Central sleep apnoea as an initial presentation of a posterior fossa juvenile pilocytic astrocytoma

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DESCRIPTION
A previously healthy patient of middle childhood presented for intermittent dull headaches without nausea, vomiting, photophobia/phonophobia and a 4-month history of snoring. Neurological examination at presentation was unremarkable. Comprehensive metabolic panel and complete blood counts with differential were normal. A sleep study performed to assess for snoring demonstrated normal sleep efficiency and sleep onset latency. Polysomnography recordings revealed an Apnoea-Hypopnoea Index of 12.3/hour, and obstructive apnoea and central apnoea frequencies were 2.1/hour and 10.2/hour, respectively. Sixty-four central apnoeas were recorded in total. These findings represented a complex sleep apnoea with predominant central sleep apnoea (CSA). MRI was performed based on the diagnosis of CSA that revealed a solid and cystic contrast enhancing mass within the right cerebellar hemisphere with obstructive hydrocephalus and surrounding oedema affecting the dorsal brainstem, middle and superior cerebellar peduncles, and cervical medullary junction (figure 1). The patient underwent gross total resection where pathology was consistent with a diagnosis of a BRAF-KIAA1549 fusion juvenile pilocytic astrocytoma (JPA). Postoperatively, the patient required rehabilitation but returned to neurological baseline without deficits and experienced no further sleep apnoea. The patient remains free of disease more than 2 years postdiagnosis.

JPA is the most common brain tumour found in the paediatric population, and most often located in the cerebellar hemispheres.1 JPA initially manifests with increased intracranial pressure over time, inducing symptoms of headache, vomiting and vision changes.2 CSA is a rare presenting feature of central nervous system (CNS) neoplasms and usually presents after diagnosis and treatment.4 CSA is characterised by repetitive periods of unstable ventilatory control during sleep and may result from cardiovascular, neuromuscular and genetic disorders or structural brain abnormalities.4 There are limited reports of CSA as a presenting feature of a CNS neoplasm in the adult population and may be associated with normal neurological, cardiovascular and metabolic findings.3 CSA is rare in the paediatric population,6 although CSA has been rarely reported as a presenting feature of CNS tumours in this age group.7–9 It is unclear whether a tumour diagnosis would have been made in our patient if only obstructive sleep apnoea (OSA) was detected during the sleep study, although OSA has been reported as a presenting feature of a brain tumour in a toddler.10

The CNS neoplasms in the adult and paediatric cases presenting with apnoea were likely impairing the brainstem neuronal network. Tumours involving the posterior fossa may influence central chemoreceptors and pontine and medullary respiratory groups, consequently producing respiration abnormalities.11 12 Changes to the cerebellar peduncles may also disrupt respiration without brainstem involvement.17 Vertebrobasilar arterial system abnormalities and subsequent arterial compression of the brainstem respiratory centres is another mechanism in which CSA may arise.14 15 CSA from vascular abnormalities is a less likely mechanism in our case since the major intracranial vessels were grossly patent, and imaging revealed no vascular compression or dilatation.

Our case represents CSA as a presenting feature of JPA in a patient of middle childhood with a normal neurological examination highlighting the significance of MRI as part of CSA evaluation, specifically in cases where no significant medical history or other risk factors of CSA development are present.

Figure 1 MRI features of posterior fossa tumour presenting with sleep apnoea. Postgadolinium MRI demonstrates a large cystic solid neoplasm with obstructive hydrocephalus (A–B) with invasion into the foramen of Luschka (arrows) and displacement of the medulla best seen on T2-weighted sequences (C). Postoperative MRI demonstrates a gross total resection (D).


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Learning points

- Central sleep apnoea (CSA) may be the presenting feature of a posterior fossa tumour in patients with a normal neurological examination.
- MRI should be considered in the diagnostic workup of patients with CSA to exclude brainstem/cerebellar pathology in the absence of risk factors.

Contributors

CH was responsible for the design and writing of the case report. MRP was responsible for the design and writing of the case report. ML was responsible for the design and writing of the case report. JRC was responsible for the design and writing of the case report.

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Case reports provide a valuable learning resource for the scientific community and can indicate areas of interest for future research. They should not be used in isolation to guide treatment choices or public health policy.

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