

# Clinicopathologic discordance: Congenital smooth muscle hamartoma clinically mimics reticulated vascular lesion



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**Key words:** congenital smooth muscle hamartoma; pediatrics.

## INTRODUCTION

First reported in 1969, congenital smooth muscle hamartomas (CSMHs) typically present as skin-colored plaques commonly located on the trunk, buttock, and proximal portion of the extremities in infants. The appearance is variable and may demonstrate hyperpigmentation, hypertrichosis, and/or epidermal changes in the form of perifollicular papules.<sup>1-3</sup> When biopsied, discrete, variably oriented hyperplastic smooth muscle bundles are found in the reticular dermis.<sup>3</sup> CSMHs are thought to originate from the *arrector pili* muscles, blood vessel walls, or from specialized smooth muscle of the genitals, given the normal distribution of smooth muscle.<sup>4</sup> We present a histologically confirmed case of a CSMH masquerading clinically as a vascular plaque.

## CASE REPORT

A healthy, full-term 20-month old girl with no significant past medical history presented for evaluation of a birthmark on the lateral aspect of her left thigh. The lesion was asymptomatic and present since birth, but the parents were concerned due to increasing size and nodularity.

Physical examination revealed an ill-defined, 7.5 cm × 5.0-cm, indurated, coarsely reticulated, violaceous plaque with subcutaneous skin-colored nodules appreciated on palpation located on the lateral aspect of her left thigh (Fig 1). Clinical examination was negative for myokymia, pseudo-Darier sign, limb length discrepancies, and other limb abnormalities.

### Abbreviation used:

CSMH: congenital smooth muscle hamartoma

Based on the clinical history and examination, the differential diagnosis included cutis marmorata telangiectatica congenita, plaque-type glomovenous malformation, tufted angioma, plaque-type non-involuting congenital hemangioma, morphea, and congenital dermatofibrosarcoma protuberans. Two punch biopsies were subsequently collected—one from the reticulated violaceous areas and the other from one of the subcutaneous nodules.

Both biopsies revealed a collection of banal-appearing smooth muscle bundles in the mid-to-deep reticular dermis. There was minimal overlying epidermal acanthosis and a subtle increase in basal layer melanin pigmentation. There was no increase in dermal blood vessels or other vascular features (Fig 2). Muscle-specific actin (Fig 3) and CD34 (Fig 4) immunostains (controls appropriate) were positive and negative in the smooth muscle bundles, respectively. The histologic differential diagnosis included CSMH, evolving Becker's nevus, and less likely multiple piloleiomyomas. Based on the clinic-pathologic correlation, the patient was diagnosed with a CSMH. Parental consent was provided in support of this case report.

## DISCUSSION

When biopsied, CSMHs will demonstrate discrete, variably oriented, hyperplastic smooth muscle

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Funding sources: None.

IRB approval status: IRB exempt.

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JAAD Case Reports 2022;26:9-12.

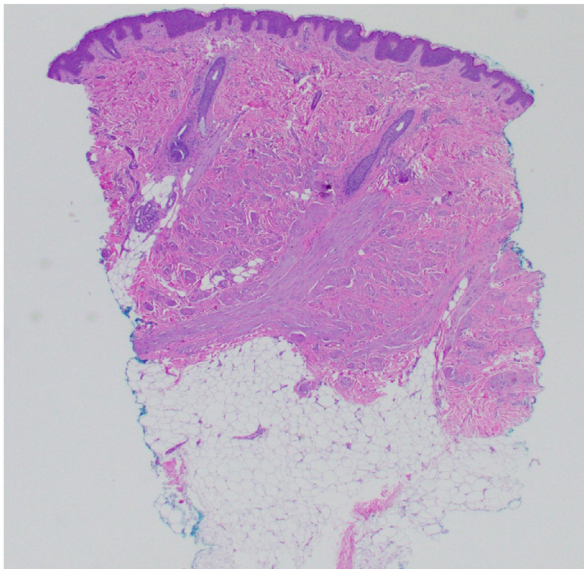
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<https://doi.org/10.1016/j.jcdr.2022.05.036>



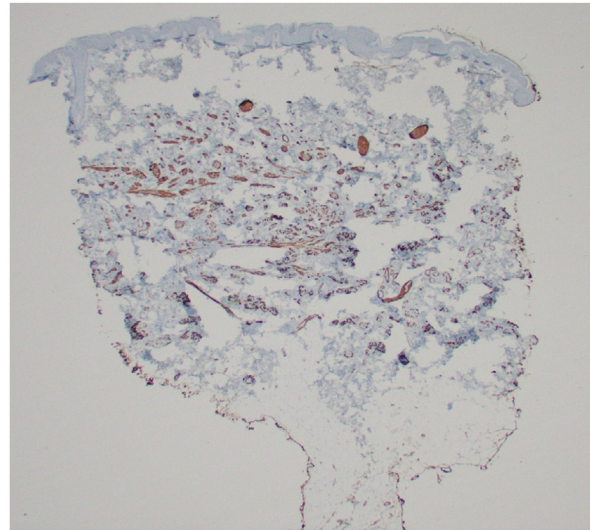
**Fig 1.** Congenital smooth muscle hamartoma. Coarsely reticulated violaceous plaque on the lateral aspect of the left thigh.



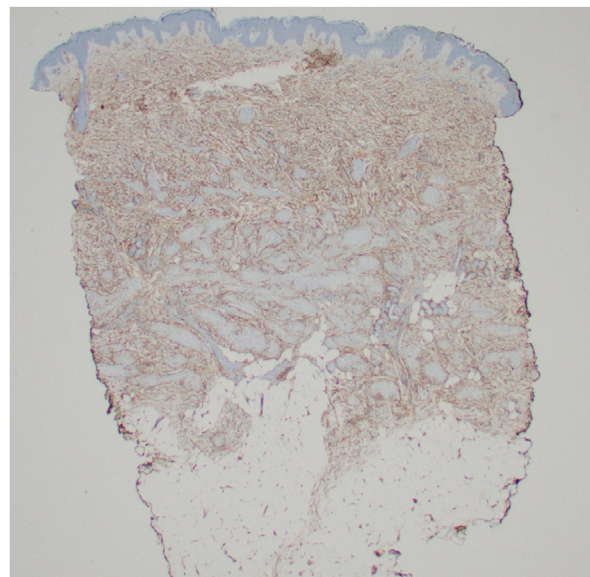
**Fig 2.** Congenital smooth muscle hamartoma. Banal-appearing smooth muscle bundles in the mid-to-deep reticular dermis with overlying epidermal acanthosis and subtle basal layer melanin pigmentation (hematoxylin-eosin staining; original magnification:  $\times 20$ ).

bundles in the reticular dermis.<sup>3</sup> Based on the normal distribution of cutaneous smooth muscle, CSMHs most likely originate from the *arrector pili* muscles, blood vessel walls, or from specialized smooth muscle of the genitals.<sup>4</sup>

Included in the histologic differential diagnosis of CSMH is a Becker nevus, and it is debated whether they are distinct entities or 2 ends of a phenotypic spectrum.<sup>5</sup> Becker nevi typically present peripubertally on the chest, back, or scapula of young men, whereas CSMHs present at the time of birth or within the first years of life. Both entities can develop overlying epidermal change; however, Becker nevi are more likely to exhibit hyperpigmentation, hypertrichosis, and epidermal changes.<sup>5</sup>



**Fig 3.** Congenital smooth muscle hamartoma. (positive muscle-specific actin immunostain; original magnification:  $\times 20$ ).



**Fig 4.** Congenital smooth muscle hamartoma. (negative CD34 immunostain; original magnification:  $\times 20$ ).

Our case is unique because it presented as a violaceous, coarsely reticulated plaque with induration—a clinical presentation that is more indicative of a vascular malformation than a CSMH or a Becker nevus. In a previous report, Fernandez-Flores et al<sup>6</sup> described a similarly presenting erythematous, reticulated macular lesion on the right thigh of a 26-year old woman. When biopsied, however, the histologic findings demonstrated 2 distinct intermingling clinical entities—CSMH and nevus flammeus.<sup>6</sup> In our case, only a CSMH was present on histology. The

**Table I.** Comparison of cases of reticulated, vascular-appearing congenital smooth hamartomas

Case report	Present study	Fernandez-Flores et al <sup>6</sup>	Vivehanantha et al <sup>8</sup>	Knöpfel et al <sup>9</sup>
Age	20 mo	26 y	3 y	2 mo
Sex	F	F	F	M
Location	Lateral aspect of the left thigh	Right thigh extending to the right buttock	Right buttock	Right lower limb
Clinical findings	Ill-defined, 7.5 × 5.0 cm, indurated, coarsely reticulated, violaceous plaque with subcutaneous skin-colored nodules appreciated on palpation. Negative for myokymia, pseudo-Darier's sign, and limb abnormalities.	30-cm erythematous, telangiectatic patch with 1-cm ulceration without elevation or hardened borders. There were no other associated abnormalities. Negative for myokymia, pseudo-Darier sign, and limb abnormalities.	Atrophic, reticulate, reddish-purple plaque in a linear distribution without any functional or developmental abnormality of the leg. Negative for myokymia, pseudo-Darier sign, and limb abnormalities.	White-to-erythematous reticulated atrophic patch to plaque in a linear distribution on the right lower limb. Positive for myokymia and transition blanching with stroking. Negative for limb abnormalities.
Histologic findings	Smooth muscle bundles in the mid-to-deep reticular dermis. Normal amount of dermal blood vessels and other vascular features. Muscle-specific actin-positive and CD34-negative.	Ulcer with abundant smooth muscle bundles in the reticular dermis and subcutis. Smooth muscle actin-, desmin-, and caldesmon-positive. Intermingled with the smooth muscle bundles were small capillary vessels and venules, which were positive for CD31, CD34, and factor VIII, whereas negative for D2-40.	Smooth muscle bundles were arranged parallel to the epidermis and were interspersed between normal adnexa, extending from the reticular dermis to the subcutis. Actin-, desmin-, and caldesmon-positive.	Numerous smooth muscle bundles in the dermis and superficial subcutis. Smooth muscle actin- and calponin-positive.
Diagnosis	Smooth muscle hamartoma	Smooth muscle hamartoma and nevus flammeus	Smooth muscle hamartoma	Smooth muscle hamartoma

reticulate vascular appearance of the CSMH may be caused by impedance in superficial blood flow from smooth muscle contraction of the hamartoma. With contraction of the hamartoma, the blood flow is impeded, and a larger content of deoxygenated hemoglobin can pool superficially, resulting in the reticulate vascular appearance because of the architecture of the cutaneous vascular network.<sup>7</sup>

To our knowledge, there have been only a few similar clinical presentations in children.<sup>8,9</sup> In these cases, the CSMHs had been present since birth and presented as a vascular-appearing lesion on the right thigh rather than on the left.<sup>8,9</sup> Table I provides a side-by-side comparison of the known cases.<sup>6,8,9</sup> By presenting this case, our hope is that more dermatologists and pathologists will consider CSMH in their differential-diagnostic considerations when confronted with a similar presentation.

**Conflicts of interest**

None disclosed.

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