Multiple Papulonodules over Face and Trunk: A Rare Case Report

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Abstract

Familial cylindromatosis (turban tumor syndrome) is a very rare neoplasm originating from eccrine or apocrine glands. It is an autosomal dominant condition, characterized by multiple cylindromas commonly presenting over face or scalp. We report a case of familial cylindromatosis diagnosed on the basis of clinical, dermoscopic, and histopathological findings in a 70-year-old female. The case is reported due to its rare occurrence in Indian scenario.

Keywords: Dermoscopy, familial cylindromatosis, jigsaw puzzle, turban tumor

Introduction

Cylindromas are benign skin appendageal neoplasms most likely originating from eccrine glands.[1,2] They can be single or multiple, commonly involving scalp, face, and neck. Solitary cylindromas occur sporadically, whereas multiple tumors are inherited in an autosomal dominant manner. Multiple lesions over scalp present as numerous small papules and/or large nodules over the scalp like a turban, hence commonly known as turban tumor. Familial cylindromatosis (FC), originally described as Ancell–Spiegler cylindroma, is a rare autosomal condition with apparently complete penetrance but variable expression characterized by multiple cylindromas over face and scalp.[3]

Case Report

A 70-year-old female born out of non-consanguineous marriage presented to us with multiple asymptomatic skin-colored to reddish raised lesions over scalp, face, and trunk since 40 years. These lesions first appeared when she was 30 years old, primarily on face and scalp, which gradually increased in size and number over the years to involve trunk. There was history of intermittent bleeding from larger lesions since 2–3 years. Similar history of lesions was also present in her grandmother, elder sisters, and daughter [Figure 1]. On cutaneous examination, there were multiple confluent skin-colored to erythematous, smooth surfaced, rounded, firm, non-tender papulonodules of varying sizes ranging from 0.5 to 5 cm in diameter with overlying telangiectasias in few lesions present predominantly over face, left half of scalp, right retroauricular, infraauricular, preauricular region, neck, chest, upper, and lower back, bilateral groin folds, and vulva [Figure 2a-g]. There was a single pedunculated nodule of size 3 × 2 cm over left side of the chin. Single well-defined, hyperpigmented, non-umbilicated nodule is present near right nasal fold. Rest of the cutaneous examination was unremarkable. General and systemic examination was normal. Based on history and clinical presentation, differential diagnoses of cylindromas, trichoepitheliomas, and spiradenomas were considered [Table 1]. Dermoscopy was performed using 3Gen DermLite DL4 (CA, USA) in 10× polarized mode. Dermoscopy of most of the nodular lesions over back revealed arborizing vessels on whitish pink background with blue dots and globules. Dermoscopy of few lesions over face and upper trunk revealed arborizing vessels on whitish pink background with ulceration and yellow

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non-homogeneous areas at places [Figure 3a-d]. Fine needle aspiration cytology (FNAC) from nodules over scalp, right nasolabial fold, and back revealed basaloid cells in clusters, acinar pattern around small hyaline globules, and lining ribbons of hyaline material. Few clusters and dispersed cells with scanty cytoplasm and oval hyperchromatic nuclei with granular chromatin seen are suggestive of cylindroma [Figure 4a and b]. Local USG of the lesions revealed multiple, round to oval, hypoechoic lesions in subcutaneous plane and normal parotid regions [Figure 5a and b]. Computed tomography of head, neck, and thorax was normal. Histopathological examination from nodule over back and face revealed epidermis showing basket weave hyperkeratosis, poorly circumscribed tumor comprising irregularly shaped islands, and cords of basaloid cells with peripheral palisading by eosinophilic hyaline bands in dermis suggestive of cylindroma [Figure 6a-c]. Routine hematological investigations were within normal limits. On the basis of clinical, dermoscopic, cytological, and histopathological findings, a final diagnosis of familial cylindromatosis was reached. Gene mapping was not done due to limited resources. The patient was referred to plastic surgery for further management.

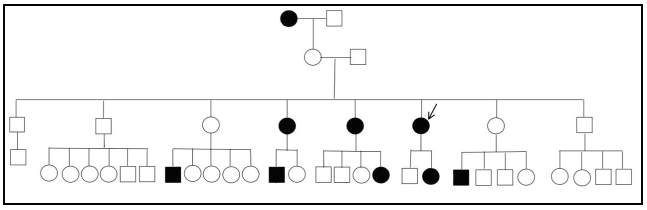

Figure 1: Pedigree chart showing similar lesions in grandmother, elder sisters, and daughter

Discussion

Ancell[4] first described cylindroma in 1842. Cylindromas are benign skin appendageal tumors originating most

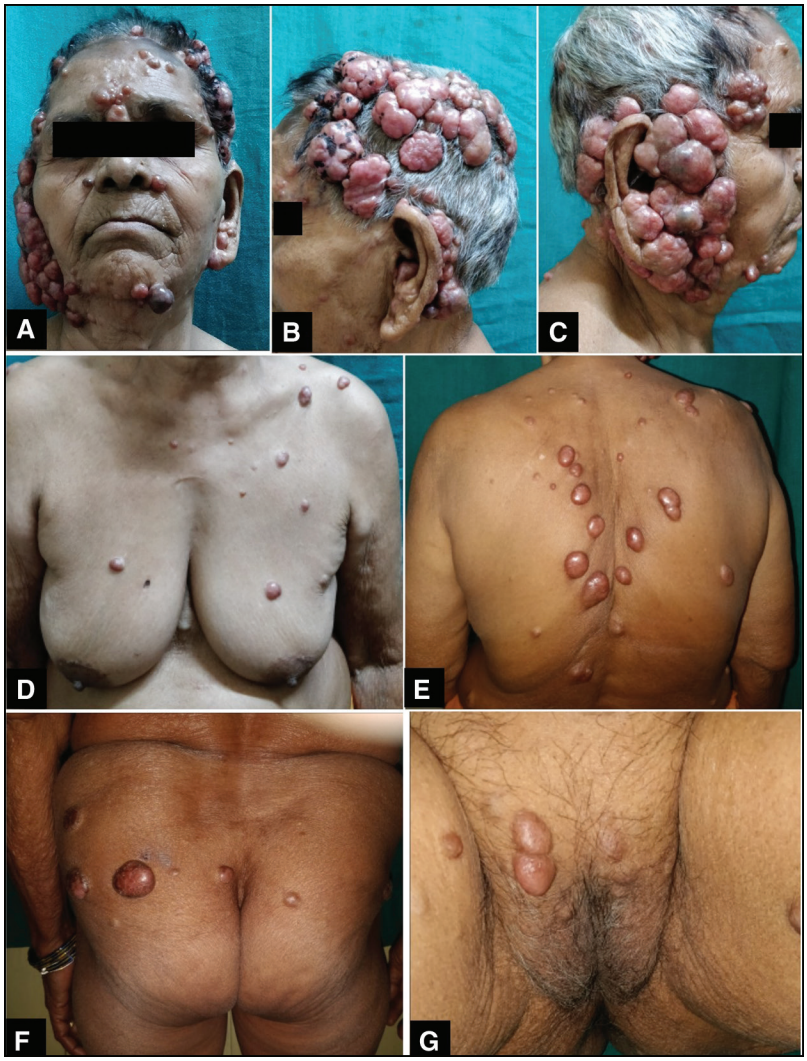

Figure 2: (a-g) Multiple confluent skin colored to erythematous, smooth surfaced, rounded, firm, non-tender papulonodules of varying sizes with overlying telangiectasias in few lesions over face (a), left half of scalp (b), right auricular region (c), neck, chest (d), upper (e) and lower back (f), bilateral groin folds and vulva (g)

Table 1: Differential diagnosis of cylindroma

	Spiradenoma	Cylindroma	Trichoepithelioma
Clinical appearance	Soft, blue, gray, or purple nodule, painful on palpation, located on head, neck or trunk.	Firm, pink, red, or blue nodule located on the face or scalp	Round, skin-colored, firm papules or nodules located on the nasolabial folds, nose, forehead, upper lip, and scalp.
Histology	Two types of tumor cells: basaloid cells contain a small, hyperchromatic nucleus with scant cytoplasm. Other cells are larger and contain a pale nucleus. Thin basal membrane. Frequent lymphocytic infiltration	Small lobules of basaloid cells arranged in a jigsaw pattern and surrounded by a prominent hyaline basement membrane. Absence of inflammatory infiltrate	Nests of basaloid cells with horn cysts in dermis. Tumor cells have minimal cytoplasm and a large hyperchromatic nuclei and show peripheral palisading. Formation of dense aggregates of fibroblastic cells referred to as papillary mesenchymal body.
Dermoscopic features	Light blue pigment with peripheral reticulate pigmentation, associated with reddish linear serpentine structures surrounded by whitish areas	Blue dots and globules associated with arboriform vessels on a whitish, salmon-pink background	Arborizing vessels, multiple milia-like cysts, and rosettes amidst a whitish background

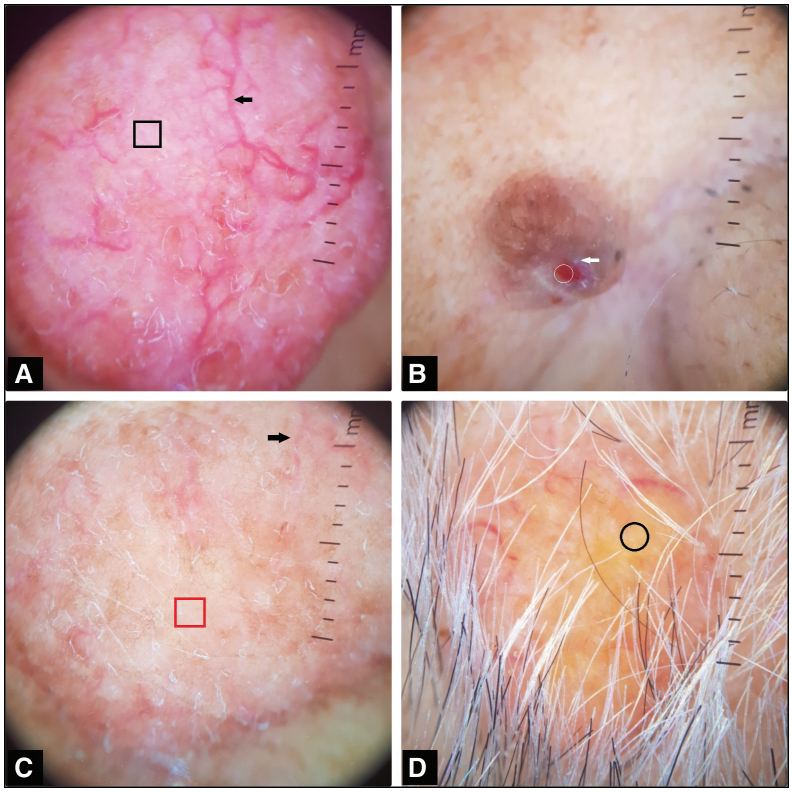

Figure 3: (a-d) Dermoscopy [3Gen DermLite DL4 (CA, USA) 10× polarized mode] of lesions over face and upper trunk revealed arborizing vessels (black arrow) on whitish pink background (black box) with blue dots and globules (white arrow). Dermoscopy of lesions over back revealed arborizing vessels on whitish background (red box) with ulceration (white circle) and yellow non-homogeneous areas at places (black circle)

commonly from folliculo-sebaceous-apocrine unit. It is usually seen in middle aged females with scalp being the most common site. Cylindroma has two types of clinical presentations: a solitary form, without family history of cutaneous cylindromas, most commonly involving skin of the head and neck.[1] Solitary (sporadic) form occurs as frequently as the multiple form. Multiple, inherited cylindromas are more common in females and occur over a wide age range, with the majority of patients in second or third decades of life, as seen in our case, which increase in size and number throughout life.[1,2] These may occur on the scalp and rarely on the trunk and the extremities.[1] Cylindroma presents as slow growing, multiple, pink to red, firm, smooth surfaced papules and nodules, often pedunculated with surface telangiectasias. Although rare, malignant transformation can be seen in multiple cylindromatosis.[5,6] Thus patients are at risk of developing tumors such as basal cell adenoma and adenocarcinoma of parotid and minor salivary glands. Multiple cylindromas can occur as a part of FC, Brooke–Spiegler syndrome (BSS) and multiple familial trichoepitheliomas.[7] All the three have been recently associated with mutations in the CYLD gene.[3] The tumor suppressor gene, cylindromatosis gene (CYLD1), is located on band 16q12-13.[8] The gene

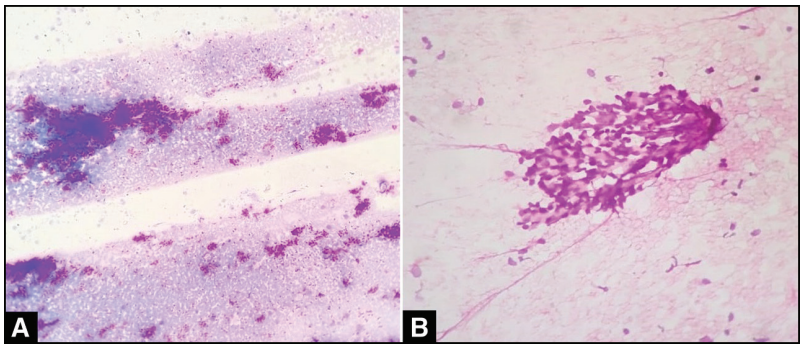

Figure 4: (a, b) FNAC from nodules over scalp, right nasolabial fold and back revealed basaloid cells in clusters, acinar pattern around small hyaline globules, and lining ribbons of hyaline material. Few clusters and dispersed cells with scanty cytoplasm, oval hyperchromatic nuclei with granular chromatin seen suggestive of cylindroma

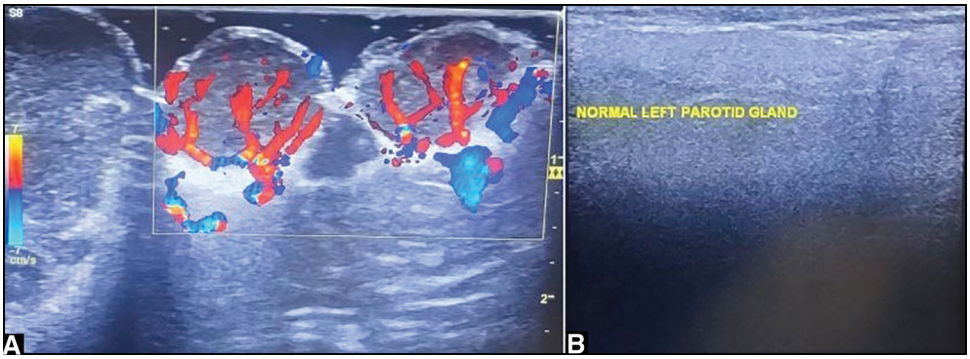

Figure 5: (a, b) Local USG of the lesions revealed multiple, round to oval, hypoechoic lesions in subcutaneous plane (a) and normal parotid regions (b)

product represses the TNFα pathway which regulates a number of antiapoptotic genes involved in proliferation of skin appendages by increasing the expression of nuclear factor κ-β.[7,8] BSS is an inherited disease characterized by multiple skin appendageal tumors predominantly cylindromas, trichoepitheliomas, and/or spiradenomas.[3,8] Histopathology of cylindroma reveals sharply circumscribed nodules within the dermis and/or subcutis composed of nests of basaloid cells arranged in a jigsaw puzzle pattern, as seen in our case. The cells are of two types: one large, with a moderate amount of cytoplasm and a vesicular nucleus arranged centrally; and the other small, with little cytoplasm and a compact nucleus arranged peripherally. Jarrett *et al.* were the first to describe dermoscopy of cylindroma.[9] On dermoscopy, the reported patterns of cylindroma consist of arborizing vessels on whitish pink background, blue dots and globules, ulceration, and yellowish non-homogeneous areas correlating to hyperkeratosis as observed in our case.[9-11] Similar dermoscopic patterns have also been reported in basal cell carcinoma.[12] The vascular patterns and color of dots and globules may help to differentiate cylindromas and nodular basal cell carcinoma. The vascular branches are more pronounced at the periphery and they extend from the periphery toward the center of the lesion in cylindromas while arborizing vessels are more pronounced towards center without any particular pattern in basal cell carcinoma.[10] Also, blue dots/globules are visible in cylindroma in contrary to gray dots in basal cell carcinoma.[11] Treatment of choice for cylindroma is surgical excision or laser ablation. Alternative treatment includes cryotherapy, electrosurgery, carbon dioxide laser, radiotherapy, and dermabrasion.[7] To prevent occurrence of new lesions, topical aspirin derivatives are currently being tried.[6] Regular follow-up is required in such cases to rule out malignant transformation. Dermoscopy can aid in the diagnosis of cylindroma in rare cases. There are very few reports of clinico-dermoscopic patterns describing multiple familial cylindromatosis in India [Table 2]. Thus we report a case of familial cylindromatosis affecting trunk along with scalp and face with strong family history and without associated other adnexal neoplasm.

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Conflicts of interest

There are no conflicts of interest.

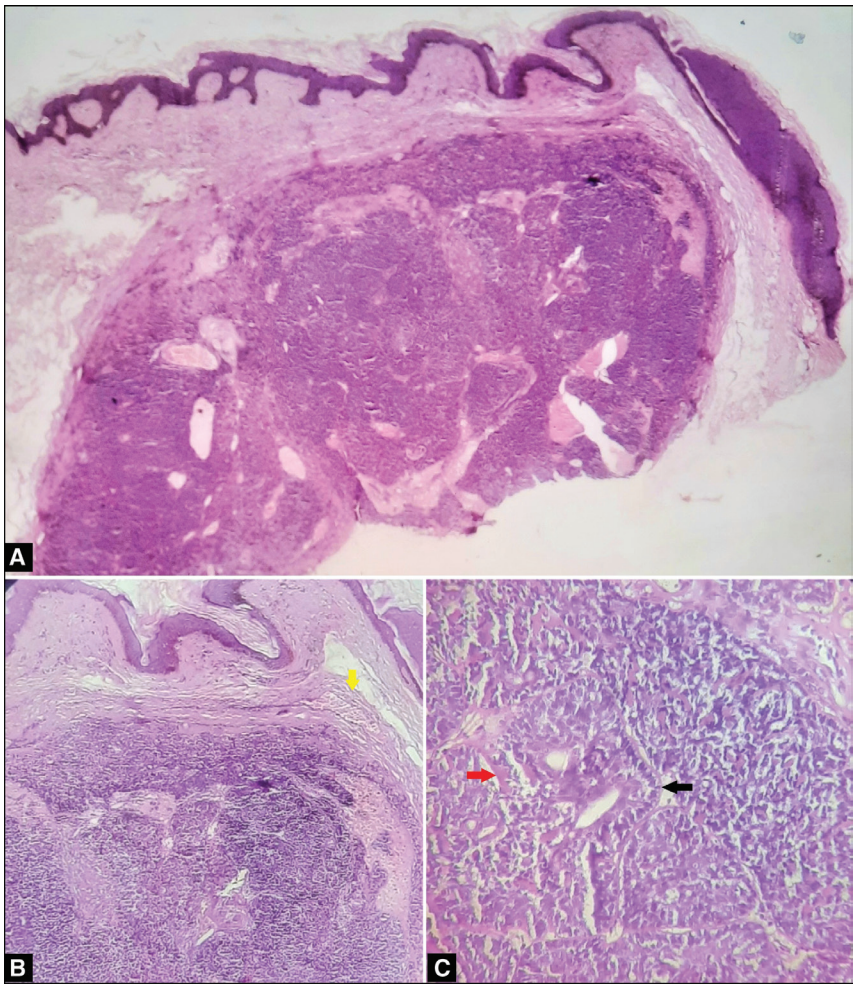

Figure 6: (a-c): Histopathological examination from nodule over back and face revealed epidermis showing basket weave hyperkeratosis (yellow arrow), poorly circumscribed tumor comprised irregularly shaped islands and cords of basaloid cells with peripheral palisading (black arrow) by eosinophilic hyaline bands (red arrow) in dermis suggestive of cylindroma

Table 2: Literature review of dermoscopic patterns of cylindromas

Sr no.	Case report	Year	Age/sex	Description	Findings on dermoscopy
1	Jarret *et al.*	2009	42/F, 45/F	Dermoscopy of BSS[9]	Areas of pink background with ill-defined arborizing vessels and ill-defined blue structures
2	Cabo *et al.*	2010	80/F	Dermoscopy of cylindroma[13]	Areas of pink background coloration, arborizing telangiectasia, blue dots/globules, and ulceration
3	Lallas *et al.*	2011	58/F	Dermoscopy of solitary cylindroma[10]	Arborizing telangiectatic vessels, with a relatively small number of branches, on a homogeneous white-pinkish background. The vessels appeared blurred and light-red-to-pinkish in color and were observed mainly at the periphery of the lesion
4	Cohen *et al.*	2014	65/M	Dermatoscopic pattern of a cylindroma[11]	Arborizing telangiectasia and several scattered white globules on a white to salmon pink background
5	Tiodorovic *et al.*	2015	43/F, 37/F	Clinical, histological, and dermoscopic findings in familial cylindromatosis: a report of two cases[14]	Arborizing vessels on a white-ivory or pinkish background, more prominent at the periphery of lesions, in some tumors blue dots and globules were also present

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