

Increasing macrocephaly in a neonate

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Infantile macrocephaly has a myriad of causes including megalencephaly, intracranial mass, and CSF disturbance.¹

An 8-week-old infant presented with increasing head circumference. Antenatal ultrasound was unremarkable, with biparietal diameter and head circumference on the 50th centile at 20 weeks' gestation. At birth, head circumference was on the 90th centile; no other abnormalities were recognized. By 8 weeks, the child had clinical macrocephaly. The head circumference was on the 99th centile with an abnormal cranial posture and lower limb hypotonia. MRI at 4 months showed a small foramen magnum with impingement on the cervical cord without cord signal abnormality or syrinx formation (figure, A). Radiographs (figure, B and C) were performed and the diagnosis of achondroplasia confirmed. The infant underwent neurosurgical referral and is at present subject to 6-monthly reviews. It is of note that cervical impingement in patients with achondroplasia usually presents at an older age, but its occurrence has been reported shortly after birth.²

Achondroplasia is a skeletal dysplasia with easily recognizable clinical and radiologic features, which, if missed, may lead to avoidable complications.²

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Received March 15, 2003. Accepted in final form June 10, 2003.

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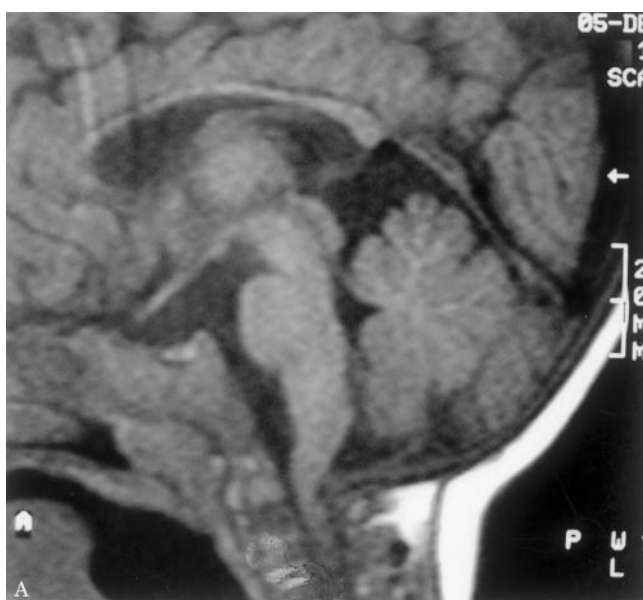


Figure A. Midsagittal T1-weighted MRI of the lower brain, showing a narrow foramen magnum, causing indentation of the cervicomedullary junction.



Figure B. Anteroposterior pelvic radiograph, demonstrating narrowing interpedicular distances and a "champagne glass" appearance to the pelvic floor due to square iliac bones, horizontal acetabular roofs, and narrow sciatic notches.



Figure C. Lateral lumbar radiograph shows sacral lordosis and anterior vertebral beaking.

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Neurology 2003;61;822

DOI 10.1212/01.WNL.0000085869.22589.A4

This information is current as of September 22, 2003

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