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Cutis marmorata telangiectatica congenita restricted to both breasts in a young female

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ABSTRACT Cutis marmorata telangiectatica congenita (CMTC) is a very rarely occurring congenital disorder with persistent cutis marmorata, telangiectasia, and phlebectasia. This disorder may be associated with cutaneous atrophy and ulceration of the involved skin. We herewith report a 20-year-old female patient with CMTC since childhood along with ulcerations on both breasts. CMTC is a benign vascular anomaly presenting with dilatation of capillaries and veins of dermis and is apparent at birth. The patient had reticulated bluish-purple skin changes over both breasts. Although it resembled physiological cutis marmorata, it was more pronounced and definitely was unvarying and permanent in pattern. A variety of vascular malformations have been described along with this disorder. Etiology is not very clear; it may be multifactorial in origin. Prognosis in uncomplicated cases is good.

Introduction

Cutis marmorata telangiectatica congenita (CMTC) was first described by a Dutch pediatrician, Von Lohuizen, in 1922, in the Netherlands, and since in his description of CMTC, less than 250 cases had been reported. CMTC is not very commonly reported disorder and is sporadic in nature. It is a congenital vascular anomaly characterized by persistent cutis marmorata, telangiectasia, phlebectasia, and sometimes, cutaneous atrophy and skin ulceration is present. A variety of associated defects have been described in up to 50% of cases [1]. It is usually a benign congenital skin lesion that is present at birth but may develop later on in life.

Case report

A 20-year-old unmarried female presented with persistent reticulated purplish erythematous to hyperpigmented lesions mainly over both breasts since childhood. The patient also had ulcerations and atrophy over both breasts where a reticulated pattern and dilated veins were prominent (Figures 1 and 2). There was no abnormality in limbs, asymmetry of the

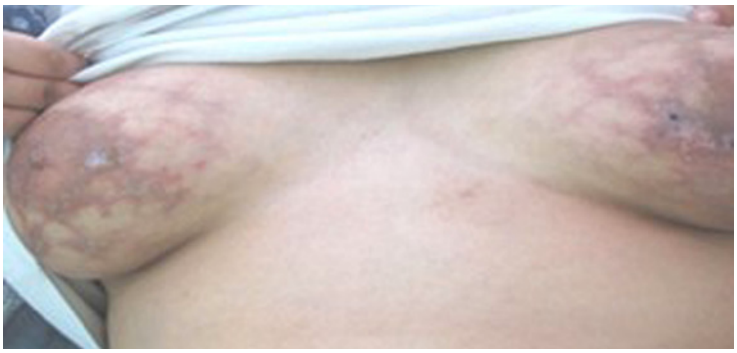

Figure 1. Reticulate erythema over both breasts with ulceration and atrophy. [Copyright: ©2014 Lunge et al.]

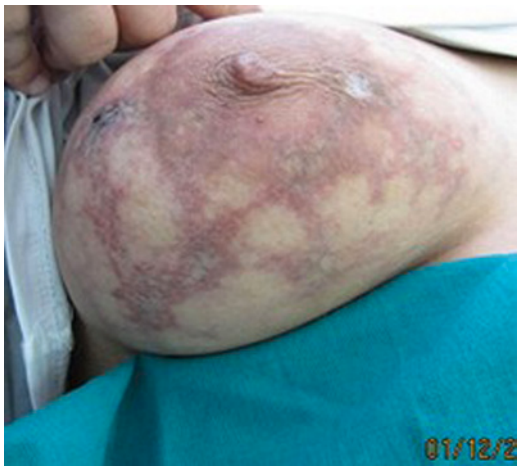

Figure 2. Right side of breast showing reticulate erythema with atrophy. [Copyright: ©2014 Lunge et al.]

body, other vascular anomalies, neurological complications, ocular changes, or syndactyly. General examination and systemic examination was within normal limits. Similar, but less pronounced, types of skin lesions were present in the patient's father and aunt, mainly over the extremities.

We considered the differential diagnoses of cutis marmorata telangiectatica congenita, physiological cutis marmorata, erythema ab agne, livedoid vasculitis, connective tissue disorders, and antiphospholipid syndrome. Complete blood count and all routine investigations were normal. Antiphospholipid antibody and ANA tests were negative. Skin biopsy from the affected area showed classic dilated capillaries and veins in the dermis (Figure 3). Similar dilated capillaries and veins were seen in the subcutaneous tissue along with proliferation of vascular channels (Figure 4).

The clinical and histopathological findings led to a final diagnosis of cutis marmorata telangiectatica congenita.

Discussion

CMTC is a very rarely reported congenital cutaneous disorder and is usually present at birth but may also appear up to two years after birth. However, since the first description of CMTC in 1922 by Von Lohuizen, less than 250 cases worldwide have been published to date.

In 1970 Petrozzi et al. [2] reported the first case of CMTC in United States. The frequency of this disorder is not known. In most cases reported, patients have associated malformations, hypoplasia of the limbs [3], or hyperplasia of the limbs.

The condition closely resembles cutis marmorata, a relatively common disorder. The benign reticular mottling of the skin seen in small children due to physiologic dilatation of capillaries and small venules in response to cold environment is the most significant difference that exists between the congenital cutis marmorata (CMTC) and physiologic cutis marmorata, a clue that can help to distinguish between these lesions. Although physiological cutis marmorata disappears with local warming, the congenital condition (CMTC) will persist despite increases in ambient temperature [4]. In our case CMTC was diagnosed on a clinical basis initially because skin lesions did not disappear with local warming [5].

The pattern of the lesions in cutis marmorata telangiectatica congenita may include prominent veins, telangiectasias, cutaneous atrophy, ulceration and hyperkeratosis of affected skin. The presence of cutaneous atrophy or skin ulceration could be useful in distinguishing between the congenital and physiologic types. Cutaneous atrophy and skin ulceration

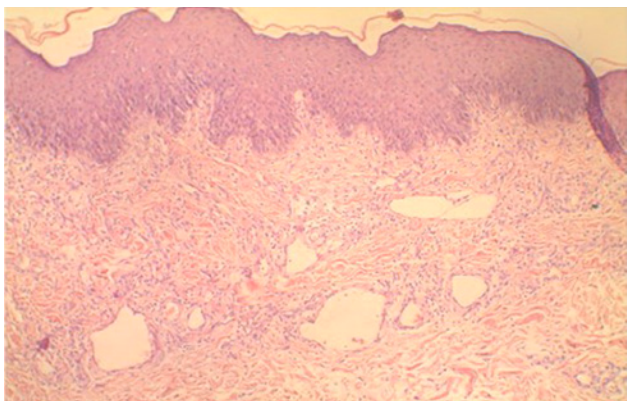

Figure 3. Dilatations of veins and capillaries in dermis. [Copyright: ©2014 Lunge et al.]

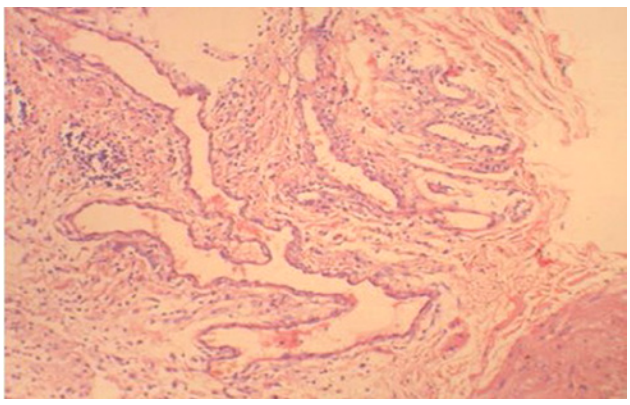

Figure 4. Proliferation of vascular channels in subcutaneous tissue. [Copyright: ©2014 Lunge et al.]

are common features of cutis marmorata telangiectatica congenita, but they are not present with physiological cutis marmorata [4]. In addition, the presence of sharp demarcation of localized lesions favors a diagnosis of the congenital type over physiological cutis marmorata, which appears more mottled and has ill-defined borders of skin lesions. Our patient had prominent veins, telangiectasias, cutaneous atrophy and ulceration along with reticulate erythema, favoring a diagnosis of CMTC.

Diagnostic criteria for CMTC helped to clearly distinguish CMTC from other vascular anomalies. A case series published in the literature by Kienast and Hoeger in 2009 [5] suggested the diagnostic criteria for CMTC that includes (1) three major criteria: congenital reticulate (marmorated) erythema, absence of venectasia, and unresponsiveness to local warming); and (2) two of the five minor criteria: fading of erythema within two years, telangiectasia, port-wine stain outside the area affected by CMTC, ulceration, and atrophy. These criteria are sufficient for the diagnosis of CMTC. Reticular erythema present at birth is a common finding in all reported cases, so it has been considered major criteria for CMTC. Physiological cutis marmorata occurs in otherwise healthy people in a cold environment; CMTC does not respond to local warming. The absence of venectasia in the affected region of skin is a very important finding and differentiates CMTC from Klippel–Trenaunay–Weber (KTW) syndrome. The presence of telangiectasia and ulcerations found in a few cases can be classified as minor criteria, but if present, strongly favor a diagnosis of CMTC [5].

Our patient meets two major criteria (congenital reticulate erythema, unresponsiveness to local warming) and three minor criteria (telangiectasia, ulceration, atrophy) and, therefore, we believe that it is a crystal clear case of CMTC.

The pathogenesis of these disorders is not very clear and the cause may be multifactorial in origin. Most cases occur sporadically, although very rare cases have suggested a possible genetic link [4]. Our case favors familial origin because the patient's father and aunt had similar lesions, though less pronounced, that were situated over both extremities. Teratogens [6] and autosomal dominant mode of inheritance with incomplete penetrance have been considered as etiologies. Some authors suggest that the lethal gene hypothesis might explain the patchy distribution of the skin lesion that occurs sporadically in cases of CMTC.

This case reports on a 20-year-old female, who had persistent cutis marmorata, telangiectasia, and phlebectasia with ulcerations over both breasts where the reticulated pattern and dilated veins are seen. Vascular anomalies, for example, Sturge-Weber syndrome [7], Klippel–Trenaunay–Weber syndrome have been associated with CMTC. The female patient reported in this article had a normal facies with no other vascular abnormality elsewhere in body, and systemic evaluations, including ophthalmological examination, were normal. She had no asymmetry of the limbs or limb growth abnormality.

The incidence of abnormalities associated with CMTC varies from 18.8 to 89% [8,9]. The association of macrocephaly with CMTC is a subgroup of distinct disorders and it may be associated with overgrowth syndrome [10].

Rare reports include atypical CMTC with retinoblastoma [11]. Interestingly, CMTC associated with meningioma or leukemia has also been reported and is very rarely associated with mental retardation, aplasia cutis congenita, multicystic renal disease, glaucoma, and Mongolian spot [12].

A review of the literature reveals existing controversies regarding gender-related prevalence of CMTC. Several series reveal that the disorder affects girls more than boys, however, statistical evaluation showed no significant difference between both. A report also suggests that boys tend to have localized disease. Our case was a young female patient with localized reticulated lesions on both breasts, uncommon and hitherto unreported.

The differential diagnosis includes capillary malformations [13], KTW syndrome, neonatal lupus erythematosus, nevus anemicus, physiological cutis marmorata, livedo reticularis associated with collagen vascular disorders, antiphospholipid antibody syndrome, nevus flammeus, and diffuse phlebectasia.

The disorder was diagnosed clinically and histopathologically. In the dermis, dilatation of capillaries and venules with a proliferation of vascular channels is an atypical histopathological finding [14]. The imaging studies were indicated only for the evaluation of any suspected congenital anomalies associated with CMTC.

The disorder is self-limiting and treatment is not necessary unless complicated with other associated anomalies, such as glaucoma, multicystic kidney disease, asymmetry of limbs, and cardiac malformations. Different approaches of treatment for CMTC include cold avoidance, vasodilators, aspirin, pentoxifylline, PUVA, and IPL [15]. In some cases consultations with orthopedics, neurosurgery, ophthalmology, and vascular cosmetic surgery may be necessary.

Conclusion

Our female patient had unique findings with restricted CMTC over both breasts, which is very uncommon.

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