Chiari I Malformation: Patient Report and a Mini Review

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CLIN PEDIATR 2006 45: 570
DOI: 10.1177/0009922806290613

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Introduction

Chiari I malformation refers to the downward herniation of the cerebellar tonsils through the foramen magnum. It may occur in pediatric patients as an incidental finding or may present with varied and unusual clinical symptoms. The case of a 17-year-old female who presented with a 2-year history of chronic back, left hip, and left leg pain is discussed. She was subsequently diagnosed with borderline Chiari I, pseudotumor cerebri, and HAIR-AN syndrome.

Case Presentation

A 17-year-old white female presented with a 2-year history of back, left hip, and left leg pain. The pain initially developed in the low back. It gradually worsened and extended to her left hip and leg. She rated her daily pain as 7 to 8 out of 10 with occasional exacerbations to 10 out of 10. Eight months later she developed new onset headaches. She had 3 to 4 headaches daily with occasional photophobia and intermittent diplopia. She subsequently developed a 1-year history of ataxia. The pain eventually prevented her from attending school or participating in her usual physical activities.

Prior evaluations by orthopedics, neurology, immunology, and rheumatology yielded a differential diagnosis that included lupus, ankylosis spondylitis, and left sciatic neuritis. She tried several medications including nonsteroidal anti-inflammatory drugs (NSAIDs), Plaquenil, Relafen, Elavil, Zonaflex, prednisone, Celebrex, Mobic, Vioxx, and ketoprofen with limited relief. Her past medical history was negative. She denied use of tobacco, drugs, alcohol, sexual activity, and physical or sexual abuse.

Her previous workups comprised: negative magnetic resonance imaging (MRI) of lumbar spine; whole body bone scan consistent with inflammatory disease involving bilateral sacroiliac joints, knee joints, tarsals of both feet and ankle joints, both shoulders and hip joints, left hip and shoulder greater than the right; negative head CT; negative electromyography and nerve conduction study and a cervical-spine MRI demonstrating cerebella tonsillar ectopia; a normal complete blood count (CBC) and differential, elevated erythrocyte sedimentation rate (ESR) (40–60), negative rheumatoid factor; positive anti-nuclear antibody (ANA) (1:160 with a homogeneous pattern); normal thyroid studies; negative antibody for *H. pylori*; and negative hepatic panel.

Upon presentation to our clinic, her physical examination was significant for obesity, depressed mood, an abnormal gait characterized by limping and bearing weight on toes of left lower extremity, facial acne, acanthosis nigricans involving her neck, axillae, and antecubital fossae, blurred right optic disc, and mild nonpitting edema over left ankle. She complained of left knee, hip, and leg pain with light touch and resisted internal and

Clin Pediatr. 2006;45:570-573
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DOI: 10.1177/0009922806290613

external hip rotation. Her neurologic examination was normal.

She was admitted for further evaluation. Her studies revealed negative plain radiographs of her back and bilateral lower extremities; minimal curvature of the lumbar spine with 4 degrees of convexity to the left; negative urine pregnancy test; normal thyroid studies, CBC, complete metabolic panel, and serum magnesium; elevated luteinizing hormone (5.7 mIU/mL) to follicle-stimulating hormone (12.4 mIU/mL) ratio = 2.2; elevated serum dehydroepiandrosterone (38.1 μg/dL); normal dehydroepiandrosterone sulfate; normal total testosterone; normal prolactin; elevated fasting insulin = 30.5 μU/mL, negative anti-DNA antibody; positive ANA titer (1:1280 with homogeneous and speckled patterns); negative anti-SM, anti-RNP, anti-SSA, and anti-SSB; positive HLA-B27; ESR = 29; negative rheumatoid factor; normal C4 (33 mg/dL); elevated C3 (170 mg/dL); negative Lyme antibody; negative spinal fluid acid fact bacilli, VDRL, glucose, and protein; elevated spinal tap opening pressure (330 mm H_2O) with a closing pressure of 230 mm H_2O; normal pap smear; negative cervical cultures; negative MRI of thoracic spine and spine survey; negative pelvic MRI; and unremarkable head MRI and MRA except for mild tonsillar ectopia with cerebellar tonsils extending approximately 6 mm through the foramen magnum.

She was evaluated by pain management, physical therapy, ophthalmology, psychiatry, pediatric neurology, neurosurgery, and nutrition. The neurologist believed her symptoms were functional. The neurosurgeons believed the borderline type I Chiari malformation was incidental and recommended no surgical treatment.

She was started on oral Diamox 250 mg twice a day (increased to 500 mg twice a day after 10 days) and Zoloft 50 mg daily. Secondary to HAIR-AN (hyperandrogenemia insulin resistance acnathosis nigricans) syndrome, metformin (500 mg twice a day) and oral contraceptives (Yasmin) were initiated. For pain management, oral Neurontin 600 mg three times a day and Etodolac 400 mg daily were started. She was continued on hydrochlorothiazide 12.5 mg orally daily for hypertension, which was well-controlled.

She was discharged after 7 days with little clinical improvement noted despite physical therapy. She did not keep her follow-up appointments. Telephonic communication with the family revealed she had not improved and that she had been reevaluated by a neurosurgeon who recommended assessment by a neurosurgeon on the West Coast. The family elected not to pursue the recommendation at this time.

Discussion

Chiari I malformation is a congenital downward herniation of the cerebellar tonsils through the foramen magnum.[1] Classic diagnostic criteria include: (1) herniation of at least one cerebella tonsil 5 mm or more below the plane of foramen magnum as defined on sagittal T1-weighted image; (2) herniation of both cerebellar tonsils 3 to 5 mm below the foramen magnum if accompanied by other definite features consistent with Chiari malformation, that is, syrinx or cervicomedullary kinking; (3) no clinical history of myelomeningocele or radiologic evidence of Chiari II malformation; (4) no prior cervical or cranial surgery; and (5) tonsillar herniation appears to be a primary dysplasia and not secondary to cerebral mass lesion.[2] A new classification based on the presence (type A) or absence (type B) of syringomyelia has been posited.[2] Type A patients had predominantly central cord symptoms, while type B patients had signs and symptoms of brainstem or cerebellar compression. Kinking of the medullocervical junction is more with type B. Type B patients had better surgical outcome after posterior fossa decompression.[2]

Etiology and Diagnosis

The etiology and pathogenesis of Chiari I malformation remains speculative. Posited etiological factors include a difficult birth delivery with local compression of the bones of the base or skull hemorrhages around the cisterna magna and brain swelling due to anoxia.[3] MRI is essential to the diagnosis.[4] Incidental MRI discovers Chiari I in one-third of patients who do not have clinical symptoms.[5]

Clinical manifestations are related to direct compression of neural tissues and abnormal cerebrospinal fluid flow at the craniovertebral junction.[5] Neurologic symptoms and signs may be related directly to the tight foramen magnum associated with the cerebellar tonsillar herniation, with compression and/or distortion of the medulla and lower cranial nerves. Long track deficits, with weakness, sensory abnormalities, hyperreflexia, and a Babinski response, may be present. Twenty percent of patients may have lower cranial nerve dysfunction with vocal cord paralysis, dysarthria, hoarseness, palatal weakness, sleep apnea, or recur-

rent aspiration. Less common presentations of brainstem/cranial nerve involvement include oscillopsia, sensorineural hearing loss, sinus bradycardia, syncope, hiccups, and hypoglycemia. The cerebellar syndrome, characterized by truncal or appendicular ataxia, nystagmus, and cerebellar dysarthric speech has been described but is less common than brainstem/cranial nerve involvement.[6]

The symptoms of Chiari I malformation can often be vague and ambiguous in children, leading to misdiagnosis.[7] A review of the records of 11 children who underwent suboccipital decompression for symptomatic Chiari type I malformation revealed that presenting complaints included neck pain, scoliosis, back pain, torticollis, motor dysfunction, and apnea.[4]

Stevens et al[8] suggested that the clinical symptoms ascribed to cerebellar ectopia could be related to the severity of the malformation and that the operative outcome could be related to the morphological findings. They determined that patients with the most severe cerebellar malformation, defined as descent of the cerebellar tonsils to or below the axis, had disabling ataxia and nystagmus more frequently. Limb weakness and muscle wasting were more frequently seen in patients with brainstem compression. Postoperative outcome was less favorable in patients with severe cerebellar ectopia (12% improved, 69% deteriorated) than in those with minor ectopia (50% improved, 17% deteriorated). Patients with distended cervical syrinx had a more favorable outcome than those without.[8]

Loder et al[9] studied the association of scoliosis with Chiari I malformation. They retrospectively reviewed 30 children with scoliosis secondary to Chiari I from three centers. Syringomyelia was present in 87% of the patients. The scoliosis was thoracic in 25, thoracolumbar in 3, and lumbar in 2; 18 curves were right and 12 were left. A positive correlation was noted between cervical lordosis and thoracic kyphosis. When cervical lordosis is greater than zero degrees or thoracic kyphosis is greater than 40 degrees (Cobb method), the clinician should suspect the presence of Chiari I malformation with or without syringomyelia.[9] Compared to adolescent idiopathic scoliosis, the patients with Chiari I and associated scoliosis had more left curves (40% vs. 0%), were more like boys (37% vs. 8%), and were younger (11.3 years vs. 14.2 years).[9] Steinbok[6] also demonstrated that scoliosis is an important and common finding in children with syringomyelia associated with a Chiari I malformation.[6,10] Progressive scoliosis was found in approximately 30% of such children.[6]

Patients with Chiari malformations may also present with vestibular system symptoms, including ataxia, nystagmus, or vertigo.[11] Patients with advanced symptoms may demonstrate oculomotor dysfunction, central vestibular nystagmus, abnormal vestibular visual interaction, and abnormal tilt suppression of postrotatory nystagmus.[11]

Pseudotumor cerebri associated with Chiari I malformation has been reported in two 12-year-old patients.[12,13] Although papilledema has been reported occasionally, a causal relationship has not been established and the mechanism for the increased pressure is not well understood.[14] The clinical distinction between Chiari I malformation and pseudotumor cerebri is sometimes difficult, as patients with pseudo tumor cerebri and Chiari I malformation usually become symptomatic in young adulthood and are usually women. However, patients with pseudotumor cerebri are usually obese, whereas patients with Chiari I malformation are not necessarily obese.[14]

Treatment and Management

Considerable debate exists about when surgery is indicated and which surgical options are best for management of Chiari I malformation with and without syringomyelia. There are at least 15 different methods and about 40 different operative methods.[3] The surgical treatment for Chiari I malformation can stabilize or slightly improve the symptoms of sryingomyelia and relieve the symptoms of brainstem compression.[15,16]

Type A patients had predominantly central cord symptoms and type B patients had primarily brainstem or cerebellar compression symptoms. The principal surgical procedure consisted of decompression of the foramen magnum, opening of the fourth ventricular outlet, and plugging of the obex. Significant improvement in preoperative symptoms and signs was observed in 45% of type A patients compared to 87% of type B patients. The presence of syringomyelia implied a less favorable outcome.[16]

The main benefit of surgical management in patients with Chiari I malformation without or without syringomyelia was prevention of disease progression.[1] Overall, surgery improved both symptoms and signs, but the improvement in symptoms was more marked. There was a positive correlation between improvement in hydrosyringomyelia and the improvement in signs and symptoms.[17]

Outcome and Prognosis

The mean interval between onset of symptoms and operation was shorter in the children (3 years 6 months vs. 7 years 1 month); preoperatively adults had more severe symptoms; and postoperatively seven of eight pediatric patients improved and one stabilized whereas two of five adults improved, one stabilized, and the disease progressed in two. Additionally, the preoperative abnormal cerebrospinal fluid flow at the craniovertebral junction and to-and-fro movement in the syrinx improved.[16]

Current opinion on the treatment of syringomyelia and Chiari malformation supports a standard bony and dural decompression of the foramen magnum region with modifications designed to maximize the restoration of cerebrospinal fluid circulation across the foramen magnum is a safe, effective operative treatment for Chiari I malformation in children.[17-20]

Summary

Children with Chiari I malformation present with varied and unusual presentations that may lead to missed or misdiagnosis and delayed treatment. Pediatric patients with chronic pain may benefit from evaluation for Chiari I malformation to ensure early treatment and prevention of progression of the disease. Chiari I malformation in patients with chronic pain may not be an incidental finding.

Acknowledgments

Written consent was obtained from the patient or their relative for publication of study.

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