CASE STUDY

Papillary Endolymphatic Sac Tumor: Catastrophic Presentation in a Child

Tumor papilar del saco endolinfático: presentación catastrófica en un escolar

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Clinical Case

This was a 9-year-old female patient from a rural area, admitted to emergency service due to rapidly progressing deterioration of the state of consciousness. In the clinical examination, she was comatose, with bilateral non-reflective mydriasis, grade IV (House-Brackmann scale) right facial paralysis, absence of right corneal reflex and bilateral extensor motor responses to pain. When asked, the relatives did not report the presence of symptoms before her admission; in fact, they denied that the patient has any symptoms referring to the vestibular-cochlear system, such as vertigo, hypoacusis, hyperacusis, weakness of the facial muscles or instability in walking.

The patient presented no other clinical signs that would suggest von Hippel–Lindau (VHL) disease.

A contrast-free cerebral computed tomography revealed a tumoral lesion of heterogeneous density with areas of calcification, which deformed and perforated the right temporal bone; the lesion extended from the temporal bone towards the cerebellopontine angle, compressing the brainstem. In addition, partial occlusion of the 4th ventricle could be seen, causing obstructive hydrocephaly. A contrasting cerebral magnetic resonance (MR) helped to define lesion borders; the MR image presented heterogeneous intensity in the T1- and T2-weighted sequences, due to the presence of solid cystic components, indicating subtle gadolinium uptake. Because of the patient’s clinical condition, she was given immediate surgical treatment (Fig. 1A and B).

To handle the hydrocephaly, an endoscopic 3rd ventriculocisternostomy was performed, with a later partial resection of the tumour using retrosigmoid approach, to decompress the brainstem and cerebellum. During this procedure, a large, pale brown encapsulated lesion of very firm consistency was identified; this lesion emerged from the temporal bone, firmly attached to the surrounding dura mater. Almost the entire lesion perforated the dura mater, compressing the cerebellum on the petrous face and displacing it posteriorly, while the protuberance and the brainstem were displaced in the anterior direction. During
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Figure 1  (A) Magnetic resonance axial image (T1 with gadolinium). The tumour can be seen occupying the right cerebellopontine angle. Extrinsic compression of the brainstem and 4th ventricle can also be seen, associated with dilation of the temporal horn of the left lateral ventricle (white arrow). (B) Axial slice of computed axial tomography of the temporal bone, where extensive destruction of the right temporal bone is shown (red asterisk).

the procedure it was impossible to identify the entrance site of the VII and VIII cranial nerves by the internal auditory canal. Likewise, it was impossible to visualise their apparent origins at the level of the brainstem. During the resection, elevated tumour vascularisation was notable, with this being reflected in intraoperative haematic losses that prevented a more radical resection.

During the postoperative period the patient did not improve in clinical condition. She remained in a profound coma for 18 days, with persistence of bilateral mydriasis and decerebrate motor responses. The patient died as a result of septic shock secondary to a nosocomial infection of the urinary tract.

Pathological and immunohistochemical studies confirmed the diagnosis of papillary endolymphatic sac tumour (PELST) (Fig. 2A–H).

Discussion

Originating in the vestibular aqueduct, a PELST is a low-grade adenocarcinoma. It can present sporadically or, as one

Figure 2  Histopathological findings of the papillary endolymphatic sac tumour. (A–C) H&E: tumour consisting of glandular acini with eosinophilic cytoplasm and other areas with clear cytoplasm. Dilated structures of cystic appearance can also be seen (A and C). (D) Immunolabeling for vimentin (+); (E) EMA (+); (F) S-100 (+ focal); (G) CD56 (+ focal), and (H) cytokeratin (+). GFAP and TTF-1 were negative.
of the manifestations, within the neoplastic spectrum that affects patients with VHL disease. These lesions mainly affect the young adult population. In patients with VHL disease, the average age at on-set is from 31 to 38 years; in individuals that do not suffer the mutation, it is diagnosed at the age of approximately 52 years; and it is only found in paediatric patients rarely.

We reviewed the articles published between January 1984 and January 2013, using PubMed, EMBASE, Lilacs, Science Direct,ovid, SCIELO and Google Scholar; in this review we found only 10 cases in patients younger than 20 years of age. In these cases, the average age at diagnosis was 13.6 years, while the distribution by sex was practically the same. This differs from the predilection for females observed in adults (female ratio of 3:2) (Table 1).

Nearly 20% of adults that have a PELST suffer from VHL disease. Nevertheless, the literature review demonstrated that VHL prevalence in paediatric patients reaches 54.5%. This suggests that in these patients, the development of the neoplasia could be earlier or more accelerated than in sporadic cases. This hypothesis is also backed by the observations of Lonser et al., who noted that the average age in sporadic cases is 20 years higher than that of the tumours associated with the mutation.

The PELST are found in Trautmann’s triangle, in an intrasosseous area located at the half-way point between the internal auditory canal and the sigmoid sinus. Due to its limited capacity, this confined space can tolerate only very small tumour volumes before producing symptoms. In the largest series published to date, Kim et al. reported an average tumour size of only 1.3 cm at diagnosis, so most of the symptoms produced are audiovestibular. Reduced auditory acuity is found in 94% of the cases, followed by tinnitus (55%), vertigo (47%), facial paralysis (32.7%), paralysis of lower cranial nerves (5%) and facial paresthesias (5%).

Serious symptoms such as acute obstructive hydrocephaly are extremely rare, although they can appear in presence of very large tumours. There was only 1 other similar case, which also began with obstructive hydrocephaly, reported by Joseph et al., who described favourable evolution in an adult who had a giant tumour that obliterated the 4th ventricle once drainage of cerebrospinal fluid was re-established with a ventricle-peritoneal derivation and the lesion was partially resected. In contrast to that case, our final result was caused by the rostrocaudal deterioration that the patient presented upon admission. This was corroborated by the clinical findings of profound coma, bilateral mydriasis and abnormal decerebrate postures, suggesting an extensive brainstem compromise, which was irreversible in spite of surgical treatment.

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Conflict of Interest
The authors have no conflicts of interest to declare.

References


