Delayed presentation of diencephalic syndrome associated with leptomeningeal dissemination in a child

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DESCRIPTION

A 7-year-old girl presented to our hospital with a history of failure to thrive since 1 year of age. Her prior workup included negative serum and stool studies to exclude gastrointestinal, metabolic and endocrinological causes. She has mild speech delay, but otherwise had achieved all her developmental milestones. On admission, she was extremely cachectic weighing 11 kg (<1st percentile) and a head circumference of 49 cm (5th percentile). Neurological examination was significant for bilateral papilloedema, optic nerve pallor and peripheral visual field deficits. MRI of the brain revealed a large contrast-enhancing suprasellar tumour with significant hydrocephalus (figure 1A). Areas of leptomeningeal enhancement along the dorsal brainstem prompted spinal MRI that showed diffuse leptomeningeal metastatic disease (figure 1). The patient underwent near total resection of the suprasellar mass where pathology was consistent with a juvenile pilocytic astrocytoma with pilomyxoid features (figure 2). Postoperatively, she exhibited signs of hypothalamic dysfunction including hypothermia, hypotension, bradycardia, diabetes insipidus and near complete visual loss.

Diencephalic syndrome was first recognised in 1951 as a constellation of severe emaciation and preserved linear growth velocity in children associated with hypothalamic tumours.¹ Treatment of hypothalamic–chiasmatic tumours with chemotherapy may result in weight gain that correlates with tumour shrinkage in some cases.² Leptomeningeal metastatic disease has been reported in children with low-grade suprasellar astrocytomas, and

Figure 2 (A) H&E-stained tumour reveals extensive myxoid background with low cellularity. (B) Adjacent areas have a biphasic appearance with abundant Rosenthal fibres and eosinophilic granular bodies (B), consistent with a diagnosis of pilocytic astrocytoma with pilomyxoid features (×40 magnification).

Learning points/take home message

- ▶ Failure to thrive may be an early manifestation of a hypothalamic tumour in the absence of enlarging head circumference.
- ▶ Diencephalic syndrome may be associated with significant neurological and endocrinological side effects at presentation and post-treatment.
- ▶ Low-grade hypothalamic/chiasmatic astrocytomas are capable of leptomeningeal dissemination that may warrant MRI spine surveillance neuroimaging.

Figure 1 (A) Sagittal T1 postgadolinium MRI brain demonstrates a large suprasellar contrast enhancing neoplasm with associated hydrocephalus. (B and C) Sagittal T1 postgadolinium MRI spine reveals multiple areas of leptomeningeal tumour spread (arrows).

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therefore a screening MRI spine is recommended at diagnosis.³ Our case highlights the importance of including diencephalic tumours in the differential diagnosis and workup of patients with failure to thrive of unknown aetiology.

Contributors All authors have contributed equally to the design and preparation of the manuscript.

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