Chiari Type III Malformation Presenting With Two Huge Encephaloceles in a New Born Infant: Case Report

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Abstract- Arnold-Chiari type III malformation constitutes the rarest type of all the Chiari malformations. This extremely rare congenital anomaly shows poor prognosis and results in either early death or development of various severe neurologic deficits in the surviving patients. It is characterized by the herniation of hindbrain contents into an encephalocele sac through a bony defect located in the upper cervical or lower occipital region. Magnetic resonance imaging (MRI) is the preferred imaging modality in the diagnosis of this rare congenital anomaly, especially due to its multiplanar capability. In this case report, we described the imaging findings of Arnold-Chiari type III malformation in a newborn male infant. He presented with two huge encephalocele sacs containing dysplastic brain tissues in the upper cervical and lower occipital regions together with various findings related to the disease. Following a right-sided shunt insertion for hydrocephalus treatment, he has undergone a surgical procedure ensuring removal of the encephalocele sacs with primary closure of the bony defects.

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Keywords: Chiari III malformation; Craniovertebral junction; Syringomyelia

Introduction

Arnold-Chiari malformation type III is an exceedingly rare congenital anomaly, characterized by caudal medullary displacement and herniation of the hindbrain contents into encephaloceles through an upper cervical or low occipital osseous defect (1,2). It was first described by an Austrian pathologist Hans Chiari in an autopsy series of 40 children presenting with hindbrain malformations and was classified into four types depending on the degree of cerebellar prolapsus (3). Among these types, Chiari type I and type II malformations are commonly encountered, where Chiari type III and IV malformations are extremely rare. Newborn infants having this type of malformation (i.e., type III) usually present with symptoms of severe respiratory failure, seizures, ataxia, spasticity and swallowing dysfunction. As a result, high mortality rates and serious neurologic deficits in the surviving patients are inevitable. Although most of the Chiari type III malformations are not able to survive, surgical reconstruction should still be recommended. Herein, we are demonstrating the computed tomography (CT) and magnetic resonance imaging (MRI) findings of a newborn male infant with Chiari type III malformation presenting with two huge encephaloceles.

Case Report

A newborn male infant was delivered by cesarean section at 36 weeks of pregnancy in our hospital. His mother was 37 years old and had no health problems. The infant has shown respiratory distress and hypotonia. On physical examination his weight was 3160 gr, head circumference was 32 cm and height was measured as 44 cm. His hematological and other biochemical parameters were found within normal limits. On palpation, two huge pedunculated masses in the upper cervical and lower occipital regions measuring 69x45x18 mm and 90x81x54 mm respectively, were

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noted. The skin covering these two masses was intact. He was first referred to cranial CT examination which was performed by using a 128-detector row helical CT scanner (Somatom Sensation 16, Siemens Medical Systems, Erlangen, Germany). Before the examination an informed consent was provided by his parents. Images were obtained with following parameters: 120 kVp, variable tube current (150-250 mAs), slice thickness 5 mm. and reconstruction interval of 2 mm. Axial parenchymal and bone window-leveled images revealed bony defects along the high cervical and low occipital regions with herniating soft tissue masses toward the extracranial region through these defects (Figure 1,2,3,4).

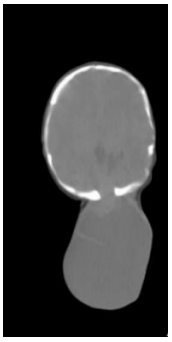

Figure 1. Axial CT bone window-leveled brain image showing midline occipital bony defect, together with soft tissue mass protruding outside through this defect

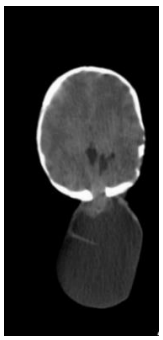

Figure 2. Axial CT parenchymal window-leveled image reveals a hypodense encephalocele sac containing brain tissue. Fronto-parietal bone scalloping is also visible

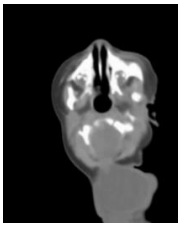

Figure 3. Axial CT bone window-leveled brain image shows midline cervical defect together with the soft tissue mass protruding outside

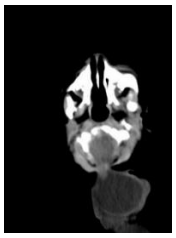

Figure 4. Axial CT parenchymal window-leveled image again demonstrates another encephalocele sac with contained brain tissue

Besides, fronto-parietal bone scalloping (the lemon sign) was observed. Thus, it was decided to investigate in further detail and an MRI examination was performed via a 1.5 tesla magnet (Avanto-SQ Engine; Siemens, Erlangen, Germany). An informed consent was again provided prior to the examination. During this examination, axial and sagittal T1 weighted, axial and coronal T2 weighted, axial and coronal FSE IR (flair), axial SWI (susceptibility weighted) and finally following intravenous gadolinium DTPA administration (0.1 mmol/kg) axial and coronal post-contrast T1 weighted images were obtained. On these images, two huge encephaloceles were detected along the upper cervical and lower occipital regions protruding outside through the bony defects. While the upper cervical encephalocele sac contained occipital brain parenchyma and the meninges, the lower occipital one did have cerebellar tissue along with the meninges. A small posterior fossa was present and the cerebellar tonsils together with the medulla oblongata showed downward herniation below the foramen magnum. A cervico-thoracal syringohydromyelia was detected as a central high T2 signal intensity cystic lesion on the related spinal cord region. Another finding that could clearly be observed on midline sagittal images was the absence of the corpus callosum. Hydrocephalus was also present (Figure 5,6,7,8). The patient immediately underwent a ventriculo-peritoneal shunt operation where a right-sided shunt catheter was inserted. One week after birth, the patient was operated where the encephalocele sacs containing dysplastic brain tissues were excised and primary closure of the bony defects was also performed.

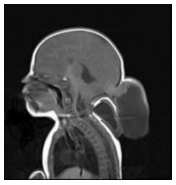

Figure 5. A sagittal T1 weighted midline MR image reveals two encephalocele sacs in the high cervical and occipital regions containing dysplastic brain tissue. Absence of the corpus callosum and a small posterior fossa are present

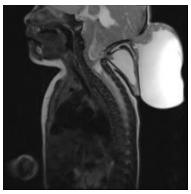

Figure 6. Sagittal T2 weighted midline MR image depicts downward herniation of the medulla oblongata and the cerebellar tonsils below the foramen magnum. Cervico- thoracal syringohydromyelia is evident. Hydrocephalus is also present

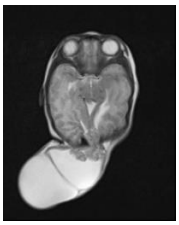

Figure 7. Axial T2 weighted MR image clearly demonstrates a huge encephalocele sac containing dysplastic occipital lobe and meningeal tissues protruding outside through a midline occipital defect

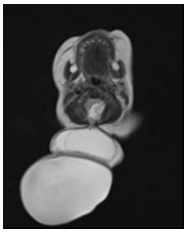

Figure 8. Axial T2 weighted MR image through a lower level shows another encephalocele sac containing the cerebellum and the meningeal tissues protruding outside through the midline bony defect at the cervical region. Posterior to this sac, the prior one is visible again

Discussion

The etiology of Arnold-Chiari type III malformation is still unclear. It is postulated that in the embryonic life, incorrect neuralization occurs during the ventricle extension process and this leads to imperfect formation of the occipital bone which results in subsequent prolapsus of the brain stem and the cerebellum (4). Other associated anomalies are cerebellar tonsillar herniation, hydrocephalus, syringomyelia, a small posterior fossa, hypoplastic tentorium cerebelli, dysgenesis of the corpus callosum, deformed midbrain, petrous and clivus scalloping and fenestrated falx cerebelli (5). MRI is the essential modality in the diagnosis of Chiari type III malformation due to its multiplanar capability and lack of ionizing radiation. Especially sagittal T2 weighted images are very helpful in terms of revealing accompanying multiple pathologies to this rare congenital anomaly. Besides, prior to corrective surgery procedures, it is crucial to give precise detailed information to the surgeons about the position of the midbrain and the brain stem in order to reduce the potential damage risk to these structures (6). In our case report, we performed both CT and MRI examinations by which the obtained findings supported the diagnosis. These included a small posterior fossa, fronto-parietal bony scalloping (lemon sign), downward herniation of the medulla oblongata and cerebellar tonsils, dysgenesis of the corpus callosum, hydrocephalus and syringomyelia. We also revealed two huge encephalocele sacs containing dysplastic brain tissues in the high cervical and low occipital regions with accompanying midline bony defects. To the best of our knowledge, this can be the first case of Arnold-Chiari type III malformation in the literature presenting with two huge encephalocele sacs simultaneously in the same patient. Hydrocephalus is commonly seen in Chiari type III patients and is thought to occur due to compression of neural structures in the posterior fossa. Hence, cerebrospinal fluid (CSF) circulation may be seriously impaired and this manifests itself as hydrocephalus and syringohydromyelia as seen in our case. It is highly recommended to follow up these patients with head circumference measurements due to increased possibility of subsequent hydrocephalus development. Ventriculo-peritoneal shunt procedure is a treatment of choice in these patients. In our case report, a right-sided ventriculo-peritoneal shunt operation was performed prior to surgery. Encephaloceles is a characteristic finding of Chiari type III malformation and may contain various amount of brain tissue within. As seen in our case, occipital lobes, cerebellum and CSF are commonly, but medulla oblongata and pons are seldomly seen in encephalocele sacs (7). The various brain tissues can be found in the encephalocele sacs, but they are considered to be nonviable (1,2,8). In Chiari malformations, the severity of the patients' symptoms has direct correlation with the amount of the herniated neural tissues to the sac (7). In Chiari type III malformations presenting with encephaloceles, the best treatment of choice is surgery for preventing meningitis and excluding the possibility of potential rupture of the sac (9). The early operative closure of the defect with the removal of encephalocele along with its contents and a following reconstruction procedure of the dura and skin is recommended (10).

In conclusion, Chiari type III malformation is a very rare entity with high mortality rates causing severe neurological deficits in surviving patients. MRI plays a

crucial role in the diagnosis and comprehensive evaluation of these patients. Appropriately timed surgical interventions can result in satisfactory outcomes and low mortality rates.

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