Hypothalamic hamartoma

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DESCRIPTION

A 9-year-old girl presented with an 8-month history of gelastic seizures and precocious puberty. Clinical evaluation including full neurological assessment was otherwise unremarkable. Endocrine tests were normal and there was no family or surgical history of note.

Hypothalamic or tuber cinereum hamartoma is a rare non-neoplastic heterotrophic lesion, which forms part of the spectrum of grey matter heterotopias. It is composed of a disorganised mass of neuronal cells within the tuber cinereum of the hypothalamus. It may occur either as a sessile or as a pedunculated lesion.

Presentation occurs in the first decade and manifests with isosexual precocious puberty owing to luteinising hormone releasing hormone secretion, gelastic seizures, hyperactivity, visual deficits and neurodevelopmental delay. Gelastic seizures are characterised by uncontrollable bursts of laughter lasting between 2 and 30 s, and affected children may rarely enter into 'status gelasticus'.[1] Hypothalamic hamartomas are typically isolated lesions but may also occur in the setting of Pallister-Hall syndrome.

They are seen as well-defined round masses on imaging, which measure up to 4 cm in diameter and project inferiorly into the suprasellar or interpeduncular cisterns. The hamartoma is isodense on CT and follows grey matter signal on both T1-weighted (figure 1) and T2-weighted sequences (figure 2). Lack of contrast enhancement is important for radiological diagnosis and distinguishes the tumour from commoner regional pathologies including hypothalamic astrocytomas, germ cell tumours and Langerhans cell histiocytosis.[2]

Treatment with luteinising hormone receptor agonists is often effective with surgery being reserved for refractory cases.

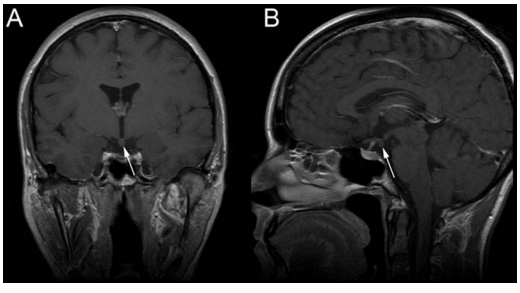

Figure 1 Coronal (A) and sagittal (B) contrast-enhanced T1-weighted sequences show a pedunculated lesion (arrows) arising from the floor of the third ventricle and projecting into the suprasellar cistern. It is isointense to grey matter, does not enhance with contrast and lies between the pituitary stalk and the mammillary bodies. The imaging features are compatible with a hypothalamic hamartoma.

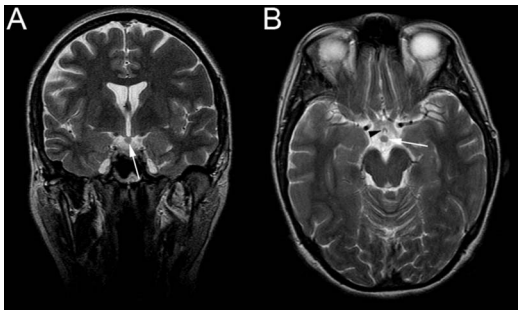

Figure 2 The lesion (arrows) also appears isointense on coronal (A) and axial (B) T2-weighted images. It lies centrally within the suprasellar cistern and is just posterior to the pituitary stalk (arrowhead).

Learning points

- Gelastic seizures are short epileptic events characterised by bouts of laughter and are classically associated with hypothalamic hamartomas.
- Hypothalamic hamartomas are isodense on CT, follow grey matter signal on MRI and do not enhance.
- Differential diagnoses include hypothalamic astrocytomas, germ cell tumours and Langerhans cell histiocytosis.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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