CHIARI MALFORMATION TYPE 1:
A NEUROSKELETAL ANOMALY

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Abstract

Chiari Malformation type 1 is a brain abnormality at the junction of the base of the skull and the spinal cervical canal. In general Chiari Malformations, the cerebellar tonsils are displaced down in to the spinal cervical canal creating compression on the cerebellum, brainstem and the cervical canal of the spine. Specifically, Chiari Malformation Type 1 is comprised of the caudal displacement of the cerebellar tonsil down the foramen magnum in to the upper cervical spinal canal. This causes CSF, or cerebrospinal fluid, flow to be disturbed and leads to other brain conditions such as hydrocephalus and most commonly, Syringomyelia. There are four categories of the disease and they are classified based on the severity of the disease and the involvement of a particular brain region. In this detailed review about the neuroskeletal brain anomaly, brain anatomy, prognosis, diagnosis, clinical presentations, and surgical treatments will be discussed to further understand the pathology and prognosis of Chiari Malformation Type 1.

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Introduction

Chiari Malformations are a congenital neuroskeletal abnormality at the junction of the base of the skull and the cervical spinal canal. The malformation contains deficits in the base of the skulls and the cerebellum in the brain. Generally in the Chiari Malformations, the cerebellar tonsils are displaced down in to the upper spinal canal, the cervical spinal canal, creating pressure and compression on the various brain regions involved: the cerebellum, the brainstem and the spinal canal. In a normal adult brain, the cerebellum and brainstem are located at the base of the skull, or the posterior fossa, above the foramen magnum. The foramen magnum is an opening at the junction of the base of the skull and the spinal canal which allows the spinal cord to pass through the opening and connect to the brainstem. In a brain effected with a form of the Chiari Malformation, the cerebellum protrudes down from the posterior fossa past the foramen magnum in to the cervical spinal canal. Although this malformation occurs during fetal development, patients often do not experience symptoms till late childhood to adulthood. Although the causes of the malformations are not widely known with skepticism present in the current literature, a predominant cause agree upon in current literature to cause that malformation is a misshapen skull or underdevelopment of the bones of the base of the skull. The formation of a misshaped skull due to the underdevelopment of the skull causes compression in the posterior fossa forcefully displacing the cerebellum down the foramen magnum in to the cervical spinal canal to relieve the pressure. Most of the Chiari Malformations are congenital and occur during fetal development; however, there can also be a late adult onset of the brain abnormality after a traumatic accident or brain trauma to increase pressure in the skull resulting in the displacement of various brain regions such as the cerebellum and the brainstem (3,5).

There are four types of Chiari Malformations and they are classified based on the severity of the disease progression and the brain regions involved in the caudal displacement in the to the cervical canal.

Chiari Malformation Type 1(CMT1)

CMT1 is the most common out of the four and is the least complex. In CMT1, the lower part of the cerebellum known as the cerebellar tonsils are caudally displaced down in to the upper spinal canal.

Chiari Malformation Type 2 (CMT2)

In CMT2, both the cerebellum and the brainstem protrude down in to the spinal canal.

Chiari Malformation Type 3 (CMT3)

CMT3 is the most complex and rare form of the malformation as the both the cerebellum and the brainstem herniate out of the skull via an abnormal opening.

Chiari Malformation Type 4 (CMT4)

Lastly in CMT4, there is a prevalence of an incomplete or underdeveloped cerebellum (3).

Brain anatomy and CMT1

To fully understand the prognosis, clinical presentations and the details of the Chiari Malformation Type 1, it is imperative to understand the anatomy of the brain and the brain regions involved in CMT1. The following brain regions are impacted in CMT1: the cerebellum, the brainstem, and the 4th ventricle in the brain. Let's begin with the cerebellum. The Cerebellum is a brain region that is located at the base of the skull or a region known as the posterior fossa of the skull. The function of the cerebellum is to coordinate motion and maintain balance. The cerebellum is divided in to two lateral hemispheres separated in the middle by a portion called

the Vermis. Under the cerebellar vermis, two small portions are found protruding known as the cerebellar tonsils. Next, the 4th ventricle is another region of importance in the posterior fossa of the skull. The 4th ventricle is a space filled with a fluid known as Cerebral Spinal Fluid or CSF. CSF is an important aspect for the protection of the brain and the spinal cord as it serves a cover and cushions both. The 4th ventricle space is located in front of the cerebellum and behind the brain stem. The cerebellum, brainstem and the 4th ventricle are all located in the posterior fossa above the foramen magnum in a normally developed brain. In Chiari Malformation type 1, there is a downward displacement of the cerebellar tonsils, by at least more than 4mm, past the opening of the foramen magnum in to the spinal cervical canal. The displacement of the tonsils in the spine increases the pressure in the skull and creates compression of the cerebellum and the spinal canal with each other. A significant consequence of the cerebellar displacement is the disruption of the CSF flow. The normal CSF flow between the intercranial space and the spinal canal is blocked creating a buildup of CSF in the brain and the spinal cord. This build up CSF in the brain and the spinal cord creates abnormal CSF filled cavities leading to other brain conditions such as Hydrocephalus (3).

Consequences and Prognosis

After the onset of the malformation itself, other diseases become associated with it due to the obstruction of the normal CSF pathway. CMT1 can progress with or without symptoms where it is mostly asymptomatic in the majority of the patients. Because of the CSF obstruction, the following brain and spine abnormalities can occur after the onset of CMT1: Hydrocephalus, Syringomyelia, and Spinal curvature. Hydrocephalus is a brain condition caused by a CSF cavity in the brain with excessive CSF build up. This causes pressure in the brain and an enlargement of

the head. Next, the most common brain abnormality caused by CMT1 is known as Syringomyelia. Syringomyelia is a condition where there is a CSF filled cyst in the central canal of the spinal cord known as a syrinx. Normally the spinal fluid flows from the 4th ventricle through the posterior fossa in to the spinal subarachnoid space. With the cerebellar tonsils displacement and obstruction of the normal CSF flow, CSF is forced from the obex in to the central canal of the spinal cord creating a syrinx. The growth of the syrinx in the central spinal cord destroys the canal and creates pain, weakness, and stiffness in the neck, back, shoulders, arms and legs. Another consequence of the syrinx destroying the central spinal cord canal is the loss of sensation in the upper torso for extreme pain and temperatures. Lastly, progression of anomalous spinal curvature can also occur with onset of CMT1 such as scoliosis, bending of spine left or right, or kyphosis, which is the bending of the spine forward or backward (3).

Clinical Presentations

Patients with CMT1 vary in the clinal presentations of symptoms, often ambiguous, inconsistent and complex, where some present with common symptoms while others have no presentations of these symptoms and are asymptomatic. The majority of the symptoms are caused by compression of the cerebellum, brainstem, and cranial nerves due to the cerebellar tonsils caudal displacement and also due to the formation of the CSF filled syrinx. Often times patients are also misdiagnosed with neuromuscular disorders, such as Multiple Sclerosis, chronic fatigue of muscles in the back and neck, and fibromyalgia due to the ambiguity and vague nature of the symptoms. Because patients with CMT1 present with headaches, and a plethora of muscle weakness throughout the body, especially the head and the neck, symptoms of CMT1 are incorrectly assumed to be chronic weakness by many physicians. Because the symptoms of

CMT1 can go unnoticed, patients do not come in to be examined for CMT1, rather an examination for another pain can reveal the abnormality in the brain via various diagnostic tests such as an MRI or a CT scan. CMT1 patients clinically present with the symptoms of occipital headaches, sever pain in the head and the neck, muscle weakness, loss of pain and temperature sensations in the upper body torso, dizziness, poor coordination and balance, dysphagia, ataxia and sensory disturbances.

Occipital Headaches: Cerebellar Compression

The most common predominant symptom is the occipital headache, which is felt at the back of the skull in the occipital lobe of the skull. The headache originates form the upper cervical region of the spine or the suboccipital area. The pain radiates from the posterior occipital region to the frontal and periorbital lobes. The headache is made worse by coughing, sneezing or any other extraneous strain felt by the skull and the brain. The headaches are most often described as build up pressure at the back of the skull and the duration of the pain can last from minutes to hours to days. Many patients report these occipital headaches as constant headaches unrelieved with medications, indicating their extreme severity and distinctness form a normal headache. Occipital, unrelieved headaches are a clear indication of a complexity in the brain that needs to be addressed.

Muscle weakness and loss of sensation: Syrinx formation

Other symptoms such as the overall muscle weakness and increased fatigue are caused by the compression of the cranial nerves in the brain and the spinal cord. Muscle weakness in the arms and the legs and the loss of sensation of extreme pain and temperatures are also caused by the development of the syrinx in the central spinal canal.

Ataxia: Improper Gait

Poor coordination, balance and ataxia are other significant symptoms experienced by many patients due to the cerebellar compression caused by the displacement of the cerebellar tonsils and the onset of hydrocephalus. The cerebellum is responsible for maintaining proper body movements and balance so disturbance of the cerebellum via compression results in impacted and improper function of the cerebellum resulting in ataxia, or loss of control of body movements. Many patients complain of dizziness along with poor balance and disturbances in the gait. Some patients, more infrequently, also encounter atrophy of the lower extremities implying the involvement of the upper and lower motor neurons.

Dysphagia and Sensory Disturbances

Another symptom encountered by many patients is a difficulty in swallowing or dysphagia which can progress rapidly and lead to aspiration. Dysphagia, along with sever occipital headaches, is another fundamental sign of compression of the cerebellum and the lower cranial nerves. Patients report difficult in swallowing both solid and liquid foods.

Sample case examples

Below are examples of patients and their clinical presentations which indicate the occurrence of Chiari Malformation Type 1. The first two cases (3) are of classical symptoms felt by patients associated with CMT1 and MRI were ordered to reveal CMT1; however, the third case, often common, is of totally unrelated symptoms and history where an MRI later revealed CMT1 with cerebellar tonsils displacement (2).

Case 1:

A 25-year old male, in a healthy condition, presents with a one-year history of worsening headaches, progressively with time, that are unresponsive to medications. The patient described the headache as throbbing and sharp starting in back of the skull at the posterior occipital region that radiates through the bi-temporal areas. He also reported hoarseness and difficulty in modulating his voice. Clinical examination revealed no neurologic deficits; however, an MRI was performed after consistent symptoms for a year. The MRI of the brain revealed approximately 5mm tonsillar herniation without the presence of a syrinx, which indicates the presence of CMT1.

Case 2:

A 45-year old woman reports acute onset of difficulty swallowing and choking at night for about two weeks. She also reported neck pain, mildly transient and transient tingling sensation of the left upper and lower extremities over the past 10 years. She had no history of severe occipital headaches, dizziness or any vision impairment. A barium swallow, a X-ray test used to visualize the esophagus, was ordered by her primary care physician, which revealed aspiration of both liquids and solids. Subsequently, an MRI was performed and cerebrovascular accident was ruled out. The MRI of the brain revealed an 18mm cerebellar tonsillar caudal displacement in to the cervical spinal canal, which is a strong indication of the presence of CMT1 with extreme displacement of the cerebellar tonsils.

Case 3:

A 4 ½ year old young girl presented with a six-month history of breast budding and transient pain of the nipples. The patient was born as full-term baby and her medical history indicates that she was treated for hypoglycemia and respiratory distress syndrome at birth. Her height was with

in the 90th and 95th percentile with her weight in the 90th percentile as well. The physical exam and neurological tests were normal. No sever neurological signs and symptoms were observed. As puberty was accelerated in the patient and began early, an MRI was performed. The MRI of the brain revealed a pineal cyst and an approximate 6mm displacement of the cerebellar tonsils, indicating the presence of CMT1(3).

Diagnostic Measures and Radiographics

To assess the presence of CMT1 in patients, appropriate diagnostics test and neurological examinations are imperative to indicate various symptoms and imaging information to indicate the presence of CMT1. Significant portions of the neurological exams include the assessment of the functions of the cerebellum, sensory evaluation and functions of the twelve cranial verves (3). As CMT1 was often unnoticed, there were infrequent diagnostic and radiological exams performed, however, more exams are being performed with the emergence of advanced neuroimaging techniques to understand the complex brain anatomy and the details of the posterior fossa. Conventional Magnetic Resonance Imaging (MRI) portrays a detailed evaluation of the posterior fossa. A midline sagittal T1 or T2 weighted image is ideal for displaying the size of the posterior fossa, the shape of the cerebellum and all its components and the size and morphology of the 4th ventricle and the brainstem. Other techniques also utilized to visualize the posterior fossa and display some abnormalities are ultrasonography and computed tomography (1).

Sample exams to test cerebellar functions, sensory evaluation, and cranial nerve tests (3).

Cerebellar functions

- Romberg
- Finger-to-nose
- Heel-to-chin
- Gait and tandem gait
- Rapid alternating movements

Sensory Evaluations

- Proprioception
- Light touch (pinprick)

Cranial Nerve Tests

I.	- smell soap, coffee, alcohol
II.	- visual acuity, visual fields, fundoscopic exam
III, IV, VI	- extra ocular movements, pupil response, to light
V	- blink reflex
VII	- smile, wrinkle brow, grimace
VIII	- hearing- finger rub, Rinne, weber's test
IX, X	- gag reflex, swallow, tongue, uvula and palate
XI	- shoulder shrug
XII	- tongue movement and symmetry

Treatments: Surgical Techniques

The most common treatment of CMT1 is a surgery performed by a neurosurgeon to decompress the foramen magnum and the posterior fossa and enlarge the foramen magnum space via a duraplasty. The purpose of the surgical intervention is to relieve the symptoms felt by both the compression of the cerebellum, brainstem and the cranial nerves and the syrinx in the spinal cord. Another goal of the surgery is to stop the progression of the syrinx by draining it via a shunt. Treatment of CMT1 depends on whether the malformation is symptomatic or asymptomatic. Patients with asymptomatic CMT1 are not considered good surgical candidates as there are no symptoms to be relieved so the surgery is not necessary. Patients with symptomatic CMT1 presenting with symptoms confirming the malformation undergo the following surgery: a posterior fossa decompression with a duraplasty. The goal of the surgery is to decompress the posterior fossa by removal of some of the foramen magnum vertebrae and to enlarge the posterior fossa space with a duraplasty, or a patch graft sewn on to the existing dura matter of the cerebral tissue, to provide additional space in the posterior fossa. The additional space after the duraplasty will allow the CSF to resume its normal flow (3).

Surgical technique

Under general anesthesia, patients are placed prone in a skeletal fixation with the elevation of the head and padding of the pressure points. Hair is shaved form the occipital area to the upper neck and perioperative antibiotics are administered. A midline incision is outlined 7 cm above the inion and extending downward in the cervical area, cervical 2, which is carried down to the pericranium and the fascia. The fascia is opened in a Y-shaped fashion displaying adequate visualization of the suboccipital region. A suboccipital craniectomy, procedure to remove a portion of the skull to relieve pressure, followed by a cervical laminectomy, removal of the

vertebrae bones, is performed on the first cervical vertebrae of the foramen magnum to decompress the foramen magnum region. The posterior fossa bone is also thinned. An incision is made to the dura to expose the tonsils. Fine electrocautery may be used to further decompress or reduce the size of the tonsils. If a syrinx is present, the syrinx is also drained via a shunt and ultimately terminated. To permit additional space to the dura of the cerebellar tissue and to allow the proper CSF flow to be restored, a patch graft or duraplasty is performed. This decompression surgery relieves most of the symptoms of CMT1 encountered by the majority of the patients as the posterior fossa is decompressed and CSF flow is normalized. There is little to none negative consequences post operation and patients observe positive results with the elimination of the majority of the symptoms (3,4).

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