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## Endolymphatic sac tumor at the cerebellopontine angle: A case report and review of literature

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## ABSTRACT

**INTRODUCTION:** Endolymphatic sac tumors may present as sporadic or may be associated with Von Hippel-Lindau disease. Patients generally present with hearing loss, tinnitus and vertigo. The tumor is highly vascular which may lead to erosion of the adjacent bony and vascular structures, resulting in heavy bleeding during surgery.

**PRESENTATION:** A twenty-five year-old female presented with a five year history of chronic ear discharge, left sided facial weakness, and recent onset of ataxia.

**DISCUSSION:** The unusual clinical presentation made management challenging, in large part due to profuse bleeding. Pre-operative embolization of the vessels supplying the tumor may reduce blood loss during surgical excision. Radiotherapy could be considered for any residual tumor.

**CONCLUSION:** The patient was diagnosed with an endolymphatic sac tumor of sporadic origin which presented at the cerebellopontine angle and was managed with a multidisciplinary approach.

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## 1. Introduction

Endolymphatic-sac tumors (ELSTs) are rarely reported in the Neurosurgical literature. These tumors are most often associated with von Hippel-Lindau disease (VHL), another rare condition. Sporadic cases of ELSTs are extremely rare. The tumor arises in the endolymphatic sac or from the endolymphatic ducts of the inner ear. It progresses gradually, often eroding adjacent structures including the internal auditory canal, labyrinth and outer ear. Generally these large tumors involve the cerebellopontine angle (CPA) and affect its complex anatomy. The larger tumors may extend up to the bulb of the jugular vein, possibly eroding vascular structures such as the venous sinuses and their associated vessels. Eroded vessels can lead to challenging excision of the tumor due to massive bleeding. Females are more frequently affected than males with symptoms that may include progressive hearing loss, vertigo, tinnitus, dizziness, imbalance or ataxia, and vermian or hemi cerebellar syndrome. Clinical progression may present as raised intracranial pressure or headache, vomiting and pupillary dilatation. Fourth ventricular compression may lead to Hydrocephalus.

ELSTs are investigated using MRI and contrast studies. A typical radiological picture of an ELST consists of mixed intensity enhancing well with contrast. It is desirable to acquire a cerebral angiogram pre-operatively to observe any involvement of the adjacent vascular structures, the vascularity of the tumor, and the possibility of pre-operative embolization. If preoperative embolization is not possible, surgical excision is considered keeping in mind the potential for having to manage heavy bleeding with the goal of excising the entire tumor. If complete excision is not possible, the residual tumor may be treated with radiotherapy or radiosurgery. Histopathologically, the tumor is commonly papillary adenoma, cystadenoma or papillary carcinoma [1–3].

## 2. Case report

A twenty-five year old female presented with a history of chronic ear discharge over a five year period, left sided facial weakness, and a recent onset of loss of coordination in the limbs and difficulty with maintaining balance for the past 6 months. The patient was mentally subnormal with microcephaly. She was deaf, attending a school for special needs children. The patient was referred to Neurosurgery by the ENT department at the King Faisal Specialist Hospital and Research Center (KFSH&RC) in Jeddah. Clinical examination was hindered by a limited ability to directly communicate with the patient. The patient's attendant provided the history and assisted with the examination. In the past several

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Fig. 1. The tumor arising from the Petrous Bone and extending all the way down to the Base of the posterior fossa.

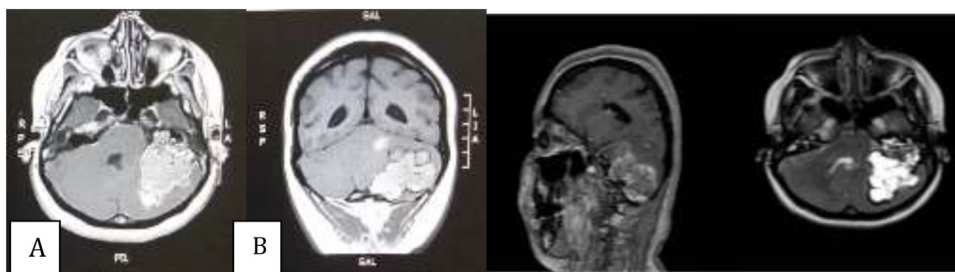


Fig. 2. Selected Images of Lesion T1 with contrast (A,B,C) and T2 axial section (D).

months, ear discharge had increased in addition to the onset of headaches and difficulty with balance.

On examination, the patient was conscious and communicated with sign language. She presented with a lower motor neuron type of left sided facial palsy, hypoesthesia of the left half of the face, ataxia including impaired gait, and positive cerebellar signs in the left sided upper and lower limbs. Involvement of the VII and VIII nerves and the sensory component of the V nerve were noted with preservation of the basal cranial nerves on the left side.

While blood work was normal, a CT scan of the brain with and without contrast revealed a mixed density lesion with enhancement on contrast arising from the left sided internal auditory canal. Erosion of the petrous bone was observed as the tumor extended through the left sided CP angle (Fig. 1) to the jugular foramen at the base of the skull. There was surrounding edema and compression of the fourth ventricle without associated hydrocephalus. The CT angiogram revealed normal anterior circulation, however, obliteration of the left transverse sinus and the left sigmoid sinus was observed. The internal jugular vein was not visualized.

A MRI, a MRA and a MRV of the lesion were obtained to provide more detailed information about the lesion, revealing a mixed intensity tumor with multiple flow voids in the center with heterogeneous contrast enhancement (Fig. 2a–d). The tumor was pushing upward on the left side occipital lobe and extending downward to the base of the skull, obliterating the left transverse sinus and compressing the left sigmoid sinus while expanding the left internal jugular vein. Complete erosion of the internal auditory meatus and canal was noted. The differential diagnosis included glomus tumor, intraosseous hemangioma and aneurysmal bone cyst.

Informed consent was obtained for a surgical intervention and the patient was prepared for left-sided retromastoid suboccipital craniotomy and excision of the tumor. The procedure was done in collaboration with ENT department.

Following general endotracheal anesthesia, the ENT team performed a retromastoid suboccipital craniotomy and mastoidec-

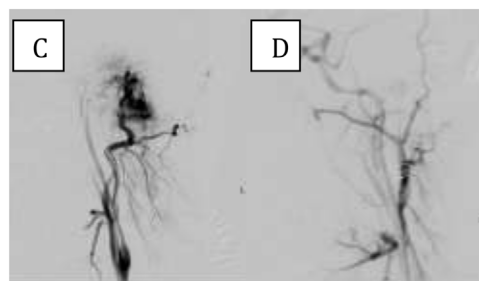
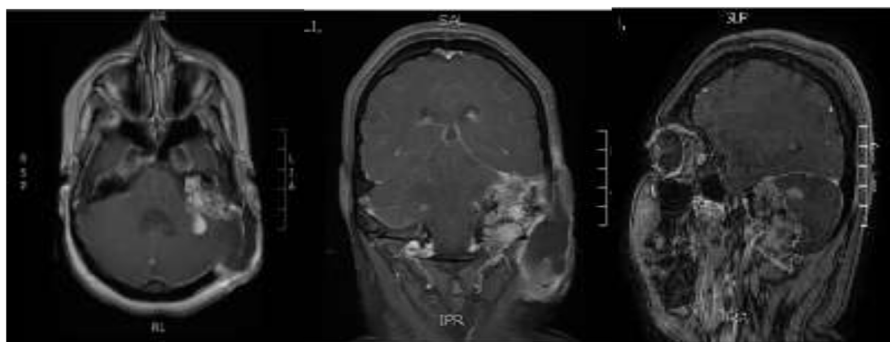


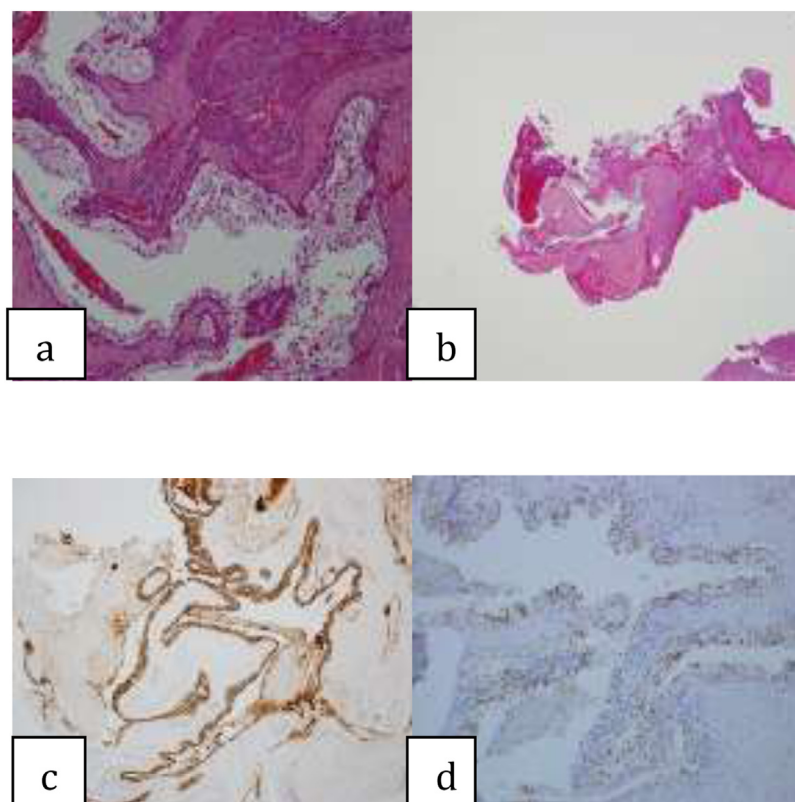
Fig. 3. Defining the Blood Supply of the Tumor and Successful Embolization resulting in complete obliteration of the blood supply of tumor.

tomy using a high speed pneumatic drill. The dura was opened and a microscope was used by the Neurosurgical team to identify the tumor, which was grayish red in color with yellowish areas of discoloration. The highly vascular lesion was removed in a piecemeal manner using CUSA and bipolar irrigation, clearing the left sided CP angle until visualization of the brain stem was possible. During dissection of the portion of the tumor attached to the petrous bone, perfuse bleeding was encountered which was controlled using neuropatties and mechanical compression with a finger. Instead of completing excision, embolization of the feeders of the remaining tumor was indicated. For this decision the wound was closed in layers and bleeding subsided.

The intubated patient was transferred to the interventional radiology suite and embolization of the tumor feeders was carried out. Selective angiography was performed for both the internal and external carotid artery on the left side which revealed that the main feeding artery for the tumor arose from the left ascending pharyngeal artery. The catheter was advanced to the tumor bed and embolization (Fig. 3a and b) was performed with PVA particles measuring 300–500 microns. During the procedure sudden extravasation was noted which was stopped with the injection



**Fig. 4.** Post operative MRI Study with Contrast showing the main bulk of the tumor in Posterior Fossa removed with no compression on fourth ventricle. The remaining tumor is mainly with the internal auditory canal, which was subjected to radiotherapy.



**Fig. 5.** a. Low magnification showing variably-sized cystic spaces. Original magnification: 40×. b. High magnification showing epithelial-lined spaces with some papillary areas. The lining epithelium is columnar cells with clear cytoplasm and small uniform nuclei that are situated mainly at the luminal side. Original magnification: 200×. c. Immunohistochemistry showing positivity for Pankeratin. d. Immunohistochemistry showing positivity for Ber-Ep4.

of Glubran and finally fibered coils were placed in the ascending pharyngeal artery and successful near complete devascularization of the tumor was achieved. A post-operative CT scan revealed a Subarachnoid Hemorrhage without significant Hydrocephalus and there was no evidence of any collection in the neck. The patient was transferred to surgical intensive care and extubated the following day before returning to the Neurosurgery ward. She was discharged from the hospital on the 9<sup>th</sup> post-operative day at which time the patient was afebrile, stable, and had normal healing of the wound without any additional neurological deficit.

At her follow-up visit, the patient was referred for radiotherapy of the residual tumor. A final MRI performed 6 months post-operatively revealed marked reduction of the tumor mass with a normal left cerebellar hemisphere and brainstem and no compression on the 4<sup>th</sup> ventricle yet residual tumor persisted within the petrous bone (Fig. 4a–c). The patient was again referred to

radiation oncology for the residual disease with MRI control and contrast follow-up scheduled after another 6 months. One year later the patient was seen in the clinic, with persistent V, VII & VIII left sided deficit. She was walking independently. MRI results showed little change from the previous 6 months. The patient was referred to plastic surgery for help with her left eye closure. Additionally, she was receiving physiotherapy for the facial muscle.

The histopathology report revealed an epithelial tumor that was consistent with an endolymphatic-sac tumor (Fig. 5a–d). Considering the association of endolymphatic-sac tumors with VHL disease, a CT scan of the abdomen and the pelvis was performed revealing no associated tumor (Fig. 6). Incidental findings included sacroiliitis and subchondral sclerosis in bilateral sacroiliac joints. The tumor was diagnosed as a sporadic type of ELST.



**Fig. 6.** No associated finding seen in abdominal CT confirming the sporadic type of ELST.

### 3. Discussion

The endolymphatic-sac tumor was first identified during a sac decompression surgery in 1984 and Heffner first used this term in 1989 [1,3]. It is a primary neoplasm of the temporal bone, which may be diagnosed in the 1<sup>st</sup>–8<sup>th</sup> decades. The median age of presentation is in the 4<sup>th</sup> decade [2]. In 2004 Lonser et al. described a 40-year-old man with VHL disease showing bilateral endolymphatic-sac tumors [3]. This type of tumor is present in 10–16% of cases of VHL disease [4,5], which occurs in 1 out of 36,000 births [6]. VHL is an autosomal dominant disease resulting from deletions or mutations in the VHL gene located on the short arm of chromosome 3 [3 p25–p26]. The normal VHL gene is a tumor suppressor gene and it is likely the diminution of the tumor suppression activity resulting from the mutation of the gene [7] that leads to the various manifestations of the disease which may include: renal cyst, renal carcinoma, pheochromocytoma, pancreatic cysts, neuroendocrine tumors and/or cystadenomas. VHL disease may have associated multiple hamangioblastomas in the brain, spinal cord and retina. Genetic pathogenesis and molecular studies of VHL disease associated ELSTs revealed mutations similar to the mutations in other neoplasms associated with VHL disease [8]. As a result, ELSTs are listed among other pathologies associated with VHL disease. Sporadic cases of ELSTs also confirmed the occurrence of somatic VHL gene mutations [9].

The ELST arises in the endolymphatic sac or ducts and is considered a tumor of the temporal bone [10]. It is slow growing, locally aggressive in nature and results in the destruction of bony structures and can extend through the CP angle to the base of the skull. The primary aim of treatment is to excise the lesion in total, but for late presenting cases, it may be nearly impossible to get rid of the entire mass [11]. In these cases, as much tumor as possible is excised without causing any iatrogenic neurological deficit and the residual tumor is subjected to radiotherapy or radiosurgery such as gamma knife. Pre-operative obliteration of the feeding vessels for this type of highly vascularized tumor with the help of embolization is sometimes performed [12].

Loss of hearing, tinnitus, dizziness, vertigo, imbalance, ataxia, and involvement of nerves V, VII, and VIII are commonly seen with ELSTs [13]. Signs of raised intracranial pressure are rarely seen in cases presenting late. While destruction of the petrous temporal bone and calcification are well documented using a CT Scan, the

tumor is best visualized using a MRI with control and contrast. Scattered areas of increased signal intensity on non-contrast T1 weighted images may be related to the presence of hemorrhage, cholesterol clefts, and proteinaceous material in large tumors. On T2 weighted images, the tumor is heterogeneously hyperintense. On gradient weighted images, it may show areas of altered gradient susceptibility secondary to foci of hemorrhage [14,15].

In a large number of cases, the main blood supply seen on cerebral angiography comes from the ascending pharyngeal artery and the stylomastoid arteries. The main blood supply originates from the external carotid artery and from the anterior inferior cerebellar arteries [16]. Angiographic information may be used for pre-operative embolization [16,17].

Histologically, these types of neoplasms are papillary or cystic with fronds lined by a single layer of cuboidal to columnar cells [3]. In fact, these tumors may be mistaken for metastatic papillary thyroid carcinoma. Immunohistochemically, these tumors are positive for cytokeratin with a minority displaying positivity for S-100 protein, GFAP synaptophysin and Leu 7 [18]. ELST behave as low grade carcinomas: they may destroy bone and brain parenchyma and recur. These tumors have been reported as metastasizing in rare situations.

Management of ELSTs includes taking a thorough history and performing a complete physical examination. Pre-operative embolization to reduce the vascularity of the tumor is also a consideration. Complete excision of the lesion is preferred if possible while maintaining normal neurological status. After confirming histopathological diagnosis, follow-up includes MRI scans with radiotherapy and gamma knife radiosurgery as options to deal with residual tumor [19]. Stereotactic Radiosurgery is an option for controlling the disease in recurrent complicated cases [20].

### 4. Conclusion

Endolymphatic-sac tumors are very rare with just 175 cases reported [21]. Despite their rarity, these tumors should be part of the differential diagnosis for tumors arising from the petrous bone and occupying the CP angle. A multidisciplinary approach is required to optimize treatment and follow-up with stereotactic radiotherapy is indicated in the presence of residual disease.

### 5. SCARE guidelines

This work has been reported in line with the SCARE criteria [22].

### Conflict of interest

All authors declare that there is no conflict of interest regarding the publication of this article.

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### Ethical approval

The Institutional Review Board (IRB) committee at King Faisal Specialist Hospital and Research Centre – Jeddah, Saudi Arabia, has given the ethical approval. With reference number RCJ/456/39.

### Consent

Written informed consent was obtained from the patient's guardian for publication of this case report and accompanying images.

**Author's contribution**

Afnan Alkhotani, Babar Butt, Mohammed Khalid, Mohammad Binmahfoodh: Management of the case.

Afnan Alkhotani, Babar Butt: Study Design, data collection and Manuscript writing, submission for publication.

Mohammad Binmahfoodh and Yousef Al-Said: Revision of manuscript.

**Registration of research studies**

Not Applicable on the case report.

**Guarantor**

Dr. Mohammed Binmahfoodh.

Dr. Afnan Alkhotani.

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