Surgical Treatment of Cervicomedullary Compression Among Infants with Achondroplasia

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Learning Objectives

In this study, we present our experience treating infants with Achondroplasia associated CMC (cervicomedullary compression). We stress the impact of surgery on the neurological and respiratory outcome, and the importance of early screening, early intervention and even prophylactic surgery.

Background

Achondroplasia is the most common form of human short-limbed dwarfism. Patients with Achondroplasia may suffer from various systemic and neurosurgical conditions. Respiratory complications are reported in high percentage of individuals with Achondroplasia. Respiratory failure is responsible for more than 50% of all deaths among children with Achondroplasia. Infants with achondroplasia may suffer from several neurological and respiratory deficits, which hinder their development and put them at tremendous risk for sudden unxpected death and SIDS in particulary.

Methods

Data was retrospectively collected for 10 infants diagnosed with Achondroplasia and who was operated in our facility for CMC. Collected data before and after surgery included thorough neurological examination, MRI, and sleep study results.

Results

Between 1998 and 2013, 10 infants diagnosed with achondroplasia related CMC were operated at our center. Ages at surgery were 4 to 23 months (12.5±6.88 months). All infants underwent a foramen magnum opening with a wide C1 laminectomy. The dura was opened only in one patient. Following surgery, seven patients (70%) had improved their neurological status. One of the patients had significant neurological deterioration.

When comparing the Apnea-Hypopnea same comment Index, seven children improved following surgery, and one child deteriorated. 60% of patients improved their sleep quality one year after surgery.

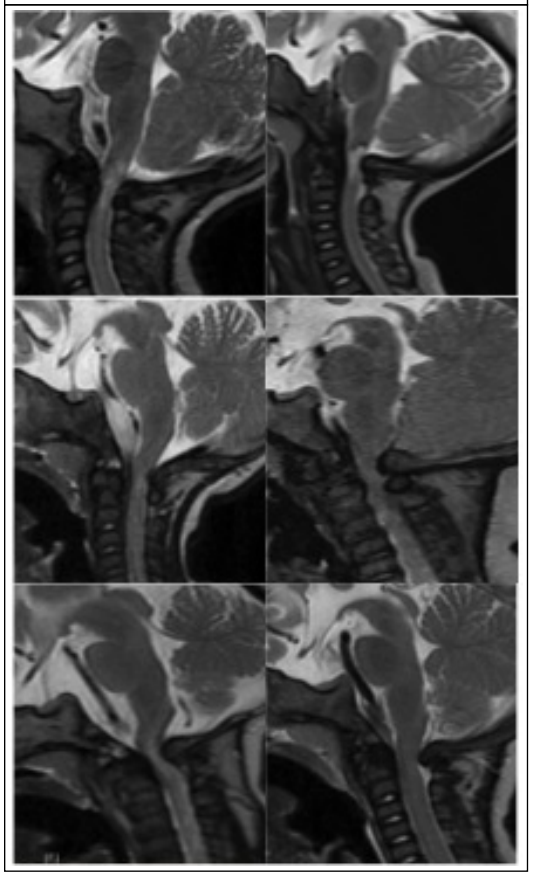
Radiological improvement, including disappearance of signal abnormality in the spine was noted in 6 infants following surgery. 4 infants had brainstem distortion before surgery, which improved following surgery. 7 patients had improved CSF flow surrounding the lower brainstem and upper cervical cord following surgery.

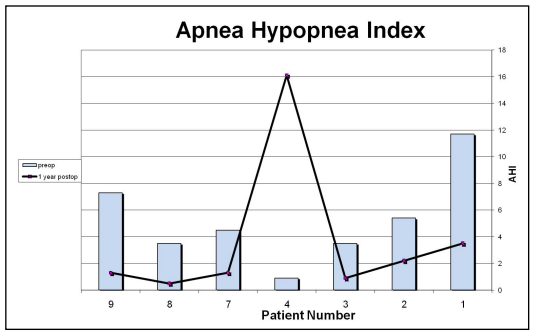
Neurologic examination before & after FMD

Patient no.	Muscle tone	Motor abilities
1	no change	no change
2	improved	improved
3	no change	improved
4	no change	deteriorated
5	no change	improved
6	no change	no change
7	no change	improved
8	improved	improved
9	no change	improved
10	no change	improved

MRI changes before & after FMD

	Abnormal T2 changes		CSF around the foramen magnum	
Patient no.	Before FMD	After FMD	Before FMD	After FMD
1	+	-	-	-
2	+	-	-	+
3	+	+	-	+
4	+	+	-	+
5	+	+	-	+
6	+	-	-	+
7	+	-	+	+
8	+	-	-	+
9	+	-	-	+
10	NA	NA	NA	NA





Conclusions

Infants with achondroplasia may have CMC even in the absence of clear neurological or respiratory signs and neurological symptoms. The higher risk of sudden death in these children may be related to CMC. Crawling neurological and respiratory damage may easily evolve without medical attention. Thus, we recommend close surveillance and early clinical and radiological screening for CMC. Once CMC is diagnosed, we recommend surgical decompression even as a prophylactic measure.