SKIN

RESEARCH LETTER

Characterization of Clinical Outcomes in Patients with Cutis Marmorata Telangiectatica Congenita

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ABSTRACT

Cutis marmorata telangiectatica congenita (CMTC) is an uncommon, congenital, cutaneous vascular disease with an unknown pathogenesis. Although considered as a benign condition, anomalies such as body asymmetry are frequently associated. Herein we present a series of patients with diagnosis of CMTC with a focus on clinical outcomes. In our series, limb length abnormalities were the most common associate anomaly, occurring in 24% of the subjects, similar to other series with rates of 33% and 27% but higher than the general population (6.7%). Importantly, dermatologists should be aware of the frequently associated anomalies in CMTC, such as leg length discrepancy, which may have serious consequences if not recognize and treated.

Cutis marmorata telangiectatica congenita (CMTC) is an uncommon, congenital, cutaneous vascular disease. The pathogenesis of CMTC remains unknown. Although considered as a benign condition, anomalies such as body asymmetry, vascular lesions, neurological and ophthalmic manifestations, hypoplasias/aplasias are frequently associated. However, whether these associations are due to reporting bias is a matter of debate. This study evaluated CMTC patients seen at our institution and examined their clinical outcomes.

The Wake Forest Baptist Health institutional review board (Winston-Salem, North Carolina) approved this single-institution retrospective medical record review. Inclusion criteria included patients 18 years or younger diagnosed with CMTC from July 2009 – July 2019. Patients with CMTC were identified using ICD-19 code: 757.32 and ICD-10 codes: Q82.8 and R23.9. Patients with physiologic cutis marmorata were excluded. Primary outcome measures were presence of associated anomalies and improvement of skin over time. Demographics collected included age, sex, and race. Information on anatomic distribution of skin lesions was also extracted.

Seventeen patients were identified using the inclusion criteria. All cases had parental reports of skin lesions present at or shortly

Table 1. Patient demographics and clinical characteristics

Patient no.	Sex	Age at diagnosis	Age at most recent follow-up	Associated signs/ symptoms	Associated anomalies	Improvement of skin over time
1	F	15 y/o	No follow-up		Raynaud's-like phenomenon	No
2	M	7 y/o	12 y/o	Varicose veins	Hypertrophy of 2nd toes (bilateral), and pectus excavatum	Yes
3	M	17 mo.	4 y/o		Leg asymmetry, port wine stain	No
4	M	8 mo.	2 y/o		Phakomatosis pigmentovascularis (CMTC + Mongolian spot), ocular melanosis, nevus spilus	Yes
5	F	4 mo.	7 mo.	Atrophy		Yes
6	M	16 mo.	22 mo.		Hamartoma	Yes
7	F	4 mo.	16 mo.		Connective tissue nevus	Yes
8	F	10 days	2 y/o	Swelling, pain, and difficulty walking	Speech delay, congenital umbilical hernia, ash-leaf spot vs nevus anemicus	Yes, but pain worsened
9	M	4 mo.	No follow-up		Leg asymmetry	Yes
10	M	2 y/o	No follow-up		Bilateral hearing loss, wheezing, dysphagia	Yes
11	F	2 mo.	15 mo.			Yes
12	M	2 mo.	No follow-up			Yes
13	M	10 mo.	16 mo.			Yes
14	F	3 mo.	6 mo.		Port wine stain	Yes
15	M	15 y/o	No follow-up	Pain, ulceration, atrophy		No
16	F	1 mo.	6 y/o	Pain	Leg circumference asymmetry (10mm), minimal limb length difference (4mm)	No
17	F	5 mo.	3 y/o	Pain, swelling, limping	Vein asymmetry	No

after birth. Both genders were equally affected (Table 1). Nine patients (53%) had only one affected limb (Table 2). Twelve cases (71%) reported fading of skin lesions over time. Four patients (24%) reported pain due to skin atrophy or limb discrepancy. Body asymmetry was present in five subjects (29%). Six subjects (35%) had another dermatosis, most commonly a port-wine stain (12%) (Table 1). Half of those who received a magnetic resonance imaging (MRI) scan (2 out of 4 subjects) had normal findings. The scans of the other two patients revealed increased number of venous structures and venous malformations with asymmetry of the greater and lesser saphenous veins.

The low prevalence of CMTC makes information on its clinical characteristics and associations limited. The rate of complications and associated abnormalities reported is highly variable ranging 18-80%. Due to different reporting methodologies and inclusion of abnormalities which are prevalent in the general population, the higher rate may be secondary to incidental or

questionable findings.(1-3) Recent reports have characterized limb length discrepancy and other congenital vascular abnormalities as the most frequent associations.(3-5)

In our series, limb length abnormalities were present in 24% of the subjects, similar to other series with rates of 33% and 27% but higher than the general population (6.7%).(4, 6) Because of possible reporting bias, the relative proportion of limb length abnormalities related to CMTC diagnosis is controversial. Limb measurement is recommended to be assessed and documented in each evaluation. Most cases are mild (<2cm difference) and do not require additional intervention other than follow-up. Discrepancies ≥2cm may require pediatric orthopedic evaluation. Overall, the orthopedic prognosis is good and only a minority of subjects require surgical intervention. (4) Also consistent with other series, most patient records (71%) noted that their lesions faded over time (3). We predict the true rate of improvement is higher due to lack of follow-up for asymptomatic courses of CMTC. Limitation of this study are its retrospective design, reliance on clinicians reporting their findings, small sample size, and limited duration of follow-up.

CMTC is a relatively benign disorder on its own, which does not usually require treatment. Health care professionals should be aware of the frequently associated anomalies, such as leg length discrepancy, which may have serious consequences if not recognized and treated. Children with CMTC on their legs should have regularly monitoring for leg length discrepancy during childhood. Improvement of marbled skin appearance occurs in the majority of patients over time, and improvement is likely higher than reported due to lack of follow-up reporting for asymptomatic courses of CMTC.

Table 2: Distribution of skin lesions

Patient no.	UE	LE	Abdomen/ Chest	Back	Face
1	X				
2	XX	XX	X		
3	XX	X	X	X	X
4	XX	XX	X		
5	X				
6		X			
7			X		
8		X			
9		X	X	X	
10	X				
11		X			
12		X	X		
13		X			
14	X		X		X
15			X		
16		X			
17		X			

XX: bilateral
LE – Lower Extremity; UE – Upper Extremity

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