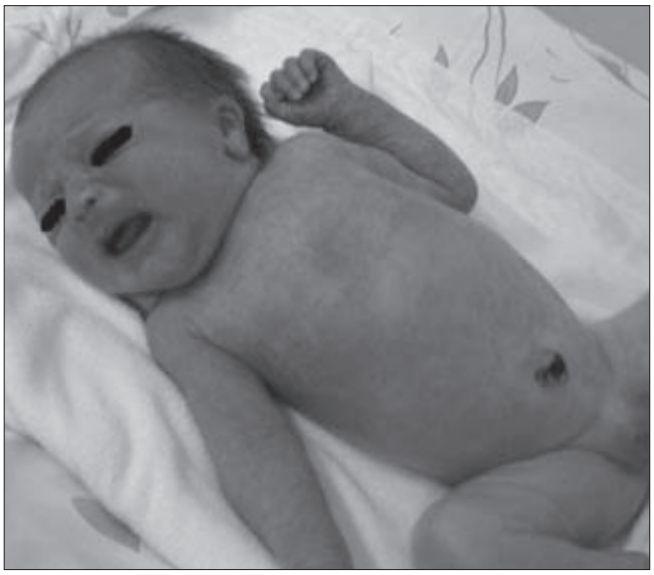

Figure 1.
persistent cutis marmorata with facial erythema

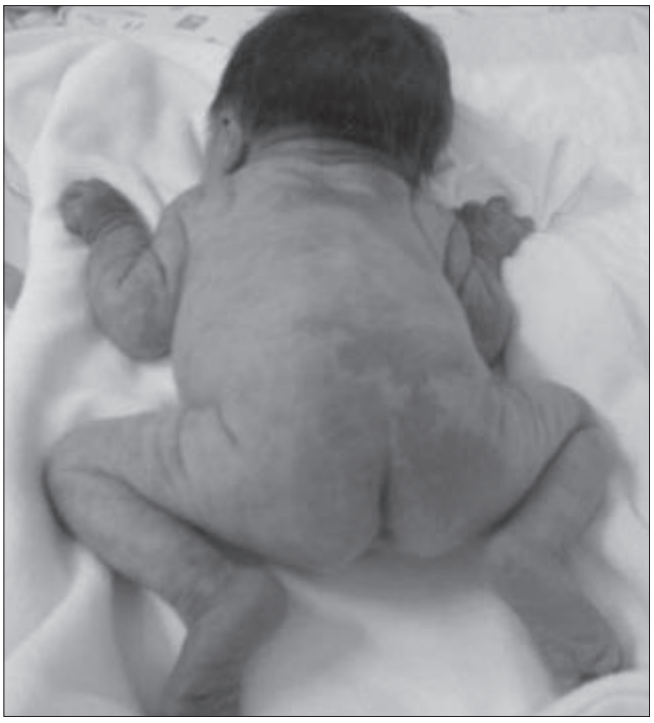

Figure 2.
erythema and telangiectasias scattered on cervicothoracic and lumbosacral spine

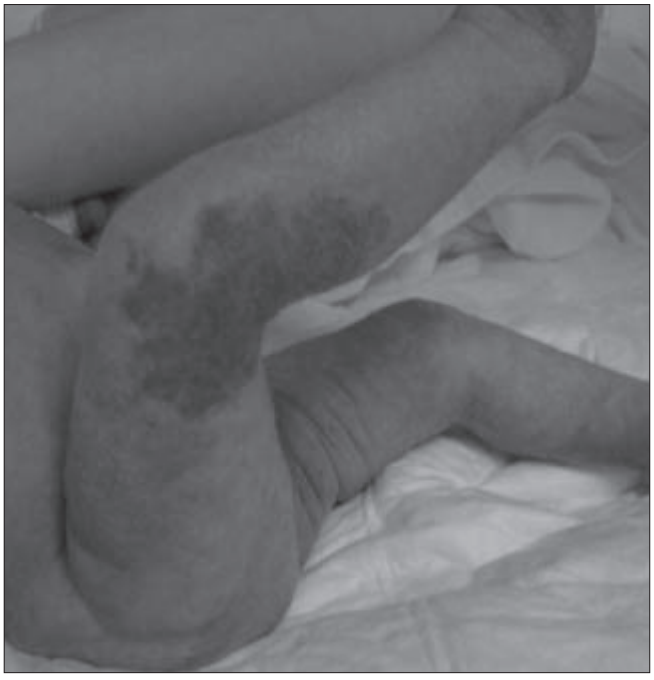

Figure 3.
teleangiectasic - erythematous patch with small areas of atrophy

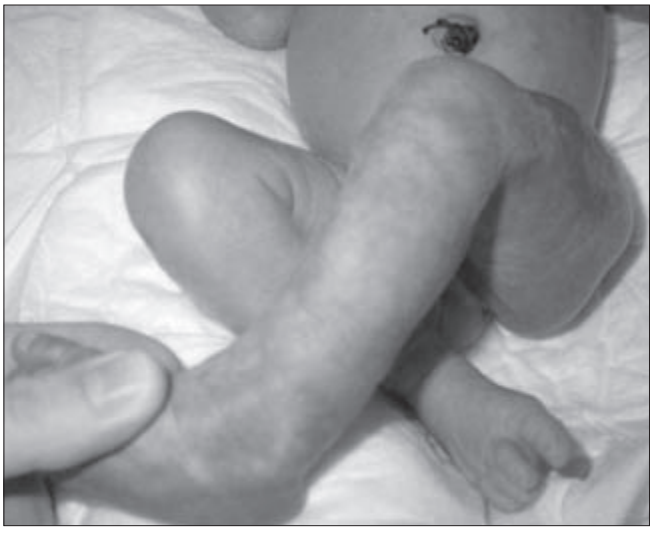

Figure 4.
diffuse livedo reticularis

ter than the left (22.5 and 17 cm versus 21 and 15 cm respectively). No significant alterations of vascular axis of the lower limbs were detected by Color Doppler ultrasound.

Neurological follow-up at 3 months of corrected age showed delayed psychomotor development: disorganization of motor and autonomic nervous system, poor spontaneous motor activity with chaotic movements of legs, hyperextension of the neck, nonspecific response to sound stimulation, frequent loss of visual contact and unsteady visual tracking in the horizontal plane. Fidgety movements were also abnormal, non armonic, slow or almost absent.

According to neurological assessment, a specific rehabilitation program was started.

At the present time, the baby continues her multidisciplinary follow-up.

Discussion

First described in 1922 by Van Lohuizen[1], CMTC is an uncommon, sporadic, congenital cutaneous disorder usually present at

birth. However, sometimes the lesions develop later, from 3 months to 2 years of age.

Clinical features include persistent cutis marmorata, telangiectasia and phlebectasia, generalized or most commonly localized, especially at the lower limbs, followed by the trunk and face.[2-6] Occasionally ulceration and atrophy of the involved skin may be present.[5,7,8] The reticulated mottling frequently becomes more prominent in cold environment but doesn't disappear with warming.[9] Our baby, as described, showed more pronounced lesions at the left leg but skin ulcerations were not present.

The frequency of CMTC is unknown and about 300 cases have been described.[10,11]

Sex-related prevalence of CMTC is controversial. Female seems to be more affected than male and may tend to have generalized disease, however the number of published cases is small and the differences are not statistically significant.[2] The case reported is a female one with generalized distribution of the lesions.

The pathogenesis of this disorder remains unclear and the cause is probably multifactorial. Most cases occur sporadically, although rare cases have a familiar recurrence.

Teratogens and autosomal dominant mode of inheritance with incomplete penetrance have been considered as etiological factors.[12] Some Authors suggest that lethal gene hypothesis (i.e., the lethal dominant gene survival by means of mosaicism) best explains the patchy distribution of the lesion in many cases. [13]

Autosomal dominant transmission was described in a family whose parent showed limited involvement compared to offsprings.[14] In 1979 Andreev reported another CMTC familial case with lesions present at birth in two sisters, one of which developed hypertension at age of 16 years.[15] Later, Toriello described typical cutaneous signs in mother and son, although the son presented some features suggestive of Adam–Oliver Syndrome (AOS).[16]

An association with elevated maternal serum human chorionic gonadotrophin level and fetal ascites has been reported.[2,17]

Review of the literature indicates that more than 50% (18.8%-89%) of the CMTC patients present associated cutaneous and/or extracutaneous anomalies.[3,5-8,10,18-24]

This variability may be explained by the lack of precise diagnostic criteria.

The first association with congenital anomalies was described by Petrozzi et al. in 1970: they found a patent ductus arteriosus and a Sturge-Weber syndrome.[18] Devilliers' statistical analysis of 35 patients indicated associated abnormalities in 80% of cases.[22] Gerritsen instead, detected 61% of associated anomalies, the most serious in patients with generalized skin involvement (28% of cases).[25] In his survey of 85 patients with CMTC, Ben Amitai described 62% of associated anomalies without a specific correlation with the extension of the involved skin.[10] Kienast conducted a prospective study of 27 cases and found associated anomalies in 56% of the patients.[3]

Cutaneous findings may include prominent veins, telangiectasias, cutaneous atrophy, skin ulcerations and hyperkeratosis.[18] In 2006 Torrelo reported two children with CMTC and extensive Mongolian spots but he hypothesized it represented a distinct form of phacomatosis pigmentovascularis.[41]

Table 1

SYSTEMIC ANOMALIES ASSOCIATED WITH CMT.

• Hemangioma
• Congenital pigmented nevus
• Cafè au lait spot
• Cutis aplasia
• Cardiac malformation
• Hypospadias
• Multicystic renal disease
• Hypothyroidism
• Elastolysis
• Phakomatosis pigmentovascularis
• Syndactyly
• Tendinitis stenosans
• Hip dysplasia
• Clubfoot
• Cleft Palate
• Micrognathia
• Scoliosis
• Hypoplasia of iliac and femoral veins
• Stenosis of iliac artery
• Imperforate anus
• Lymphohistiocytosis

Table 2

KIENAST'S SUGGESTED DIAGNOSTIC CRITERIA FOR CMTC.

Major criteria
• Congenital reticulate (marmorated) erythema
• Absence of venectasia
• Unresponsiveness to local warming
Minor criteria
• Fading erythema within 2 years
• Port wine stain outside the area affected by CMTC
• Telangiectasia within the affected area
• Skin ulceration within the affected area
• Atrophy within the affected area

Our patient had face erythema, most pronounced in the upper 2/3, a large teleangiectasic-erythematous patch that affected the front right hemithorax (Fig.1), erythema and telangiectasias scattered on cervicothoracic and lumbosacral spine (Fig.2), an extensive teleangiectasic-erythematous patch on the lateral surface of the right leg, at the level of knee, with a few small areas of atrophy (Fig.3); upper limbs and contralateral lower limb showed livedo reticularis more evident during environmental temperature exposure. Foot sole, especially on lateral side, appeared erythematous (Fig.4).

Body asymmetry, either hypertrophy or hypoplasia, particulary of the limbs, is the most common extracutaneous finding in CMTG. Devilliers noted asymmetry in 43% of their patients with CMTC, the majority had (mainly limb) hyperplasia or hypoplasia, but also facial asymmetry occurred.[22] Kienast found body asymmetry in 33% of cases, limited at the affected areas.[3] These rates were lower than reported by Pehr and Moroz who found a prevalence of 68% in their review.[22] Other skeletal defect have been reported, such as syndactyly, hip dysplasia, clubfoot, cleft palate and tendinitis stenosans.[8,22,27,28]

Table 3

DIFFERENTIAL DIAGNOSES.

Physiological cutis marmorata	Present during few weeks of life, fine symmetric pattern over the trunk and extremities, disappearing with local warming
Klippel-Trénaunay Syndrome	Vascular malformations associated with venectasia and soft-tissue hypertrophy
Sturge-Weber Syndrome	Facial capillary malformations (port-wine stain), ocular and neurological anomalies (glaucoma, leptomeningeal vascular anomalies, comitial crises, motor de motor deficit and leaning disability)
Adam-Oliver Syndromes	CMTC, cardiac malformations, limb defects, aplasia cutis congenita of the scalp and abnormalities of cranium
Bockenheimer's disease	Diffuse phlebectasia, hamartomatous malformation involving the deeper veins, gradual onset during childhood, irregular painful venous dilatation, usually affecting one limb
Divry Van Boageart Syndrome	Corticomeningeal angiomatosis, visual field defects,seizures and "marbled skin"

At birth, our newborn had not body asymmetry which became gradually evident during follow-up. Unlike most limb discrepancies due to vascular malformations, asymmetry here involved relative growth retardation of the affected leg.

Ocular anomalies are rather uncommon and may include congenital glaucoma and congenital retinal detachment.[29,30] Most frequent disorder is congenital glaucoma: up to 2007, 15 cases have been published.[10,30-38]

Petrozzi and Rahn first described congenital glaucoma in a CMTC patient with nevus flammeus overlying the involved eye and mental retardation.[18]

In 1989 Picascia described two patients (of 22 with CMTC) with ocular anomalies: left cataract and glaucoma in a three month old infant and another patient with bilateral glaucoma and nevus flammeus involving the whole face.[19] Also in other cases, congenital glaucoma was detected in patients with vascular anomalies in the periocular skin, indeed it seems to affect the area of distribution of the ophthalmic branch of trigeminal nerve.[18,19,31,33,34,36,37] Congenital glaucoma is usually unilateral: on 15 reported cases 11 had unilateral forms, 2 bilateral ones and in the remaining 2 cases laterality was unspecified. The time of diagnosis is normally soon after birth or in infancy, though Murphy discovered a congenital glaucoma in a nine years old patient with CMTC during his follow-up.[38]

The etiology of this ocular defect is not yet well defined. It appears to be caused by a similar pathologic mechanism as in Sturge-Weber Syndrome, probably due to a neural crest migration disorder; however some Authors describe as cause an abnormal filtration in the anterior angle or increased episcleral venous pressure.[31,35-39] In 1990 Shields described a case of CMTC associated with bilateral total retinal detachment which produced leukocoria simulating retinoblastoma.[30]

According to previous cases reported[38], in our patient trabeculotomy successfully controlled the intraocular pressure, without prolonged postoperative hypotony and suprachoriodal hemorrhage and, moreover, it was not necessary to use glaucoma drainage implant surgery to date.

Additional vascular anomalies, especially capillary malformations such as port-wine stain, were reported frequently.[25,40-44] According to this, in Kienast's study were found 15% of vascular anomalies, 50% of which were capillary malformations.[3]

Neurological abnormalities have been described, such as neonatal hypotonia, developmental delay, mental retardation and seizures. The overall incidence of psychomotor retardation in patient with CMTC has been reported between 0% and 22%, and the presence of neurologic abnormalities may be a diagnostic feature of macrocephaly-CMTC syndrome.[6-8,10,19,22-25] Devilliers found neurological abnormalities in 14% of patients with CMTC (macrocephaly, hydrocephalus, psychomotor retardation, seizures, cerebral atrophy, agenesis of corpus callosum and dilated brain ventricles); one of this fulfilled the diagnostic criteria of Macrocephaly-CMTC Syndrome.[22]

Three patients of Kienast's prospective study were preterm infants born at 27, 33 and 34 weeks of GA.[3] Previously, prematurity in CMTC was described only in other two cases: the first one, a male born at 31 weeks of GA delivered by cesarean section due to placenta previa totalis; the second one a male born at 33 weeks of GA delivered by cesarean section due to maternal hypertension. Both presented typical skin lesions since birth, they had no other systemic or dermatologic disorders with normal laboratory and instrumental evaluation. Their follow-up demonstrated an improvement of the lesions at 4 and 3 months after discharge respectively.[44]

A wide variety of other systemic anomalies are reported in patients with CMTC.[8,11,20,26,42,45-49] (Table 1)

Diagnosis is clinical. Histopathology is often nonspecific or show swollen endothelial cells, dilated capillaries and veins in the dermis or venous lakes.[49,50] Imaging studies are indicated only for the evaluation of suspected congenital anomalies.[49,51]

In 2009 some Authors proposed diagnostic criteria for CMTG based on the presence of all three major criteria and at least two or more minor criteria. (Table 2)

However, the diagnostic validity of these criteria has not been confirmed yet and further studies are needed for this purpose.[3]

Differential diagnoses include Klippel-Trénaunay, Sturge-Weber, Divry-Van Bogaert and Adam-Oliver syndromes.[3,19,27,38] (Table 3)

Association of macrocephaly with CMTC is a distinct disorder and may be associated with overgrowth syndrome, developmental delay and connective tissue defects.[40,52-60] Livedo reticularis and telangiectasia may also be initial signs of neonatal lupus eritematosus.[61] Persistence of skin lesions with local warming and presence of atrophy or skin ulceration distinguishes CMTC from physiological cutis marmorata. The latter may be persistent in children with Down syndrome, de Lange syndrome, homocystinuria and.[8,60]

Prognosis is good with improvement of skin lesions especially during the first two years of life and treatment is usually not required.[22]

Laser therapy has been tried in several patients with persistent CMTC and variable outcomes have been reported.[20]

Conclusion

Actual frequency of the CMTC is likely to be greater than known since it is a benign disorder which tends to self limiting in a relatively short time.

An initial complete examination and multidisciplinary follow-up should be assessed in order to evaluate related abnormalities and enable to distinguish the other conditions that may mimic CMTC itself.

The variety of clinical presentation and associated anomalies has not yet allowed the identification of precise diagnostic criteria that would be able to distinguish CMTC from other vascular malformations.

Future prospective studies are needed to assess the diagnostic validity of proposed criteria.

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