

Pediatric Neurosurgery

SEVERE CRANIOCERVICAL JUNCTION STENOSIS IN ACHONDROPLASIA:

Two Pediatric Cases and Their Surgical Indications

BOUDJELOUD Yasmina¹, HABCHI Nawel¹, MAHI Naziha¹ and TLIBA Souhil¹

1: Department of Neurosurgery, Frantz Fanon University Hospital, Blida, Saad Dahlab University Blida 1, Algeria

***Corresponding Author:** BOUDJELOUD Yasmina, Department of Neurosurgery, Frantz Fanon University Hospital, Blida, Saad Dahlab University Blida 1, Algeria

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ABSTRACT

Introduction: Achondroplasia predisposes patients to craniocervical junction stenosis, which may compress the cervicomedullary junction (hypotonia, central apneas, sudden death). The objective is to present two pediatric cases assessed using the AFMS (Achondroplasia Foramen Magnum Score) and to discuss the surgical indication.

Materials and Methods: Two children with genetically confirmed achondroplasia underwent clinical assessment, MRI with AFMS scoring, and polysomnography.

Results:

Case 1 (girl, 16 months): severe axial hypotonia and inability to maintain head control. MRI: anteroposterior (AP) foramen magnum diameter = 7 mm, obliteration of the posterior CSF spaces, cervicomedullary compression, and no signal abnormality → AFMS 3.

Case 2 (boy, 3 years): falls and central apneas requiring ventilatory support. MRI: AP diameter < 7 mm, obliteration of the anterior and posterior CSF spaces, cervicomedullary compression, no signal abnormality, and moderate ventriculomegaly without intracranial hypertension → AFMS 3.

Both children are symptomatic with AFMS ≥ 3 and are being considered for cervicomedullary decompression at a multidisciplinary conference.

Conclusion: In symptomatic children with achondroplasia, an AFMS ≥ 3 (AP diameter ≤ 7 mm, obliteration of the CSF spaces) supports a surgical indication, even in the absence of spinal cord signal abnormality. Moderate ventriculomegaly is not a contraindication to decompression.

Keywords: Achondroplasia – AFMS – Foramen magnum – surgical decompression – child

INTRODUCTION

Achondroplasia is the most common constitutional skeletal dysplasia, with an incidence of approximately 1 in 25,000 births. It is caused by an activating mutation of the FGFR3 gene, resulting in impaired endochondral ossification [1]. One of the most feared complications is foramen magnum stenosis, secondary to hypoplasia of the skull base. This stenosis may compress the cervicomedullary junction, presenting with axial hypotonia, central apneas, psychomotor delay, and, in severe cases, sudden death, with the risk estimated at up to 7.5% [2].

The AFMS (Achondroplasia Foramen Magnum Score), described by Cheung et al. in 2021, is a validated tool for stratifying the severity of stenosis. It ranges from 0 (normal) to 4 (compression with T2 hyperintensity). Stage 3 – compression without signal abnormality – is considered a surgical threshold [3].

The objective of this study is to present two pediatric cases of achondroplasia with severe foramen magnum stenosis and to discuss the surgical indications in light of recent data from the literature.

MATERIALS & METHODS

We report two cases of children with genetically confirmed achondroplasia who were managed in the Department of Neurosurgery at Blida University Hospital. Clinical data (age, sex, neurological signs, head circumference, polysomnography) and radiological data (brain and spinal MRI with measurement of the anteroposterior diameter of the foramen magnum and calculation of the AFMS) were collected. The presence of ventriculomegaly and possible hydrocephalus was assessed. Surgical indications were discussed at a multidisciplinary meeting involving neurosurgeons, pediatricians, ENT specialists, and rehabilitation physicians.

The AFMS was assessed according to the classification of Cheung et al. [3] Fig. 1: stage 0 (normal), stage 1 (narrowing with preserved CSF), stage 2 (narrowing with loss of CSF without spinal cord deformation), stage 3 (spinal cord compression without T2 hyperintensity), and stage 4 (compression with T2 hyperintensity).

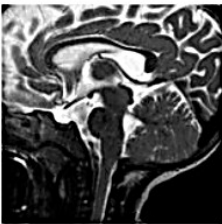
AFMS0	AFMS1	AFMS2	AFMS3	AFMS4
Normal foramen magnum	Constitutional narrowing of the foramen magnum with preserved CSF (no cord distortion)	Narrowing of the foramen magnum with loss of CSF space surrounding the cord	Loss of the CSF space with cord compression	Cord compression and signal changes (Myelomalacia)
				

Fig. 1: AFMS according to Cheung MS et al. Arch Dis Child 2021; Guidelines AAP & Best Practice Consensus 2024

At birth 15 to 20 mm; Achondroplasia 9 to 11 mm; Severe stenosis ≤ 7 mm

RESULTS

Case 1: a 16-month-old girl, Fig. 2A, was referred for psychomotor delay. The diagnosis of achondroplasia was confirmed by genetic testing. Clinical examination revealed macrocephaly (head circumference 54 cm) and severe axial hypotonia, with inability to maintain head control or sit unsupported. Nocturnal snoring was present in association with adenoid hypertrophy. Polysomnography was negative. Sagittal T2-weighted MRI showed severe foramen magnum stenosis, with an anteroposterior diameter measured at 7 mm, complete obliteration of the posterior CSF spaces, and clear cervicomedullary compression. There was no intramedullary T2 hyperintensity and no associated hydrocephalus. The AFMS was stage 3.

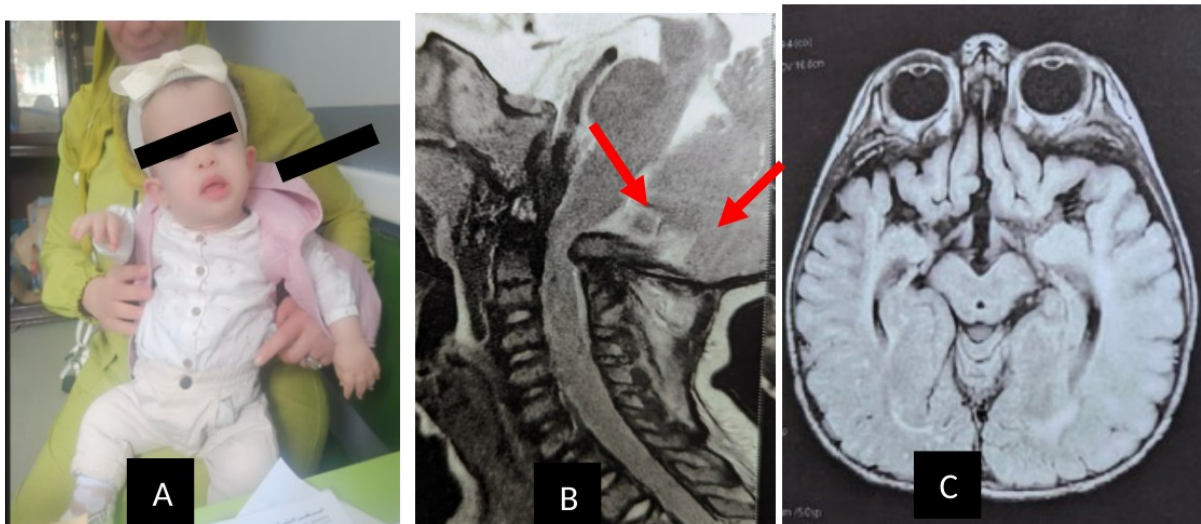


Fig. 2: A, Case 1, a 16-month-old girl; B, sagittal T2-weighted MRI of the cervicomedullary junction showing compression (red arrow) without spinal cord signal abnormality; C, absence of ventricular dilatation.

Case 2: a 3-year-old boy followed for achondroplasia (Fig. 3A) presented with unexplained falls while walking and abnormal motor fatigability at the end of the day. Polysomnography demonstrated severe central sleep apnea syndrome requiring nocturnal assisted ventilation. MRI showed an AP diameter of less than 7 mm, with complete circumferential obliteration (anterior and posterior) of the subarachnoid spaces. The cervicomedullary compression was severe. There was no T2 hyperintensity. Moderate, stable ventriculomegaly was noted, without signs of intracranial hypertension. The AFMS was stage 3, bordering on stage 4.

Source photograph
unavailable

A



B



C

Fig. 3: A, Case 2, a 3-year-old boy; B, sagittal T2-weighted MRI showing an AP diameter of less than 7 mm, with complete circumferential obliteration (anterior and posterior) of the subarachnoid spaces. The cervicomedullary compression is severe without T2 hyperintensity; C, moderate ventriculomegaly. Panel A: source photograph unavailable.

DISCUSSION

Foramen magnum stenosis in achondroplasia is a common and potentially serious complication. Early screening is essential. In the literature, the median age at radiological diagnosis is 9.5 months [4]. Clinical examination alone lacks sensitivity for predicting severe stenosis, hence the recommendation for routine MRI before 6 months of age in every infant with achondroplasia, in accordance with AAP recommendations [5].

The AFMS, validated by Ando et al. in 2026, is a tool that supports surgical decision-making. In their cohort, the mean anteroposterior diameter in operated patients was 5.2 mm, compared with 10.7 mm in non-operated patients. The surgical threshold was set at 7.6 mm [4]. Our two patients, with diameters of 7 mm and less than 7 mm, are below this threshold.

An essential point is the potential pitfall of relying on T2 signal changes. The review by La et al. (2025) emphasizes that the absence of T2 hyperintensity (AFMS stage 3) should not lead to postponement of surgery. Waiting for T2 hyperintensity to appear amounts to accepting irreversible neurological injury [6].

Regarding ventriculomegaly, Kashanian et al. (2020) showed that 70% of children experience improvement or stabilization of ventriculomegaly after bony decompression alone, and that only 14% require secondary ventricular shunting [7]. This finding supports our approach: in the presence of moderate ventriculomegaly without signs of intracranial hypertension, primary CSF diversion should not be performed.

Surgical safety is a major concern. Cracchiolo et al. (2024) showed that 3D neuronavigation allows accurate screw placement without complications, owing to reliable identification of the often dysplastic course of the vertebral arteries [8].

Finally, a systematic review of the literature (2023) including 153 operated patients reported symptomatic improvement in 91% of children, confirming the effectiveness of decompression when appropriately indicated [9].

Limitations of our study: this is a retrospective, single-center series of two cases. Prospective studies with larger numbers of patients are needed to validate our conclusions.

Perspectives: the identification of genetic or molecular biomarkers could enable earlier screening and personalized treatments [10].

CONCLUSION

Foramen magnum stenosis in achondroplasia is a severe complication that warrants early, multidisciplinary management. MRI with calculation of the AFMS is the reference examination. An anteroposterior diameter ≤ 7.6 mm or an AFMS ≥ 3 , associated with clinical signs (hypotonia, falls, central apneas), constitutes a surgical indication for cervicomedullary decompression. The absence of T2 hyperintensity should not lead to postponement of surgery. In the presence of moderate ventriculomegaly without intracranial hypertension, decompression alone is the initial procedure, with shunting reserved for failures. Surgery is effective, with a 91% rate of symptomatic improvement in published series.

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