VOLUME - 9, ISSUE - 7, JULY - 2020 • PRINT ISSN No. 2277 - 8160 • DOI : 10.36106/gjra

Original Research Paper

Dermatology

CUTIS MARMORATA TELENGIECTATICA CONGENITA (CMTC): A CASE REPORT.

Dr Sanket Vashist	Department of Dermatology, venerology and leprosy, Regional Hospital, Dharamshala, Himachal Pradesh.
Dr Aashutosh sharma	Department of pediatrics, Zonal Hospital, Dharamshala, Himachal Pradesh.*Corresponding Author

KEYWORDS :

Introduction

Cutis marmorata telangiectatica congenita (CMTC) is a rare capillary malformation with sporadic occurrence and unknown exact aetiology which is characterised by persistent reticulated marbled erythema which may be associated with skin atrophy, ulcerations, limb asymmetry, variable systemic involvement and can mimic few rarer syndromes. Thorough systemic evaluation and subsequent follow up are necessary, even in cases where initially no extra cutaneous associations might have been detectable.

Case Report

A six months otherwise healthy female infant was presented to the hospital by parents with complaints of reddish reticulated lesions over the left lower limb and progressive difference in girth of both lower limbs, with relative decreased girth of the left lower limb, apparently noticed since past 2 months. She was born out of normal vaginal delivery with uneventful perinatal period and no gross developmental delay. The reddish lesions were persistent even on warming. Dermatological examination showed the presence of reticulated erythematous vascular appearance of skin of left lower limb resembling physiological cutis marmorata with difference in girth of about 1.3 centimetre at middle thigh level. Few similar but less prominent lesions were also present on upper part of the other limb. No temperature difference, bruits or other skin changes were noted (figures:1,2,3,4).

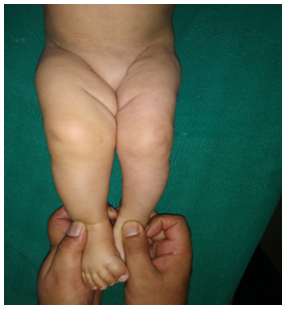

Figure 1 "

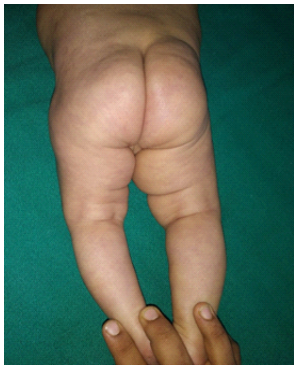

Figure 2

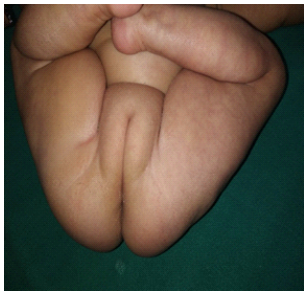

Figure 3

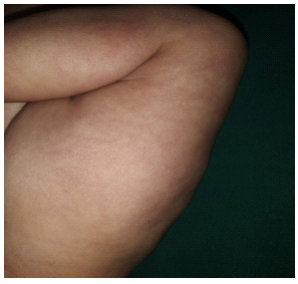

Figure 4

Patient was evaluated by paediatrician and no systemic abnormality was detected. Orthopaedic examination was also normal except for difference in girth of both limbs. Ophthalmological examination was also normal. Ultrasound colour doppler of both the lower limbs was also normal. The patient was diagnosed as a case of Cutis Marmorata Telengiectatica Congenita (CMTC), with no frank systemic involvement and advised regular follow up at 3 monthly intervals.

DISCUSSION

Cutis Marmorata Telengiectatica Congenita (CMTC), eponymously known as Van Lohuizen syndrome[1] is a relatively rare sporadic congenital capillary malformation characterized by persistent cutis marmorata, with or without skin atrophy and ulcerations along with other systemic involvement in some cases. Although few cases have been reported in the past the variable presentations and mimicking conditions make it an interesting entity requiring thorough evaluation. The exact etiology is unknown but associations with teratogenicity[2], fetal ascites , raised maternal beta human chorionic gonadotropin hormone [3], GNA11 [4,5,6] and ARL6IP6 gene[7,8] mutations are few proposed mechanisms.

In a review of 485 patients [9], 206 patients (42.5%) had associated anomalies. Body asymmetry in 37.7%, neurological defects in 10.1%, most commonly developmental delay and seizures and ophthalmological complications in 9.9% of patients mostly congenital glaucoma were the most

common associations. Most commonly it can be confused with physiological cutis marmorata which is a benign self-resolving condition.

Megalencephaly-capillary malformation and megalencephalypolydactyly – polymicrogyria -hydrocephalus (MPPH) syndromes are related syndromes with some overlapping features with megalencephaly being a distinctive finding.[10]

Other relatively less common conditions which need to be excluded include port-wine stain, Klippel–Trénaunay syndrome, Parkes Weber syndrome, Congenital livedo reticularis primary or with associated Down's syndrome, neonatal lupus erythematosus, antiphospholipid antibody syndrome, vasculopathies or autoimmune connective tissue disorders , Macrocephaly–capillary malformation, Adams–Oliver syndrome , Phakomatosis pigmentovascularis with cutis marmorata telangiectatica congenita, Bockenheimer syndrome etc.

Kienast and Hoeger [11] have proposed the presence of all three major criteria in addition to two minor criteria for diagnosing CMTC.

Major criteria- Congenital reticular (marmorated) erythema Absence of venectasia Unresponsiveness to local warming
Minor criteria- Fading of erythema within two years Telangiectasia within affected area Port-wine stain Ulceration within affected area Atrophy within affected area.
It is mainly a clinical diagnosis requiring multidisciplinary approach to rule out systemic involvement or mimicking conditions. The prognosis of the condition depends on systemic involvement. In uncomplicated cases the improvement in skin appearance is seen over few years.

CMTC without systemic involvement is a benign condition in which skin lesions improve over few years but a patient presenting with CMTC should be thoroughly evaluated to rule out systemic involvement and other rarer disorders with similar presentation. Parents must be counselled and regular follow up especially for glaucoma, neurodevelopmental delays and locomotor disability.

REFERENCES

1. Van Lohuizen C. Über eine seltene angeborene Hautanomalie (Cutis marmorata telangiectatica congenita). Acta Dermatol Venereol. 1922;3: 202–11.
2. Bhargava P, Kuldeep CM, Mathur NK. Cutis marmorata telangiectatica congenita with multiple congenital anomalies: Further clues for a teratogenic cause. Dermatology 1998;196: 368-70.
3. Chen CP, Chen HC, Liu FF, Jan SW, Chern SR, Wang TY, et al. Cutis marmorata telangiectatica congenita associated with an elevated maternal serum human chorionic gonadotrophin level and transitory isolated fetal ascites. Br J Dermatol 1997;136: 267-71.
4. Sassalos TM, Fields TS, Levine R, Gao H. Retinal neovascularization from a patient with cutis marmorata telangiectatica congenita. Retin Cases Brief Rep. 2018.
5. Thomas FP, Guergueltcheva V, Gondim FA, Tournev I, Rao CV, Ishpekova B, et al. Clinical, neurophysiological and morphological study of dominant intermediate Charcot-Marie-tooth type C neuropathy. J Neurol. 2016;263(3): 467–76.
6. Kumar A, Zastrow DB, Kravets EJ, Beleford D, Ruzhnikov MRZ, Grove ME, et al. Extracutaneous manifestations in phacomatosis cesioflammea and cesiomarmorata: case series and literature review. Am J Med Genet A. 2019; 179(6):966–77.
7. Abumansour IS, Hijazi H, Alazmi A, Alzahrani F, Bashiri FA, Hassan H, et al. ARL6IP6, a susceptibility locus for ischemic stroke, is mutated in a patient with syndromic cutis marmorata telangiectatica congenita. Hum Genet. 2015;134(8):815–22.
8. Dereure O. Cutis marmorata telangiectatica congenita: mutations in a susceptibility gene involved in cerebrovascular accidents. Ann Dermatol Venereol. 2016;143(1):96–7
9. Bui, T.N.P.T., Corap, A. & Bygum, A. Cutis marmorata telangiectatica congenita: a literature review. Orphanet J Rare Dis; 2019(14): 283.
10. Mirzaa GM, Conway RL, Gripp KW, LermanSagie T, Siegel DH, deVries LS, et al. Megalencephaly-capillary malformation (MCAP) and megalencephalypolydactyly-polymicrogyria-hydrocephalus (MPPH) syndromes: Two closely related disorders of brain overgrowth and abnormal brain and body morphogenesis. Am J Med Genet Part A.2012; 158A:269–91.
11. Kienast AK, Hoeger PH. Cutis marmorata telangiectatica congenita: a prospective study of 27 cases and review of the literature with proposal of diagnostic criteria. Clin Exp Dermatol 2009;34:319-23.