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Case Report

A Rare Case of Chiari Malformation Type 4

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Abstract: The Chiari malformations are a family of conditions characterized by developmental or, less commonly, acquired displacements of the cerebellum. The original 19th century description by Hans Chiari delineated 4 types, but only types 1 and 2 are more than just curiosities. In his initial description, Chiari classified the hindbrain malformations into type I, II and III and then latter added type IV malformation. Type IV is a very rare type. It is characterized by cerebellar hypoplasia or aplasia and tentorial hypoplasia. There is no hindbrain herniation in this type. We report a case of a 6 year old male patient who presented to us with a 6 year history of an occipitocervical mass and inability to stand and walk for one year and a 5 month history of headache and vomiting. CT scan of the brain showed a midline posterior fossa bone defect with a meningocele with active obstructive hydrocephalus and hypoplastic cerebellum without hindbrain herniation. A diagnosis of a posterior fossa congenital anomaly (Chiari 4) with obstructive hydrocephalus and occipito-cervical meningocele was made. Ventriculo-peritoneal shunt was inserted three days post admission. Patient was then electively taken to theatre five months later for repair of the occipitocervical meningocele.

Keywords: Chiari Type IV, Hydrocephalus, Meningocele, Hindbrain

1. Introduction

The Chiari malformations are a family of conditions characterized by developmental or, less commonly, acquired displacements of the cerebellum. The original 19th century description by Hans Chiari delineated 4 types, but only types 1 and 2 are more than just curiosities. [1]

In 1883, John Cleland described a case of hindbrain malformation found during autopsy. Hans Chiari, an Austrian pathologist, performed post-mortem examination of forty cases between 1891 and 1896 and gave a detailed description of hindbrain malformations. [2]. Chiari described these malformations as congenital anomalies of the hindbrain characterised by downward elongation of the brain stem and cerebellum into the cervical portion of spinal cord. [3] In his initial description, Chiari classified the hindbrain malformations into type I, II and III and then latter added type IV malformation. [4] Type IV is a very rare type. It is characterised by cerebellar hypoplasia or aplasia and tentorial hypoplasia. There is no hind brain herniation in this type [5].

2. Case Presentation

6 year old male patient who presented to us with a 6 year history of an occipito-cervical mass and difficulty in standing and walking, and a 5 month history of headache and vomiting.

This was a chronically unwell first born child who was delivered via a normal vaginal delivery. The mass was noticed at birth and child was put on a course of antibiotics with no improvement. This was a planned and booked pregnancy. Mother did not have any antenatal ultrasound scan and the antenatal period was uneventful. She did not suffer from any infections and did not take any medications. There is no family history of birth defects or any malignancies. Mother did not have any chronic illnesses.

After birth child was otherwise well except that he generally had delayed developmental milestones. He walked at the age of 4 years and at the same period developed slurred speech and was having broad-based staggering gait. These symptoms and signs progressively got worse until the age of 5 when he could now only manage about 2 steps before he would fall down. Right around about the same time child started complaining of severe early morning headaches which were associated with projectile vomiting independent of meals. Mother noted also that the child was developing a divergent squint. With these symptoms she went to a local clinic from where he was then immediately referred to our hospital.

On examination, he was ill looking with generalised pallor and dehydration, apyrexial, Level of consciousness 14/15 and pupils were 3mm bilaterally reactive to light. He had bilateral 6th nerve palsies. He had blurred vision. He had nuchal rigidity and was globally hypotonic, hyporeflexic, could stand and walk with difficulties or sit unsupported. There was a fluctuant occipitocervical pulsatile ovoid mass of 5 x 4 cm in its widest diametres, located over the midline. Power was 4/5 across all muscle groups. Planter reflexes were up going bilaterally and had positive dysdiadochokinesia and dysmmetria. Patient also had a slurred speech; however he had normal cough and gag reflexes.

Patient was resuscitated and then investigations were done. Full blood count revealed anaemia with HB of 7, 9 gm/dl

CT scan of the brain showed a midline posterior fossa bone defect with a meningocele and active obstructive hydrocephalus and a hypoplastic cerebellum without hindbrain herniation.

A diagnosis of a posterior fossa congenital anomaly (Chiari 4) with obstructive hydrocephalus and occipitocervical meningocele was made.

Patient received one unit of packed cells then had a ventriculo-peritoneal shunt inserted three days post admission, after which the signs and symptoms of raised intracranial pressure resolved. Patient was then electively taken to theatre five months later for repair of the occipito-cervical meningocele.

Postoperatively, patient developed CSF leak which resolved spontaneously after a day. Two days postoperatively, patient could now sit, stand and walk with an almost normal gait unaided without falling down.

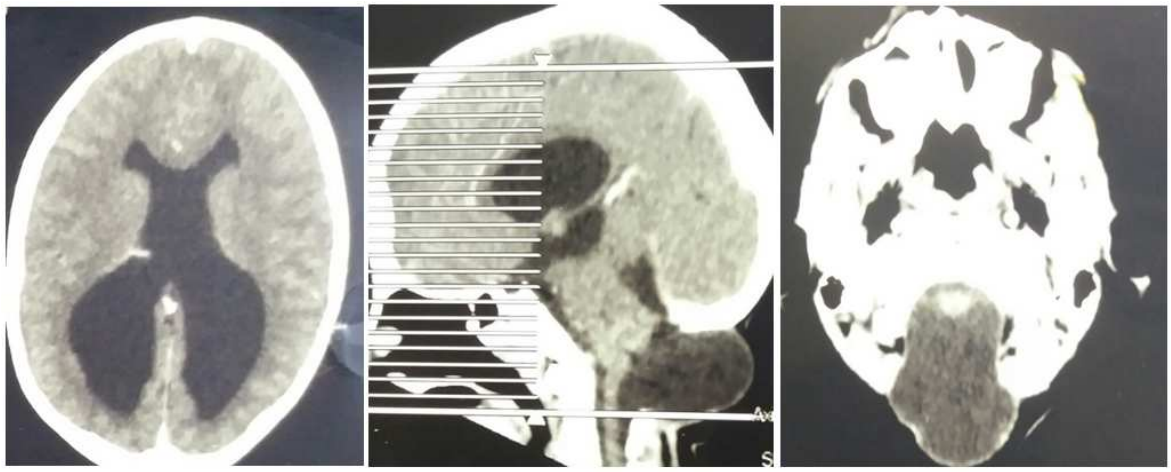

Figure 1. *A-axial ct scan showing hydrocephalus. b-saggittal image showing hypoplastic cerebellum and occipitocervical meningocele. c-axial ct scan at the skull base showing the meningocele.*

Figure 2. *D and e are showing intraoperative images of position and appearance of the occipitocervical meningocele, marking of the incision and the vp shunt which was inserted prior. f is showing the sac of the meningocele opened exposing the cervical cord and aprt of the tonsils.*

Figure 3. *G, H and i are showing patient on day 3 post op. patient standing and walking without assistance.*

3. Discussion

Chiari malformations are generally regarded as a pathological continuum of increasingly severe hindbrain mal developments [6]. It is a very rare type. It is characterised by cerebellar hypoplasia or aplasia and tentorial hypoplasia. There is no hind brain herniation in this type. Other types of Chiari malformation include Chiari 0 and Chiari 1.5 types. Chiari 0 includes minimal or no hind brain herniation but the headache and other symptoms of Chiari malformation are present. Chiari 1.5 includes patients with tonsillar herniation without brain stem elongation or fourth ventricle deformation. [5]. Chiari type 4 is the most severe form and the rarest. The cerebellum fails to develop normally which was the case in our patient. There may be other associated malformations of the brain and brainstem. Most babies born with this malformation do not survive infancy [7]. Because of the lack of cerebellar tonsillar herniation, some researchers do not consider this condition a form of Chiari malformation [8].

The exact cause of the developmental failures leading to Chiari Malformations is unknown. Chiari Malformations affect individuals of every race and ethnicity, but some studies suggest that females are more often affected versus males. The abnormally small posterior fossa may lead to the growing brain to be pushed down into the foramen magnum. Exposure to harmful substances or not enough vitamins and nutrients during fetal development, post-natal osseous growth disturbances, or a genetic association are hypothesized causes of the the small posterior fossa [9] A number of studies suggest that genetic factors play an important role in Chiari Malformations. Research has been conducted on familial aggregation and a higher degree of concordance in monozygotic twins. [10]

To date, no single gene has been linked to Chiari Malformations. Another study was conducted using MRI images from 23 families with 71 affected individuals. The heritability and cranial morphology correlations were measured among the individuals [9]. A linkage of over 10,000 single-nucleotide polymorphisms (SNPs) across the genome were used to identify regions of linkage to Chiari Malformation. A SNP is the most common variation in the human genome and occurs approximately once every 100 to 300 bases [11] the study suggested linkage to Chromosome 9 and [12] based on the results of the studies, it is suggested that Chiari Malformations

Originate as a disorder of Para-axial mesoderm [10]. The diagnosis of Chiari Malformations is related to the patient's symptoms. Some patients experience symptoms specific to Chiari Malformation while others experience a gradual onset of symptoms that are often misdiagnosed with another condition. Some patients do not experience any symptoms at all. In this case the finding is incidental as the patient had neuroimaging for another reason. Adult patients often go 5 years before being properly diagnosed with Chiari Malformation [12]. The diagnosis of Chiari Malformations is established with diagnostic imaging. The diagnostic imaging modality of choice is MRI. [13] An encephalocele is a congenital hernia of intracranial contents which protrude from a cranial defect. The intracranial contents which extrude to the exterior from the defect may include cerebrospinal fluid (CSF), meningeal structures, or brain tissue [14, 15] our patient had a meningocele instead. The type of encephalocele can be described according to the anatomic localizationof the defect [14]. Nearly 75% of encephaloceles are located in the occipital region, while 13–15% are located in the frontal ethmoidal region and 10–12% in the parietal or sphenoidal region [16, 17]

Encephaloceles occur less frequently than spinal dysraphisms. Hydrocephalus may accompany encephaloceles, while anomalies of other organs and extremities can also be observed [18, 19] The size of the brain tissue within the sac, the presence of hydrocephaly and microcephaly, and other congenital abnormalities are factors that influence the prognosis of patients with encephaloceles. [20] According to the literature, the mortality rate is nearly 33.3% in patients with encephaloceles [21], however, the mortality rate according to kotil et al was 29% of cases. Hydrocephaly and infection are the most frequent complications encountered during the postoperative period [20]. But our patient did not suffer from any postoperative complications. Meckel-Gruber syndrome is a rare autosomal recessive condition that is characterized by an occipital encephalocele, cleft lip or palate, microcephaly, microphthalmia, abnormal genitalia, polycystic

kidneys, and polydactyly. Congenital anomalies of the posterior fossa, including the Dandy-Walker syndrome, the Chiari malformation, and encephalocele, are prominently associated with ataxia because of their destruction or replacement of the cerebellum. [22]

Chiari Malformation treatment is dependent upon the individual's symptoms.

Patients with Chiari Malformation that do not experience symptoms are often not treated at all. Conservative treatments such as pain medications, massage therapy or activity reduction usually suffice in individuals with mild symtoms. Patients with either no symptoms or mild symptoms are checked regularly using MRI to monitor the progression of the condition. A thorough search of literature did not reveal any case of Chiari type 4 with occipitocervical meningocele. From our experience with this patient we recommend occipitocervical decompression of such patients which includes a midline durotomy and opening of the arachnoid membrane with adhesiolysis as well, followed by a watertight dural closure. Fifty percent or less of people diagnosed with Chiari Malformation actually undergo surgery

4. Conclusion

Chiari Malformation findings are becoming more common as the use of diagnostic imaging has increased. Sagittal MRI images are used to help diagnose and classify the different types of Chiari malformations. Researchers are working to narrow down the genetic aspects that could possibly be the cause of this condition. The symptoms of Chiari Malformations depend on which type and vary from person to person. An occipital headache is the most common symptom among patients with Chiari Malformation. Some patients do not experience any symptoms at all, while others range from mild to severe. Conservative treatments should be considered for patients with mild symptoms and for patients with chiari type 4 with occipitocervical meningoceles, posterior fossa decompression via repair of the meningocele helps in improving symptoms and ultimately quality of life of such patients with this extremely rare form of chiari malformation.

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