Epidermal choristoma of the tongue: a case series and review of the literature

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Keywords: pigmentary disorders, dermatopathology, mucous membrane disorders

Conflict of interest disclosures: No conflicts of interest to disclose from any authors.

Consent for Publication: All authors provided consent for publication.

Informed Consent: All parents provided written consent to publish photos.

Word Count: 1090, Figure Count: 2, Table Count: 1

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Abstract

Epidermal choristoma is a rare, benign, congenital lesion in which islands of ectopic skin are found within the oral cavity. They present as pigmented macules or papules on the tongue. Histological appearances are characteristic and benign, and observation of the lesion rather than complete excision should be considered as a reasonable management option. We present three cases, review the current literature and recommend observation of the lesion rather than compete excision should be considered as a reasonable management option.

Introduction

Epidermal choristoma is a rare, benign, congenital lesion that often presents as a concerning melanocytic macule or papule on the tongue. Epidermal choristomas are ectopic skin diagnosed by histology demonstrating normal stratified squamous epithelium with hyperkeratosis, hypergranulosis, basilar pigmentation, and submucosal skin adnexal structures, such as hair follicles and sebaceous glands. The first case of epidermal choristoma of the tongue was reported in 2005 in a newborn, with only four additional cases reported since.[1] Here, we present three new cases of epidermal choristoma of the tongue with a focus on conservative management.

Case 1

A 7-week-old Asian male, born without complications at term, was referred to dermatology for evaluation of tongue lesions. He presented with clustered, asymptomatic pigmented macules on the left tongue, present and stable since birth. He was otherwise well and there was no family or past medical history of note. The largest macule measured approximately 1.0 x 0.5 cm (Figure 1A). He also had lumbosacral and right leg dermal melanocytosis consistent with congenital dermal melanocytosis. Biopsy from the largest tongue lesion demonstrated pigmented squamous epithelium with hyperkeratosis, a prominent granular layer, and submucosal skin adnexal structures, including sebaceous glands and abortive hair follicles (Figure 2A). This confirmed the diagnosis of epidermal choristoma of the tongue. Further excision was deferred to preserve tongue tissue. He has been followed in clinic with no evolution of the lesion at the age of 3 years old.

Case 2

A 21-month-old Asian male was referred to dermatology for brown lesions on his tongue. On evaluation, he had a cluster of pigmented macules on the right tongue (Figure 1B), present and stable from birth, and asymptomatic. He was otherwise well and there was no family or past medical history of note. The largest macule measured 0.3 x 0.3 cm. He also had congenital dermal melanocytosis on the back in the midline and three café-au-lait macules. Biopsy of one tongue lesion confirmed all characteristic histological findings of epidermal choristoma of tongue (Figure 2B). He was managed conservatively without further excision and the clinical appearance remained stable at the age of 3 years old.

Case 3

A 6-year-old male of Asian descent presented with a congenital, stable and asymptomatic tongue lesion. He was otherwise well and there was no family or past medical history of note. On examination he had a light brown, slightly lesion on the left side of the tongue, measuring approximately 2.0 x 1.1 cm, with extension onto the left lateral margin and with no induration or ulceration (Figure 1C). Histopathological assessment revealed normal oral squamous mucosa adjacent to stratified squamous epithelium with a well-formed granular layer, dense lamellar keratin, and submucosal sebaceous units (Figure 2C). Complete excision of the lesion was performed due to preference by the family, who decided on definitive treatment over potential low risk adverse consequences and close monitoring. Post-operative review confirmed good aesthetic outcome and complete functional recovery of the tongue. There has been no regrowth at 7 years of age.

Discussion

Epidermal choristoma is a rare congenital malformation characterized by ectopic epidermis with well-differentiated adnexal structures in the oral cavity. Choristomas represent normal cells in abnormal locations and are common throughout the body, including the oral cavity.[2, 3] Oral choristoma can be composed of numerous types of tissue, including respiratory, glial, follicular, osseous, and cartilagenous.[4, 5, 6] An epidermal choristoma is a rare sub-type of oral choristoma.

Clinical features of the three cases presented here and all five previously reported cases are summarized in Table 1. Importantly, there have been no reports of malignant transformation.[7, 8, 9]

All eight reported cases have presented in males. Epidermal choristoma present as brown pigmented tongue macules which can be single or multiple and can be present either unilaterally or in the midline. On the tongue, color varies from light brown to dark brown. An epidermal choristoma has previously also been described arising from the midline of the maxillary gingiva, presenting as a large pedunculated flesh colored polypoid mass.[4]

Several causes of oral pigmentation, which can be distinguished by thorough history, examination and histopathology.[10] Congenital malignant melanomas within the oral cavity have not been described in the literature. Similar to epidermal choristoma, congenital melanotic macules present with pigmented macules visible on the tongue at birth which grow proportionately with the child and follow a benign course.[10] Owing to their similar clinical appearance and course, these lesions may be only distinguishable by histopathology. Congenital melanotic macules have increased melanin in the stratum basale but without the additional presence of adnexal structures which are observed in epidermal choristomas.[10,11] Congenital melanocytic nevi can be easily distinguished by nests of nevus cells on histology but have to date only rarely been described in the oral cavity and affecting the buccal, palatal and gingival mucosa rather than the tongue.[12,13,14] Familial syndromes may cause oral hyperpigmentation, however this would be unlikely to present in the neonatal period; a thorough history including family history, and full physical examination should be undertaken.[15, 16] Peutz-Jeghers syndrome presents with mucocutaneous pigmentation from the first year of life, but most commonly affects the lips, perioral and buccal mucosa.[17.]

The pathogenesis of epidermal choristoma is unknown. The presence of this lesion in association with other pigmentary birthmarks in patients 1 and 2 is potentially interesting. Future studies could investigate whether epidermal choristomas have any mutations previously described in mosaic hyperpigmentation disorders.

Total excisional biopsy has been chosen in a majority of previous reported cases. This approach may make sense for smaller, singular lesions which are being biopsied for diagnostic reasons; however, there is no evidence that it is necessary for larger or clustered lesions. In patients 1 and 2 in this study the families and medical team agreed to observe the epidermal choristomas after biopsy to preserve tongue tissue.

Clinicians should consider epidermal choristoma when presented with oral pigmented lesions in a newborn. Biopsy is key in differentiating these lesions from other oral pigmented lesions, some of which may have malignant potential. If histopathology confirms diagnosis of epidermal choristoma, observation rather than complete excision should be considered as a reasonable management option to preserve the tongue.

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Figure Legends

Figure 1: Clinical features of epidermal choristoma. (A) Patient 1 – 6 month old with three pigmented tongue macules present at birth (B) Clinical image of patient 2 with six pigmented tongue macules (C) Clinical image of patient 3 light brown, hyperpigmented tongue lesion

Figure 2: Composite of histology images. (A) Histology of largest lesion from patient 1 demonstrated stratified squamous epithelium: a pigmented basal layer, hypergranulosis, hyperkeratosis and abortive hair follicle (white arrow) with sebaceous glands (black arrow) in the submucosa (H&E, x200). (B) Histology from patient 2 demonstrated a pigmented basal layer, hypergranulosis, hyperkeratosis, and sebaceous glands (black arrow) in the submucosa (H&E, x200). (C) Histology from patient 3 also demonstrated diagnostic features of epidermal choristoma of the tongue, including stratified squamous epithelium with dense lamellar keratin and sebaceous glands (black arrow) in the submucosa (H&E, x200).

***Table 1**:* Summary of all reported cases of oral epidermal choristomas.

Author	**Sex**	**Race**	**Age at Presentation**	**Location**	**Size of Largest Lesion (cm)**	**Examination**	**Management**
Azorin *et al* (2005)	Male	Caucasian	1 month	Left dorsal/lateral tongue	1.0 x 0.8	3 clustered dark-brown pigmented papules	Total Excisional Biopsy
Chi *et al* (2010)	Male	Unknown	56 years	Left dorsal tongue	0.3 x 0.3	1 brown pigmented macule	Total Excisional Biopsy
Chi *et al* (2010)	Male	Unknown	32 years	Right buccal mucosa	1.1 x 0.8	1 firm, leathery, brown-grey-white plaque	Observation

Yoshioka *et al* (2012)	Male	Japanese	2 months	Midline maxillary gingiva	1.0 0.6	x	1 soft, elastic, pedunculated, polypoid mass	Total Excisional Biopsy
Curto-Barredo *et al* (2015)	Male	Caucasian	1 month	Left dorsal tongue	0.4 0.3	x	1 light-to-dark brown pigmented macule	Total Excisional Biopsy
Hughes *et al* (2020)	Male	Indian	7 weeks	Left/middle dorsal tongue	1.0 0.5	x	3 clustered dark-brown pigmented macules	Observation
Hughes *et al* (2020)	Male	Indian	21 months	Right dorsal tongue	0.3 0.3	x	6 clustered dark-brown pigmented macules	Observation
Hughes *et al* (2020)	Male	East Asian	6 years	Left dorsal/lateral tongue	2.0 1.1	x	1 light brown hyperpigmented plaque	Wide Local Excision