CASE STUDY

Erdheim–Chester Disease in a Sinonasal Location

Enfermedad de Erdheim-Chester de localización nasosinusal

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A female patient aged 26, with no other history of morbidities, presented with a 6-month history of symptoms of progressive occipital headaches and vomiting, photophobia, and diplopia with no impaired visual acuity. In addition, she later suffered from medial convergent deviation in the right eye.

She was admitted to hospital with paralysis of the sixth left cranial nerve. The CT scan with contrast showed a mass with a neoplastic type appearance with its epicentre in the sphenoid sinuses and clivus extending to ethmoid cells, maxillary sinuses, the masticator space, infratemporal fossa, pterygopalatine fossa and nasopharynx, with major bone compromise (Fig. 1a and b). The MRI scan showed the presence of a tumour at the base of the skull which compromised the sphenoids and the clivus, which extended towards both temporal fossae and the soft tissues of the pterygopalatine fossae (Fig. 1c and d).

The patient was referred to the Otolaryngology Department of the University of Chile Clinical Hospital for evaluation and study. A nasal endoscopy disclosed erythematous mucosa at choana level, with no suspicion of tumour from the imaging.

An endoscopic biopsy was performed under general anaesthesia of the rhinopharynx and both sphenoid sinuses.

During surgery through the nose, on reaching the sphenoids, abundant inflammatory avascular soft tissue was observed. This was easily removed on dissection (Fig. 2). The biopsy concluded with the presence of morphological and immunohistochemical findings consistent with Erdheim–Chester disease (ECD) type histiocytosis (Fig. 3a and b).

Discussion

Histiocytosis refers to a group of alterations in cellular proliferation where there is an accumulation and infiltration of immune cells and histiocytes, the latter being monocytes, macrophages, dentritic dermal cells or interstitial and Langerhans’ cells.

They are classified into 3 types depending on the predominant cell type. Class I is Langerhans cell histiocytosis, class II are non-Langerhans cell histiocytosis and class III are malignant histiocytosis.

Class II includes ECD.

To date fewer than 400 cases have been referred to in world literature4; and this rare disease is frequently fatal. It is characterised by xanthomatose infiltrating of the tissues composed of foamy histiocytes.4,5 ECD has no hereditary component and it usually clinically presents in adult males with a mean age of 53, within an age range of 7–78 years. It has the ability to infiltrate several organs, generating varied clinical presentations.3-5 The etiology of this condition has not yet been determined, but it is postulated that abnormal monocyte activity occurs.6

Although the disease is normally asymptomatic, it usually presents by compromising long bones, leading to pain and
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Figure 1  Contrasted CT scan, sagittal and axial cross-sections, which show an extensive tumour at the base of the skull measuring 3.4×4×2.4 cm, with soft tissue density compromising the sphenoid sinuses and the clivus extending to ethmoid cells, maxillary sinuses, the masticator space, infratemporal fossa, pterygopalatine fossa and nasopharynx, with major bone compromise (a and b). MRI imaging showed the presence of a tumour at the base of the skull which compromised the sphenoids and the clivus, and which extended towards both temporal fossae and the soft tissues of the pterygomaxillary fossae (c and d).

showing up as bone sclerosis in imaging.² It may also begin with fever, weight loss and fatigue.³ Neurological symptoms such as diabetes insipidus and cerebellum syndromes occur in a third of ECD cases.³,⁷,⁸ Other clinical conditions include bilateral exophthalmos, neurological compromise and the compromising of the lungs, heart, kidneys and skin.³ Diagnosis can pose a challenge since ECD usually presents symptoms which are common to Langerhans cell histiocytosis, but ECD commences at a later age.¹ Immuno-histochemical testing stained positive for CD68 and negative for S-100 and CD1a³,⁶ proteins.

Histopathological differential diagnosis must encompass Langerhans cell histiocytosis, mastocytosis, myeloid metaplasia amyloidosis and CNS lymphoma, among others.⁹ Generally speaking, the majority of patients usually present with a lesion in the brain MRI, with the hypothalamus–pituitary gland axis being most frequently involved, followed by orbital and meningeal compromise.¹⁰ It has been reported that up to 80% of patients presented with compromising of the skull bones, the facial bones, and with paranasal sinus compromise.¹⁰ In these patients, CT scans showed thickening of bone tissue or invasion of

Figure 2  Images obtained during endoscopic surgery of the sphenoid sinus. Abundant inflammatory avascular soft tissue may be observed. This was easily removed on dissection.
a soft tissue mass and the MRI scan disclosed a lesion with hypointense markings on T1 and T2. A minority of these patients presented sinonasal symptoms and for these patients diagnosis was made from rhinosinus biopsy.

There is currently no cure for ECD. Corticosteroids may be used for the management of symptoms but their effects diminish in the long term.

Positive findings from radiotherapy have been described for pain management in cases where there is infiltration of long bone, but lesions may recur. The use of radiotherapy in brain lesions has not been successfully reported in literature.

The use of interferon alfa has demonstrated its effectiveness in patients with early diagnosis, but results will depend on each patient and the level of multisystemic compromise.

Lastly, surgical procedures may be used as an initial method of diagnosis, whenever the diagnosis of systemic disease is not possible. Resection may be performed on patients with well contained intracranial lesions, combined with neurological symptoms.

Conflict of Interest

The authors have no conflict of interests to declare.

References


