

Achondroplasia: Brain and Skull Manifestations on MRI Brain

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Abstract: Achondroplasia is the most common cause of congenital dwarfism. It is autosomal dominant disturbance in epiphyseal chondroblastic growth and maturation⁽¹⁾. Patients are of normal intelligence. Usually they have normal motor function. However a few present with mild to moderate motor symptoms. . Not all patients presents with neurological findings and hence MRI is usully not done. As skull base forms by enchondral ossification, its growth can be effected with increasing age, leading to stenosis of the foramen magnum, that can cause compression over medulla/cervical cord. We present a case of 12 month old girl child who presented to the pediatric OPD with chief complains of difficulty in walking. The patient was subjected to clinical examination & conventional radiological investigations which revealed rhizomelic shortening of lower limbs, trident hand and hyperplastic vertebrae. The MRI revealed stenotic foramen magnum which was causing cervico-medullary kinking, short clivus, small foramen magnum and relative straight course of straight sinus

Keywords: Achondroplasia, trident hand, rhizomelic shortening,

I. Introduction

Achondroplasia is the most common cause of congenital dwarfism. It is autosomal dominant disturbance in epiphyseal chondroblastic growth and maturation⁽¹⁾. Patients are of normal intelligence. Usually they have normal motor function. However a few present with mild to sever motor symptoms ranging from gait disturbances to quadriparesis to quadriplegia⁽²⁾. As skull base forms by enchondral ossification, its growth can be effected with increasing age, leading to stenosis of the foramen magnum, that can cause compression over medulla/cervical cord. Compression of medulla may result in quadriparesis, sleep apnoea, and even sudden death. Interruption to normal CSF flow can produce hydrocephalus. MRI brain is usually not performed in patients with Achondroplasia and patient may present to us in later stages with dire symptoms. So one has to be wary of consequences that can be caused by stenosis of foramen magnum which can be avoided by in time MR imaging of brain and craniovertebral junction. We present a case of 12 month old girl child who presented to the pediatric OPD with chief complains of difficulty in walking. The patient was subjected to clinical examination & conventional radiological investigations which revealed rhizomelic shortening of lower limbs, trident hand and hyperplastic vertebrae. The MRI revealed stenotic foramen magnum, which was causing cervico-medullary kinking, short clivus, small foramen magnum and relative straight course of straight sinus

II. Case Report

Clinical History and Examination

A 9 month girl came with chief complaints of difficulty in walking and enlarged head. Examination & history revealed Rhizomelic dwarfism apparent at birth, Frontal bossing and depressed nasal bridge and Prominent Buttocks-Lumbar Lordosis. Provisional diagnosis of achondroplasia was made and patient was subjected to radiological evaluation.

Imaging Findings

Conventional Radiography

- Hand AP skiagram revealed Trident hand(- the 1st, 2nd, 3rd & 4th) fingers being almost equal in length & the hand is short with stubby fingers, with a separation between the middle and ring fingers^(1,2,3).
- Dorso lumbar spine skiagram revealed bullet shaped hypoplastic vertebrae.
- Lower limb skiagram Short femur-rhizomelic shortening, metaphyseal flaring, V shaped growth plates (chevron sign), long & bowed fibula⁽¹⁾.

MRI Brain

Multiplanar MR images of the brain were obtained using T1W, T2W & FLAIR sequences in Axial, Sagittal, coronal and diffusion images.

T1, T2 & FLAIR Weighted Sagittal Images Show:

1. Cranial Vault

A large cranial vault with relatively short skull base with prominent forehead, depressed nasal bridge. Narrowing of the foramen magnum with cervicomedullary kinking. AP cranio-cervical diameter is 10mm (normal→30-43mm). Short Clivus^(1,2).

2. Cisternal Spaces

Suprasellar cistern is enlarged and cisterna magna is small. Bilateral lateral, 3rd & 4th ventricles appear mildly enlarged⁽¹⁾.

3. Brain Findings

- Hyper intensities are seen around lateral ventricles on FLAIR and T2W images due to trans ependymal seepage of CSF due to spinal canal stenosis at the level of foramen magnum.
- The straight sinus shows a more vertical course with tentorial angle of approximately 78.2 degrees (Normally it should measure between 27 and 52 degrees.) Elevation of brain stem^(1,4).
- Cavum velum interpositum is seen⁽⁵⁾.

III. Conclusion

Achondroplasia is usually easily diagnosed on conventional radiology via radiographs of the skull, limbs and spine however usually brain MR imaging is not done, thereby missing the narrowed foramen magnum and its compression over cervico-medullary region which can present later with severe motor, respiratory complications. Timely investigation can help early diagnosis of compression over medulla/cervical cords and basilar invagination and thereby a timely surgical decompression can be done, with resection of the posterior aspect of the foramen magnum, the posterior arch of C1 and duroplasty⁽⁶⁾.

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Figures

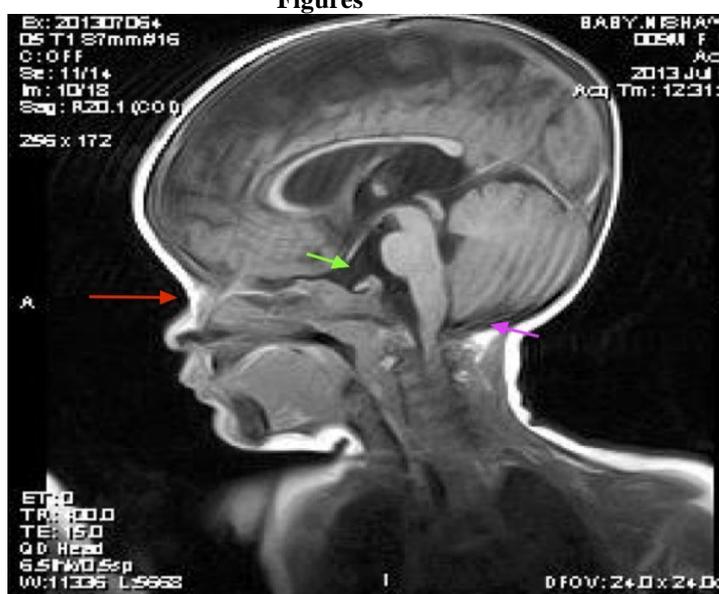


Figure 1 : T1 weighted mid sagittal image shows large cranial vault with relatively short skull base with prominent forehead and a depressed nasal bridge (red arrow), Enlarged suprasellarcistern (green arrow) & small cisterna magna (pink arrow).



Figure 2 : T2 FLAIR image shows narrowing of the foramen magnum with cervicomedullary kinking (red arrow) and a short clivus (green arrow).

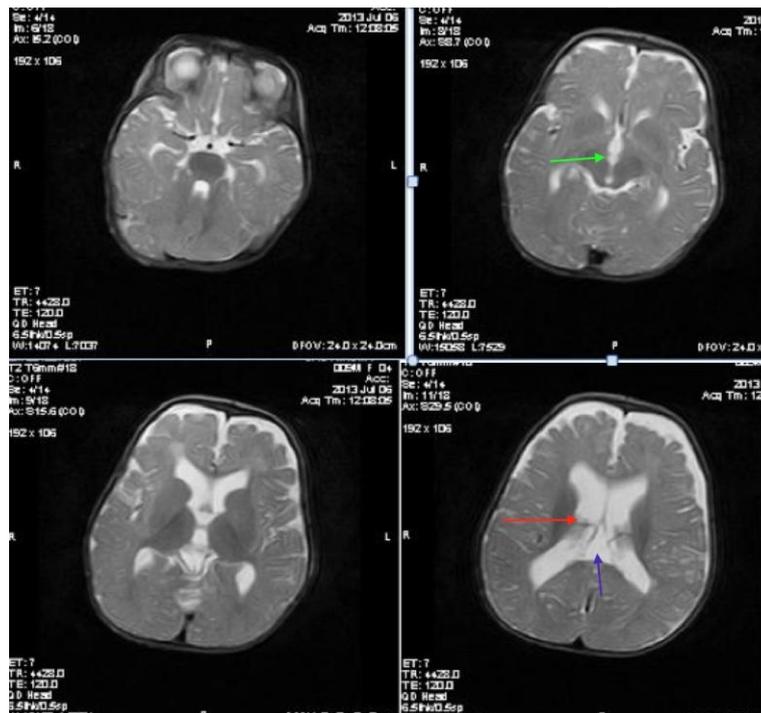


Figure 3: Axial T2 weighted images show enlarges lateral (red arrow) and 3rd ventricle (green arrow) and Cavum velum interpositum (purple arrow).

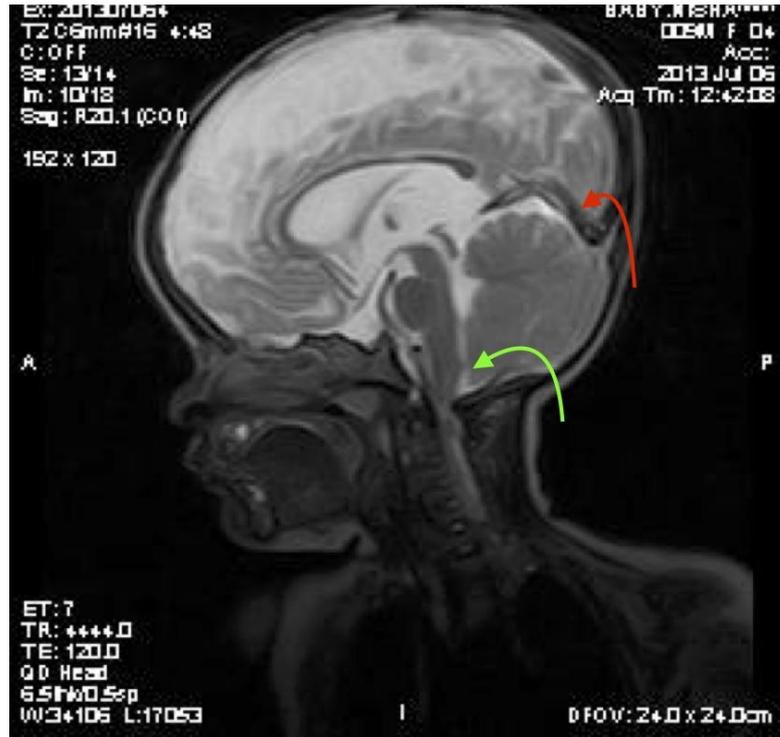


Figure 4: T2 weighted image show a relative vertical course of straight sinus with tentorial angle (78.2 degrees approx) (red arrow) with elevation of brain stem & small cisterna magna (green arrow)