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Arnold Chiari Malformation Type 1: A case report

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Abstract: Arnold-Chiari malformation is a constellation of congenital anomalies related to the hindbrain and base of the brain. It consists of cerebellar tonsils herniation through the foramen magnum into the cervical spinal canal. Chiari malformation is a rare entity and its etiology is not clearly known but said to be genetic. We report a case of Arnold Chiari Malformation type 1 with holocord syrinx.
Keywords: Arnold, Chiari, Congenital malformation, Syringomyelia, Syrinx.

INTRODUCTION

Chiari malformation is a rare disease with prevalence rates of 0.1-0.5% with a female predominance.[1] Chiari malformation is of four types, in which chiari 1, commonly diagnosed in adulthood, is least severe and most common type of malformation. Chiari 1 malformation is characterized by abnormal protrusion of the cerebellar tonsils from the base of the skull through the foramen magnum into the cervical spinal canal. The cerebellar tonsils frequently are stretched out like a peg. Medulla may be flattened or twisted with slight posterior displacement. The fourth ventricle & vermis cerebelli may be slightly deformed or can be normal. [2] Patients with Chiari I may present with a headache and neck pain which are exacerbated by Valsalva maneuver & cough. It is less frequently associated with hydrocephalus. It can lead to syringomyelia and central cord symptoms such as dissociated sensory loss and hand weakness results due to altered flow of CSF through the foramen magnum. [3]

CASE REPORT

A 30-year-old male presented with complaints of bilateral burning type calf pain and neck pain since five-month for which he was admitted in hospital and traction was given for five days. For next three months, the patient was asymptomatic. After 3 months, he again developed tingling sensation in both lower limb associated with bilateral lower limb pain and low back pain. The patient also complained of restricted movement of the back, difficulty in walking and gait ataxia. The patient also gives a history of right sided limbs weakness, difficulty in doing overhead activities with the weakness of hand grip, difficulty in getting up from lying position and climbing stairs.

Central nervous system examination

Higher mental function & Speech were normal. Cranial nerves normal, Built: wasting of small muscles of both hands, Tone: Increased tone in all limbs,

Power:

Group of muscles	Right	Left
Shoulder	3/5	4/5
Elbow	3/5	4/5
Wrist	4/5	5/5
Hand	3/5	4/5
Hip	3/5	4/5
Knee	3/5	4/5
Ankle	4/5	5/5

Reflexes:

Joints	Right	Left
Shoulder	3/5	4/5
Elbow	3/5	4/5
Wrist	4/5	5/5
Hand	3/5	4/5
Hip	3/5	4/5
Knee	3/5	4/5
Ankle	4/5	5/5

Sensory system: Decreased pinprick sensation over left and right half of body over trunk till T2.

MRI cervical spine showed small cerebellar fossa with hypoplastic cerebellum and tonsillar herniation with a large syrinx involving cervical and thoracic cord - Arnold Chiari Malformation type 1.

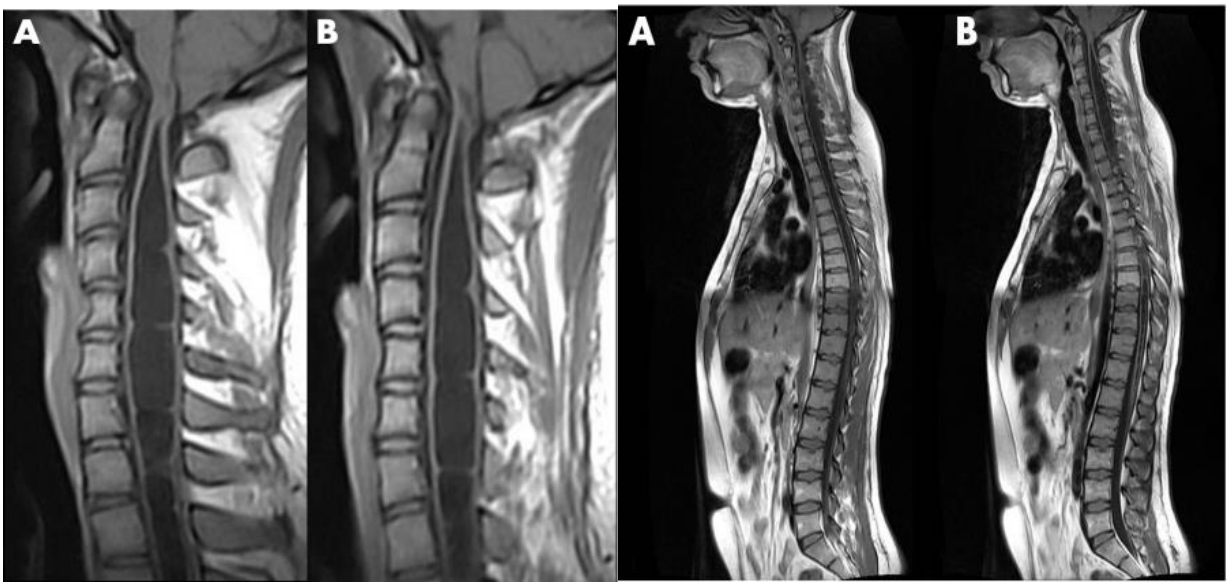

Fig-1:

Treatment: Patient went for C1-C2 distraction and lateral mass fixation, followed by use of Philadelphia collar for 6 months.

DISCUSSION

Chiari malformation was first described by Hans Chiari in 1891. Dr. Arnold made an additional contribution to the type 2 Chiari malformation and hence in his honor, his name was included to Chiari malformation. Chiari classified malformation into four types of abnormalities. Chiari's type 1 consist of cerebellomedullary descent without meningomyelocele while in type 2, it is associated with meningomyelocele. Type 3 is cerebellar herniation with occipitocervical or high cervical meningomyelocele and type 4 consist of hypoplasia of cerebellum. [7]

Chiari malformation has been suggested to have genetic basis based on familial aggregation analysis.[4] Current studies have linked Chiari malformation to chromosomes 9 and 15.[5] It is suggested that Chiari type 1 arises as a result of para-axial mesoderm disorder, which leads to small posterior fossa formation. As a result, cerebellum development takes places in this small posterior compartment which leads to overcrowding and not only results in abnormal protrusion of cerebellar tonsils but also its impaction with foramen magnum. This theory has been found to be consistent with the association of Chiari 1 malformation with other hereditary mesodermal connective tissue diseases i.e Ehlers-Danlos syndrome.[6] Pathophysiology leading to symptoms of Chiari 1 are as follows: (A) compression of cerebellum, (B) compression of upper spinal cord and medulla, and (C) CSF flow disruption through foramen magnum due to which patient may present with a headache, progressive cerebellar ataxia, progressive spastic quadriparesis, downbeating nystagmus, segmental atrophy and sensory loss in the hands and arms with or without pain. Lower cranial nerve & nuclear dysfunction with myelopathy may occur due to compression of medulla and cord. Symptoms such as dysmetria, ataxia, disequilibrium and nystagmus may result due to cerebellum compression. Pain being the

most common symptom arise due to CSF flow disruption through the foramen.[3,7]

CONCLUSION

Chiari 1 malformation is a rare entity and we recommend that before surgical intervention is done, the severity of clinical signs & symptoms must be correlated with radiographic tonsillar herniation. Surgery is recommended in the symptomatic patient while in a mildly symptomatic patient, the decision of surgery lies on the results of CSF flow across foramen magnum which is done using phase contrast cine MRI. After surgical treatment, syringomyelia improves and rarely requires shunting.

Surgical treatment is not indicated in asymptomatic patients of Chiari 1 malformation without syringomyelia who are diagnosed on MR imaging. In the asymptomatic patient, if there is a significant radiographic abnormality, then such patient should be educated about his condition and need of medical help if the patient develops symptoms in the future.

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