Case report

Neuro-folliculo-sebaceous cystic hamartoma is a unique entity

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Folliculosebaceous cystic hamartoma (FSCH) is a distinct type of cutaneous hamartoma of pilosebaceous origin that usually occurs on the face. For FSCH, other parts have been reported such as the genital area, and the trunk. A 50-year-old woman presented with an asymptomatic dome-shaped scalp nodule. The clinical diagnosis was pilar cyst or tumor. Histopathological assessment showed FSCH with absolute neural component as the only mesenchymal stroma, leading to the diagnosis of folliculosebaceous cystic neural hamartoma. To the best of our knowledge, absolute neural stroma in FSCH has not been reported previously in the literature.

Key words: hamartoma, folliculosebaceous cystic hamartoma, neuro-folliculo-sebaceous cystic hamartoma.

Introduction

Folliculosebaceous cystic hamartoma (FSCH) is an uncommon, rarely reported non-neoplastic lesion which arises from the hair follicle [1, 2]. Folliculosebaceous cystic hamartoma was first described by Kimura *et al.* [1] in 1991 as a rare cutaneous hamartoma composed mainly of follicular, sebaceous and mesenchymal elements [1, 2]. Folliculosebaceous cystic hamartoma does not have peculiar clinical features [3, 4]. Clinicians always suggest cyst-like cutaneous lesion, soft tissue neoplasm, sebaceous gland hyperplasia or melanocytic nevus [3, 4].

It always presents as a painless, slowly growing single firm papule or nodule with a predilection for the central face [3, 4]. Other parts such as the vulva, the nipple, the scrotum, the ear or scalp have been reported for FSCH [5, 9].

Case report

A healthy 50-year-old woman presented with a 3-month history of a painless, non-itchy, firm, skin-colored, scalp dome-shaped nodule that gradually increased in size, with a recent focal cystic sensation. Clinical diagnosis included pilar cyst or other tumors with cystic changes. Excisional biopsy of the nodule was performed. Gross morphology showed a dome-like tissue piece measuring 0.7 × 0.5 × 0.5 cm (Fig. 1, inset) covered by intact skin firm in consistency. The cut section was greyish white in color. The hematoxylin and eosin (HE) stained sections showed epidermis with unremarkable changes. The dermis displayed a delineated non-capsulated lesion composed of infundibular structure with a central cyst attached to sebaceous lobules via sebaceous ducts and filled with orthokeratotic keratin (Fig. 1). The pilosebaceous unit was embedded in a mesenchymal stroma composed mainly of neural tissues and few collagen bundles (Fig. 2). Also rudimentary hair structure and a dilated sweat gland with apocrine change were noted. There were no mature adipose tissue, blood vessels or inflammatory cells in the mesenchymal stroma. Clefts were noted between the mesenchymal epithelial units of the lesion and the rest of the dermal stroma. Immunohistochemical (IHC) confirmation for absolute neural mesenchymal stroma was done, as the neural elements displayed

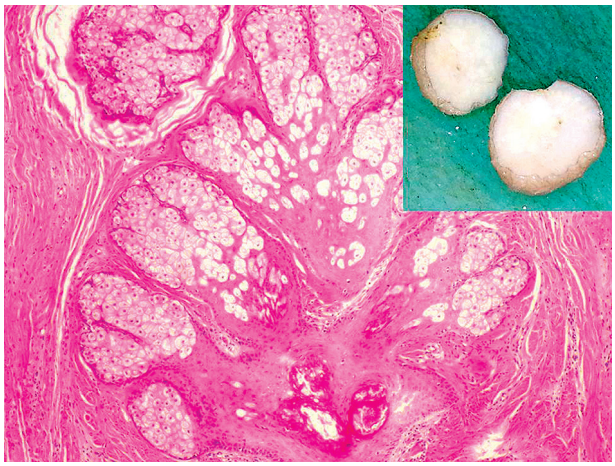

Fig. 1. Non-capsulated dermal lesion composed of infundibular structure with central cyst attached to sebaceous lobules via sebaceous ducts and filled with orthokeratotic keratin. Clefts are visible between the mesenchymal epithelial units and the surrounding altered mesenchymal stroma and at the periphery between the altered stroma and the adjacent compressed dermal fibrous tissue (HE 100×). Inset: Gross morphology showed dome-like tissue piece covered by intact skin firm in consistency. Cut section was greyish white in color

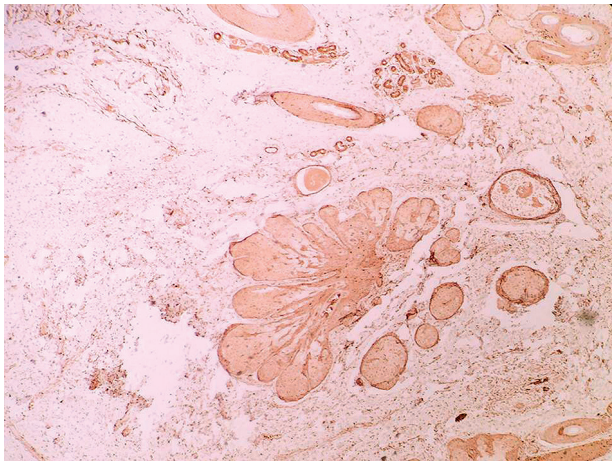

Fig. 3. Scanning view of FSCH shows cystic follicles surrounded by spindle mesenchymal stroma (S100 40X)

S100 positivity (Fig. 3) and smooth muscle actin (SMA) negativity (Fig. 4).

Based on these findings we proposed a new nomenclature of neuro-folliculo-sebaceous cystic hamartoma for this peculiar entity of FSCH with absolute neural mesenchymal stroma and absence of other mesenchymal compositions.

Discussion

Cutaneous hamartomas are defined as tumor-like lesions showing abnormal growth of resident normal epithelial and mesenchymal elements [10]. In these groups of malformation, one component can predominate over the other [10]. A predominant mesenchymal stroma was observed in fibro-folliculoma (FF),

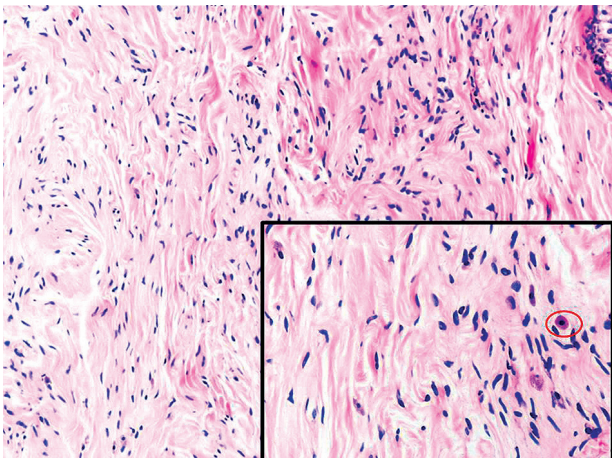

Fig. 2. Haphazardly arranged neural mesenchymal stroma reminiscent of mature peripheral nerve components. Inset: High power view of spindle-shaped cells with occasional curvilinear or shredded carrot nuclei and mast cell (circle) (HE 100× and 400× for inset)

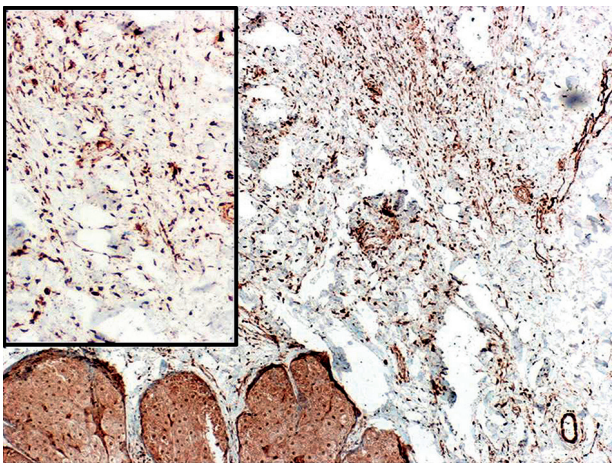

Fig. 4. The stromal spindle-shaped cells show strong diffuse positivity for S100. Inset: Closer view (IHC for S100 100× and inset 200×)

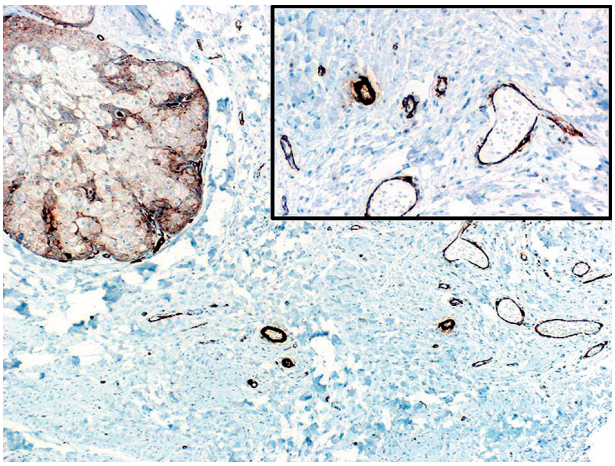

Fig. 5. The stromal spindle-shaped cells show diffuse negativity for SMA. Inset: Closer view (IHC for SMA 100× and inset 200×)

trichodiscoma (TD), and trichogenic myxoma (TM) [10]. The mesenchymal components that are reported in these hamartomatous lesions include fibroblastic or fibrous with mucinous components in TD and FF or a pure mucinous stroma in TM [10]. However, an equal proportion of epithelial and stromal structures has been demonstrated in tricho-folliculoma (TF), fibrous papule of skin (FP), neuro-follicular hamartoma (NFH), and FSCH [10]. Other kinds of hamartoma are composed of either well-developed terminal or vellus hair follicles in TF and FP or disturbed pilo-sebaceous units in TF, NFH and FSCH, intermixed with collagenous, fibroblastic, or sclerotic stroma [10].

Cutaneous hamartomatous lesions with a mature peripheral nerve component have been reported in the literature in a case of mature port wine stain [11, 12], NFH [13], and FSCH [14] as a component among other hamartomatous elements [10].

Only two previous reports mentioned the presence of neural proliferation in the stroma of FSCH [15, 16]. One report demonstrated presence of aberrant nerve bundles lacking the normal IHC expression of neuropeptides in the stroma of FSCH [15]. The second report found neural proliferation in the stroma of a case of FSCH without confirmation by IHC or detailed description [16].

The proposed nomenclature for this lesion with absolute neural mesenchymal is neuro-folliculo-sebaceous cystic hamartoma (NFSCH). Folliculosebaceous cystic hamartoma could be considered as a prototype for various hamartomatous lesions, and their nomenclature is based on the components of mesenchymal stroma. The classic FSCH has various mesenchymal components in different proportions. However, FSCH with absolute mesenchymal composition perhaps could have the peculiar name of the mesenchymal component.

Suarez-Peñaranda and associates suggested that presence of smooth muscle or neural elements is exceptional in FSCH [17]. Smooth muscle was the only mesenchymal component that was reported in one case, leading to the diagnosis of folliculosebaceous smooth muscle hamartoma [10].

The previous mentioned reports support our proposal of NFSCH as a subtype of FSCH with absolute neural stroma.

The differential diagnosis of NFSCH is NFH. Neuro-folliculo-sebaceous cystic hamartoma is similar to NFH in composition, but there are several histological differences between these two entities. The haphazardly arranged cystic infundibular structures are a characteristic finding of FSCH and a distinguishing feature. However, NFH lacks the haphazard cystic infundibular structures.

Two previous reports mentioned neural proliferation in the stroma of FSCH. However, our report described absolute presence of neural elements that is confirmed by IHC. To the best of our knowledge, absolute neural mesenchymal stroma in FSCH has not been reported previously in the literature. In conclusion, we propose the nomenclature for this rare hamartomatous lesion with absolute neural mesenchymal stroma to be NFSCH. Neuro-folliculo-sebaceous cystic hamartoma is a subtype of FSCH with absolute neural mesenchymal stroma.

The authors declare no conflict of interest.

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