Case Report

Cutis Marmorata Telangiectatica Congenita: Clinical Features in 7 Cases

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Cutis marmorata telangiectatica congenita (CMTC) is a sporadic congenital vascular anomaly usually presents at birth as a localized or generalized reticulated, erythema and telangiectasia. The pathogenesis of CMTC is unknown. Additional anomalies have been frequently reported in association with CMTC. This article describes the characteristics of the clinical presentations in a series of 7 children with CMTC in HRH Princess Maha Chakri Sirindhorn Medical Center, Srinakharinwirot University, Thailand. Both genders were almost equally affected (3 male/4 female). The lower limbs were involved in 7 patients (100%), the trunk in 3 patients (42.9%), and the upper limbs in 1 patient (14.3%). There were 2 patients (28.6%) who presented with the involvement of both trunk and limbs. The mean of the body surface area involved was 17% (2-50). Asymmetry of affected limbs were found in 2 patients (28.6%). On follow-up, improvement of reticulated lesions was noted in all patients. The prognosis of uncomplicated cases is good. No specific treatment is needed. The skin lesions usually disappear gradually.

Keywords: *Cutis marmorata telangiectatica congenita, Benign vascular anomaly*

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Cutis marmorata telangiectatica congenita (CMTC), a rare sporadic congenital vascular anomaly, usually presents at birth. CMTC resembles cutis marmorata, a physiologic response to cold in infants. However, it does not resolve with warming of the skin[1]. Etiology of CMTC is still unclear. The proposed hypotheses include environmental factors, autosomal dominant inheritance, multifactorial cause, and genetic factors[2,3]. Diagnosis is made solely on the clinical presentations. It is characterized by reticulated mottling, telangiectasia, infrequent ulceration, and skin atrophy.

Various anomalies associated with CMTC have been reported. The frequency varies between 20 to 80 percent. Asymmetry of the body, other vascular anomalies, glaucoma, macrocephaly and mental or psychomotor retardation were frequently described[2,4-6]. It is still unclear whether the associations occur as incidental findings or due to reporting bias[7]. We report a series of 7 neonates with CMTC at Department of Pediatrics, Faculty of Medicine, Srinakharinwirot University, Thailand, describing the characteristics of clinical presentations.

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Case Report

CMTC is defined as a congenital cutaneous pattern of localized or generalized reticulated mottling telangiectasia. Seven neonates, 3 males and 4 females, born between 2009 and 2015 presented with this characteristic vascular anomaly. We recorded the gender, distribution of cutaneous lesions, other associated anomalies, and limb discrepancy. All seven patients were followed-up until the resolution of the skin lesions.

According to the data collected from the seven term neonates with CMTC, we found that both genders were almost equally affected (3 boys and 4 girls). The skin lesions were found at birth or shortly thereafter. All of them did not have a family history of similar vascular birthmark. The mean area of involvement was 17% of body surface area (range from 2 to 50%). The lower-limbs were involved in all 7 patients (100%), the trunk in 3 patients (42.9%), and the upper limbs in 1 patient (14.3%). There were 2 patients (28.6%) who presented with the involvement of both trunk and limbs (Table 1). Skin atrophy was

found in all 7 patients.

Body asymmetry or limb atrophy in the affected limbs were found in 2 patients (28.6%). Other associated anomalies such as neurological involvement, ocular involvement, syndactyly, and skin ulceration were not found. During follow-up period, the improvement of reticulated lesions were noted in all of our patients within the first 2 years (Fig. 1).

Discussion

We report a series of 7 patients with CMTC. CMTC can either be generalized or localized. It is not clearly stated regarding the frequency of the localized forms of CMTC, but it tends to be more common. In a study by Amitai et al, localized forms and generalized forms occur in 60% and 40%, respectively[4]. Complete covering of the entire skin has not yet been reported to date[8]. Preferentially, they affect the limbs and the trunk. Palms, soles, and mucous membranes are usually spared[5]. When the lesion is localized, it tends to be unilateral and not cross midline[1]. In all of our patients (100%), CMTC involve the lower limbs. Only 3 of the cases involve the trunk (42.9%) and 1 case involves the upper limbs (14.3%). These results correspond with previous reports regarding common areas of involvement of CMTC.

Cutaneous findings of CMTC usually present at or within a few days after birth. From previous literatures, CMTC were represented equally in both genders as also shown in our series[2,4,7].

Several anomalies were reported in associated with CMTC in a wide range of 20% to 80% of patients[1,4,9,10]. However, it is still controversial on the accuracy of these rates. In 2009, Kienast and Hoeger reviewed types of associated anomalies from previous literatures. Of 215 patients with CMTC, body asymmetry was the most frequent associated anomaly found, accounted for 25.1% of all anomalies. Other vascular anomalies, skin atrophy, neurological involvement, ocular involvement, syndactyly, and skin ulceration were found, respectively[4,7]. In another smaller case series, similar associated anomalies were presented and body asymmetry was also found to be the most common finding with a frequency of 43%[2]. In our series, body asymmetry was found in 2 out of 7 cases (28.6%). The circumferences of the affected limb of both cases are smaller compared to the unaffected limb. The frequency is similar to the previous literatures[4]. No other associated anomalies are found in our patients. However, these associations cannot be grouped into certain syndromes. No known mechanism can explain the association between the occurrence of each anomaly and CMTC. Therefore, associated anomalies of CMTC may occur sporadically and may not depend on the extent of areas of skin involvement.

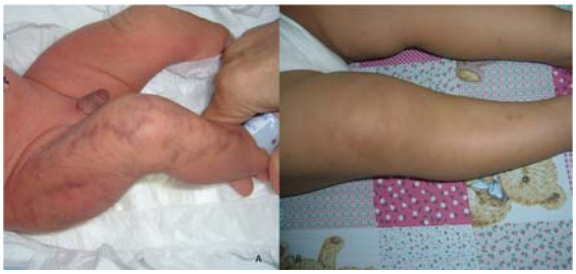

Fig. 1 Clinical presentation of cutis marmorata telangiectatica congenita; (A) at birth and (B) at 2 years old.

Table 1. Patients with cutis marmorata telangiectatica congenita

	Gender	Extent of lesion (% BSA)	Area involvement	Associated anomalies	Limb atrophy/ leg length discrepancy	Improvement of skin lesions with age
Case 1	Female	10	Left leg, trunk	None	No	Yes
Case 2	Female	2	Right leg	None	No	Yes
Case 3	Female	5	Right leg	None	No	Yes
Case 4	Male	10	Right leg	None	No	Yes
Case 5	Female	50	Trunk, arm, forearm, leg	None	Limb atrophy	Yes
Case 6	Male	2	Left leg	None	No	Yes
Case 7	Male	40	Left leg, trunk	None	Limb atrophy	Yes

BSA = body surface area

CMTC requires no specific treatment. Prognosis is good in cases with cutaneous lesions alone or with minor anomalies. Spontaneous resolution usually occurs within 2 years[2,11]. The common associated anomalies such as limb length discrepancy should be followed-up and orthopedic consultation is recommended for consideration of surgical intervention[12]. Upon follow-ups, cutaneous lesions of our patients gradually disappear into complete resolution.

Conclusion

CMTC is a sporadic congenital vascular anomaly with characteristic erythematous reticulated pattern affecting mainly the limbs and trunk. Associated anomalies occur sporadically, which body asymmetry is the most common finding. The prognosis of uncomplicated cases is good. No specific treatment is needed. The skin lesions will gradually disappear.

What is already known on this topic?

CMTC is an uncommon, sporadic congenital vascular anomaly. Additional anomalies have been often reported in association with CMTC.

What this study adds?

The prognosis for uncomplicated cases is excellent.

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Potential conflicts of interest

None.

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Cutis marmorata telangiectatica congenita: ลักษณะทางคลินิกในผู้ป่วย 7 ราย

แพรว วิโรจน์ไพศาลกุล, ฐิตาภรณ์ วรรณประเสริฐ, อรุชา ตรีศิริโชติ

Cutis marmorata telangiectatica congenita เป็นภาวะที่มีความผิดปกติของหลอดเลือดตั้งแต่กำเนิดที่พบได้ประปราย มีลักษณะเป็นผื่นคล้ายตาข่ายเฉพาะที่หรือกระจายทั่วตัวสีแดงและมีหลอดเลือดฝอยขยายตัว ยังไม่ทราบพยาธิกำเนิดที่แน่ชัด มีรายงานพบความผิดปกติอื่นๆ ร่วมด้วย ในบทความฉบับนี้ได้รายงานลักษณะทางคลินิกของผู้ป่วย Cutis marmorata telangiectatica congenita 7 ราย ในโรงพยาบาลศูนย์การแพทย์สมเด็จพระเทพรัตนราชสุดาสยามบรมราชกุมารี คณะแพทยศาสตร์ มหาวิทยาลัยศรีนครินทรวิโรฒ ประเทศไทย ผลการศึกษาพบผู้ป่วยสัดส่วนใกล้เคียงกันทั้งสองเพศ (เพศชาย 3 คน เพศหญิง 4 คน) พบผื่นที่บริเวณขาในผู้ป่วยทั้ง 7 ราย (ร้อยละ 100) ที่บริเวณลำตัว 3 ราย (ร้อยละ 42.9) ที่บริเวณแขน 1 ราย (ร้อยละ 14.3) มีผู้ป่วย 2 ราย (ร้อยละ 28.6) ที่มีผื่นทั้งบริเวณลำตัวและขา ค่าเฉลี่ยของพื้นที่ผิวของผื่นเป็นร้อยละ 17 (2-50) มีผู้ป่วย 2 ราย (ร้อยละ 28.6) บริเวณขาที่มีผื่นมีขนาดของขาไม่เท่ากัน ในผู้ป่วยที่ศึกษา เมื่อติดตามการรักษา พบว่าผู้ป่วยทุกคนมีผื่นจางลง โดยสรุปการพยากรณ์โรคดีในผู้ป่วยที่ไม่มีความซับซ้อนของโรค การให้การรักษายังไม่มีความจำเป็น ผื่นจะค่อยๆ จางลงได้