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Myxoma of the ear lobe in a 23 month-old girl with Carney complex

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Abstract

Myxomas of the ear are extremely uncommon, especially in young children. A 23-month-old girl born to a family with known Carney complex, a condition that predisposes to multiple myxomas, presented with a large, cauliflower-like mass located on the back of her right ear.

Histopathologically, the lesion was composed of scattered spindle-shaped or stellate cells with abundant associated mucin and a very sparse inflammatory infiltrate containing lymphocytes and neutrophils. The patient was a carrier of a protein kinase A regulatory subunit 1A (*PRKARIA*) mutation; *PRKARIA* mutations cause Carney complex in most patients with this rare disorder.

This is the earliest presentation of an ear lobe myxoma reported in the literature.

Keywords

Ear tumors; protein kinase A; *PRKARIA* mutation; pediatrics; Carney complex

INTRODUCTION

The disorder known as Carney complex is characterized by the development multiple skin lesions, myxomas, endocrine tumors, schwannomas and other neoplasms. Skin lesions include lentigines, café-au-lait spots, and Spitz, blue, and conventional melanocytic nevi (1). Skin myxomas also occur and typically increase in number after puberty; they are usually associated with cardiac myxomas and myxoid lesions in other tissues (i.e. the breast, genitalia, and elsewhere) (1). Typical sites for skin myxomas in Carney complex include the eyelids, external ear canal, and nipples (2). Grossly, skin myxomas typically appear as spherical, circumscribed, gray-white, glistening masses.

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Disclosure

The authors have no conflict of interest to disclose

Herein, we report the case of a 23-month-old girl with a large nodular ear lobe myxoma. It presented as a highly irregular, cauliflower-shaped mass and represented the first presentation of Carney complex in this patient. This is the earliest reported ear lobe myxoma in the literature.

Subject & Methods

Case presentation—A 23-month-old twin born after 32 weeks of gestation to a 22-year old mother with Carney complex presented to the National Institutes of Health (NIH) Clinical Research Center (CRC) for a large skin lesion located on the back of her right ear. Her mother was diagnosed with Carney complex at age 13 years and had multiple heart myxomas, endocrine tumors, and a lower spine schwannoma. There were no other relatives with any history suggestive of Carney complex in the family. The patient's physical examination, including examination of her head, eyes, nose, throat, neck, thyroid, lungs, cardiovascular system, abdomen and neurological system, was within normal limits. On examination of her skin, she did have a few freckles on her abdomen and her extremities. She also had a very prominent exophytic lesion behind her right ear. It was approximately 2 × 1 cm and had a soft, fleshy, mamillated and papillomatous appearance with dark necrotic-appearing areas scattered on the superior surface (Fig. 1A-B). There was no pus, oozing or bleeding noted out of this friable lesion. She underwent surgery with successful excision. The proliferation was found to have developed from an area of the skin just posterior to the auricular cartilage of the right ear.

Results

DNA studies

Analysis of a peripheral DNA sample confirmed the presence of the pathogenic *PRKARIA* mutation (c. 43_58del16/p.Leu15fs104X). This was the mutation of the mother; interestingly, the patient's twin brother (who remains asymptomatic) was also a carrier of the same mutation.

Histopathology—Scanning magnification revealed an exophytic lesion surfaced by a hyperkeratotic epidermis. There was a 2 mm, circumscribed, un-encapsulated nodule in the dermis (Fig 2A). The lesion was composed of scattered spindle-shaped or stellate cells with associated abundant mucin and small blood vessels (Fig. 2B&C). A very sparse inflammatory infiltrate containing lymphocytes and neutrophils was also present. An Alcian-blue stain was performed and highlighted the intra-lesional mucin (Fig 2D). The adjacent dermis showed fibrotic changes. The necrotic-appearing areas corresponded microscopically to areas of inflamed granulation tissue (not shown). The myxomatous lesion itself was intact and had no associated necrosis.

Discussion

Carney complex represents a rare and often fatal syndrome with an associated high prevalence of cutaneous lesions. Factors that permit early recognition, such as the large ear mass in this patient, are important so that affected patients can be appropriately investigated for potentially life-threatening components of the complex, such as cardiac myxomas (3). Ear, eyelid, and other cutaneous myxomas are detected prior to the cardiac myxoma in the majority of patients (4). Importantly, the ear canal and temporal bone lesions often are accompanied by hearing loss (5) while myxomas of the middle ear may cause facial palsy (6).

Cutaneous myxomas present as papules or nodules that usually have a smooth surface and are white, skin-colored, opalescent, or pink. They appear between birth and the fourth decade. Although extremely rare in the general population, myxomas of the external ear have been described in 14% of patients with Carney complex (7). The gender distribution is equal, with an age of onset ranging from birth to 41 years. Affected patients may have both ear canal and external ear lesions or even bilateral lesions. Recurrences after simple excision can be seen. The lesions have been described as mucoid, ranging from 3 mm to 2 cm in greatest dimension, and are composed of scattered stellate and spindle cells set in a myxoid, capillary-rich matrix (7). The external appearance of our patient's lesion resembled those of trichogenic myxomas and trichofolliculomas, which have been previously described in a girl who developed generalized subcutaneous nodules and polypoid ear lesions during her first decade of life (8). However, our patient's lesion was composed of scattered spindle-shaped or stellate cells and small blood vessels with associated mucin, and therefore it met the specific histopathologic criteria for a true cutaneous myxoma.

Carney complex is inherited in an autosomal dominant manner and its diagnosis usually relies on clinical diagnostic criteria. Approximately 70% of individuals diagnosed with Carney complex have an affected parent and the remaining 30% have a *de novo* mutation. Each child of an individual with Carney complex has a 50% chance of inheriting the mutation. The disease is genetically heterogeneous and has been mapped to 2p16 (CNC2) and 17q22–24 (CNC1) (9). Sequence analysis of the *PRKARIA* coding region, available on a clinical basis, has a mutation detection rate of approximately 62% (10). Patients with a *PRKARIA* mutation are more likely to have pigmented skin lesions, myxomas, and thyroid and gonadal tumors; they also present earlier with these tumors (10).

The patient described in our report presented with an unusual, large ear tumor and this represented her first manifestation of Carney complex, a diagnosis that was confirmed by genetic analysis. Clinicians and pathologists encountering similar tumors should consider Carney complex as a potential diagnosis.

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Figures 1 A&B.

A prominent exophytic mass (2×1 cm) behind the patient's right ear has a cauliflower-like appearance and focally shows dark, necrotic-appearing areas scattered on its superior surface.

Figure 2.

A. Scanning magnification view shows a small, circumscribed but un-encapsulated nodule in the dermis (40x magnification); **B.** and **C.** The unencapsulated lesion is composed of scattered spindle-shaped or stellate cells with associated mucinous stroma and small blood vessels (100x and 400x magnification); **D.** Alcian-blue stain highlights the intra-lesional mucin (100 × magnification)