An uncommon folliculosebaceous cystic hamartoma on the lower extremity

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BACKGROUND: Folliculosebaceous cystic hamartomas (FSCH) are a recently recognized cutaneous hamartoma comprised of follicular, sebaceous and mesenchymal elements. Only nine cases have been previously reported. The authors present a case of FSCH on the lower extremity in addition to a literature review.
OBJECTIVE: To further characterize the clinical, histological and immunohistological features of FSCH.
METHODS: The clinical, histological and immunohistological features of FSCH were evaluated and the literature reviewed.
RESULTS: This lesion uncommonly presents as exophytic papules or nodules on the leg, and most commonly presents on the central part of the face. Clinical differential diagnoses included nevus, neurofibroma and adnexal tumour. The diagnostic microscopic features of infundibular cystic structures with attached sebaceous glands and characteristic stroma were present in all lesions. Many of the stromal spindle cells were CD34-positive.
CONCLUSION: The FSCH is a recently recognized hamartoma comprised of follicular, sebaceous and mesenchymal elements. Although reported only rarely, its incidence is likely much higher. In the present patient, the FSCH was located in femoral region, which is very rare.

Key Words: *Exophytic papules; Folliculosebaceous cystic hamartoma; Hamartoma; Immunohistological features; Nodules; Sebaceous glands*

Un hamartome kystique folliculosébacé rare du membre inférieur

HISTORIQUE : Les hamartomes kystiques folliculosébacés (HKFS) sont des hamartomes cutanés récemment catégorisés qui sont composés d'éléments folliculaires, sébacés et mésenchymateux. Seulement neuf cas ont été déclarés jusqu'à présent. Les auteurs présentent un cas d'HKFS de l'extrémité inférieure conjointement avec une analyse bibliographique.
OBJECTIF : Mieux caractériser les aspects cliniques, histologiques et immunohistologiques de l'HKFS.
MÉTHODOLOGIE : On a évalué les aspects cliniques, histologiques et immunohistologiques de l'HKFS et procédé à une analyse bibliographique.
RÉSULTATS : Dans de rares cas, cette lésion se manifeste sous forme de papule ou de nodule exophytique de la jambe et, plus souvent, du milieu du visage. Le diagnostic différentiel clinique inclut un nævus, un neurofibrome et une tumeur des annexes cutanées. Le diagnostic microscopique révèle des structures kystiques infundibulaires accompagnées de glandes sébacées et d'un stroma caractéristique sur toutes les lésions. Bon nombre des cellules fusiformes du stroma étaient positives au CD34.
CONCLUSION : L'HKFS est un hamartome cutané récemment catégorisé composé d'éléments folliculaires, sébacés et mésenchymateux. Bien qu'on le déclare rarement, son incidence est probablement beaucoup plus élevée. Chez le patient présenté, l'HKFS se situait dans la région fémorale, ce qui est très rare.

Folliculosebaceous cystic hamartomas (FSCH) are unique, uncommon cutaneous entities that are considered a hamartoma on the basis that they are composed of multiple tissue elements, including ectodermal and mesodermal components. The epithelial components include adnexal and folliculosebaceous cystic proliferations, while the mesenchymal component exhibits variable fibroplasia, and vascular and adipose tissue proliferations. The name 'folliculosebaceous cystic hamartoma' describes the major attributes of the lesion, which include a follicular cystic structure with multiple attached sebaceous lobules, and stromal mesenchymal changes, such as lamellar fibroplasia, and increased adipose and vascular tissue. Since the first descriptions by Kimura et al (1), who described five cases, fewer than 20 cases have been reported (2,3).

Clinically, FSCH lack distinctive features, and the clinical diagnosis in all reported cases has included diagnoses other than FSCH, such as intradermal nevi, sebaceous hyperplasia, basal cell carcinoma, lipomas, dermoid cysts of the nose and neurofibromas. However, cases diagnosed clinically as dermoid cysts of nose did not show a central dimple in the lesion or hairs emerging from it, features characteristic of dermoid cysts of nose (4).

CASE PRESENTATION

A two-year-old girl was brought by her parents to the plastic surgery clinic at Numune State Hospital, Erzurum, Turkey, because of a mass on her leg (Figure 1). Because the lesion had its onset six months after birth, the authors considered it to be a congenital variant. Sebaceous material could be expressed from the papules, and a few papules had a central terminal hair. Compared with the surrounding skin, there was a decrease in the density of terminal hairs (7.6 cm in diameter). For cosmetic reasons, her family requested that this mass be removed. The lesion was totally excised, with 0.5 cm margins. The defect was closed by cutaneous advancement flaps with wide undermining. Histological examination of a lobule demonstrated an

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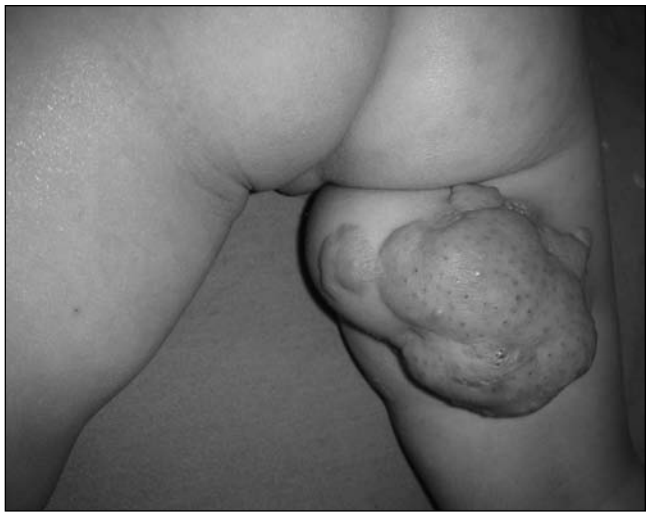

Figure 1) *Preoperative view of the leg of patient with a folliculosebaceous cystic hamartoma*

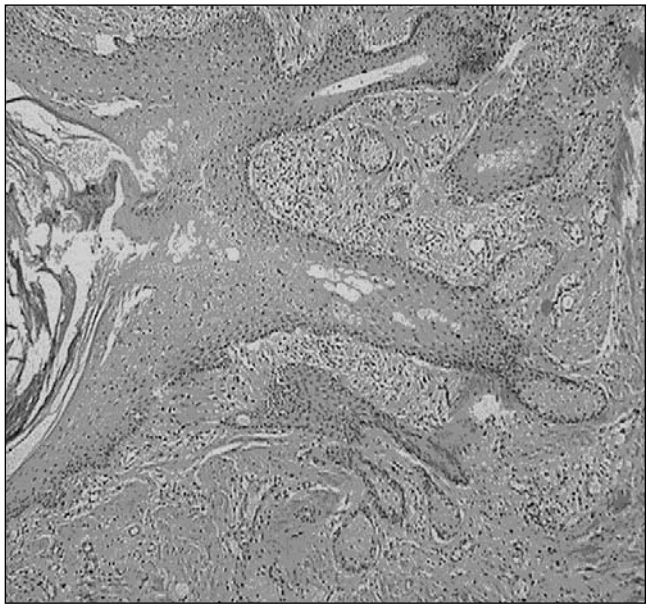

Figure 2) *Cystic dilated follicular structures consisting of stratified squamous epithelium that contains infundibular-type keratinization and mature sebaceous lobules. These cystic dilated follicular structures were connected with the overlying epidermis through the sebaceous ducts (hematoxylin and eosin stain, original magnification ×40)*

irregularly shaped cystic structure within the reticular dermis. It was lined by stratified squamous epithelium that resembled that of the follicular infundibulum, and contained laminated orthokeratotic keratin and sebum (Figure 2). The cyst was connected with the overlying epidermis through a single pore, resembling that of a normal hair follicle. Hair shafts, however, were not present. Radiating from the cyst were multiple sebaceous structures. In some histological sections, lobules containing normally maturing sebocytes were noted to be connected to the central cystic space via sebaceous ducts. The fibrous stroma investing the tumour was composed of thin collagen bundles, and normal-appearing capillaries and venules.

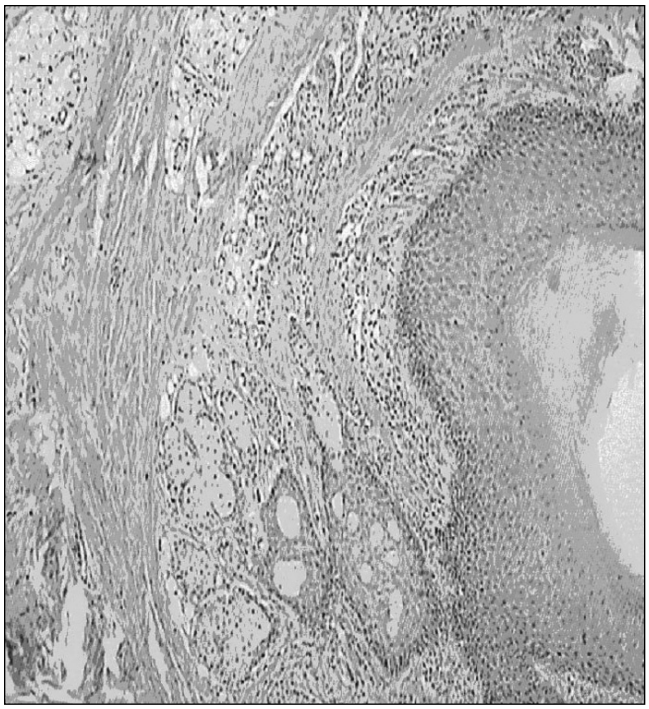

Figure 3) *Cleft formation is seen between the cystic dilated folliculosebaceous unit and the stroma (hematoxylin and eosin stain, original magnification ×40)*

There were clefts both within the stroma and between the stroma and the cystic tissue, but no aberrant or rudimentary vellus hair follicles, apocrine glands or structures associated with the inferior segments of hair follicles were observed (Figure 3). There were no connections noted between the sebaceous lobules and the overlying epidermis.

DISCUSSION

The major entity in the differential diagnosis of FSCH is sebaceous trichofolliculoma (5), and before 1991, there are examples of FSCH that were labelled as a sebaceous trichofolliculomas (6-8). Both tumours occur most commonly on the nose, but in contrast to papulonodular configuration of FSCH, sebaceous trichofolliculomas present as depressed ostia that often contain terminal or vellus hairs (9-12). The histological correlate is a large central pore lined by a squamous epithelium and surrounded by multiple rudimentary sebaceous follicles. When the histological features of FSCH and sebaceous trichofolliculomas are compared, several differences are noted. A connection to the surface epithelium can be seen in both structures, but it is observed more frequently in sebaceous trichofolliculomas. Multiple refractile hair shafts within the dilated follicular structure is a finding characteristic of sebaceous trichofolliculomas but not FSCH (1-5,9,13-16). The stroma of an FSCH contains thin bundles of collagen, adipocytes, CD34+ spindle cells (Figure 4), an increased number of capillaries and venules and occasionally, a proliferation of nerve fibres (1,4,13). This mesenchymal component may even represent a considerable portion of the hamartoma (14). Such stroma is not found in sebaceous trichofolliculomas (17). In 1990, Nomura and Hata (8) described a genital variant of a sebaceous trichofolliculoma in 22-year-old man. In this

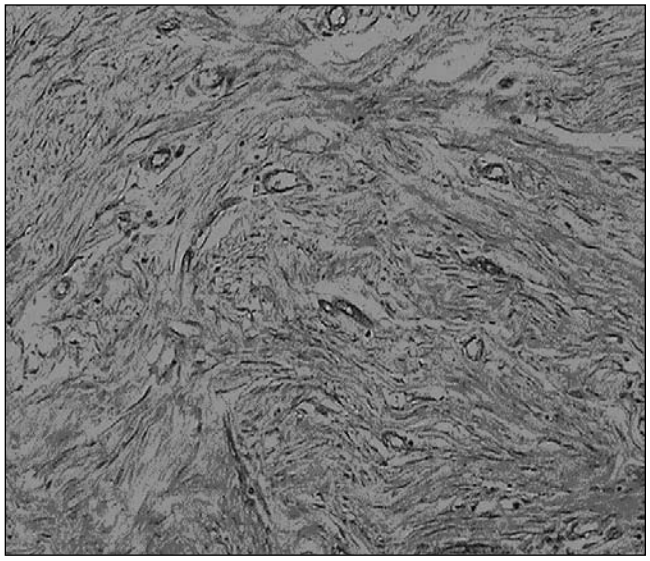

Figure 4) *Spindle stromal cells and immunoreactivity on vascular walls stained with CD34 (original magnification ×100)*

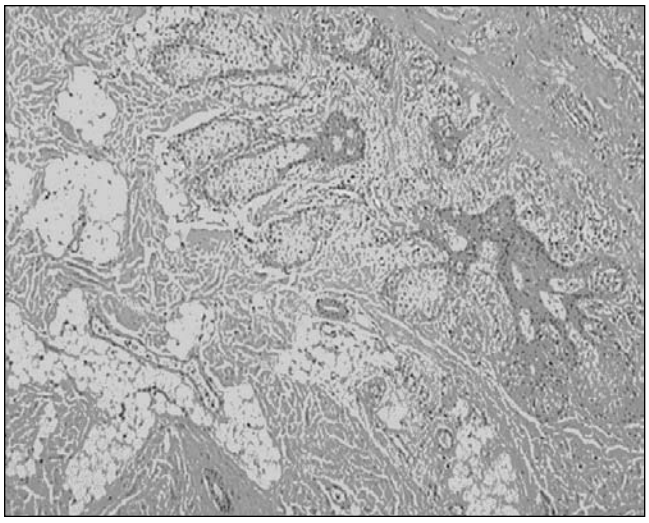

Figure 5) *Mature fat cell groups and fibrillar rough collagen elements around the folliculosebaceous structures (hematoxylin and eosin stain, original magnification ×40)*

patient, the lesion was present only on the lower part of the body, similar to our patient. Multiple soft, skin-coloured papules and nodules were present on the scrotum and penis, and several had coalesced into multilobulated plaques.

In our patient, centrally, nodules were generally yellow in colour, and in some, a hair was present. Histologically, irregularly shaped cystic structures were seen in the reticular dermis, some of which were connected to the surface of the skin. They were lined by squamous epithelium and filled with laminated orthokeratotic keratin material. Multiple sebaceous follicles were noted to radiate from these cystic structures (Figure 5). In the photomicrographs, the stroma is noted to contain clefts as well as adipocytes.

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