

## Cutis Marmorata Telangiectatica Congenital: A Case Report

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### Abstract

**Introduction:** Cutis marmorata telangiectatica congenita (CMTC) is a sporadic congenital skin vascular abnormality, present at birth or shortly thereafter. The prognosis is generally good.

**Case report:** we report the case of a preterm female newborn with cutis marmorata pattern.

**Conclusion:** CMTC is a rare disease whose determinism remains unknown. The treatment required a multidisciplinary intervention including dermatologist, pediatricians and psychologists.

**Keywords:** Cutis marmorata telangiectatica congenita; Diagnosis; Prognosis

### Introduction

Cutis marmorata telangiectatica congenita (CMTC) is a rare vascular skin abnormality. It was recognized the first time in 1922 by VON LOHUIZEN. It is also named VON LOHUIZEN disease.

It is a benign angiomatous abnormality that manifests as dilation of superficial capillaries and veins usually seen at birth. The skin lesions are typical, characterized by the presence of localized or generalized red or purple reticulated and anastomosed macules, frequently asymmetrical [1]. These skin lesions may be associated with ulceration or atrophy.

### Case Report

A female premature new-born at 35 weeks of consanguineous (second degree) healthy parents was admitted immediately after birth in the Neonatal Intensive Care Unit (NICU) of Maternity and Neonatology Center of Tunis (MNCT).

Her mother was 24 years old, gravida 4 para 4. The pregnancy was mono-chorionic bi amniotic complicated by fetal death of the first twin and spontaneous preterm labour.

The new-born was eutrophic (weight=2800 g). Head circumference was within normal limits (33.5 cm). The hemodynamic and respiratory states were stables.

On the skin, we noted multiple bruises and purple macules anastomosed making a reticulated appearance as a spider's web, those skin lesions cover the entire trunk but did not affect the face and did not disappear during the warming of the baby. Limits between normal and involved skin were clear. The baby had facial dysmorphic features as microretrognathia and low seat ears.

The cranial ultrasound and the abdominal Doppler ultrasound were normal. Thyroid function was also normal. Karyotype didn't objective any deletion.

Given the constellation of clinical findings, Cutis Marmorata Telangiectatica Congenita (CMTC) was diagnosed (Figure 1).

### Discussion

Cutis marmorata telangiectatica is a rare vascular malformation; about 300 cases were reported in literature [1]. Lesions can be present at birth as was our case. Sometimes, they appear few days or weeks later. Occurrence of the skin changes at birth was found in 95% of cases in most series of CMTC. In these studies, an equal sex distribution was observed.

The pathogenesis of CMTC is unknown, all reported cases were sporadic, there was not familial cases recognized as some theories proposing that CMTC may have a genetic support [2,3]. In literature, only one case of CMTC associated to a macrocephaly was reported by Stoll in 2003 with a novo translocation t(2;17)(p11;p13) which disrupted one or more genes entailing skin lesions but also other features: mental retardation, macrocephaly and facial dysmorphism [4].

Lesions are mostly localized touching mainly limbs, primarily legs and the affection could be also unilateral. According to the results found in the larger series, the average of percentage of cases with generalized lesions of CMTC was 30% versus 70% of localized anomalies. The face was involved in the distribution of the skin changes only in some cases, while mucous membranes, palms and soles are always spared [2,5].

The skin anomalies are often characteristic to make the diagnosis, so that diagnostic tests are not necessary to confirm the pathology. Histopathologic examination of biopsy specimens may show a nonspecific increase in the number and the size of capillaries and veins [5]. Reticulated vascular pattern, phlebectasia, focal cutaneous atrophy and ulceration could be seen.



**Figure 1:** Lesions of cutis marmorata telangiectatica.

Syndrome/Criteria	CMTC	M-CM
Major criteria	<ul style="list-style-type: none"> <li>- congenital marmorated erythema</li> <li>- absence of venectasia</li> <li>- non-vanishing after warming</li> </ul>	<ul style="list-style-type: none"> <li>- Macrocephaly</li> <li>- Capillary malformation (both required)</li> </ul>
Minor criteria	<ul style="list-style-type: none"> <li>- fading of erythema within 2 years</li> <li>- telangiectasia</li> <li>- port-wine stains outside the area affected by CMTC</li> <li>- ulceration</li> <li>- atrophy</li> </ul>	<ul style="list-style-type: none"> <li>- Asymmetry or overgrowth</li> <li>- Developmental delay</li> <li>- Midline facial capillary malformations</li> <li>- Neonatal hypotonia</li> <li>- Syndactyly or polydactyly</li> <li>- Frontal bossing</li> <li>- Joint hypermobility or hyperelastic skin</li> <li>- Hydrocephalus (at least 2 criteria)</li> </ul>

**Table 1:** Major and minor criteria for the diagnosis of CMTC and M-CM.

Despite its typical appearance, there are some differential diagnoses of CMTC namely the physiological cutis marmorata resulting from the low temperature of the skin and it disappears after warming. We should also think about klippel Trenaunay syndrome characterized by hypertrophy of soft tissues, venectasia and nevus flammeus [6], Adams Oliver syndrome that associates a congenital skin aplasia, limbs abnormalities and CMTC, Down syndrome and Lange syndrome may be observed with cutis lesions but in these cases the rest of the clinical findings help to make the right diagnosis.

Kienast and Hoeger, in one of their prospective study on CMTC published in 2009 including 27 cases, suggested clinical criteria to help in the distinction of CMTC from the other vascular malformations. Diagnostic criteria were classified on 3 major criteria and 5 minor criteria (Table 1) [3]. Our case meets the 3 major criteria.

In 25% of cases reported in the literature there were associated abnormalities to the vascular lesions such as glaucoma, macrocephaly, syndactyly, renal hypoplasia, body asymmetry and psychomotor or mental retardation, developmental delay, hypothyroidism, hypospadias

[7-10]. This association is without correlation with the extent of lesions.

Occurrence of vascular complications such as vessel stenosis and cerebral vascular malformations in addition to the vascular skin changes makes some authors postulate that CMTC is a generalized vasculopathy [11,12].

Association of macrocephaly and CMTC (M-CMTC) create a particular syndrome that does not share with isolated CMTC the same prognosis in view of risk of occurrence of sudden death, cardiac arrhythmia, apnea and association with unusual cerebral manifestations, therefore, monitoring of the head circumference is very important in babies with CMTC. MRI scan findings show in most cases structural cerebral abnormalities namely megalencephaly, cortical dysplasia, asymmetry of the cerebral hemispheres and abnormally increased signal of white matter [11,13].

Dakar Rucker Wright et al, based on their own experience with 112 patients suffering from M-CMTC, suggest major and minor criteria (Table 1) for the diagnosis of M-CMTC and proposed the name of macrocephaly-capillary malformations (M-CM) in view of the frequent association of macrocephaly not only with CMTC but rather with reticular shaped capillary malformations [14].

Assessment of patients with CMTC in the larger series showed tendency to the spontaneous resolution or improvement of skin lesions in 50% cases within 2 years.

There is no curative solution to persistent CMTC up to day.

## Conclusion

CMTC is a rare skin disease whose determinism remains unknown. Other congenital anomalies can be seen. The diagnosis of the CMTC is based on the clinical findings. Diagnostic tests like skin biopsy are not necessary. The treatment required a multidisciplinary intervention including dermatologist, paediatricians and psychologists. The prognosis depends on the associated anomalies but it is generally good. Some cases have spontaneous resolution.

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