Endolymphatic Sac Tumor: A Case Report

Tumeur du sac endolymphatique: A propos d'un cas

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Received: 18 May 2021; Accepted: 22 September 2021; Published online: 31 October 2021

ABSTRACT

Objective: Endolymphatic sac tumor (Heffner tumor/ELST) is benign, indolent but invasive arising in the posterior petrous ridge. ELST could occur sporadically or as a part of von Hippel–Lindau (VHL) disease. Morphologically, these tumors showed a papillary and glandular architecture. We report a case of endolymphatic sac tumor to discuss the diagnostic and therapeutic features of this rare entity.
Observation: A 58-year-old woman presented with one-year history of slowly progressing unilateral hearing loss, tinnitus and vertigo. The patient was diagnosed with an endolymphatic sac tumor and was managed with a radiation therapy.
Conclusion: Endolymphatic sac tumor (ELST) is a rare low-grade adenocarcinoma of the skull base. Diagnosis is based on clinical, radiological, and pathological correlation and the optimal treatment is surgical resection with security respected margins. Radiotherapy remains controversial. A long term follow up is recommended.
Key-words: Endolymphatic sac tumor; Papillary tumor; Von Hippel-Lindau disease.

RÉSUMÉ

Objectif: La tumeur du sac endolymphatique (tumeur de Heffner) (ELST) est une tumeur bénigne, indolente mais invasive située dans la partie postérieure de l'os pétreux. ELST peut être sporadique ou s'intégrer dans une maladie de von Hippel – Lindau (VHL). Morphologiquement, ces tumeurs présentent une architecture papillaire et glandulaire. Nous rapportons un cas d'une tumeur du sac endolymphatique afin de discuter les particularités diagnostiques et thérapeutiques de cette entité rare.
Observation: Une femme de 58 ans s'est présentée avec une hypoacousie unilatérale à progression lente, des acouphènes et des vertiges évoluant depuis 1 an. La patiente a été diagnostiquée d'une tumeur du sac endolymphatique et a été traitée par une radiothérapie.
Conclusion: La tumeur du sac endolymphatique (ELST) est un adénocarcinome de bas grade rare de la base du crâne. Le diagnostic repose sur une corrélation clinique, radiologique et pathologique et le traitement optimal est une résection chirurgicale avec des marges de sécurité respectées. La radiothérapie reste controversée. Un suivi à long terme est recommandé.
Mots-clés: Tumeur du sac endolymphatique; Tumeur papillaire; Maladie de von Hippel-Lindau.

INTRODUCTION

Endolymphatic sac tumors (ELST) is slow-growing, locally aggressive and low-grade malignant tumor that originates from the epithelium of the endolymphatic duct or sac which located in the posterior petrous bone [1,2]. This tumor may be presented as sporadic or may be associated with von Hippel-Lindau (VHL) disease in 11%-16% of the reported cases [1,3]. It was first described by Hassard et al. in 1984 as an adenoma of the endolymphatic sac [1,4,5,6]. They described a highly vascular, reddish, lobular mass eroding the sheath of the endolymphatic sac removed during decompression surgery of the sac in a patient presumed having Meniere's disease sac [5]. In 1989, ELST was defined by Heffner as individual adenomatous temporal bone tumors. According to the recently published World Health Organization tumor classification, ELST is a very uncommon papillary epithelial neoplasm arising within the endolymphatic sac or endolymphatic duct. This tumor shows a very high association with von Hippel-Lindau syndrome (VHL) [1]. Although it is a benign tumor involving the temporal bone, its pattern of growth is often invasive. In advanced cases, it can invade the posterior and/or middle cranial fossae [6]. This tumor is a hypervascular lesion that may erode the adjacent bony and vascular structures. The earliest manifestations of ELST are often sensorineural hearing loss and vertigo. Surgery is the treatment of choice for small ELST. Patients with residual mass or unresectable tumor are treated with radiotherapy or radiosurgery. We present a rare case of an endolymphatic sac tumor in a patient with VHL disease, and we will discuss the clinical, radiologic, and pathologic features of that lesion.

CASE REPORT

A 58 years old female patient was admitted in March 2018 to our Otolaryngology Department with one-year history of progressive sensorineural hearing loss in the left ear, non-pulsatile tinnitus, aural fullness and disequilibrium. No complaints of nausea, vomiting, or cerebellar signs were reported. The family medical history disclosed the presence of symptoms of von-Hippel–Lindau (VHL) disease. Her mother, brother and three nephews had history of resection of cerebellar hemangioblastomas and evidence of genetic VHL disease.

Otoscopic examination revealed bilateral normal tympanic membranes and clear external auditory canals. Pure tone audiometry showed left side mild sensorineural hearing loss. Computed tomography (CT) found a low-density lesion with enhancement on contrast arising from the left side of the cerebellopontine angle region. Erosion of the left petrosal bone and mastoid air cells was observed, extending to the internal auditory meatus (figure1). On MRI study, a 32 × 18 × 26 mm hyper vascular lesion was detected in the petrous part of the left temporal bone and mastoid air cells. The mass was hypointense on T1 and hyperintense on T2 with heterogeneous contrast enhancement (figure2). The differential diagnosis included glomus tumor, intraosseous hemangioma and aneurysmal bone cyst. Moreover, a work-up was performed to confirm the von Hippel–Lindau syndrome. An ophthalmologic examination done detected multiple hemangioblastomas of the retina and an ultrasonography of the abdomen revealed bilateral renal cysts. Genetic study was not performed in our case. With these findings a provisional diagnosis of ELST associated with VHL disease was made. The patient was planned for surgical resection after embolization, but she refused surgery. Therefore, she was treated with intensity modulated radiotherapy. She is followed regularly for 36 months with routine MRI surveillance. There were no several complaints and MRI display no radiological signs of progression, the tumor size was stable.

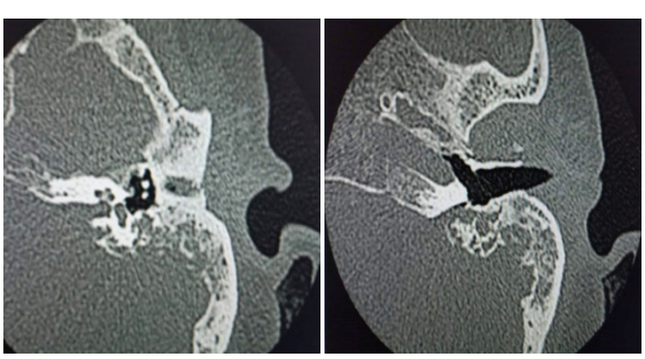

Figure.1. Axial CT images demonstrate an osteolytic lesion along the medial petrosal bone, in the location of the left endolymphatic sac. The bone window shows extensive destruction of the petrous bone, extending to the internal auditory canal and mastoid air cells.

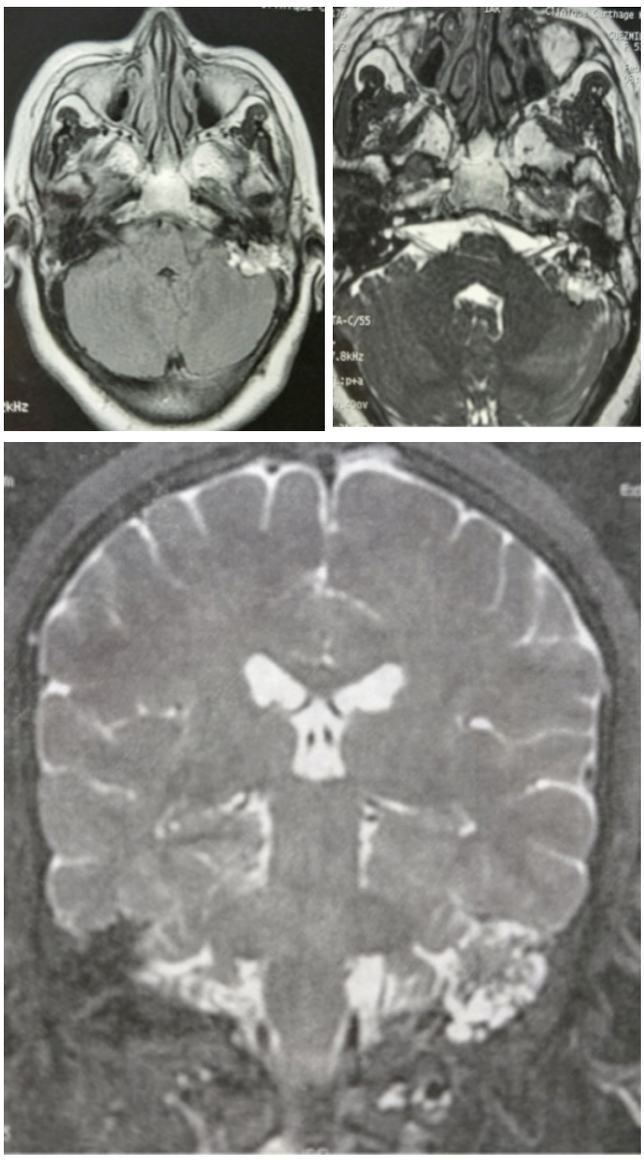

Figure.2. Axial MR images reveal a heterogeneous mass lesion involving the left endolymphatic sac and petrosal bone with extension into the left internal auditory canal. The mass is hypointense on T1 and hyperintense on T2 with heterogeneous enhancement.

DISCUSSION

According to the recently published World Health Organization tumor classification, ELSTs were classed as aggressive papillary middle ear and Heffner's tumors. This tumor was first reported in 1984 when Hassard operated for suspected endolymphatic hydrops and accidentally discovered a soft cyst [4]. In 1989, Heffner concluded that these tumors were all low-grade adenocarcinomas and were likely to originate from the endolymphatic sac [1]. ELSTs are very rare and are considered as primary neoplasm of the temporal bone [4,7].

These tumors are almost found in adults, the peak incidence is in the third and fourth decades with a wide age range [4]. Some studies report a female predominance [5].

The majority of the endolymphatic sac tumors are

sporadic in nature whereas some tumors are associated with von Hippel–Lindau (VHL) disease [1,3,5,7]. It has been reported that 11%-16% of this type of tumor arose in VHL disease known patient. This association was first established by Manski and al and approximately 30% of those patients would develop bilateral tumors [1,3,5].

VHL is an autosomal dominant disease with a variable expression (penetrance 95%) which is characterized by an inherited genetic abnormality of the VHL gene, located on the short arm of chromosome 3 (3p26-p25) [1,4,7]. The incidence of VHL disease is 1/39,000. It is a multisystem disorder characterized by multiple hemangioblastomas of the retina or central nervous system, renal or pancreatic cysts, renal cell carcinoma, pheochromocytomas, and other visceral tumors. If genetic tests confirm the presence of VHL syndrome, the patients must have annually a brain and spine MRI, ophthalmoscopy, urinary catecholamine measurement, and an abdominal CT scan [1,4].

The ELSTs are considered as a tumor of the temporal bone that arise in the endolymphatic sac or ducts [3,7]. They are slow growing, aggressive, and hyper vascular tumors that are capable of extensive destruction of the adjacent bone [8]. The endolymphatic sac is an anatomic structure that lies along the posterior and medial petrous temporal bone. As the tumor grows, it may involve the supra and infralabyrinthic, medial mastoid, and posterior tympanic cavities.

The clinical manifestations depend on tumor extension. Cochleovestibular dysfunction is the most common presenting symptom for ELST [3]. Patients often present with the gradual onset of sensorineural hearing loss and tinnitus and initially they were often misdiagnosed as Meniere's disease [3,5]. Vertigo and aural fullness are the next most common symptoms affecting 70% and 37% of patients, respectively [3]. Facial nerve palsy is seen once the tumor becomes large and involves the facial bone canal [2,6].

Early diagnosis and treatment are helpful in preserving hearing since the resection of small ELSTs could be performed safely without hearing damage, but most of the tumors are usually large when diagnosis was done [1]. In the present series, the duration was approximately at an average of five years from the first presentation.

The imaging specificity of ELST is the presence of a retrolabyrinthic mass associated with osseous erosion. Classically in Computed tomographic (CT) scans, ELST is a heterogenous enhancing lesion centered in the posterior region of the temporal bone, with surrounding destructive bone changes of 'moth-eaten' pattern [8]. Prominent intratumor (central) calcific speculation and posterior rim calcification are often seen [4,8,9]. Magnetic resonance imaging (MRI) usually reveals a heterogeneous mass which is hyperintense on contrast enhanced T1 and T2 sequences due to hypervascularity [2,5]. The heterogeneous appearance is owing to intraparenchymal bleeding, cysts, cholesterol crystals, calcifications, and flow voids due to vessels [1,2]. Early stage ELSTs involves the internal acoustic canal, the sigmoid sinus, and the medial mastoid bone. Advanced lesions could affect the posterior fossa (the area of most frequent spread), the anterior cavernous sinus, the superior middle cranial fossa (usually with destruction of semicircular canals), and the inferior jugular foramen [6].

There are no universally accepted staging recommendations. The later classification proposed by Schipper and al in 2006, distinguishes type-A, locally confined, tumors without temporal bone erosion or infiltration of the dura mater; type-B tumors showing evidence of infiltration of the osseous labyrinth and sensorineural hearing loss; and type-C tumors further invading the sigmoid sinus and jugular bulb [2].

Angiography demonstrates the hypervascularity of the tumor due to arterial blood support, which could necessitate embolization [6]. All ELST are hyper vascular [3,8]. The blood supply of these hyper vascular tumors arises predominantly from the external carotid system, but larger tumors also recruit supply from the internal carotid artery and from the vertebral arteries [6]. On histopathologic examination, endolymphatic sac tumors are commonly papillary adenoma, cystadenoma or papillary carcinomas that originate in the epithelium of the endolymphatic sac [7]. These tumors were considered as low-grade adenocarcinoma because most of the reported cases describe their benign clinical course, with locally invasive nature [9]. It has been proven that this tumor is a non-metastatic malignant tumor [1,4].

Microscopically, the general character of ELST is a papillary, cystic, and glandular neoplasm containing eosinophilic colloid-like material that usually showed positive reactions for PAS staining [5]. The histopathologic differential diagnosis for ELST includes middle ear adenoma or adenocarcinoma, jugular-tympanic paraganglioma, vascular tumors like hemangioma, meningioma,choroid plexus papilloma, metastatic thyroid carcinoma, renal cell carcinoma, and bony lesions like chondrosarcomas or eosinophilic granuloma [2,8].

Currently, the mainstay treatment of ELST is surgical resection [3,4]. Pre-operative embolization to reduce the vascularity of the tumor and facilitate complete resection is also an option if the tumor is highly vascularized. Very little data are available concerning the efficacy of non-surgical methods as primary therapy. Different skull base surgical procedures were carried out according to the different size and extension of the tumors. Due to the development of surgical techniques and the application of intraoperative neurophysiological monitoring, the risk of cranial nerve palsy was reduced [1]. Therefore, once these ELSTs are detected, they could be successfully resected with preservation of hearing and even improvement of other associated audio vestibular dysfunction.

With recent increasing use of ear endoscopy surgeries,

endoscopy was introduced in surgical management of petrous apex and lateral skull base lesions, with advantages of providing wider visualization of previously inaccessible anatomical sites [10].

The type of approach depends on the presence of functional hearing, tumor size and localization. Sporadic tumors usually show worse hearing function and have a more delayed diagnosis [2].

Radiation therapy for ELSTs remains controversial [1,6]. The role of adjuvant radiation therapy remains to be defined, as does that of gamma knife surgery [6]. Aggressive characteristics of the tumor could give credence to radiotherapy. In Friedman's retrospective survey, after long-term follow-ups, only two of three patients with subtotal resection, who received radiotherapy postoperatively, had no recurrence. However, radiotherapy before surgery is not recommended because it can't control tumor growth [1]. Therefore, radiation therapy is exclusively indicated as a palliative treatments for tumors that cannot be resected completely [7]. It has a 50% cure rate with large or residual tumors [10].

Remission may last for years, but local recurrence after surgery, likely secondary to incomplete resection, can occur. Nonetheless, most authors have described ELSTs as non-metastasizing tumors. Two reports, however, have described metastasis from an ELST [4]. This case demonstrates a difficult diagnosis of endolymphatic sac tumor, due to its rarity, non-specific presentations and atypical radiological findings. The optimal management of patients with these locally aggressive tumors includes early diagnosis, surgical excision and long-term follow-up to detect recurrent disease [6].

CONCLUSION

ELST is a rare tumor of the temporal bone. It may be an aggressive or low-grade malignant tumor, with high rate of local recurrence and bone invasion. The association between ELST and VHL disease has increased our knowledge about this rare tumor. Early surgery resection is the best treatment strategy for long-term disease-free survival. Radiotherapy is indicated as palliative treatments for tumors that cannot be resected completely. Lastly, it has been suggested that regular follow-up seems mandatory with sequential radiology.

Compliance with ethical standards

Conflict of interest: The authors stated that there is no conflict of interest.

Funding Statement: The authors received no specific funding for this work.

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