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Solitary asymptomatic lobular perianal nodule on an infant

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Abstract

Folliculosebaceous cystic hamartoma (FSCH) is a benign lesion that presents as a solitary papule or nodule that typically affects the face and scalp of adults. A few reports have observed a congenital presentation. We present an infant boy exhibiting a tumor with overlap features between an FSCH and fibrofolliculoma in the perianal region that was first noticed at 6 months of age. The histological examination showed a hamartomatous benign proliferation of hair follicles and disordered sebaceous glands, which is consistent with the infundibular structures and radiating sebaceous glands that are typically observed in previously reported cases of FSCH. Sebaceous differentiation is a hallmark feature of FSCH. Folliculosebaceous cystic hamartoma is believed to be a late-stage form of trichofolliculoma (TF). Another consideration is that FSCH and TF are two distinct entities set apart by their degree of sebaceous or follicular differentiation and development of the mesenchymal stroma.

Keywords: benign neoplasm, dermatopathology, folliculosebaceous cystic, hamartoma

Introduction

Folliculosebaceous cystic hamartoma (FSCH) typically presents as an asymptomatic flesh-colored solitary papule or nodule with a pedunculated or dome shape that is approximately 0.5-2cm in size [1]. It is commonly found on the face or scalp, with a strong predilection for the nose [1,2]. Rarely, it is found on the genitalia [1,3]. The growth is usually found in adults, although there have been a few reports of congenital cases [4]. Folliculosebaceous cystic hamartoma is generally not associated with any systemic conditions or co-morbid skin diseases [2]. A few previous reports have observed a co-occurrence of rosacea [5], nevus lipomatosus superficialis [6,7], and a port-wine stain [8]. Histopathological examination of a skin biopsy can confirm the diagnosis of FSCH. Our patient exhibited a tumor with overlap features between a FSCH and fibrofolliculoma. Herein, we compare the key features that distinguish FSCH from neoplasms with a similar clinical appearance that may affect the perianal region.

Case Synopsis

A 6-month-old boy with a normal birth history and development was referred for evaluation of a perianal lesion. Physical examination found a 0.5cm×0.5cm flesh-colored lobulated papule with a pedunculated base in the right gluteal cleft. The lesion had been present at birth and had grown in proportion to the patient's size. The patient was reevaluated as a 1-year-old and the lesion had progressively increased in size to 1.8cm×1.2cm (**Figure 1**). The growth was asymptomatic and had no tenderness, visible hairs, bleeding, or pruritus. It did not affect the child's bowel movements. Owing to its progressive growth however, it was surgically excised. Histopathologic examination of the biopsied lesion revealed a hamartomatous benign proliferation of hair follicles and disordered sebaceous glands (**Figure 2**). Vellus hairs radiated from some of the hair follicles. The mesenchymal changes around the follicular units included a

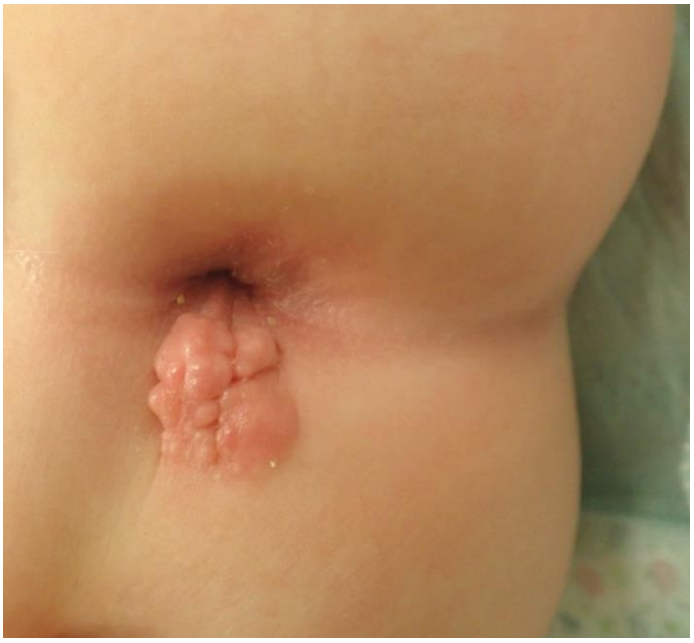

Figure 1. *Initial presentation of the flesh-colored verrucous and lobulated papule with a pedunculated base measuring 0.5cm×0.5cm located in the patient's right gluteal cleft.*

proliferation of small venules and collagen bundles. Based on the histological findings, a diagnosis of a benign follicular proliferation with features of FSCH and fibrofolliculoma was made.

Case Discussion

The initial clinical differential diagnosis in this case included an epidermal nevus, smooth muscle hamartoma, perianal rhabdomyosarcoma, or condyloma. Epidermal nevi can be isolated birthmarks or present as syndromes that affect many organ systems [9]. Keratinocytic epidermal nevi are usually linear or whorled and may have a verrucous appearance [9]. Smooth muscle hamartomas can present as indurated plaques and congenital forms may be accompanied with hyperpigmentation and hypertrichosis [10]. Perianal rhabdomyosarcoma has a poor prognosis and often presents with lymphadenopathy, rapid growth, and recurrence after excision [11]. There was low clinical suspicion for a condyloma since there was no parental history of human papillomavirus infection (HPV) infection. The diagnosis of FSCH is dependent on histological findings and the key features that distinguish FSCH are an infundibular cystic structure with radiating sebaceous structures and a mesenchymal stroma [2]. Occasionally, there are rudimentary hair follicles present [2]. Although most cases have a copious amount of sebaceous hyperplasia, the anal location present in this case might be the reason for a decreased number of such elements, compared to most facial examples of this entity.

The main histopathologic tumor in the differential is a trichofolliculoma (TF). Trichofolliculoma appears as a flesh-colored nodule on the head or neck that typically has a central umbilication embedded with tufts of hair [12]. Histologically, TF is characterized by a large central dilated follicular infundibulum radiating small hair follicles in different stages of development [12]. Fibrofolliculoma and trichodiscomas are closely related benign adnexal

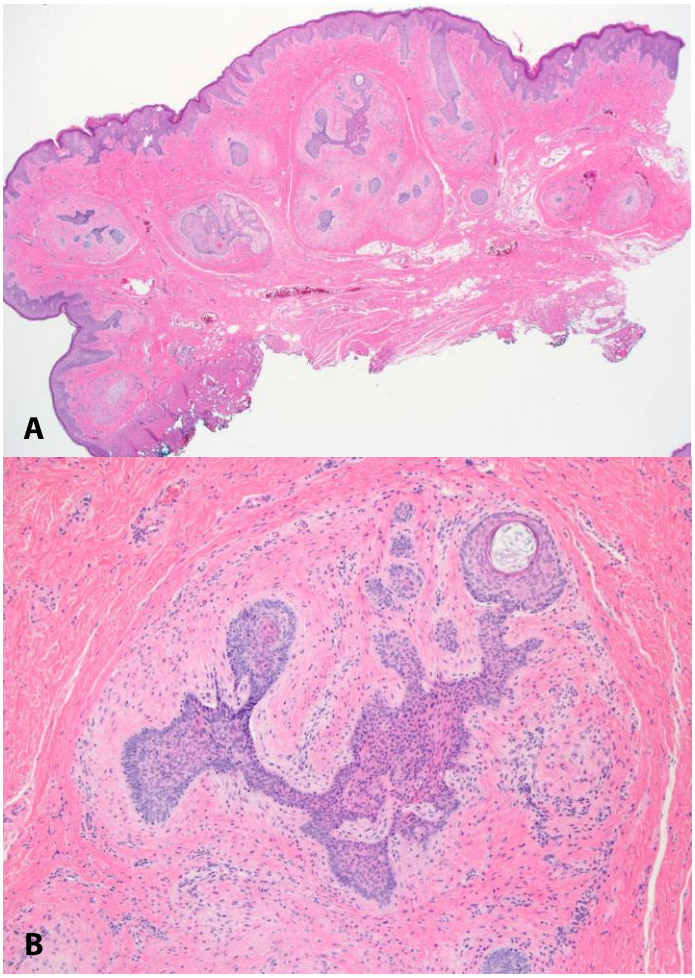

Figure 2. *H&E stain of the excised lobulated nodule in the perianal region.* ***A)*** *Low magnification shows multiple distinct regions of hair follicle proliferation with disordered radiating sebaceous glands, 50×.* ***B)*** *High power magnification shows a hair follicle surrounded by fibrous and perifollicular stroma, 100×.*

tumors seen in patients with Birt-Hogg-Dube syndrome [13]. As opposed to the current case, the lesions are frequently located on the face. In fibrofolliculomas, there are hamartomatous hair follicles with cords and thin columns of mantle-like epithelial components often showing a fenestrated pattern and perifollicular connective tissue containing variable amount of mucin. The stroma is more prominent in trichodiscomas. Abnormal sebaceous glands, as seen in the current case, can be rarely present. Folliculosebaceous cystic hamartoma was previously regarded as a sebaceous trichofolliculoma and there is considerable debate on whether FSCH is an independent entity or a late-stage TF. Schulz et al. propose that the expanded stroma found in sebaceous TF and FSCH arises from the regression of secondary hair follicles [14]. The argument against this is that there are congenital FSCH that do not arise from precursor lesions [15]. Another theory is that mesenchymal stem cells within the stroma promote infundibular and sebaceous differentiation [15]. A case series that compared FSCH and TF observed that FSCH lesions had an upregulation of nestin which promoted a variety of mesenchymal components [16]. A case series of TF observed that sebaceous differentiation was found in advanced forms of TF, but the follicular and sebaceous elements had independent cycles [16]. Since follicular and sebaceous elements originate from the same follicular germ structure, it is expected that some TF will feature partial sebaceous differentiation and conversely, some FSCH will have partial follicular differentiation [17]. Other entities in the histological differential diagnosis are pilar sheath acanthoma (PSA) and dilated pore of Winer. Pilar sheath acanthoma typically appears on the upper lip of adults and has a dilated central cavity and epithelial lobules similar to the outer root sheath of a hair follicle [18]. A dilated pore of Winer is an enlarged pore similar to an open comedo that is also commonly found on the head and neck of adults. It characterized by a central cystic cavity with keratinaceous debris lined by infundibular epithelium [18].

Generally, FSCH and fibrofolliculoma can be managed with surgical excision for cosmetic concerns and recurrence is unlikely [2]. Our patient had no recurrence of the tumor at a one-year follow-up. One case report described successful removal of a genital lesion with CO_2 laser ablation followed by acitretin therapy [1]. Retinoids may be effective because they reduce the size and secretion of sebaceous glands and normalize follicular keratinization [1].

Conclusion

The classic presentation of FSCH is a solitary papule or nodule that usually affects the scalp or face of adults [1]. Folliculosebaceous cystic hamartoma is characteristic for predominantly having malformed sebaceous differentiation [16]. Folliculosebaceous cystic hamartomas can be successfully managed with surgical excision and there is a low likelihood of recurrence [2]. This is a novel case that features an atypically located FSCH in the perianal region of an infant, which has not been reported before. This tumor also shows some overlap features with a fibrofolliculoma. This unique presentation highlights the importance of careful histopathologic review to differentiate FSCH from other benign or malignant neoplasms.

Potential conflicts of interest

The authors declare no conflicts of interest.

References

1. Brücher J-J, Franke I, Ulrich J, Gollnick H, Leverkus M. Giant genital variant of folliculosebaceous cystic hamartoma: successful management by CO2 laser and acitretin therapy. *Br J Dermatol.* 2007;157:833-835. [PMID: 17711522].
2. Suarez-Peñaranda JM, Vieites B, Ramírez-Santos A, et al. Clinicopathological and immnuohistochemical findings in a series of folliculosebaceous cystic hamartoma. *J Cutan Pathol.* 2009;36:251-256. [PMID: 18715254].
3. Bolognia JL, Longley BJ. Genital variant of folliculosebaceous cystic hamartoma. *Dermatol Basel Switz.* 1998;197:258-260. [PMID: 9812032].
4. Selçuk ÖT, Osma U, Süren D, Eyigör H, Yılmaz D, Sezer C. Nasal

folliculosebaceous hamartoma with vascular-mesenchymal overgrowth in an infant. *J Cranio-Maxillo-fac Surg Off Publ Eur Assoc Cranio-Maxillo-fac Surg*. 2013;41:242-244. [PMID: 23127544].
5. Yamamoto T, Ohkubo H, Nishioka K. Folliculosebaceous Cystic Hamartoma Associated with Rosacea. *J Dermatol*. 1993;20:712-714. [PMID: 8300942].
6. Brasanac D, Boricic I. Giant nevus lipomatosus superficialis with multiple folliculosebaceous cystic hamartomas and dermoid cysts. *J Eur Acad Dermatol Venereol*. 2005;19:84-86. [PMID: 15649197].
7. Kang H, Kim SE, Park K, Son SJ, Song KY. Nevus lipomatosus cutaneous superficialis with folliculosebaceous cystic hamartoma. *J Am Acad Dermatol*. 2007;56:S55-S57. [PMID: 17224390].
8. Yang C-H, Shen S-C, Yeh J-T, Tsai Y-L, Hong H-S. Folliculosebaceous cystic hamartoma arising within a port-wine stain. *Clin Exp Dermatol*. 2005;30:509-511. [PMID: 16045680].
9. Asch S, Sugarman JL. Epidermal nevus syndromes: New insights into whorls and swirls. *Pediatr Dermatol*. 2018;35:21-29. [PMID: 29044700].
10. Lau SK, Koh SS. Cutaneous Smooth Muscle Tumors: A Review. *Adv Anat Pathol*. 2018;25:282-290. [PMID: 29649005].
11. Gökdemir G, Ekmen S, Gungor S, Singer R. Perianal Rhabdomyosarcoma: Report of a Case in an Infant and Review of the Literature: A Polypoid Anal Mass as Embryonal Rhabdomyosarcoma. *Pediatr Dermatol*. 2013;30:97-99. [PMID: 22352996].
12. El-Komy MH, Abdelkader HA. Congenital trichofolliculoma: a very rare presentation. *Dermatol Online J*. 2020;26. [PMID: 32898407].
13. Tong Y, Schneider JA, Coda AB, Hata TR, Cohen PR. Birt-Hogg-Dubé Syndrome: A Review of Dermatological Manifestations and Other Symptoms. *Am J Clin Dermatol*. 2018;19:87-101. [PMID: 28695430].
14. Schulz T, Hartschuh W. Folliculo-sebaceous cystic hamartoma is a trichofolliculoma at its very late stage. *J Cutan Pathol*. 1998;25:354-364. [PMID: 9765020].
15. Wu Y-H. Folliculosebaceous cystic hamartoma or trichofolliculoma? A spectrum of hamartomatous changes inducted by perifollicular stroma in the follicular epithelium. *J Cutan Pathol*. 2008;35:843-848. [PMID: 18494824].
16. Misago N, Kimura T, Toda S, Mori T, Narisawa Y. A Revaluation of Folliculosebaceous Cystic Hamartoma: The Histopathological and Immunohistochemical Features: *Am J Dermatopathol*. 2010;32:154-161. [PMID: 19755908].
17. Misago N, Ansai S, Fukumoto T, et al. Chronological changes in trichofolliculoma: Folliculosebaceous cystic hamartoma is not a very-late-stage trichofolliculoma. *J Dermatol*. 2017;44:1050-1054. [PMID: 28370423].
18. Ho J, Bhawan J. Folliculosebaceous neoplasms: A review of clinical and histological features. *J Dermatol*. 2017;44:259-278. [PMID: 28256760].