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CASE REPORT

Endolymphatic sac tumour

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Abstract

We present a case of a papillary tumour at the cerebellopontine angle in a 41-year-old man. He presented with left-sided facial and ear pain associated with dizziness, nystagmus and hearing loss. CT scan of the temporal bone showed a destructive tumour at the left cerebellopontine angle. Surgical excision was performed and the diagnosis of the endolymphatic sac tumour was made. Endolymphatic tumour is a low grade adenocarcinoma that originates from the endolymphatic sac. The definitive diagnosis requires a combination of clinical features, radiological finding and pathological correlation.

Keywords: endolymphatic sac tumour, papillary adenocarcinoma, von Hippel-Lindau

INTRODUCTION

Endolymphatic sac tumour (ELST) is a slow growing tumour which widely invades the petrous bone. It is extremely rare in the general population and has an association with von Hippel-Lindau disease. Because of the rarity of this tumour, this lesion may be overlooked by clinicians. We present a case of endolymphatic sac tumour in a 41-year-old man who was clinically and radiologically diagnosed as glomus tumour and review the available literature on its clinicopathological features.

CASE REPORT

A 41-year-old man presented with left-sided facial paralysis and intermittent left ear discharge for two years. He also complained of dizziness, left ear hearing loss and left sided headache. Physical examination revealed House Brackmann Grade VI left lower motor neuron facial nerve palsy. Otological examination showed left pulsatile healed tympanic membrane without any discharge. Meanwhile, pure tone audiography confirmed profound sensorineural hearing loss of his left ear. Other ENT examination revealed insignificant findings.

High Resolution Computed tomography (HRCT) scan was performed and showed an enhancing lesion at the left cerebellopontine angle causing destructive changes of the mastoid and petrous bone with dehiscent of the left carotid canal (Fig. 1). The radiological diagnosis at that time was glomus jugulare paraganglioma. Magnetic resonance imaging (MRI) was ordered to delineate the soft tissue mass clearer. He then underwent tumour excision via translabyrinthine and transsigmoid approaches by the combined team of neurosurgeon and ENT surgeon. Intraoperatively, the tumour was at left cerebellopontine angle measuring approximately 3x6cm. The tumour eroded the posterior wall of the middle ear and involved the sigmoid sinus. Total excision of the tumour was attended. He was complicated by cerebrospinal fluid leak postoperatively. Fortunately, the leak was resolved day five postoperatively with insertion of the lumbar drain. He was then discharged uneventfully.

Pathology

Histopathological examination showed endolymphatic sac tumour. On hematoxylin and eosin staining, the tumour showed papillary projections with thin fibrovascular core. The papillae were lined by a single layer of low columnar and cuboidal epithelium. (Fig. 2a) Areas of glandular structures with similar epithelial linings and eosinophilic secretion are noted. (Fig. 2b) The neoplastic cells showed

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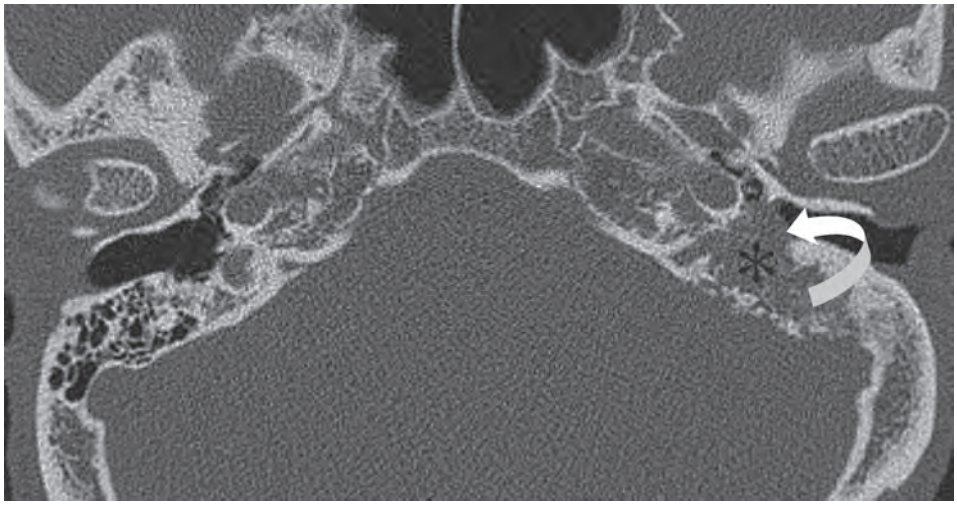

FIG. 1 Axial plain CT scan in bone setting at the level of mastoids showing soft tissue lesion occupying the left mastoid air cells with loss of the expected bony trabeculae within (*). The soft tissue density within the mastoids has eroded the medial wall of the temporal bone extending into the external and middle ear canal (curved arrow).

monomorphic nuclei with no mitotic figures or atypia seen. The adjacent stroma shows foci of foamy macrophages, cholesterol granuloma and rich proliferation of small capillaries.

Immunohistochemical study was performed and the tumour showed positivity towards cytokeratin, epithelial membrane antigen (EMA) and focal positivity towards glial fibrillary acidic protein (GFAP). Stains for thyroid transcription factor- 1 (TTF-1), smooth muscle actin and prostate specific antigen were negative.

He was followed up by both neurosurgical and ENT team. Repeated MRI of the brain two months postoperatively revealed residual tumour and he was referred to oncologist for radiotherapy. He underwent 30 fractions of external beam radiotherapy. His latest CT scan in January 2011 showed no residual tumour seen. Currently, he is still under ENT clinic follow-up.

DISCUSSION

Neoplasm arising from the endolymphatic sac was first described by Hassard.[1] Subsequently, the term low grade adenocarcinoma of endolymphatic sac origin was coined by Heffner who found similarities in the anatomical

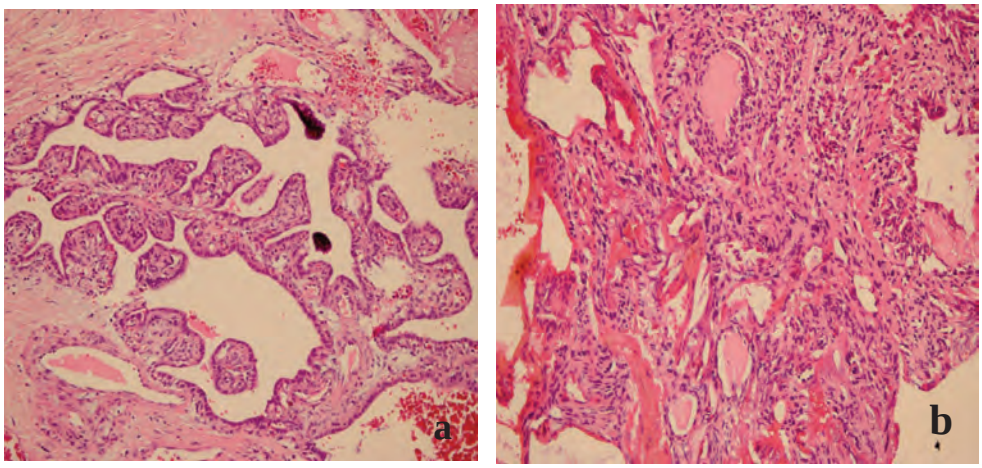

Fig. 2a Endolymphatic sac tumour showing papillary structures lined by a single layer of cuboidal epithelium. Fig. 2b Endolymphatic sac tumour showing glandular structures filled with eosinophilic material. (H&E x 200)

location and histological features of the normal endolymphatic sac to this tumour.[2]

ELST is reported to be more common in females with a mean presenting age of 45 years. Sensorineural hearing loss, tinnitus and vertigo are the typical presenting symptoms in patients with ELST as seen in our patient. Cranial nerve paralysis including facial nerve palsy and cerebellar disorders develops as the tumour extends into the jugular foramen and cerebellopontine angle.[3] This neoplasm behaves as a slow growing tumour which is locally invasive and exhibits bone destruction. Earlier reported cases of ELST showed no metastasis but recently ELST metastasis to the spine has been observed.[4]

The majority of ELST are sporadic while around 15% of patients have an autosomal dominant inherited disorder, von Hippel-Lindau disease. Patients with von Hippel-Lindau disease have a germline mutation in the VHL tumour suppressor gene that is responsible for their genetic susceptibility to various neoplasms. Apart from ELST, von Hippel-Lindau disease is associated with haemangioblastoma of the central nervous system, choroid plexus papilloma, renal cell carcinoma, phaeochromocytoma and papillary cystadenoma of the epididymis.[5]

CT scan typically shows a retrolabyrinthine mass which displays temporal bone erosion as the tumour expands. Radiological differential diagnosis includes paraganglioma, endolymphatic sac tumour, metastasis from occult primary tumours, chondrosarcoma and cholesterol granuloma.[6]

Grossly, these tumours appear reddish or bluish, hypervascular and are soft in consistency. Under the microscope, two main growth patterns may be observed as in our patient. The first growth pattern shows colloid filled cysts lined by a single layered low cuboidal epithelium with relatively minimal stroma. Occasionally, this thyroid-like area may be the predominant histological feature in the biopsy. The second growth pattern is characterized by abundant papillary and solid architecture with vascularized stroma. The stromal component of ELST may show areas of haemorrhage with foamy, giant cell or hemosiderin-laden macrophages.[7]

Definitive diagnosis of ELST may be difficult to reach due to the rarity of this tumour and its various architectural presentations. Possible differential diagnosis includes metastatic follicular thyroid carcinoma, metastatic renal cell carcinoma, metastatic prostate adenocarcinoma, paraganglioma and choroid plexus papilloma. Thus the use of immunohistochemical studies is essential. ELST expresses cytokeratin and glial fibrillary acidic protein while it is negative for specific markers for metastases including thyroglobulin and prostate specific antigen.[7]

Surgery remains the treatment of choice for ELST. However, total excision of the tumour is often not possible due to its aggressive growth pattern. Recurrences could occur years after the primary surgery.[2]

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