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Case Report

Heterotopic gastrointestinal cyst within the submandibular space in an adult: A case report and review of the literature [☆]

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ARTICLE INFO

Article history:

Received 8 March 2022

Revised 9 May 2022

Accepted 12 May 2022

Keywords:

Heterotopic gastrointestinal cyst

Enteric duplication cyst

Lingual cystic choristomas

Gastric cystic choristoma

Foregut duplication cysts

Submandibular cyst

ABSTRACT

Heterotopic gastrointestinal cysts (HGIC) are rare congenital cysts that arise anywhere along the gastrointestinal tract. HGICs are infrequently reported in the oral cavity; 0.3% of HGICs are reported in the tongue and even more rarely in the submandibular space. Oral HGICs are more common in children with only 13 reported cases in adults. In the present report, we discuss the differential diagnoses of the submandibular space lesions and describe a rare case of a very large submandibular space HGIC in an adult patient.

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Introduction

Heterotopic gastrointestinal cysts (HGIC) are congenital cysts that arise from ectopic undifferentiated endodermal cells any-

where along the gastrointestinal tract [1]. They are often referred to as enteric duplication cyst, lingual or gastric cystic choristomas, or foregut duplication cysts; however, we have chosen the term heterotopic gastrointestinal cyst to refer to these cysts. They are histologically composed of one or a combination of gastric, squamous, intestinal, and respiratory epithelium [2]. They are most commonly found in the small intestine; however, 0.3% of HGICs have been reported in the tongue and even more rarely in the submandibular space [3].

[☆] Competing Interests: None.

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<https://doi.org/10.1016/j.radcr.2022.05.034>

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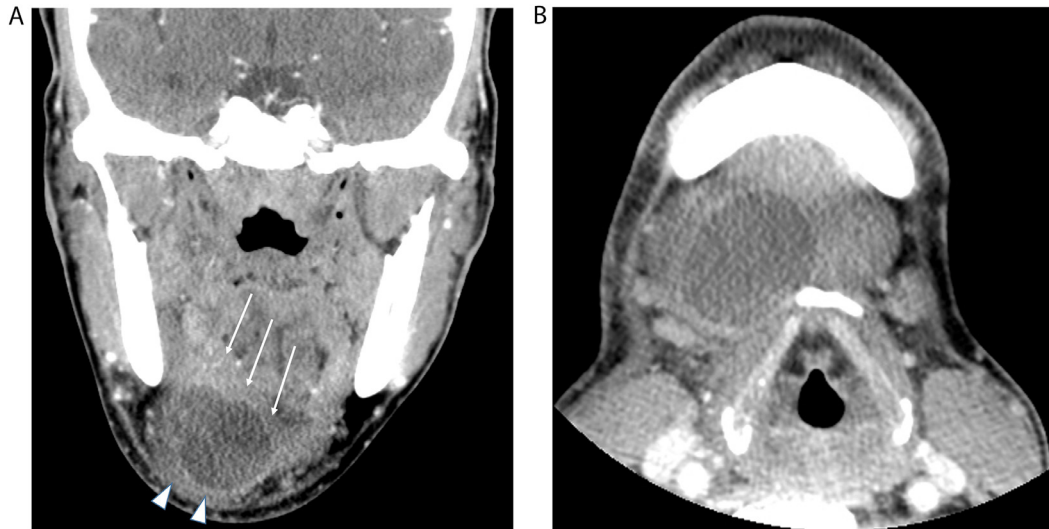


Fig. 1 – Coronal (A) and axial (B) postcontrast facial bone CT: Well-defined, hypodense lesion in the right submandibular space with mild rim enhancement, measuring approximately 3.8 cm. Arrows: mylohyoid muscle. Arrowheads: anterior belly of digastric muscle.

HGICs most commonly manifest in the first decade of life, with a male predilection [3]. Although a definitive etiology has not been established, it is hypothesized that islands of undifferentiated endoderm are separated and entrapped in the gastrointestinal tract during the third to fourth week of embryogenesis when the primitive stomach is located anatomically by the stomodeum [2,3]. Preoperative imaging, usually MRI, narrows the differential diagnosis and assists with surgical planning. Given the low recurrence rate of this lesion, surgical excision is both diagnostic and curative. In the present report, we describe a rare case of a very large submandibular space HGIC in an adult patient.

Case report

A 51-year-old transgender (male to female) patient presented to the emergency department for evaluation of right submandibular region swelling and subjective odynophagia, dysphagia, and dyspnea. The patient reported a 1-year history of submandibular swelling with self-limiting acute exacerbations, frequently associated with upper respiratory infections. Previous exacerbations were successfully treated with antibiotics according to the patient. The patient's medical history was notable for HIV and seizure disorder, well controlled with HAART and Levetiracetam, respectively.

On presentation, the patient was afebrile, normocardic, and normotensive, and the basic metabolic panel and complete blood count were normal. Physical examination revealed a large, well defined, soft, nontender right submandibular space swelling without erythema or warmth. No neurologic deficit was present. Intraoral examination demonstrated elevation and firmness of the submandibular space, on the right, causing tongue elevation. The airway was patent, although the uvula was deviated to the contralateral side. Clear

saliva was expressed from the bilateral submandibular ducts. The patient had poor oral hygiene; several carious teeth were noted.

CT imaging identified a well-defined, somewhat oval-shaped hypodense lesion in the right submandibular space with mild rim enhancement, measuring approximately 3.8 cm (Figs. 1 A and B). The lesion was located above the anterior belly of the digastric muscle and inferior to the mylohyoid muscle. There was asymmetric thickening of the platysma secondary to surrounding edema.

MRI on the same day showed a well-defined mass in the right submandibular space. The lesion was hypointense on T1WI, hyperintense on T2WI, with abnormal thin rim enhancement on post contrast images (Figs. 2A and B, and 3A and B). There was a thin septum within the lesion. The mass contained solid, nonenhancing sediment in its inferior portion. The submandibular gland was displaced dorsally by the mass (Fig. 4). There was associated surrounding soft tissue edema.

The patient was taken to the operating room for excision of the lesion via a transcervical approach, under general anesthetic. The lesion was identified in the submandibular space superior to the anterior belly of the digastric muscle and inferior to the mylohyoid muscle (Fig. 5). The lesion had a smooth mucosal lining and purulence was encountered around the lesion. During dissection, the lesion was lysed, resulting in clear fluid discharge. The lesion was excised in its entirety; the specimen was placed in 10% neutral buffered formalin and sent for histopathologic examination. Biochemical analysis did not reveal elevated amylase levels and staphylococcus was cultured. Microscopic examination showed a large unicystic process lined by undulating gastric type mucosal epithelium composed of columnar foveolar epithelium and underlying gastric glands consistent with HGIC (Figs. 6A-C)

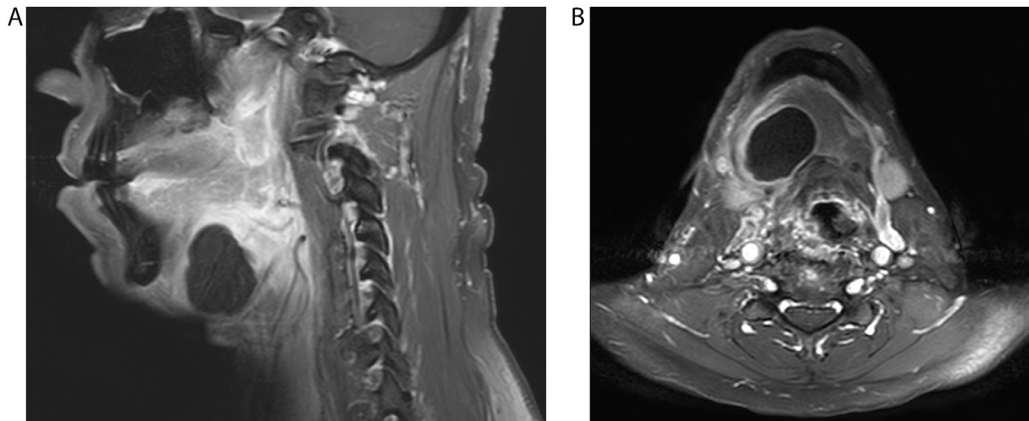


Fig. 2 – Sagittal (A) and axial (B) postcontrast T1-weighted facial bone MRI with fat suppression: The lesion is hypointense with associated surrounding abnormal soft tissue enhancement.

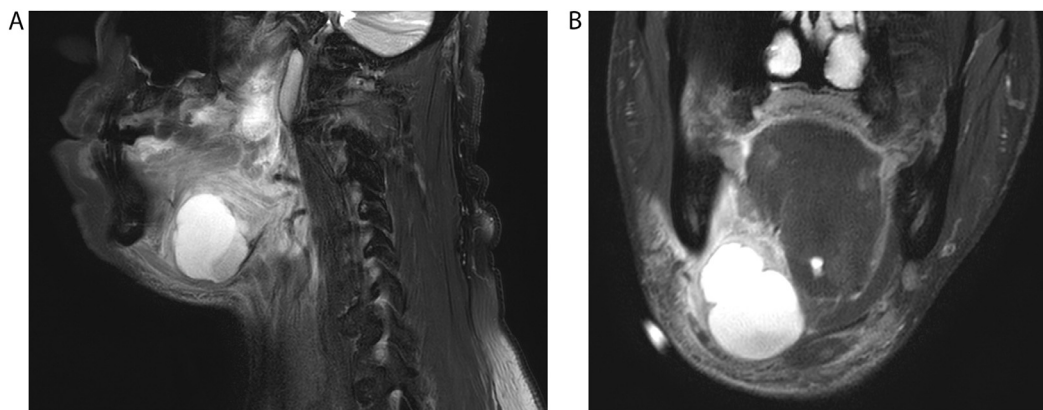


Fig. 3 – Sagittal (A) and coronal (B) T2-weighted, facial bone MRI with fat suppression: The lesion is hyperintense with associated surrounding soft tissue edema.

Postoperative antibiotics were administered for 5 days, and the postoperative course was uneventful. One-year follow-up examination showed no return of swelling, and no further imaging was performed.

Discussion

This case highlights the differential diagnosis of submandibular space lesions as clinical presentation of the HGIC can mimic other masses in the head and neck. The differential diagnosis for this benign cystic submandibular lesion includes plunging ranula, suppurative lymphadenitis, epidermal inclusion cyst, and duplication cyst. Other differential diagnosis may include dermoid cyst and thyroglossal duct cyst which are often midline.

Ranulas (mucoceles) are benign, acquired cyst-like lesions of sublingual gland origin that occur in the floor of the mouth. They are classified as simple ranula, which is confined to the sublingual space, or plunging ranula (also known as diving ranula or cervical ranula) which extends either to the poste-

rior edge of mylohyoid muscle or directly passes through a defect in the mylohyoid muscle. On CT, uncomplicated ranulas appear as thin-walled cystic spaces with central fluid attenuation. Ranulas have low signal intensity on T1WI, are hyperintense on T2WI and may show thin rim enhancement on post-contrast sequences [4,5].

Epidermal inclusion cysts are well-circumscribed lesions, most commonly located on the scalp, face, neck, trunk, and back. Epidermoid cysts contain no fat, manifest earlier in life, and are usually found during infancy [8]. The pathogenesis is by proliferation of squamous epithelium within a confined space in the dermis or subdermis and contain keratin debris. Epidermal inclusion cysts are thought to occur due to traumatic or surgical implantation, occlusion of the pilosebaceous unit, congenital rest of cells, or infection due to human papillomavirus type 57 or 60 [6,7].

Dermoid cysts are similar lesions lined by epithelium; however, they differ from epidermoid cysts in that they contain skin appendages such as sebaceous glands and hair follicles within the cyst wall [8]. Dermoid cysts occur in the second and third decades of life as slow-growing masses in the submandibular or sublingual space. Typically, dermoid cysts are

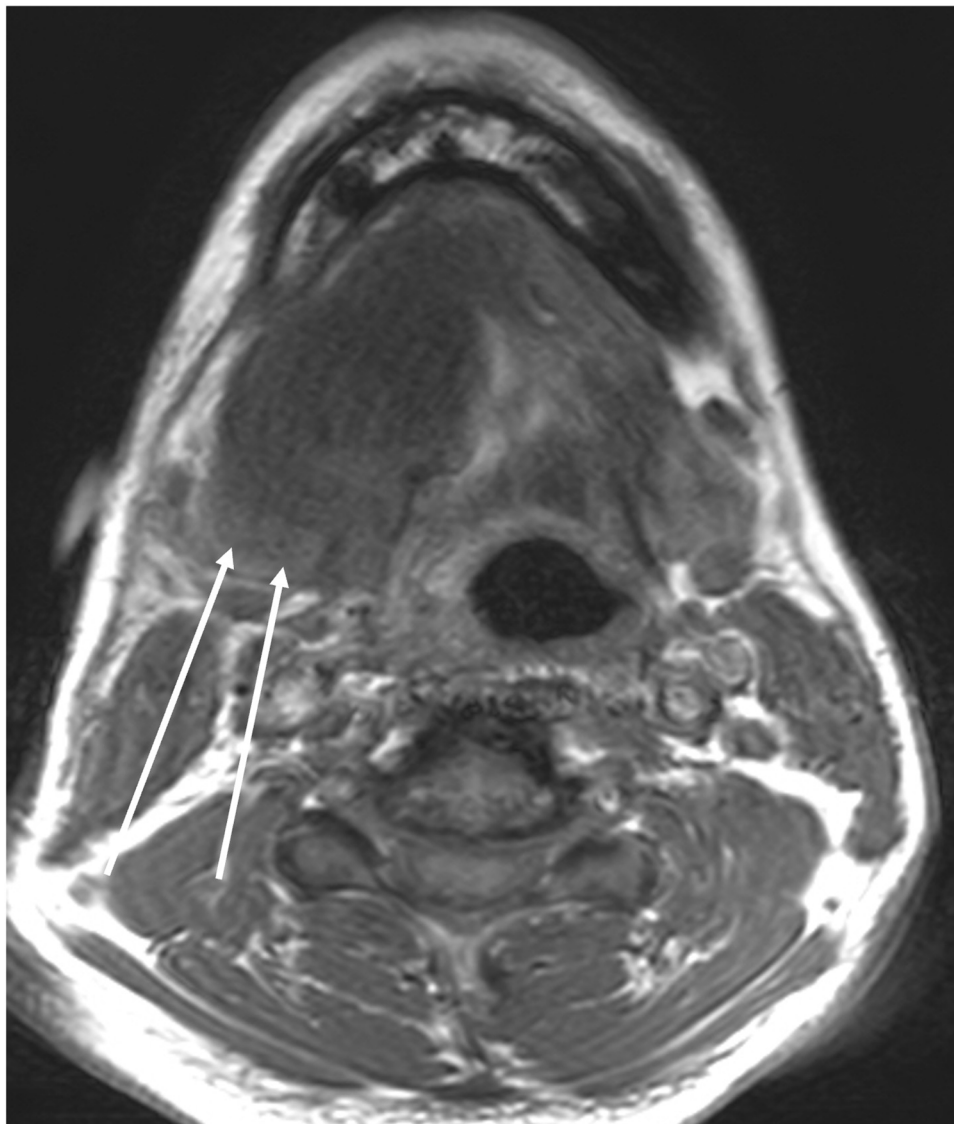


Fig. 4 – Axial T1-weighted, facial bone MRI without fat suppression: The lesion is hypointense, displacing the submandibular gland (arrows) dorsally.

midline and contain fat. On CT, dermoid and epidermoid lesions have the density of water, and margins are usually thin or sclerotic. On MR, they demonstrate low to intermediate signal on T1WI, are hyperintense on T2WI, and do not enhance [6,7].

Thyroglossal duct cysts are the most common congenital neck cyst, accounting for almost 70% of congenital neck lesions. They are typically midline lesions and generally present during childhood as a growing, painless, fluctuant cervical mass. Patients may remain asymptomatic until the cyst becomes infected, and therefore, the cyst can present at any age [9]. On CT, they are thin-walled with smooth margins, well-defined with homogeneous fluid density, and are located anteriorly in the midline or paramedial, typically within 2 cm of the midline [9]. Rim enhancement may be seen. On MRI T1WI, they have variable signal intensity. Hyperintense signal may be seen due to previous hemorrhage, infection, or high protein

material [10]. If uncomplicated, they can be hypointense on T1WI due to low protein content. On T2WI, they are typically hyperintense and on postcontrast MRI, uncomplicated lesions demonstrate no significant enhancement. However, thin peripheral enhancement may be seen.

HGICs are rare congenital lesions that occur anywhere from the oral cavity to the rectum [11]. They most commonly occur in the thorax, followed by the abdomen [12]. HGICs can be lined by a variety of epithelium such as gastric mucosa, ciliated respiratory-type epithelium, stratified squamous epithelium, and simple cuboidal epithelium [1]. There are several theories which attempt to explain the embryogenesis of HGICs; however, none fully accounts for the various types of epithelium found in these lesions [13]. The most widely accepted theory is that during fetal development there are islands of undifferentiated endoderm that are entrapped along the gastrointestinal tract [2,3]. During the third to fourth week

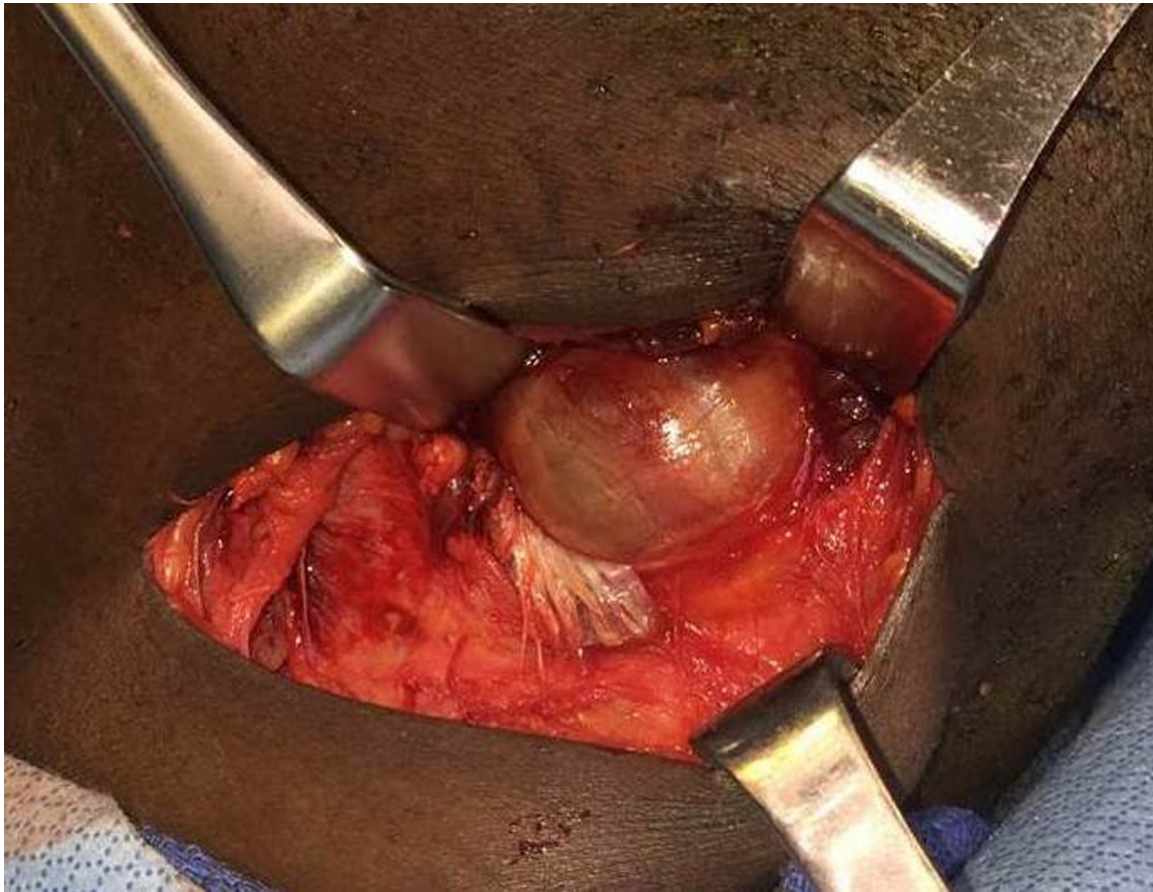


Fig. 5 – Intraoperative photo: The lesion is superior to the anterior belly of digastric muscle and superficial to the mylohyoid muscle.

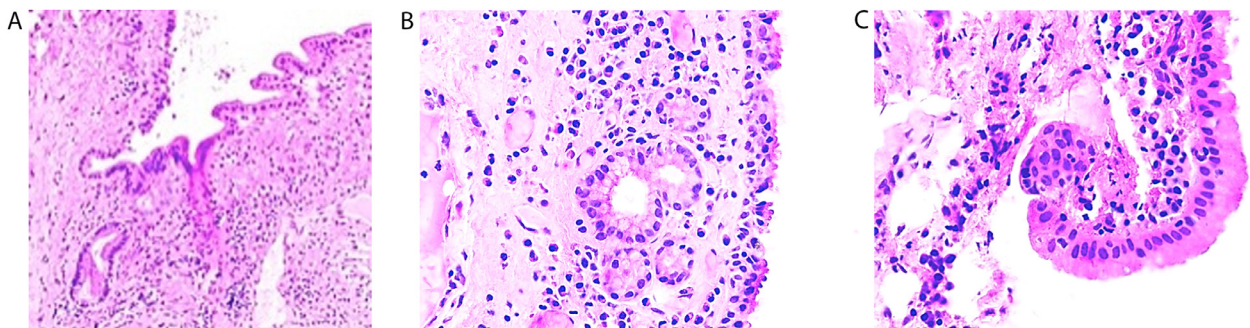


Fig. 6 – Histopathologic images of the floor of mouth heterotopic gastrointestinal cyst. (A) H&E stain, 10x magnification. Cystic process lined by undulating epithelium reminiscent of gastric mucosa and pits. (B) H&E stain, 20x magnification. Gastric epithelium and underlying glandular elements. (C) H&E stain, 40x magnification. Gastric foveolar cells lining the cyst.

of gestation, the primitive stomach contains undifferentiated endoderm, is located in the midneck, and is in close anatomic proximity to structures that will develop into the oral cavity. This may explain the etiology of oral HGICs [2].

Rarely, HGICs can be found in the head and neck; from 57 to 90 reported cases are in the oral cavity [14,15]. This wide range reported is a result of the varied nomenclature and the

many classification systems used for HGICs, all of which have shortcomings and fail to account for all reported cases.

Head and neck HGICs are most commonly diagnosed in the first decade of life with a predilection for males [3,14]. The oral cavity is the most common head and neck site, with the tongue representing 0.3% of all reported cases [16]. HGICs of the head and neck have only been reported in 13 adults. Most

Table 1 – Differential diagnosis.

	CT	T1WI	T2WI
Enteric duplication cyst	Well-circumscribed spherical or ovoid masses of variable attenuation, depending on the amount of internal proteinaceous content.	Variable signal intensity, ranging from low (similar to fluid) to high (due to high protein content).	Hyperintense signal intensity due to high fluid content.
Ranula	Thin-walled cystic spaces with central fluid attenuation	Low signal intensity	Hyperintense and may show thin rim enhancement on postcontrast sequences
Epidermoid and dermoid cyst	Density of water and margins are usually thin or sclerotic	Low to intermediate signal	Hyperintense and do not enhance
Thyroglossal duct cyst	Thin-walled with smooth margins, well-defined with homogeneous fluid density, and are located anteriorly in the midline or paramedial, typically within 2 cm of the midline. Rim enhancement may be seen.	Variable signal intensity. Hyperintense signal may be seen due to previous hemorrhage, infection, or high protein material. If uncomplicated, they can be hypointense due to low protein content.	Hyperintense and on postcontrast MRI, uncomplicated lesions demonstrate no significant enhancement. However, thin peripheral enhancement may be seen.

Table 2 – Summary of HGIC.

Etiology	It is hypothesized that islands of undifferentiated endoderm are separated and entrapped in the gastrointestinal tract during the third to fourth week of embryogenesis when the primitive stomach is located anatomically by the stomodeum [2,3].
Incidence	0.3% of HGICs have been reported in the tongue and even more rarely in the submandibular space [3].
Gender ratio	Male predilection [3].
Age predilection	HGICs most commonly manifest in the first decade of life [3].
Risk factors:	-
Treatment	Surgical excision
Prognosis	Curative with surgical excision.

oral HGICs are from 1 to 3 cm in size, ranging from less than 1 to 9 cm [2]. Only 30% of individuals are symptomatic, with symptoms of difficulty feeding, swallowing, and/or breathing, depending on lesion size and location [2].

On CT, HGICs are generally well-circumscribed spherical or ovoid masses of variable attenuation, depending on the amount of internal proteinaceous content [17]. They have variable signal intensity on T1WI, ranging from low (similar to fluid) to high (due to high protein content). These lesions commonly demonstrate hyperintense signal intensity on T2WI due to high fluid content [18]. Fluid-fluid levels have been described in the literature (Fig. 3 A).

Though typically benign, histologic findings of malignant transformation and adenocarcinoma have been reported in esophageal, bronchial and hepatic HGICs [19]. In 2007, Volchok et al. described the first case of adenocarcinoma arising in a lingual HGIC. Therefore, given the possibility of malignant degeneration, early surgical excision is indicated. Excision with complete removal of the cyst lining has shown to be curative [1,14] (Tables 1 and 2).

HGICs of the head and neck usually involve the tongue or submandibular space in neonates and children and are exceedingly rare in adults. Infection and malignant transformation have been reported but are rare [1]. Preoperative imaging, usually MRI, can narrow the differential diagnosis and assist with surgical planning. Surgical excision is both diagnostic and curative.

Authors' contribution

Dr Elliot Saleh: Writing - original draft, Writing - review & editing, Visualization

Dr Mohammad Mansouri: Writing - original draft

Dr Patrick J. Nolan: Writing - review & editing, Supervision

Dr Rober Kelsch: Writing - review & editing, Visualization

Dr Keivan Shifteh: Writing - review & editing, Supervision

Teaching point

In conclusion, HGIC are very rare, but should be considered in the differential diagnosis of a congenital head and neck lesion.

Patient consent

Patient signed informed consent regarding publishing their data and photographs.

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