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Cutis Marmorata Telangiectatica Congenita with Limb Hypertrophy- A Case Report and Elaboration of its Salient Features.

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Abstract: *Cutis Marmorata Telangiectatica Congenita (CMTC) is an uncommon, sporadic, usually congenital, cutaneous vascular disorder of unknown aetiology. Clinically characterised by persistent cutis marmorata , telangiectasia, cutaneous atrophy and ulceration. CMTC have been found to be associated with many anomalies like limb asymmetry, cutis aplasia congenital, hemangiomas , pigmented nevi , macrocephaly , glaucoma etc. Diagnosis is made on clinical grounds. The prognosis of CMTC is mostly benign with half of patients demonstrating resolution of fixed reticulated erythema over two years but because of associated anomalies patients should be properly examined and investigated. Here, we are reporting a 3 month old girl child with fixed reticulated erythematous lesion distributed in a generalized pattern with limb hypertrophy present since birth.*

Keywords: *Cutaneous vascular malformations, cutis marmoratatelangiectaticacongenita*

I. Introduction

Cutis MarmorataTelangiectaticaCongenita(CMTC) is an uncommon [1], cutaneous vascular malformation of unknown aetiology which mostly presents at birth[2]. It is sporadic in nature but some familial cases have been reported[3,4]. Some studies have shown an autosomal dominant pattern of inheritance [3]. CMTC is also known as Van Lohuizen's syndrome because the first case of CMTC was described by Van Lohuizen in 1922 in a female child[5]. The fixed reticulated cutaneous erythema does not disappear on warming the skin surface as seen in physiological cutis marmorata[1]. The pattern of distribution of this persistent cutis marmorata can be localised or generalised[2,6]. These skin lesions can be accompanied by ulceration[7] , underlying subcutaneous tissue atrophy (being more common) or hypertrophy[1] , leading to limb asymmetry.
The percentage association of Cutis MarmorataTelangiectaticaCongenita with other congenital anomalies ranges from 20-70% [8-11]. The most common being cutis aplasia, cleft palate and developmental delay[8,10,11]. Other anomalies are macrocephaly[12], hypothyroidism[10],glaucoma[8,13]and anogenital malformation[10,11]. Diagnosis is based on clinical findings[1,14].Histopathology shows dilated capillaries and veins, and venous lakes in dermis and subcutis[1]. The histopathology is nonspecific hence is not considered as diagnostic. The course of disorder is usually benign with fading of cutaneous lesions and in long term spontaneous resolution occurs in most cases[1,14]. Here we are reporting a case of CMTC with limb hypertrophy which has been rarely reported.

II. Case report

A 3 month old female child visited our out-patient department with complaints of fixed mottled reddish lesions on skin of whole body and swelling of left thigh which were present since birth. The patient was a second child born to non-consanguinous parents. The patient was a term baby with normal birth weight born by normal vaginal delivery. The child was a product of uneventful pregnancy. The developmental milestones were normal. There was no family history of such lesions.

On physical examination, persistent fixed erythematous reticulated cutaneous lesions were present in a generalised manner [Fig.1&3] on scalp, abdomen and whole of back(which crossed the mid-line), on buttocks, on both upper and lower limbs. Some of the skin lesions had underlying venous prominences and atrophy [Fig.2]. The anthropometric measurements were within normal range except for difference in the girth of right

and left thighs, which on proper examination revealed hypertrophy of left thigh [Fig.2&3]. Systemic examination including neurologic and ophthalmologic examination of child were normal.
Laboratory investigations included complete blood count, thyroid profile, abdominal ultrasonography which came out to be normal. On the basis of these findings diagnosis of cutis marmoratatelangiectaticacongenita was made.

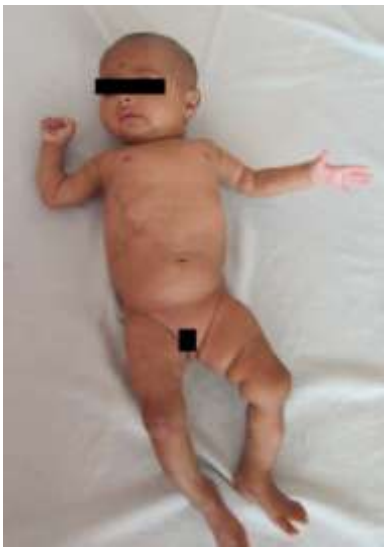

Figure 1 showing fixed reticulated erythematous lesion on abdomen ,upper & lower limbs

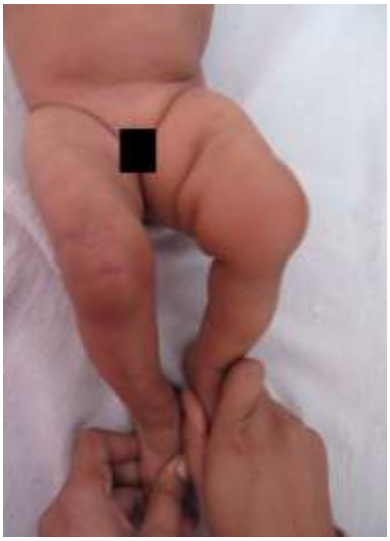

Figure 2 showing left thigh hypertrophy and Cutaneous atrophy on right knee and vein prominence on right thigh

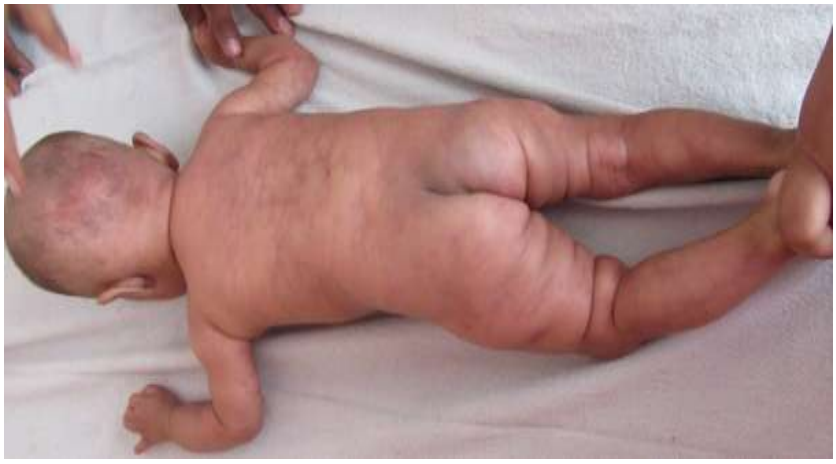

Figure 3 generalised persistent cutis marmorata on scalp, back of whole trunk, posterior aspect of upper & lower limbs

III. Discussion

Cutis MarmorataTelangiectaticaCongenita is a characterised by presence of persistent cutis marmorata, telangiectasia with underlying cutaneous atrophy and ulceration[14].Though cutaneous atrophy is more common occasionally hypertrophy [1] can also occur as seen in this case. David et al showed CMTC mostly presents at birth[8] which is also true in our case but it can also present in first few days after birth[2]. One study showed that all children were delivered after an uncomplicated pregnancy[7], same is true in our patient. Many studies showed a female preponderance which matched with our case[8,15]. Amitai et al[15] reported localised variety of CMTC is more common than generalised variety. Moreover, localised variety of CMTC with lower limb involvement was common in females and the generalised variety with distribution on head, neck and trunk found to becommon in males in his study [15]. Contrary to this, in our case, it is a female child with generalised distribution making it a rare case to report.

Apart from limb asymmetry which is the most common associated anomaly as observed in our case, other anomalies like aplasia cutis congenital, cleft palate, macrocephaly, hypothyroidism, glaucoma, hemangioma, pigmented nevi as mentioned before were all absent according to history, physical examination and laboratory investigation.

The closest differential diagnosis of CMTC are physiological cutis marmorata which occurs due to prolonged exposure to cold but disappears after warming of skin surface[1], Klippel-Trenaunay-Weber syndrome ,presents with a vascular lesion, varicosities of veins and hypertrophy of soft tissues often difficult to distinguish from CMTC and requires a long term follow-up to differentiate between the two[8] and Bockenheimer's syndrome characterised by progressivedevelopment of many large painful venous ectasias mostly in a single limb, but its onset is in childhood unlike CMTC[8]. Other diiferential diagnosis include homocystinuria, Cornelia de Lange's syndrome and down's syndrome which can be distinguished on the basis of clinical features[1,14]. Sometimes neonatal lupus erythmatosus may present with cutis marmorata skin lesion but can be easily differentiated by serological tests of mother and neonate[1,14].

IV. Conclusion

As there are variation in presentation of CMTC with serious associated anomalies, many yet not discovered, each and every patient of CMTC must undergo a thorough history taking, physical examination and laboratory investigation.

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