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Cranial features of achondroplasia presenting with headaches

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A young man presented to the emergency department with increasingly frequent headaches. A magnetic resonance imaging scan was ordered to rule out space-occupying lesions. This revealed intracranial findings pathognomonic for achondroplasia (Figure 1). The lateral and third ventricles and the aqueduct of Sylvius were enlarged, and the straight sinus had a near-vertical course. The combination of magnetic resonance imaging and clinical findings ruled out obstructive hydrocephalus and were diagnostic of achondroplasia.

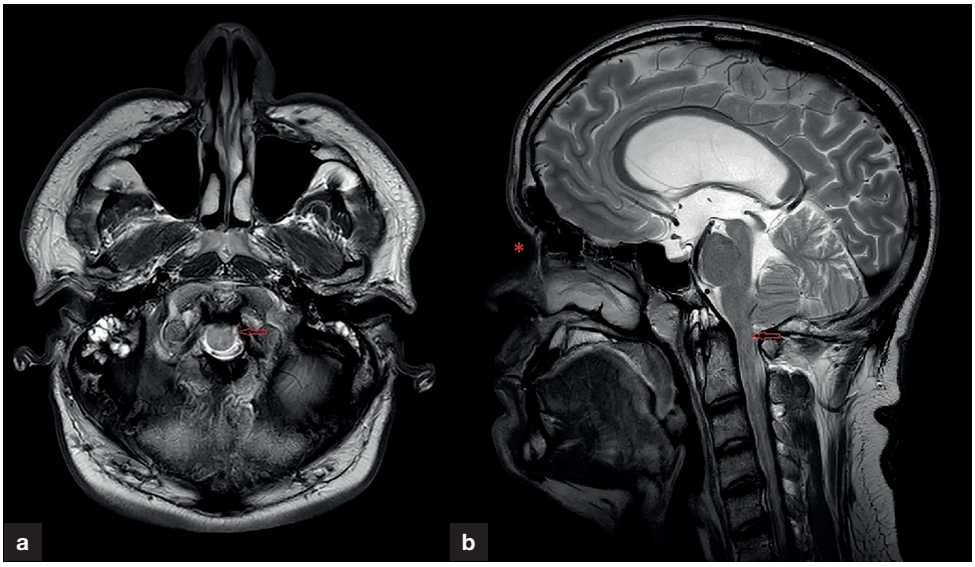

Figure 1. a. Axial T2-weighted magnetic resonance imaging at the level of the foramen magnum, showing severe stenosis of the foramen (arrow) with effacement of surrounding CSF spaces. b. Kinking at the cervico-medullary junction (arrow) is also noted on sagittal T2 weighted sequence. Note also the frontal bossing with a depressed nasal bridge, also known as midfacial retrusion (asterisk).

Achondroplasia is a common form of short-limbed dwarfism, resulting from a dominant, gain-of-function mutation in the fibroblast growth factor receptor-3 gene. This prematurely suppresses chondrocyte proliferation, thus stunting structures arising from endochondral ossification (Pauli, 2019). As this includes the base of the skull and most long bones, it is characterised by a markedly short stature and neuro-symptomatology proportional to the nature and degree of cranial malformation. Early use of exogenous growth hormone may stave off stunting, but symptom relief (eg decompression of the foramen magnum) is the standard treatment (Seino et al, 2000; Pauli, 2019).

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