

**Response: "Best Practices in the Evaluation and Treatment of Foramen Magnum Stenosis in Achondroplasia during Infancy" and "Is there a Correlation between Sleep Disordered Breathing and Foramen Magnum Stenosis in Children with Achondroplasia?"**

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To the Editor,

Thank you for the opportunity to respond to Dr. Pauli's letter. Both my co-authors and I are indebted to the seminal work of Dr. Pauli on the issue of foramen magnum stenosis in infants with achondroplasia: it formed the basis for much of our current understanding of this complex problem. The intent of these papers was to generate a dialogue that would inspire better understanding of foramen magnum stenosis, and ultimately, better clinical care of our patients.

The first paper for which Dr. Pauli has expressed concerns is “Best Practices in the Evaluation and Treatment of Foramen Magnum Stenosis in Achondroplasia during Infancy”. (White et al., 2015) This work is the product of a *modified* Delphi method from a workgroup composed of highly experienced and qualified clinicians. The format adopted is well accepted in the literature. The core of this workgroup included eleven clinicians, all of whom are well-published in the care of patients with skeletal dysplasias (I would encourage the readership perform their own PubMed searches of each individual), with an average clinical experience of 18.9 years (range 8-41 years) caring for children with skeletal dysplasias.

Thirteen individuals were invited and two declined due to scheduling conflicts. Selection of these individuals was based on perceived interest and expertise, as well as amenability to equipoise. The methodology chosen is well accepted in the medical literature, having modifications made from its original design to resolve issues regarding health care problems. Anonymity was maintained in several ways. During the first round, the panelists completed the survey electronically through Survey Monkey. In the second round (face-to-face meeting) each panelist received a sheet with the distribution of the other panelists' responses, but each panelist knew only his/her own rating. The second survey was administered after the second round anonymously. A face-to face interaction during this process is recommended (Brook, 1994; Fitch et al., 2001) and has been used in multiple studies (Gurvitz et al., 2013, Mangione-Smith et

al. 2011). Consensus of 80% is considered the standard for most Delphi processes, and in this case was agreed *a priori*. A lack of consensus agreement on all statements underscores an absence of coercion within the group.

The formulation of statements was performed in a way to provide clarity of purpose. Combined statements such as “all infants should have some kind of neuroimaging (CT and/or MRI)” were purposely avoided to be clear that the authors do not agree with either of these suppositions. With regard to this example, there was 100% consensus that computerized tomography (CT) scan is not recommended as a screening modality, while there was 91% consensus against *routine screening* by magnetic resonance imaging (MRI). A rationale was provided for making both recommendations. Nonetheless, combining these statements would have mathematically produced 91% consensus against both modalities ( $100 \times .91$ ).

Regarding concern about our use of the literature, this methodology uses the best available evidence. Levels of Evidence or a Cochrane review is not necessary for this process (Fitch et al., 2001) As for the results and tables, we summarized the data in percentages as reflected by the panel’s ratings. This methodology emphasizes group responses and not individual responses; although we agree with Dr Pauli that showing whole numbers might help the reader in the understanding of the results.

Ultimately, it appears that the primary concern is with the recommendations in regard to infant screening for foramen magnum stenosis, particularly with regard to imaging studies. To be fair, the recommendations of this group differ not only from his opinion but with others, including the American Academy of Pediatrics (Trotter and Hall, 2005). We understand that this opinion may be controversial. It is, nonetheless the consensus opinion of the authors that routine imaging of all infants with achondroplasia presents more risk than benefit. It is the exact intent of this work to make clear recommendations for the less initiated provider. We have made it clear that first line screening should include a thorough history and physical exam (with delineation of what this means) and polysomnography, and that MRI scan should be performed when any concerns arise from these two evaluations. Our reasoning for this sequence is clarified in the discussion. It is the fear of our work group that universal MRI screening puts infants with achondroplasia at unnecessary risk due to sedation for the exam and unnecessary surgical intervention due to over-reading of MRI scans.

It was not the intent of the primary author to present these two papers in tandem, this was an editorial decision. The goal of the second paper, "Is there a correlation between sleep disordered breathing and foramen magnum stenosis in children with achondroplasia?" (White et al., 2015) was to elucidate the role of screening sleep studies in the diagnosis of foramen magnum stenosis, and specifically the correlation between central sleep apnea and foramen magnum stenosis. The authors stand by their detailed assessment of the literature;

literature that is far from perfect in demonstrating any true physiologic correlation between central sleep apnea and foramen magnum stenosis. Sleep apnea, both obstructive and central, and foramen magnum stenosis are common in infants with achondroplasia. The causes of central sleep apnea in infants with or without achondroplasia are complex. It is not our contention that sudden infant death in achondroplasia does not occur, nor that sleep apnea is not the cause. We merely question the true physiology of this process.

It is unfortunate that Dr. Pauli underestimates the aptitude of the readership with his suggestion: "Taken on face, these two articles together might lead some to conclude that neither polysomnography nor neuroimaging need to be done routinely in babies with achondroplasia." On the contrary, both articles emphasize that polysomnography and neuroimaging are critical in the assessment of infants and children with achondroplasia. We simply suggest a more judicious understanding and application of these modalities.

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