Introduction

Rhabdomyomas are rare, benign tumours that are topographically divided into cardiac and extracardiac types. Cardiac rhabdomyomas occur more frequently in children and are mostly associated with phacomatoses. The extracardiac type is further subdivided morphologically and clinically into adult, fetal, and genital types, and rhabdomyomatous mesenchymal hamartoma. The adult type appears more frequently in the head and neck region, and it is believed to originate from the skeletal muscle of the third and fourth branchial arches. Most patients are between 40 and 70 years old. We report a case of pyriform sinus rhabdomyoma, together with a review of the literature.

Case report

A 62-year-old man was referred for evaluation; he had a low-grade fever and throat pain that had started 48 hours previously. Clinical examination showed erythema of the oropharyngeal mucosa with bilateral slight enlargement of some lymph nodes, suggestive of viral pharyngitis. Flexible endoscopy incidentally revealed the presence of a submucosal mass located in the right pyriform sinus; vocal cord function was normal, and there were no signs of infection.

A contrast-enhanced computed tomography (CT) of the neck was performed and showed a well-demarcated diffusely hyperattenuating submucosal mass that involved the right pyriform sinus. The mass narrowed the airway lumen and extended from the right pyriform sinus to the right true vocal cord. Magnetic resonance imaging (MRI) of the neck was additionally performed a few days later. MRI showed a mass surrounded by soft tissue, approximately 2 cm in diameter, that occupied and obscured the right pyriform sinus, with no cartilage erosion and no expansion to the parapharyngeal space (Figure 1). The mass appeared slightly hyperintense relative to muscle on T1- and T2-weighted MRI images, and there was mild, diffuse enhancement after intravenous administration of gadolinium.

The patient underwent direct laryngoscopy under general anesthesia, and the mass was removed transorally with a carbon dioxide laser. Microscopically, the tumour demonstrated features of adult-type rhabdomyoma.

The postoperative course was uneventful, and the patient was discharged on the second postoperative day. One year after surgery, the patient had no signs of recurrence.
was asymptomatic, and endoscopy and MRI of the hypopharynx did not reveal signs of recurrence (Figure 2).

**Discussion**

Extracardiac rhabdomyomas are uncommon neoplasms of mesenchymal origin; they comprise less than 2% of all striated tumours. Nearly 90% of adult rhabdomyomas are found in the head and neck region, where they present as a solitary, slowly growing mass. However, approximately 3-10% of adult rhabdomyomas are multifocal. The most common sites are the floor of the mouth, tongue, palate, lips, cheeks, and larynx, and the parapharyngeal and submandibular spaces. Rhabdomyoma usually affects people after the fourth decade of life, with a male-to-female ratio of 3:1. Rarely, rhabdomyomas have been found in the extremities, esophagus, stomach, and mediastinum.

The clinical presentation of rhabdomyoma usually includes a slowly growing, painless cervical mass. The associated symptoms are dysphagia, hemoptysis, hearing loss, and hoarseness. In some cases, the tumour may be the cause of airway obstruction and sleep apnea, whereas in about 10% of patients, the tumour is asymptomatic.

The microscopic features of rhabdomyoma are extremely characteristic, and mimic those of skeletal muscle. The tumour consists of polyhedral-to-elongated intensely stained by eosin cells with bland nuclei, intermingled with cells with clear cytoplasm. The latter may be univacuolated or multivacuolated, possibly as a consequence of glycogen or fat accumulation, which dissolves during tissue processing. The immunohistochemical features of rhabdomyoma include cytoplasmic positivity for muscle-specific actin, desmin, myoglobin, and myo-D1, while the immunoreactivity for vimentin, S-100 protein, Leu-7, and smooth muscle actin varies. The differential diagnosis includes granular cell tumours, well-differentiated rhabdomyosarcoma, extra-adrenal paraganglioma, and hibernoma. Differential diagnosis between rhabdomyoma and rhabdomyosarcoma is of the utmost importance. Rhabdomyosarcoma appears as a rapidly growing mass, and it is usually associated with pain and paresthesia. The histopathology of rhabdomyosarcoma includes the presence of spindle or rounded cells, with very occasional cross-striations, and atypical nuclei.

Diagnosis by fine-needle aspiration (FNA) may be challenging, and granular cell tumours are commonly confused with adult rhabdomyoma. Both of these tumours have abundant eosinophilic granular cytoplasm and round, bland nuclei that are centrally or eccentrically located. However, a correct diagnosis can be achieved if the cytopathologist is
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aware of the possibility of adult rhabdomyoma and uses a limited panel of immunostains, including desmin, myoglobin, and S-100.

In the preoperative assessment, CT and MRI scans are frequently used. The submucosal location, as well as the absence of invasion into surrounding tissues, may help to distinguish adult rhabdomyoma from malignant lesions. On T1- and T2-weighted MRI, rhabdomyoma is isointense or slightly hyperintense compared to muscle. Nevertheless, imaging alone may not clearly differentiate rhabdomyomas from other neoplasms.

Surgical excision is the cornerstone of treatment for adult rhabdomyoma. Various approaches have been used, depending on tumour location. Transcervical approach is the most common technique used for excision of tumours of the lower parapharyngeal space, since it provides direct access to the parapharyngeal space with adequate exposure of the neurovascular structures. The transoral approach obviates the need for dissection through healthy tissue to access the tumour, thus minimizing blood loss. In addition, the simultaneous use of a carbon dioxide laser provides precise and bloodless cutting, with a better view of the surgical field. Recurrence rates vary from 10% to 42%. The tumour may recur even 35 years after the first excision. Inadequate removal and multifocality are the most common factors associated with recurrence.

A PubMed search of the literature revealed that since 1948, only ten cases of adult rhabdomyoma involving the hypopharynx have been reported. Eight of these patients were male, and most of the tumours were excised with an external surgical approach. Transoral laser excision was used in only two cases, including our patient. A transoral approach with the use of a carbon dioxide laser is an effective procedure for the total, precise, and bloodless resection of rhabdomyomas of the hypopharynx, limiting the need for an external transcervical approach.

In conclusion, rhabdomyoma represents a rare benign pathology of the hypopharynx. Thorough endoscopy and MRI are valuable tools in the diagnosis of this tumour. The characteristic microscopic and immunohistochemical features of rhabdomyoma allow it to be easily differentially diagnosed from other benign tumours. Total excision is the cornerstone of treatment, and transoral laser resection is an effective procedure in patients with rhabdomyoma of the hypopharynx.

References


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