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What is This?
Brief Communication

An Unusual Presentation of Chiari I Malformation

Capi Seeger Scheidler, MS, PT1,2, and Meredith R. Golomb, MD, MSc3

Abstract

The most typical symptom of Chiari malformation type I in children is headache. The authors describe a 14-year-old girl who presented with a 3-year history of gait decline and no headache, which is very unusual. After surgery to correct the Chiari I malformation, the patient’s gait improved; however, she went on to develop decreased hand use with joint deformities. She was diagnosed with a probable connective tissue disorder. Patients with connective tissue disorders are at increased risk for developing Chiari I malformation. The authors discuss the possible reasons for the unusual presentation of the Chiari I malformation and possible mechanisms. The unusual presentation of Chiari I delayed this young patient’s diagnosis and treatment.

Keywords

connective tissue disorder, headache, pediatric, gait, joint, atrophic scarring, foot, hand

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Chiari malformation type I is characterized by the herniation of the cerebellar tonsils through the foramen magnum.1-5 With the introduction of magnetic resonance imaging, the diagnosis of Chiari malformation type I has been made more frequently in the pediatric population, including those patients with more subtle signs and symptoms.3-7 The most typical symptom of Chiari malformation type I in children is headache.1-7 Other documented common symptoms include neck pain, nausea, vertigo, ataxia, scoliosis, decreased balance and coordination, and weakness.1-3,5,7 Presentation with gait decline alone is very unusual.

This report describes a 14-year-old girl with Chiari I malformation who presented with a 3-year history of gait decline and no history of headache. When she went on to develop decreased hand use with joint deformities, she was diagnosed with probable connective tissue disorder. Possible mechanisms underlying this rare presentation of a Chiari I malformation are explored.

Case Report

This child presented to an orthopedic surgeon at age 10 years with complaint of a limp for the past year. She also had increased wear on her right shoe and foot pain when running for an extended time. Evaluation revealed an “antalgic gait” with significant Trendelenberg on the right side that was noted in gait but not in one leg stance, strength of 5/5 in all hip movements, mild femoral antversion, difficulty standing on toes, normal sensation to light touch, 2+ and symmetrical ankle and knee reflexes, and negative Babinski reflexes. Radiographs of the spine and hips were negative for scoliosis and hip joint abnormalities. Magnetic resonance imaging of the lumbar spine revealed mild annular bulges at L4-L5 and L5-S1 levels and was otherwise unremarkable. The physician prescribed physical therapy to increase strength.

Sixteen months later, she returned with complaints of pain in her feet and difficulty walking. Evaluation revealed bilateral foot pain, bilateral pes cavus feet, bilateral femoral antversion, normal sensation to touch and pinprick, and abnormal gait. She had 3+ knee jerks, 2+ ankle jerks, and negative Babinski bilaterally. She had beginning development of hallux valgus and hammertoe deformity of the second through fifth toes. Operative and nonoperative options to address the foot pain were discussed, and she was referred to a neurologist.

One month later, a neurologist evaluated her and documented a gradual onset of symptoms, no pain or swelling, complaints of foot pain after walking long distances, stiffness in legs after prolonged sitting, walking up and down stairs sideways, inability to run, limping, and fear of falling. The patient reportedly could not “get down on the floor.” She had bilateral

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hyperreflexia of 3+ in her legs. Laboratory evaluations revealed positive antinuclear antibody screen and SSA (Ro) antibody; erythrocyte sedimentation rate and liver function tests were unremarkable.

She was referred to rheumatology at our hospital. Physical findings were similar to those documented on the neurology examination. Mucous membranes were moist, with no sign of sicca syndrome. Testing for anti-DNA antibody, C3 complement, C4 complement, thyroid function tests, renal function tests, aldolase, creatine kinase, and urinalysis were unremarkable.

The patient was then referred to neurology at our hospital. At this point she was 12.5 years old, and her family reported gradually worsening gait over the past 3 years. Her mother described her gait as “that of a seventy- or eighty-year-old.” The patient had difficulty getting up from the floor independently. She complained of pain on the medial surfaces and soles of her feet after walking for extended periods. She had developed a bunion, and her toes had begun to flex. She complained of cramps at the tops of her thighs in the past week. She denied pain elsewhere and denied problems with upper-extremity weakness or fine motor skills. Both she and her family were certain that she had not experienced any significant headaches. Further history revealed that she had lost the ability to ride a bike in the past year. On examination the patient revealed 5/5 strength in the upper and lower extremities. She reached well for a target with either hand. Her gait was slightly stiff and shuffling with a narrow base of support. She had decreased temperature and sharp sensation over the dorsal and plantar surfaces of the feet. She had very brisk reflexes in the upper (3+) and lower (3+/4-) extremities. When either wrist was rapidly extended, the ipsilateral shoulder jerked. She denied any pain with this reflex, which was present bilaterally. She had 2 beats of clonus at both ankles and positive Babinski reflexes. Magnetic resonance imaging of the cervical spine and brain that evening revealed a Chiari I malformation with tonsillar descent of about 9 mm. The ventricles were of normal size; there was no syrinx. The cerebrospinal fluid flow study showed diminished flow posteriorly at the level of the foramen magnum (Figure 1). The patient was evaluated by a neurosurgeon,
who concluded that although she did not have the classic symp-
tom of Chiari I malformation, that is, headache, the symptoms
she was experiencing were progressive and Chiari was the only
potential identified etiology.

Two months later, she underwent a Chiari decompression
that included a suboccipital craniectomy and C1 laminectomy
with expansile duraplasty. Two days after surgery, she was able
to stand on one leg for 10 seconds bilaterally and squat to the
floor to pick up objects. In a surgical follow-up appointment
1 month later, she had much improved gait and tandem gait.
She had bunionectomies 6 and 11 months later.

At age 13 years 9 months, she returned complaining of
progressive deformation of fingers of both hands and weak-
ness. Physical exam revealed swan neck deformities of fingers
of both hands and weak grip. Grip strength revealed 2.67 (norm
56.8) on the right and 5.0 (norm 50.9) on the left. Gait was
improved. Reflexes were brisk: 3+ in bilateral biceps, brachial
radialis, knees and ankles. She had the same unusual reflex of
shoulder jerk with wrist extension, present bilaterally, and was
noted to have atrophic scars from her bunionectomy surgeries.
Rheumatoid factor, antinuclear antibody, sedimentation rate,
and thyroid stimulating hormone were unremarkable. No syr-
inx was seen on magnetic resonance imaging of the cervical
spine. Nerve conduction study of the upper extremity was unre-
markable. She was referred to genetics over concern for con-
nective tissue disorder because of the combination of
ataxia, or scoliosis. This is a very unusual presentation of
Chiari malformation type 1.

Discussion

This report describes an adolescent girl with Chiari I malforma-
tion and probable connective tissue disorder who presented
with several years of gait decline and no history of headache,
neck pain, upper-extremity weakness or ataxia, nausea, vertigo,
ataxia, or scoliosis. This is a very unusual presentation of
Chiari malformation type 1.

While genetic testing did not reveal a clear diagnosis, her
joint hypermobility, skin laxity, atrophic scars, high palate,
and micrognathia all suggested a connective tissue disorder.
People with connective tissue disorders appear to be at
increased risk for Chiari I malformation. In a series of 2813
patients with Chiari I malformation, 12.7% had connective
tissue disorders.9 Connective tissue disorders are caused by
abnormalities of structural proteins such as collagen and elas-
tin, which may create excess joint mobility. Children with
various types of joint hypermobility syndrome have symp-
toms including arthalgia, joint deformity, gait abnormality,
problems with handwriting, and limitations of physical activ-
ities.10 This patient’s joint abnormalities associated with con-
nective tissue disease may have contributed to her
presentation with years of gait decline.

Gait decline has been described as an unusual presenting
symptom for Chiari I malformation in combination with other
symptoms, though, to our knowledge, gait decline alone has not
been described as a presentation of Chiari I. Chiari malforma-
tion type I is characterized by the herniation of the cerebellar
tonsils through the foramen magnum. The main clinical fea-
tures of cerebellar involvement include incoordination, imbal-
ance, and ataxia.1-5

This patient’s unusual lack of headache with Chiari malfor-
mation type I might be explained with the diagnosis of probable
connective tissue disease. Headache in Chiari malformation
type I is thought to be caused by increased pressure resulting
from blockage or occlusion of cerebrospinal fluid flow in the
posterior cranio cervical junction.11,12 This increased pressure
is caused by the containment of the cerebrospinal fluid by the
dura mater. The dura mater is made up of connective tissue.
It is a thick layer that is primarily composed of collagen.13 In
some connective tissue diseases, the collagen in the dura might
be hypermobile, thus decreasing the tensile strength of the dura
and thus decreasing the overall pressure gradient. This may
decrease the possibility of headache. It must be noted, however,
that a previous patient who had the connective tissue disorder
Ehlers-Danlos syndrome with Chiari malformation type I
presented with chronic headaches.14 Since headaches are com-
mon in patients with Ehlers-Danlos syndrome,14,15 there could
have been other factors contributing to headaches in that
patient. Our patient’s lack of anxiety disorder and depressive
disorder may have played a role. Anxiety and depression are
associated with chronic pain.16 Our patient’s mother described
her as “not a worrier,” although she did have understandable
anxiety concerning her illness. She had no signs of depression
on the Patient Health Questionnaire.

In this case, surgery relieved our patient’s cerebellar
symptoms, improving her ability to walk, get up off the floor,
walk up the stairs, ride a bike, and walk in tandem. However,
she had progressive symptoms of connective tissue disorder
including hypermobility in the hips, knees, and ankles, which
can continue to challenge stability in gross motor skills. These
symptoms along with weakness and deformity of the joints in
her hands brought her back for further evaluation and a diagno-
sis of probable connective tissue disorder.
Patients with connective tissue disorders have long delays in diagnosis, with consultation of multiple specialists. This has a significant effect on the patient’s quality of life not only in terms of finances and time, but also in the pursuit of incorrect therapies, the delay of treatment, and preventable continued progression of the disease state.\textsuperscript{15} While we were unable to make an exact diagnosis, our patient had the clinical signs of a connective tissue disorder. There is increasing awareness of medical disorders associated with Chiari I such as connective tissue disease. In this case, the unusual presentation of Chiari I malformation was an additional factor that contributed to several years of delay in diagnosis and treatment. As more is learned about disorders associated with Chiari I, it is hoped that the diagnoses of associated conditions can be made earlier.

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Author Contributions
CSS wrote the manuscript. MRG supervised CSS, providing feedback and editing on all drafts.

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Ethical Approval
The family consented to the publication of this case and approved the paper in final form.

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