OXFORD

Oxford Medical Case Reports, 2023, **1**, 17–19

https://doi.org/10.1093/omcr/omac148

Case Report

Massive nasopharyngeal rhabdomyosarcoma in an adult patient: a rare case report

Christopher Seifen *, Christoph Matthias and Karoline O'Brien

Department of Otolaryngology, Head and Neck Surgery, University Medical Center Mainz, Mainz, Germany

*Correspondence address. Department of Otolaryngology, Head and Neck Surgery, University Medical Center Mainz, 55131 Mainz, Germany. E-mail: kim.seifen@unimedizin-mainz.de

Abstract

Rhabdomyosarcoma (RMS) is a highly invasive malignant soft tissue sarcoma that rarely affects adults. The head and neck region accounts for most adult RMSs. Here, we report a case of a 24-year-old Caucasian woman who was diagnosed with persistent hoarseness and a painless left-sided neck mass. Physical examination and image studies showed a massive tumor of the nasopharynx, extending from the left-sided skull base to supraglottic structures. Histopathologic evaluation revealed the diagnosis of a poorly differentiated RMS. Due to the primary tumor size and involvement of crucial structures, extensive surgical excision was not amenable. Thus, the patient was treated with radiotherapy and chemotherapy. Although very rare, nasopharyngeal RMSs should be considered in the differential diagnosis of neck masses in adult patients. This case report illustrates the difficulty in the diagnosis and treatment of rare head and neck malignancies and encourages its reporting.

INTRODUCTION

Rhabdomyosarcoma (RMS) was first mentioned by Weber in Virchow's Archives in 1854 and describes a malignant soft tissue neoplasm of skeletal muscle origin [1]. RMS constitutes more than 50% of all soft tissue sarcomas in children but is exceedingly rare in adults. Less than 1% of all adult malignancies are soft tissue sarcomas, of which RMS accounts for only 3% [2]. The most common histologic variants encountered in children and adolescents are embryonal and alveolar subtypes, whereas the pleomorphic subtype and RMS not otherwise specified are more frequently seen in adult patients [3]. RMS can arise in a variety of anatomic sites throughout the body but the most common primary tumor sites include the head and neck region, followed by the genitourinary and extremity primaries [4]. Routinely, patients are diagnosed with an asymptomatic mass or show symptoms that are associated with mass effects of the primary tumor site [4]. Diagnosis is made by image studies including computed tomography (CT) scan or magnetic resonance imaging (MRI) as well as tissue biopsies of the suspected mass. Treatment of RMS is multimodal and includes surgery, radiotherapy and chemotherapy. However, RMS of the head and neck region are rarely amenable to extensive surgical excision due to proximity of vital structures. The outcome of adults with RMS appears to be worse than that of children with a 5-year overall survival rate of 27% versus 61%, respectively [4]. To date, only a handful of cases of nasopharyngeal RMS in adult patients have been reported in the English literature. We report a case of an aggressive pulmonary metastasized RMS of the nasopharynx in an adult patient who was initially misdiagnosed and treated conservatively.

CASE REPORT

A 24-year-old Caucasian woman was referred to our outpatient department for persistent hoarseness and a painless left-sided neck mass. Both symptoms had developed 6 weeks ago and progressively worsened. Additionally, cervical radiculopathy occurred and was treated with non-beneficial physical therapy. Apart from being a premature born twin, status post-appendectomy and status post-tonsillectomy, the patient's past medical and family histories were unremarkable. In physical examination, a painless, hard and non-mobile left-sided neck mass could be found in level II. With ultrasound of the neck a non-specific hypoechoic mass could be correlated (Fig. 1). Endoscopic examination showed the left-sided palatal arch and pharyngeal wall to be protracted and unilateral paralysis of the left vocal cord. The patient's tongue pointed leftwards and showed muscle fasciculation. Laboratory testing showed normal blood count and non-elevated inflammation parameters. A contrast-enhanced MRI of the head and neck was performed and revealed a 6.1 cm × 3.1 cm × 7.0 cm well-circumscribed, homogeneous, contrast-enhanced tumor extending from the left-sided skull base to supraglottic structures (Fig. 2). Also, two suspicious lymph nodes were found in the left-sided neck in level II. Additionally, a contrast-enhanced CT angiography showed the lateralization of the carotid artery and indentation of the internal jugular vein but ruled out infiltrative tumor growth and bone erosion (Fig. 3). Naso- and oropharyngeal tissue biopsies of the tumor plus ultrasound-guided tissue biopsies of the suspicious lymph nodes were performed. Histopathological evaluation was performed in our pathology department plus a referral center and revealed the diagnosis of a poorly differentiated

Received: October 3, 2022. **Revised:** November 24, 2022. **Accepted:** November 28, 2022

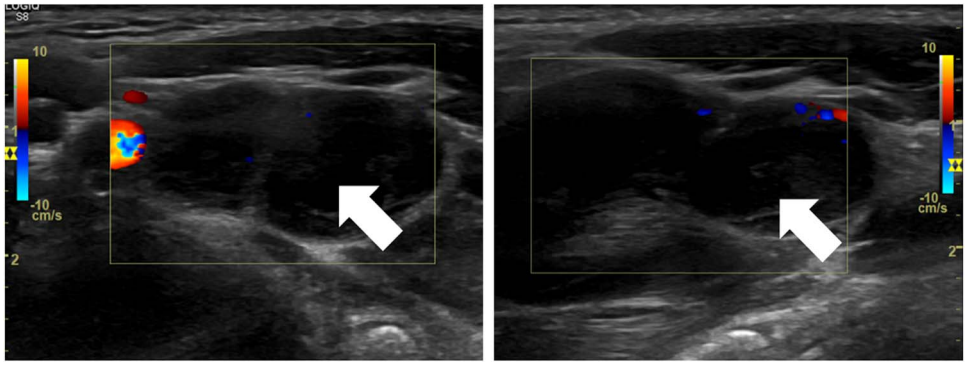

Figure 1. Ultrasound of the left-sided neck showed a non-specific hypoechoic mass. Left: Transverse view. Right: Sagittal view. The arrows point to the non-specific neck mass.

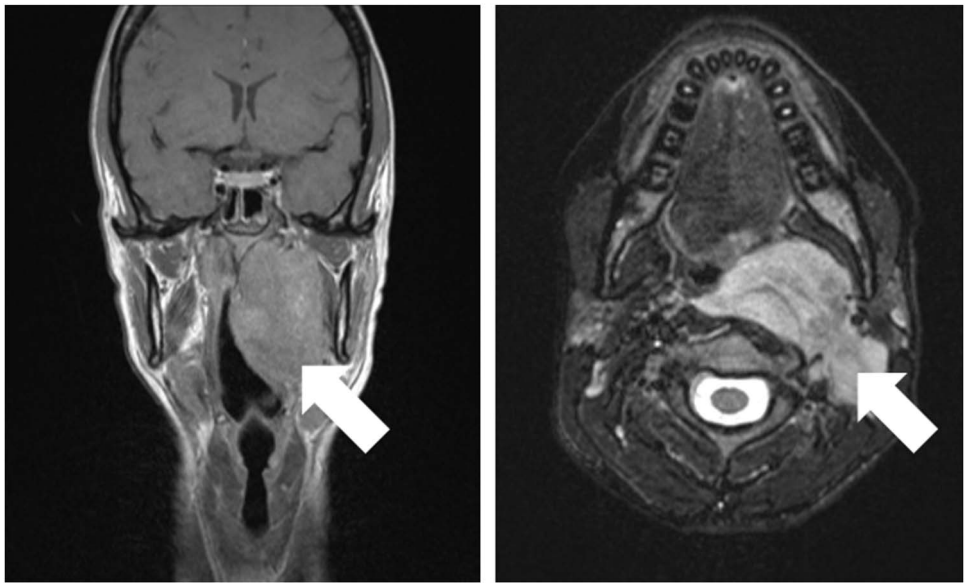

Figure 2. Contrast-enhanced MRI of the head and neck showed the full extent of nasopharyngeal RMS. Left: coronar view, T1-weighted. Right: axial view, T2-weighted. The arrows point to the tumor.

RMS, whereas patterns of alveolar RMS or Myo-D1 mutant could not be identified. The case was discussed in an interdisciplinary tumor board. In conclusion, the tumor was defined as a poorly differentiated RMS of the nasopharynx with bilateral pulmonary metastases (T4N1M1). Rapidly emerging symptoms as recurrent syncope and increasing paralysis of affected cranial nerves IX, X and XII favored emergency irradiation of the parapharnygeal space and skull base, which was initiated 1 month after the first consultation of the patient. The patient received a total dose of 54 Gray (Gy) in 30 fractions (1.8 Gy/fraction), followed by three courses of chemotherapy according to CWS guidance. At the time this case report was prepared, the patient received the last course of chemotherapy.

DISCUSSION

Anatomically, RMS of the head and neck are differentiated as parameningeal or nonparameningeal. Parameningeal RMS include RMS of the nasopharynx, paranasal sinuses, nose, middle ear, mastoid, infratemporal fossa and pterygopalatine fossa, whereas nonparameningeal RMS include RMS of the orbit, parotid gland, oral cavity, oropharynx, larynx and scalp [5]. The head and neck region accounts for most RMS, with the orbit being the most frequent primary tumor site, followed by the oral cavity. In contrast, RMS rarely presents originating from the nasopharyngeal space as seen in our patient [4]. In our case, when the patient was admitted at our institution, she had asymmetric endoscopic findings and experienced hoarseness as well as a painless left-sided neck mass. A variety of clinical symptoms can occur with soft tissue carcinomas mostly lacking a cardinal sign. Therefore, most patients have an advanced disease at the stage of initial presentation. RMS exhibit fast and aggressive tumor growth, reaching large dimensions, and are typically painless but associated with generalised metastases and high rates of recurrence [3, 4]. The most common site of metastasis is the lung, followed by metastases to the bone marrow. Other common metastases are found in bone or distant nodes [6]. RMS often is difficult to diagnose because of its resemblance with other malignancies of the head and neck region. To date, no clear risk factors for RMS have been identified. Compared with other soft tissue carcinomas, prognosis of RMS is relatively poor but depends on clinical staging and the anatomical site of the tumor [2, 3]. When such lesion is suspected, tissue biopsy analysis is the key to diagnosis, and should be performed as soon as possible. A careful histological examination is required to differentiate RMS

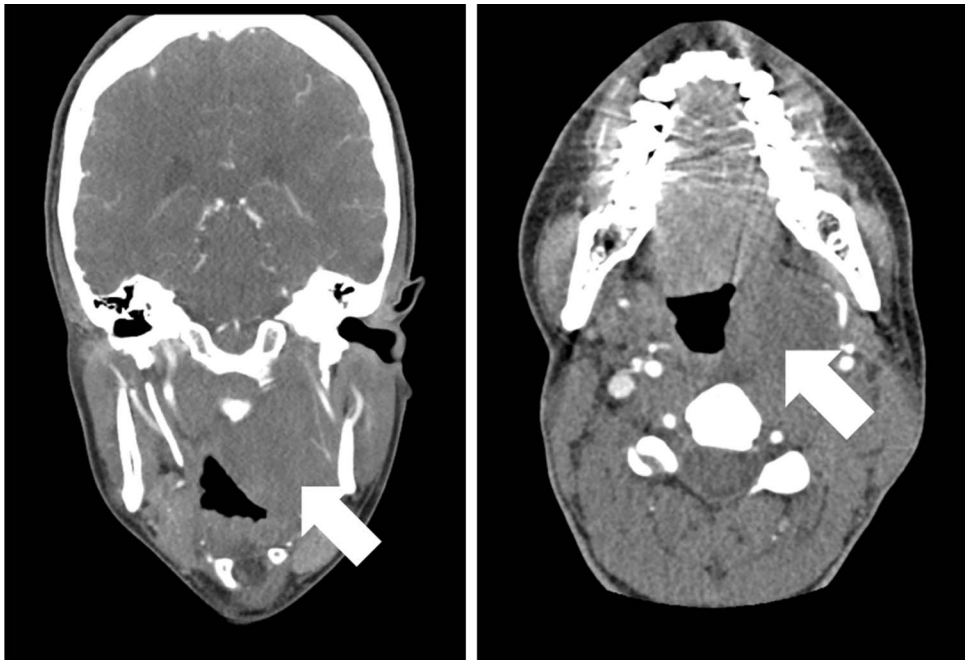

Figure 3. Contrast-enhanced CT angiography showed lateralization of the carotid artery and indentation of the internal jugular vein but ruled out infiltrative tumor growth and bone erosion. Left: coronar view. Right: axial view. The arrows point to the tumor.

from other more frequent and aggressive lesions affecting the concerned site. For the head and neck region, the embryonal subtype is most commonly found and frequently occurs in children and adolescents [3, 4]. Once a diagnosis is made, a combined, multimodal therapeutic approach involving surgery, radiotherapy and chemotherapy is needed for the management of RMS. The basic goal of RMS therapy is locoregional control, and prevention or treatment of systemic metastases [7]. To the best of our knowledge, this is the first reported case of pulmonary metastasized poorly differentiated RMS of the nasopharynx in an adult patient. This case report demonstrates the importance of a multidisciplinary diagnostic approach in cases of rare malignancies. Early image studies and timely biopsies should be of notice when RMS is suspected to accelerate the start of a targeted and appropriate therapy. Physicians should be aware of various atypical presentations of rare malignancies, so that further measures can be taken. Nasopharyngeal RMS, although very rare, should be considered in the differential diagnosis of neck masses in adult patients.

FUNDING

None.

CONFLICT OF INTEREST STATEMENT

None declared.

GUARANTOR

Professor Matthias, Professor of Otolaryngology, Head and Neck Surgery, University Medical Center Mainz, 55131 Mainz, Germany.

PATIENT CONSENT FORM

Written informed consent was obtained from the patient for publication.

REFERENCES

1. Weber CO, Virchow R. Anatomische Untersuchung einer hypertrophischen Zunge nebst Bemerkungen über die Neubildung quergestreifter Muskelfasern. *Virchows Arch* 1854;**7**:115–25. https://doi.org/10.1007/BF01936232.
2. Ferrari A, Dileo P, Casanova M, Bertulli R, Meazza C, Gandola L, *et al.* Rhabdomyosarcoma in adults. A retrospective analysis of 171 patients treated at a single institution. *Cancer* 2003;**98**:571–80. https://doi.org/10.1002/cncr.11550.
3. Sultan I, Qaddoumi I, Yaser S, Rodriguez-Galindo C, Ferrari A. Comparing adult and pediatric rhabdomyosarcoma in the surveillance, epidemiology and end results program, 1973 to 2005: an analysis of 2,600 patients. *J Clin Oncol* 2009;**27**:3391–7. https://doi.org/10.1200/JCO.2008.19.7483.
4. Egas-Bejar D, Huh WW. Rhabdomyosarcoma in adolescent and young adult patients: current perspectives. *Adolesc Health Med Ther* 2014;**5**:115–25. https://doi.org/10.2147/AHMT.S44582.
5. Al-Khateeb T, Bataineh AB. Rhabdomyosarcoma of the oral and maxillofacial region in Jordanians: a retrospective analysis. *Oral Surg Oral Med Oral Pathol Oral Radiol Endod* 2002;**93**:580–5. https://doi.org/10.1067/moe.2002.123860.
6. Oberlin O, Rey A, Lyden E, Bisogno G, Stevens MC, Meyer WH, *et al.* Prognostic factors in metastatic rhabdomyosarcomas: results of a pooled analysis from United States and European cooperative groups. *J Clin Oncol* 2008;**26**:2384–9. https://doi.org/10.1067/moe.2002.123860.
7. Abbas A, Awan S. Rhabdomyosarcoma of the middle ear and mastoid: a case report and review of the literature. *Ear Nose Throat J* 2005;**84**:780–4. https://doi.org/10.1177/014556130508401214.