Three Cases of Congenital Smooth Muscle Hamartomas

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Congenital smooth muscle hamartomas (CSMHs) usually appear at birth as skin-colored or slightly hyperpigmented patches or plaques on the trunk or extremities and are often covered by and increased amount of vellus hair. Histopathologically, it represents a proliferation of smooth muscle bundles within the dermis[1,2].

We herein report on three cases of localized CSMHs; Case 1 was a 2-month-old boy who presented with a skin-colored, 6 × 5 cm-sized plaque with folds, hypertrichosis and pseudo-Darier's sign on the face, an unusual site. But there was no hyperpigmentation. Case 2 and 3 were both 1-month-old boys who presented with typical hyperpigmented, hairy plaques on the lateral aspect of the left thigh. (Ann Dermatol (Seoul) 19(1) 38~42, 2007)

Key Words: Congenital smooth muscle hamartoma, Hypertrichosis, Pseudo-Darier's sign

INTRODUCTION

Congenital smooth muscle hamartoma (CSMH) is a rare cutaneous abnormality which is characterized by a benign proliferation of intersecting dermal smooth muscle bundles that may or may not be associated with hair follicles. It presents clinically as a congenital hairy patch or plaque, with or without hyperpigmentation, and it is usually seen as a single lesion on the trunk or proximal extremities. Gerdsen et al.[3] have proposed a clinical classification of CSMH into four types; classical localized CSMH, patchy follicular variant, multiple CSMH and diffuse CSMH.

To date, 5 cases of congenital smooth muscle hamartomas have been reported in the Korean dermatologic literature[4-8] (Table 1), but only 1 case has reported classic localized CSMH. We herein report on three cases of classic localized CSMHs; Case 1 was a 2-month-old boy who presented with a large, thick-haired plaque with skin folds on the right infraorbital area, which is not predilection site of CSMH. Case 2 was a 1-month-old boy who presented with a hairy, hyperpigmented patch consisting of three small lesions on the lateral aspect of the left thigh. Case 3 was also a 1-month-old boy who presented with a hyperpigmented, slightly atrophic plaque with hypertrichosis on the left outer thigh.

Received April 26, 2006
Accepted for publication January 22, 2007

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CASE REPORT

Case 1

A 2-month-old boy presented with a folded skin lesion on the right infraorbital area which had been present since birth. Physical examination revealed a circumscribed, hairy, flesh-colored, 6 × 5 cm sized plaque with skin folds (Fig. 1). The initial diagnosis was congenital melanocytic nevus. But a pseudo-Darier's sign was elicited upon rubbing the lesion. His medical history was uneventful and there was no family history of similar lesions. Laboratory studies including a complete blood cell count, blood chemistry, urinalysis, chest-X-ray and EKG were within normal limits or they were negative. He had

Table 1. Summary of Reported Cases of Congenital Smooth Muscle Hamartoma in the Korean Literature

Year	Case	Journal	Age/sex	Location	Type	Hyper trichosis	Pseudo Darier's sign
1987	Jung et al.	Korean J Dermatol	F/5 yr	Left elbow	Classic localized	×	×
1991	Oh et al.	Korean J Dermatol	M/7 yr	Back and four extremities	Multiple	○	○
1998	Lee et al.	Korean J Dermatol	M/4 mo	Both lower extremities	Multiple	○	○
2000	Kwon et al.	Ann Dermatol	F/10 yr	Right upper arm	Patchy follicular variant	×	×
2002	Choi et al.	Korean J Dermatol	F/8 mo	Lumbosacral area	Classic localized	○	○
Case 1			M/2 mo	Right infraorbital area	Classic localized(?)	○	○
Case 2			M/1 mo	Left thigh	Multiple	○	○
Case 3			M/1 mo	Left thigh	Classic localized	○	○

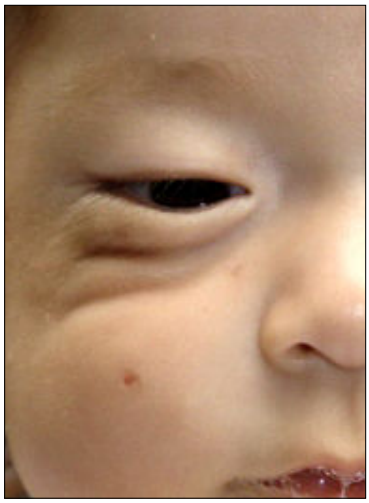

Fig. 1. Thick, hairy, 6 × 5 cm sized plaque with skin folds on the infraorbital area.

no other developmental anomaly and his height and body weight were within the normal range for his age group. Multiple biopsies were performed on the lesion.

Histopathologically, numerous and various sized bundles of smooth muscle were scattered throughout the dermis, and they were mostly unattached to the hair follicles (Fig. 2A). Smooth muscle actin staining confirmed the presence of smooth-muscle bundles (Fig. 2B). A diagnosis of congenital smooth muscle hamartoma was made, and the patient underwent a partial resection of the folds and was treated by laser for hair removal.

Case 2

A 1-month-old boy presented with a large hyperpigmented patch on the left thigh which had been present since birth. Examination revealed a circumscribed patch with hyperpigmentation and hypertrichosis on the lateral aspect of the left thigh, which consisted of three small, 3 × 3 cm sized lesions (Fig. 3). A pseudo-Darier's sign was positive. There was no significant medical history and no family history of similar lesions. The histopathologic examination revealed typical findings of CSMH. When he was seen about 1 year later, there was no specific change except for an enlarged size plus loss of the patch reaction to friction.

Case 3

A 1-month-old boy presented with a pigmented, slightly atrophic plaque on the left thigh which had

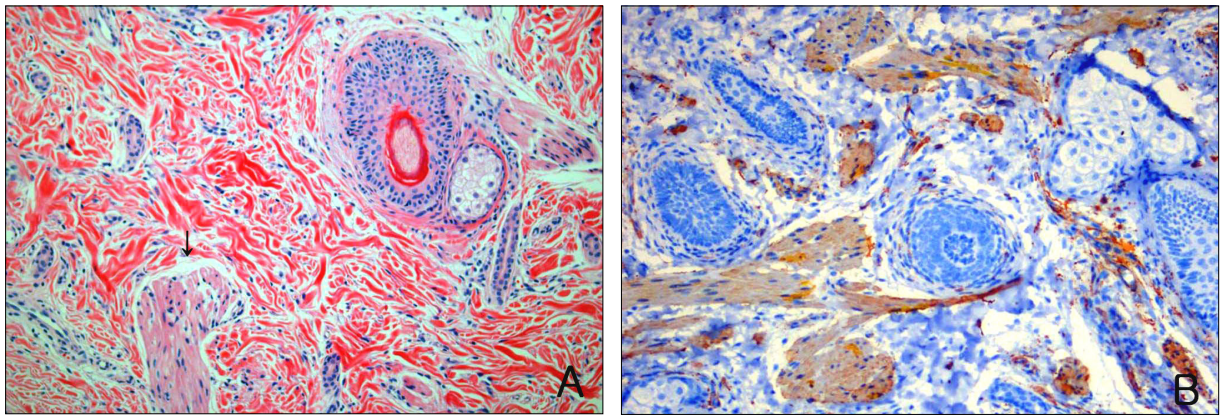

Fig. 2. Case 1. (A) Numerous, well-defined bundles of smooth muscle fibers (arrow) in the dermis (H&E, × 100). (B) Immunohistochemical stain shows numerous thick, red-colored, smooth muscle bundles (Smooth muscle actin stain, × 100).

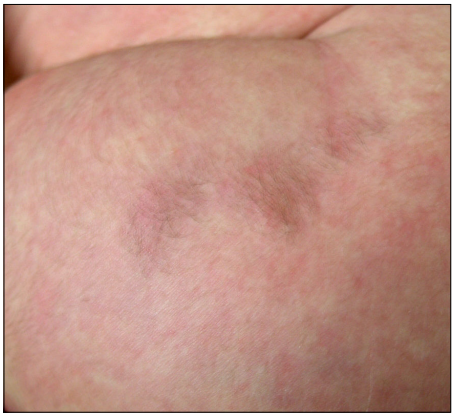

Fig. 3. Case 2. Three small, hairy and hyperpigmented lesions on the lateral aspect of the left thigh.

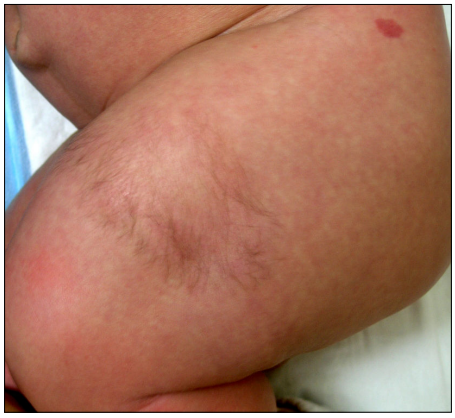

Fig. 4. Case 3. A large, hairy, pigmented, slightly-depressed plaque on the lateral aspect of the left thigh.

been present since birth. The examination revealed a 6 × 5 cm-sized, slightly-atrophic, hairy, pigmented plaque on the left thigh (Fig. 4). A pseudo-Darier's sign could not be elicited. The patient was otherwise healthy and had normal growth parameters. The clinical differential diagnosis included CSMH, Becker's nevus and congenital melanocytic nevus. Histopathologically, numerous and various-sized bundles of smooth muscle were scattered throughout the dermis. A proliferation of red- colored smooth muscle bundles in the dermis was noted by smooth muscle actin staining. A diagnosis of congenital smooth muscle hamartoma was made and the patient received no further therapy.

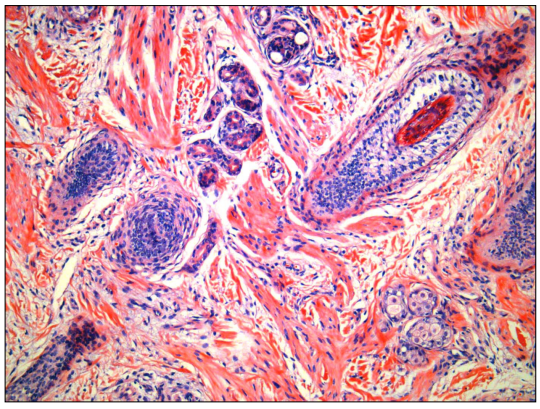

Fig. 5. Case 3. Numerous, well-defined bundles of smooth muscle fibers extending in various directions (H&E, × 100).

DISCUSSION

Congenital smooth muscle hamartomas are benign proliferations of smooth muscle bundles within the dermis. They can be congenital or acquired, but most cases are congenital lesions. CSMHs develop from the excessive production of pilar smooth muscle (arrectores pilorum) at the time of maturation of the mesoderm during fetal development[9-11], and acquired smooth muscle hamartomas have been reported most frequently in association with a Becker's nevus[9,12,13]. Although some authors[2,14,15] have tried to relate the congenital and acquired types of smooth muscle hamartomas, it is still preferable to consider the congenital form as a separate and distinct clinopathologic entity[14,15]. CSMH has been regarded as excessively rare, but this once rare lesion is now being seen with an increased frequency[1,12,16]. The estimated prevalence is about 1:2,600 to 1:2,700 live births[1,14] with a slight male predominance[1,2].

Gerdsen et al.3 have proposed a clinical classification of CSMH into four types. Type 1, classical localized CSMH is the most common, and exhibits a hairy, hyperpigmented plaque with a positive pseudo-Darier's sign. Type 2, patchy follicular variant, is a less common sub-entity that is characterized by a circumscribed annular patch with multiple follicular papules. There is no hyperpigmentation and the hair pattern changes are less prominent. Type 3, multiple CSMH, is associated with underlying nevus lipomatosus. The 4th type is a diffuse CSMH. Our cases are all classic localized CSMHs, although the occurrence on the face is unusual.

Most CSMHs usually present as a single lesion that is a darkly-hyperpigmented or flesh-colored patch or plaque, and they are commonly found on the trunk, particularly in the lumbosacral region, but other sites including the chest, extremities, abdomen, upper back and rarely the face like in case 1 have also been reported[1,14]. Although it has been occasionally described as a patch with perifollicular papules without prominent hair[11], hypertrichosis or prominent overlying hair is usually present[1,14], whose density is unchanged and the diameter and length[14] is increased. The pseudo-Darier's sign in CSMH is a transient induration of the skin that appears when the lesion is rubbed and the induration is due to the stimulation of the arrector pili muscles. A positive pseudo-Darier's sign is present in up to 80% of cases, and this may be helpful in the differential diagnosis of the disease[1,18].

Histopathologically, numerous thick, well-defined bundles of smooth muscle fibers are scattered throughout the dermis and they extend in various directions. The overlying epidermis often shows increased basal pigmentation[19]. In addition to the marked hyperpigmentation, 50% of the patients have hyperkeratosis and papillomatosis that is reminiscent of Becker's melanosis[2].

The clinical differential diagnosis should include Becker's melanosis (Becker's pigmented hairy nevus), congenital melanocytic nevus, piloleiomyoma, epidermal nevus, connective tissue nevus, and folded skin with lipomatous nevus[11,19].

CSMH may be grossly similar to Becker's melanosis. The CSMHs in 50% of patients have similar epidermal changes to those seen in Becker's nevus, such as hyperkeratosis, papillomatosis and hyperpigmentation. Therefore, there are certain difficulties in differentiation of the two. Yet CSMH appears at birth, and there is prominent vellus hair in cases with hypertrichosis. Moreover the hyperpigmentation, skin induration and hypertrichosis diminish over time. In contrast, Becker's melanosis appears at childhood or puberty, and hyperpigmentation always precedes the hypertrichosis; these lesions contain thick terminal hairs and the aforementioned signs tend to increase[1,2,18]. However, since similar smooth muscle proliferations may be also observed in Becker's melanosis[1,15,17], it has been suggested that CSMH could be at one end of a spectrum of dermal smooth muscle proliferative disorders, and that Becker's melanosis could be at the opposite end. Although this proposed continuum is an attractive attempt to relate these two types of lesions, it is probably preferable to consider them as separate and distinct clinicopathologic entities. A positive pseudo-Darier's sign and the presence of smooth muscle hyperplasia in the dermis can be helpful in distinguishing CSMH from congenital melanocytic nevus (CMN). The differentiation between two disorders is most important especially prior to surgical treatment. In case 1, the clinical diagnosis at the time of the first visit was CMN and multiple biopsies were performed to delineate the boundary of the lesion for definite surgical excision. CSMH does not appear to have any malignant potential[1]; therefore, unnecessary surgical treatment is of no therapeutic value other than for cosmetic reason, and it may result

in permanent deformity[1,2,18].

Folded CSMH, also called "Michelin tire man", is caused by underlying nevus lipomatosus and appears as excessive symmetric circumferential folds of skin affecting the neck, forearms, or lower extremities. Although it may coexist with smooth muscle hamartoma, folded skin has also been observed in overlying smooth muscle hamartoma just as in our patient (case 1). Histopathologic examination of the folded skin lesion is helpful when differentiating between the two conditions.

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