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Nahin Hussain, Sushil Beri

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CASE REPORT

Chiari malformation type 1 in children, varied clinical manifestations

Nahin Hussain, Sushil Beri

Abstract:
Chiari malformations type 1 is characterized by herniation of the hindbrain tissue below the foramen magnum. It's more frequently recognised now due to better imaging modalities. The symptoms are typically subtle and evolve slowly. These could be related to the compression of neural structures at the cranio-cervical junction. We describe 5 children diagnosed with Chiari 1 malformation over last 1 year. Clinical features varied from snoring to spastic quadriplegia. Symptoms improved after surgery in four children. These cases highlight the varied clinical spectrum associated with it.

Key words: *Diamond Blackfan, anaemia*
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Introduction

Chiari malformations (CM) are regarded as a pathological continuum of hindbrain maldevelopments characterized by downward herniation of the cerebellar tonsils [1]. Chiari malformation Type 1(CM1) is defined as downward herniation of the cerebellar tonsils of a distance of at least 5 mm through the foramen magnum. Classical symptoms are defined only in late adolescents or adults. With the advent of MRI, more number of children are diagnosed in early years with signs and symptoms that were not recognised before. The presentation is highly variable with subtle and nonspecific symptoms which can be different from those in adults. Thus, timely recognition may be delayed if CM1 is not included in the differential diagnosis. We describe the clinical presentation of all the children who were diagnosed with CM1 in our hospital.

Methods

We conducted the retrospective review of all the patients diagnosed with CM 1 in our department over the last 1 year. Clinical notes were reviewed. Presenting symptoms, signs, neuroimaging and post operative details were identified along with other relevant features.

Nahin Hussain, Susil Beri

Deptartment of Paediatric Neurology, Leicester Royal Infirmary, Leicester, UK

Corresponding Author: ***Nahin Hussain, MD***

Department of paediatric neurology, Leicester Royal Infirmary, Infirmary Square, Leicester, LE1 5WW, United Kingdom
Tel: 00441162587760
e-mail: dr_nahin@yahoo.com

Results

Five children were diagnosed with CM 1 over last 1 year which included 3 boys and 2 girls. The mean age was 10.2 years with range from 4-14 years. Duration of symptoms varied from 6 months to 2 years. All five children presented with different clinical symptoms ranging from headache, snoring, blurring of vision, weakness and ataxia. One child was diagnosed on MRI done for other reasons but was clinically asymptomatic. Examination findings varied according to the clinical symptoms from being normal to spastic quadriplegia.

Table1: Clinical features of children diagnosed with Chiari 1 malformation

	Case 1	Case 2	Case 3	Case 4	Case 5
Age (years)	13	14	10	10	4
Sex	F	M	M	M	F
Presenting features	'Snoring' for last 1 year, Mild occipital headache on awakening	Occipital headache, shoulder weakness and ataxia for 6 months	Headache, blurring of vision and intermittent vomiting for 6 months,	Headaches with migrainous features for 2 years	Admitted to PICU after falling from 20 feet
Examination	Enlarged tonsils	Mild proximal weakness in upper limbs and ataxia	Mild spastic quadriplegia with ataxia, downbeating nystagmus	Normal	No neurological deficit
Investigations	CM 1* 12 mm caudal herniation of cerebellar tonsils (Fig 1)	CM 1	CM 1	CM 1	CM 1 7 mm caudal herniation of cerebellar tonsils
Others	Polysomnogram: central sleep apnoea				
Progress	Improved after decompression surgery	Complete recovery after surgery	Improved after surgery	Complete resolution after surgery	Managed conservatively

*CM1: Chiari malformation type 1

One child (case 1) had a polysomnogram which showed features of central sleep apnoea. All of them were found to have CM 1 on the MRI brain and were referred for neurosurgical opinion. Four children were operated and all had posterior fossa decompression surgery. Two children (case 2 and 4) had complete resolution of their symptoms postoperatively and are doing well. The other two children (cases 1 and 3) also had significant improvement. One child (case 5) is under regular follow up and is currently asymptomatic.

Table 1 describes the clinical findings and outcome of all 5 children diagnosed with CM1.

Discussion

The clinical manifestations of Chiari I malformation (CM 1) are extremely variable and usually correlate with the age of presentation. The symptoms and signs may be directly related to the compression of neural structures by the herniation of cerebellar tonsils at the cranio-cervical junction or with associated syringomyelia [2]. In a review of 39 patients with symptomatic Chiari I malformation done by Nohria [3], of whom 30 were younger than 19 years, the most common symptom was occipital/neck pain and weakness. This is usually associated with episodic exacerbations caused by Valsalva maneuver like laughing, or coughing (cough headache) and is often felt like electric shock. Other symptoms described are focal neurological deficits, pyramidal signs and sensory abnormalities. Down beating nystagmus is considered characteristic of lesions at the level of the foramen magnum.

Sleep related disorders associated with CM 1 are well described in literature [4, 5]. Symptoms can vary from simple snoring, bilateral vocal cord palsy to stridor and sleep apnea. Both obstructive and central types of sleep apnea are described [6]. Dauvilliers et al found a 70% prevalence of sleep apnoea syndrome in patients with CM 1 [5]. Sleep apnoea is also more common in children less than 6 years of age [7]. Craniocervical MRI is recommended when a young child is diagnosed with central sleep apnoea without an obvious cause. Case 1 in our series presented with 'snoring' and was thought be obstructive due to enlarged tonsils. Polysomnography confirmed central sleep apnoea and MRI showed CM1.

There is a higher incidence of associated anomalies with CM1 and could include occult

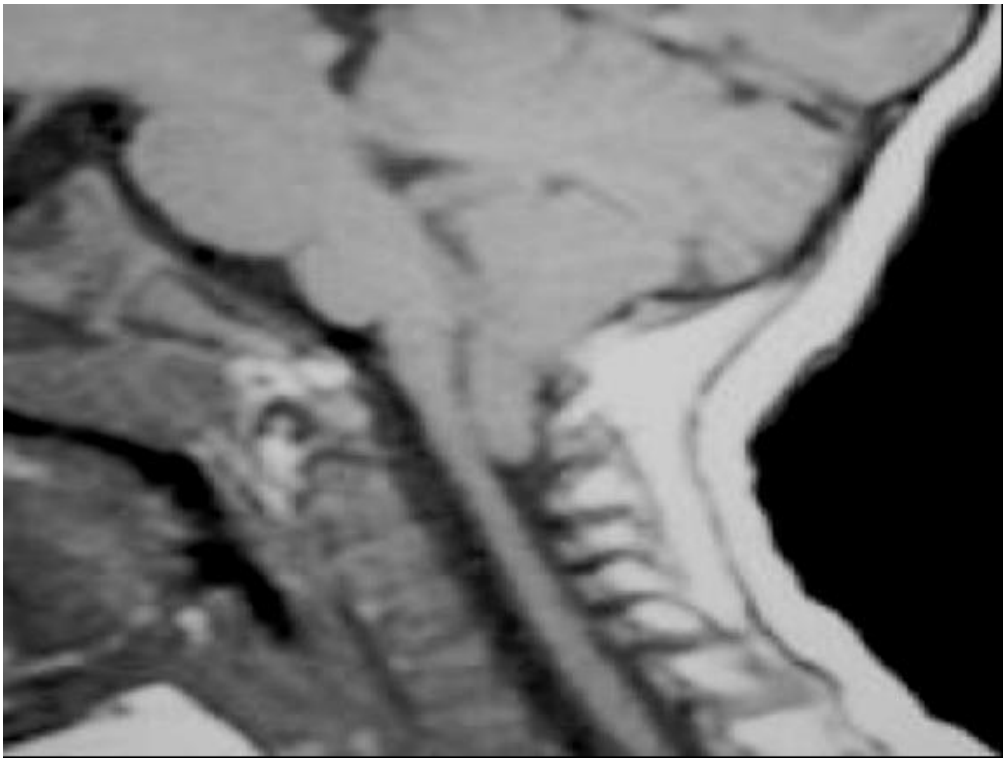

Figure 1: MRI brain shows Chiari 1 malformation with caudal herniation of the cerebellar tonsils 12 mm below the foramen magnum (case 1)

spinal dysraphism, scoliosis and syringomyelia. MRI is currently the investigation of choice for the diagnosis of craniovertebral malformations. None of our patients were found to have any associated abnormalities or hydrocephalus.

Symptomatic patients should undergo neurosurgical evaluation as a treatment option. Posterior fossa decompression is the most commonly performed surgery for this group of patients. Outcome usually depends on preoperative symptoms and associated anomalies. Bindal et al [1] found that patients with Chiari I malformation without syringomyelia have marked improvement after decompression, while those with syringomyelia stabilized or improved slightly. Symptoms lasting longer than 24 months, ataxia, nystagmus and muscle atrophy are usually predictive of a less favourable outcome [8]. Four patients in our series underwent surgery and posterior fossa decompression. Two of them had complete resolution of their symptoms and other had significant improvement. One child was asymptomatic and is under regular follow up, though hasn't required any surgical intervention.

These cases highlighted the varied clinical presentation of CM 1 in children. The spectrum is still expanding as more children are diagnosed with previously unrecognised symptoms. Some children can be asymptomatic as it's picked up on the brain scan done for various other reasons. The neurosurgical evaluation should be considered in symptomatic children as it can often be helpful to control the symptoms.

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