Desmoid-Type Fibromatosis of the Tongue

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Abstract Desmoid-type fibromatosis rarely occurs in the oral tongue. Diagnostic imaging, including ultrasound, is useful for delineating the extent of the lesion and can aid in treatment planning. However, definitive diagnosis is made based on the characteristic histologic features, which include cytologically bland spindle cells with surrounding collagenous matrix and minimal mitotic activity. Immunohistochemical analysis of these lesions reveals that they are classically positive, at least focally, for smooth muscle actin, calponin and muscle-specific actin. These features are exemplified in this sine qua non radiology-pathology correlation article.

Keywords Desmoid · Fibromatosis · Ultrasound · Pathology

Case Presentation

A 25-year-old male presented to the outpatient clinic for chief complaint of tongue fullness, but no other significant past medical or surgical history. On physical exam, a submucosal mass on the underside of the tongue was

minimally tender to palpation. Further workup, including CBC and inflammatory markers, such as ESR and CRP, was negative.

Radiographic Features

Doppler ultrasound was performed, which showed a well-defined, heterogeneous, but predominantly solid, hypoechoic, and hypovascular mass within the oral tongue (Fig. 1). The differential considerations for this unusual lesion included minor salivary gland neoplasms, sarcomas, and lymphoma.

Diagnosis and Treatment

Following the ultrasound, the patient was referred to otolaryngology for further evaluation and underwent trans-oral biopsy with pathology revealing squamous mucosa with dense fibrosis and diffuse chronic inflammation. Subsequently the patient underwent excision of the mass. Intraoperatively a 2 cm dense mass in the right anterior tongue was identified which appeared to be submucosal. The mass was excised in its entirety and sent to pathology (Fig. 2). Microscopic evaluation revealed a bland, fasciculated spindle cell proliferation with pale eosinophilic cytoplasm and oval nuclei with occasional small nucleoli; numerous compressed thin-walled blood vessels were noted and the spindle cells infiltrate entrapped surrounding skeletal muscle (Figs. 3 and 4). The margins were negative. Immunostains were performed, which revealed strong and diffuse nuclear staining for beta-catenin and negative staining for S-100 and smooth muscle actin. The pathological findings were consistent with desmoid-type fibromatosis.

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Fig. 1 The transverse Doppler ultrasound image shows a heterogeneous, but predominantly solid nonvascular mass within the oral tongue

Fig. 2 Gross specimen photograph shows a smooth and rubbery white-tan well-circumscribed mass abutting the inked margins

Fig. 3 Photomicrograph shows bland spindle cells arranged in sweeping fascicles with infiltration into surrounding skeletal muscle and intermixed medium sized blood vessels ($\times 4$)

Fig. 4 Photomicrograph shows that the lesional cells have pale eosinophilic and fibrillary cytoplasm, oval nuclei with occasional small nucleoli, indistinct cytoplasmic borders, and intermixed stromal collagen ($\times 40$)

Discussion

The differential diagnosis of submucosal tongue lesions in the pediatric population is broad and includes inflammatory/reactive processes, developmental, benign tumors and tumor-like conditions, and malignant neoplasms [1]. Benign tumors and tumor-like conditions include hemangioma (and vascular malformations), fibroma, granular cell tumors, neurofibroma, lipoma, teratoma, and leiomyoma. Malignant tumors of the tongue are most often of epithelial origin, with squamous cell carcinoma being most common. Otherwise, submucosal neoplasms most frequently originate from the minor salivary glands and most frequently are adenoid cystic carcinoma and mucoepidermoid carcinoma [1], while floor of mouth lymphomas and oral tongue sarcomas are rare, representing $<1\%$ of all lingual neoplasms [2, 3].

CT and MR imaging play an important role in the evaluation and work up of tongue lesions, as they provide good anatomic detail and are effective in delineating the extent of the lesion and relationship to surrounding structures, with MRI providing superior soft tissue resolution [1, 4, 5]. Besides CT and MRI, ultrasonography (US) is an effective tool in the evaluation of oral cavity lesions; advantages include lack of ionizing radiation and can be performed quickly with minimal discomfort to the patient [1]. In addition, US can be used to provide guidance for diagnostic or therapeutic purposes, including aspiration/drainage or biopsy. It is important to note however that determining the etiology of tongue lesions is usually difficult based on imaging alone, thus the patient's history and physical examination should always be considered when interpreting the imaging findings.

Desmoid-type fibromatosis (aggressive fibromatosis) is a rare entity characterized by a fibroblastic proliferation arising from connective tissue, fascia and muscle aponeurotic structures [4–7]. It is usually seen in adolescents and young adults with peak incidence at 25–35 years of age. Females are more commonly affected and there exists no racial predilection [4, 7, 8]. Multiple etiologies have been implicated with this entity, including genetic, endocrine (associated with high estrogen status as seen in oral contraceptive use and young females during or following pregnancy), trauma, and as a sequelae of prior surgery; however, sporadic cases are common as well [4–9]. It is categorized as extra-abdominal, abdominal and intra-abdominal regions based on location of origin [4, 6, 8]. The head and neck is not a common site for these lesions [6], with the tongue being an extremely rare location [8].

Imaging features vary depending on the histologic composition of the tumor. On ultrasound, these lesions usually appear ill-defined and hypoechoic with variable vascularity on color Doppler imaging. CT demonstrates an ill-defined infiltrative soft tissue mass with variable attenuation and mild enhancement on post-contrast sequences [4, 5]. MRI demonstrates a heterogeneous soft tissue mass extending along fascial planes and sometimes encasing adjacent vascular and neuronal structures; signal intensity is variable depending on the extent of collagen present with low signal on T1 and T2 usually implying mature collagen/fibrosis; in fact a helpful and distinguishing imaging feature of desmoid-type fibromatosis on MRI is nonenhancing low signal intensity bands on T1 and T2 weighted images representing these areas of fibrosis [4, 5]. Variable enhancement is usually apparent, analogous to CT. MRI is the imaging modality of choice to assess the extent of tumor, including evaluation of the adjacent vascular and neural structures [4, 5].

Gross pathologic features include a firm glistening mass resembling scar tissue; the margins are usually poorly circumscribed with peripheral extension of fibrous septae [4, 5]. Microscopically, it is characterized by a monotonous proliferation of cytologically bland spindle cells with surrounding collagenous matrix and minimal mitotic activity [5]. Immunohistochemical analysis of these lesions reveals that they are classically positive, at least focally, for smooth muscle actin, calponin, and muscle-specific actin [5]. Additionally, approximately 70–80 % of these lesions show aberrant nuclear and cytoplasmic expression of β -catenin. These tumors are usually negative for EMA, keratins, caldesmon, CD34, and S-100. As stated above, the APC gene and β -catenin gene CTNNB1 have been implicated in the pathogenesis of desmoid-type fibromatosis; both of these genes are part of the Wnt signaling pathway and mutations affecting those genes result in stabilization of β -catenin which leads to accumulation within the

nucleus and cytoplasm [5, 6], thus leading to an abnormal staining pattern for β -catenin within the nucleus and cytoplasm, as opposed to the normal pattern of staining which is only cytoplasmic. Somatic mutations in CTNNB1 have been linked to sporadic cases of desmoid-type fibromatosis [5].

Various treatments have been reported for the lesions including surgical resection, radiation, and chemotherapy. Complete surgical resection is the treatment of choice and can be potentially curative [5, 7]. However, this is sometimes not feasible depending on the extent of tumor and location such as the head and neck, where critical structures and delicate anatomy can be major limiting factors. In addition, because of the locally aggressive and infiltrative nature, recurrence is common after surgical resection [5, 6]. Nevertheless, the well-defined nature of the fibromatosis in this case enabled successful complete surgical resection. Radiation has yielded mixed results and sometimes is not feasible due to location and associated toxicity; therefore, radiation therapy is usually reserved for cases with positive surgical margins or recurrence after surgery [5]. Chemotherapeutic agents have shown some positive results in lesions that are unresectable or those that recur after surgery. Various agents, both non-cytotoxic and cytotoxic, have been used; non-cytotoxic agents include Tamoxifen and non-steroidal anti-inflammatory drugs while cytotoxic agents include Methotrexate, Doxorubicin, and Vinblastine [5].

References

1. Aydil U, Kizil Y. Tongue ultrasonography in the diagnosis of a submucosal tongue infection mimicking a neoplasm: a case report. *Gazi Med J*. 2012;23:65–8.
2. de Fatima Correia-Silva J, Duarte E, Lacerda J, de Sousa S, Mesquita R, Gomez R. Alveolar soft part sarcoma of the tongue. *Oral Oncol Extra*. 2006;42:241–3.
3. Leroy X, Delobelle A, Lefebvre J, Cabaret V, Bloget F, Vilain M. Epithelioid sarcoma of the tongue. *J Clin Pathol*. 1997;50:869–70.
4. McDonald E, Yi E, Wenger D. Best cases from the AFIP: extraabdominal desmoid-type fibromatosis. *Radiographics*. 2008;28:901–6.
5. Wong S. Diagnosis and management of desmoid tumors and fibrosarcoma. *J Surg Oncol*. 2008;97:554–8.
6. Ferenc T, Sygut J, Kopczynski J, Mayer M, Latos-Bielenska A, Dziki A, Kulig A. Aggressive fibromatosis (desmoid tumors): definition, occurrence, pathology, diagnostic problems, clinical behavior, genetic background. *Pol J Pathol*. 2006;57:5–15.
7. Gonzales-Garci R, Escorial-Hernandez V, Munoz-Guerra M, Rodriguez-Campo F, Reina-Duran T, Nieto-Llanos S, Naval-Gias L. Fibromatosis of the tongue: a case report. *Oral Surg Oral Med Oral Pathol Oral Radiol Endod*. 2005;100:E31–4.
8. Hoos A, Lewis J, Urist M, Shaha A, Hawkins W, Shah J, Brennan M. Desmoid tumors of the head and neck—a clinical study of a rare entity. *Head Neck*. 2000;22:814–21.
9. Horn C, Thaker H, Tampakopoulou D, de Serres L, Keller J, Haddad J Jr. Tongue lesions in the pediatric population. *Otolaryngol Head Neck Surg*. 2001;124:164–9.