# Foramen magnum stenosis in Achondroplasia

- Sleep apnea in patients with achondroplasia associated with foramen magnum stenosis
- Dynamic MRI in the Evaluation of the Cervical Spine in Pediatric Patients With Achondroplasia
- Potential efficacy of vosoritide for foramen magnum stenosis in a patient with achondroplasia
- An infant with achondroplasia worsening of the foramen magnum stenosis during early vosoritide treatment
- TYRA-300, an FGFR3-selective inhibitor, promotes bone growth in two FGFR3-driven models of chondrodysplasia
- Multidisciplinary Management of Acute Tetraparesis in an Infant with Achondroplasia, with a Focus on Anesthetic Strategies: A Case Report
- Flexion-extension cervical MRI imaging in pediatric patients with achondroplasia unsupervised by neurosurgery or radiology, is it safe?
- Characteristics of sleep disordered breathing in children with achondroplasia

Foramen magnum stenosis is a serious, and potentially life-threatening complication of achondroplasia. The foramen magnum is smaller in infants with achondroplasia, compared with the general population, and both restricted growth in the first 2 years and premature closure of skull plate synchondroses can contribute to narrowing. Narrowing of the foramen magnum can lead to compression of the brainstem and spinal cord, and result in sleep apnea and sudden death.

## Diagnosis

The timely acquisition of effective neuroimaging that can help to guide clinical management is essential. Wright et al. propose imaging protocols and follow-up strategies for evaluating the neuroanatomy of these children and to effectively identify potential neurological complications, including compression at the cervicomedullary junction secondary to foramen magnum stenosis, spinal deformity and spinal canal stenosis. When compiling these recommendations, emphasis has been placed on reducing scan times and avoiding unnecessary radiation exposure. Standardized imaging protocols are important to ensure that clinically useful neuroimaging is performed in children living with achondroplasia and to ensure reproducibility in future clinical trials. The members of the European Society of Pediatric Radiology (ESPR) Neuroradiology Taskforce and the European Society of Neuroradiology pediatric subcommittee, together with clinicians and surgeons with specific expertise in achondroplasia, wrote an opinion paper. The research committee of the ESPR also endorsed the final draft. The rationale for these recommendations is based on currently available literature, supplemented by best practice opinions from radiologists and clinicians with subject-specific expertise [1)]

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There is a lack of clarity in the literature on the timing of regular monitoring for foramen magnum stenosis, which assessments should be carried out and when regular screening should be ceased. The European Achondroplasia Forum (EAF) is a group of clinicians and patient advocates, representative of the achondroplasia community. Members of the EAF Steering Committee were invited to submit suggestions for guiding principles for the detection and management of foramen magnum stenosis, which were collated and discussed at an open workshop. Each principle was scrutinised for content and wording, and anonymous voting held to pass the principle and vote on the level of agreement. A total of six guiding principles were developed which incorporate routine clinical monitoring of infants

and young children, timing of routine MRI screening, referral of suspected foramen magnum stenosis to a neurosurgeon, the combination of assessments to inform the decision to decompress the foramen magnum, joint decision making to proceed with decompression, and management of older children in whom previously undetected foramen magnum stenosis is identified. All principles achieved the ≥ 75% majority needed to pass (range 89-100%), with high levels of agreement (range 7.6-8.9). By developing guiding principles for the detection and management of foramen magnum stenosis, the EAF aim to enable infants and young children to receive optimal monitoring for this potentially life-threatening complication [2)]

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A study aimed to describe the incidence and severity of FMS in an unselected, sequential series of infants using a novel MRI score and retrospectively correlate severity with clinical examination and cardiorespiratory sleep (CRS) studies.

The Achondroplasia Foramen Magnum Score (AFMS) was developed and scores were retrospectively correlated with clinical and CRS data over a 3-year period.

Results: Of 36 infants (M:F, 18:18), 2 (5.6%) did not have FMS (AFMS0); 13 (36.1%) had FMS with preservation of the cerebrospinal fluid (CSF) spaces (AFMS1); 3 (8.3%) had FMS with loss of the CSF space but no spinal cord distortion (AFMS2); 13 (36.1%) had FMS with flattening of the cervical cord without signal change (AFMS3); and 5 (13.9%) had FMS resulting in cervical cord signal change (AFMS4). Mean Total Apnea and Hypopnea Index (TAHI) for AFMS0-4 was 3.4, 6.41, 2.97, 10.5 and 25.8, respectively. Severe TAHI had a specificity of 89% but only a 59% sensitivity for AFMS3-4. Neurological examination was normal in 34/36 (94%) patients. Overall, 9/36 (25%) infants required neurosurgery with minimal surgical complications.

Conclusions: Clinical examination and CRS have a low sensitivity for predicting the effects of foramen stenosis on the spinal cord. Routine screening with MRI using AFMS can aid in detecting early spinal cord changes and has the potential to reduce infant morbidity and mortality [3)].

# Test

What is the significance of foramen magnum stenosis in achondroplasia?

a) It is a cosmetic issue with no medical consequences.

b) It can lead to compression of the brainstem and spinal cord, resulting in severe health problems.

c) It only affects adults with achondroplasia.

d) It is completely unrelated to achondroplasia.

What contributes to the narrowing of the foramen magnum in infants with achondroplasia?

a) Growth spurt during adolescence

b) Restricted growth in the first 2 years of life and premature closure of skull plate synchondroses

c) Diet and nutrition

d) Lack of physical activity

How can foramen magnum stenosis be diagnosed?

a) By physical examination alone

b) Through a blood test

c) By acquiring effective neuroimaging

d) By measuring head circumference

Why is standardized imaging protocol essential for children with achondroplasia?

a) It helps in diagnosing achondroplasia itself.

b) It ensures that clinically useful neuroimaging is performed and reduces unnecessary radiation exposure.

c) It is required for insurance purposes.

d) It helps in determining the child's future height.

How did the European Achondroplasia Forum (EAF) contribute to the management of foramen magnum stenosis?

a) By recommending surgery for all cases

b) By providing guidelines for the detection and management of foramen magnum stenosis

c) By developing a new drug

d) By organizing awareness campaigns

What is the Achondroplasia Foramen Magnum Score (AFMS) used for?

a) To assess the severity of sleep apnea in achondroplasia patients

b) To diagnose achondroplasia in infants

c) To evaluate the severity of foramen magnum stenosis in infants with achondroplasia

d) To measure head circumference

What does a high Total Apnea and Hypopnea Index (TAHI) indicate in relation to foramen magnum stenosis?

a) It suggests that the patient has no stenosis.

b) It indicates severe foramen magnum stenosis.

c) It has no correlation with foramen magnum stenosis.

d) It indicates a need for dietary changes.

What is the sensitivity of clinical examination and CRS (cardiorespiratory sleep studies) for predicting the effects of foramen magnum stenosis on the spinal cord?

a) High sensitivity

b) Low sensitivity

c) No sensitivity

d) Moderate sensitivity

How can routine screening with MRI using AFMS benefit infants with achondroplasia?

a) It helps in cosmetic improvements.

b) It has no benefits.

c) It aids in detecting early spinal cord changes and can reduce infant morbidity and mortality.

d) It is only useful for diagnosing other medical conditions.

What percentage of infants required neurosurgery in the study mentioned in the text?

a) 0%

b) 10%

c) 25%

d) 50%

Answers:

b) It can lead to compression of the brainstem and spinal cord, resulting in severe health problems. b) Restricted growth in the first 2 years and premature closure of skull plate synchondroses c) By acquiring effective neuroimaging b) It ensures that clinically useful neuroimaging is performed and reduces unnecessary radiation exposure. b) By providing guidelines for the detection and management of foramen magnum stenosis c) To evaluate the severity of foramen magnum stenosis in infants with achondroplasia b) It indicates severe foramen magnum stenosis. b) Low sensitivity c) It aids in detecting early spinal cord changes and can reduce infant morbidity and mortality. c) 25%

1)
Wright J, Cheung M, Siddiqui A, Lucas J, Calder A, Argyropoulou MI, Arthurs OJ, Caro-Dominguez P, Thompson D, Severino M, D'Arco F; members of the ESPR Neuroradiology Taskforce; members of the ESNR Pediatric Subcommittee. Recommendations for neuroradiological examinations in children living with achondroplasia: a European Society of Pediatric Radiology and European Society of Neuroradiology opinion paper. Pediatr Radiol. 2023 Sep 6. doi: 10.1007/s00247-023-05728-0. Epub ahead of print. PMID: 37674051.
2)
Irving M, AlSayed M, Arundel P, Baujat G, Ben-Omran T, Boero S, Cormier-Daire V, Fredwall S, Guillen-Navarro E, Hoyer-Kuhn H, Kunkel P, Lampe C, Maghnie M, Mohnike K, Mortier G, Sousa SB. European

Achondroplasia Forum guiding principles for the detection and management of foramen magnum stenosis. Orphanet J Rare Dis. 2023 Jul 27;18(1):219. doi: 10.1186/s13023-023-02795-2. PMID: 37501185.
3)
Cheung MS, Irving M, Cocca A, et al. Achondroplasia Foramen Magnum Score: screening infants for stenosis [published online ahead of print, 2020 Sep 3]. Arch Dis Child. 2020;archdischild-2020-319625. doi:10.1136/archdischild-2020-319625