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Case Report

Chiari Malformation Type III: A Rare Case Report

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Abstract

Chiari malformations (CM) are intricate brain deformities first described and characterized by Hans Chiari in 1891 [1]. They constitute a group of disorders involving abnormalities in the posterior fossa and posterior brain. There are seven types of CM: 0, I, 1.5, II, III, IV, and V [1]. Chiari malformation type III (CM III) is an exceedingly rare condition characterized by a low occipital or upper cervical encephalocele and downward displacement of the brainstem. Type III carries a grim prognosis, marked by a high mortality rate and severe neurological and developmental deficits in surviving patients [2]. We present the case of a newborn female with a lumbo-sacral mass, clubfoot, and dysmorphic facial features. An MRI revealed an open lumbosacral myelomeningocele associated with an occipital encephalocele and downward protrusion of the cerebellum, fitting within the framework of a Chiari type 3 malformation.

Keywords: *Chiari Malformation Type III; MRI; Encephalocele; Amygdaloid Protrusion*

Introduction

Chiari malformation type III is the rarest among Chiari malformations [3], typically reported as sporadic cases, underscoring the anomaly's rarity [4]. It is often associated with a bleak prognosis [3], initially documented by Hans Chiari during the autopsy of 40 children with posterior brain dysplasia in 1981 [5]. Anatomically, Chiari type III is characterized by the presence of a high cervical or occipital encephalocele, along with several abnormalities observed in type II. These include a shallow posterior cranial fossa, caudal migration of cerebellar tonsils, medullary kinking, beaked tectum, and hydrocephalus [6]. We present the case of a 15-day-old female newborn with an open lumbosacral myelomeningocele associated with an occipital encephalocele and an amygdaloid protrusion, fitting the profile of a Chiari type 3 malformation.

Observation

We present the case of a 15-day-old female newborn delivered vaginally following a full-term pregnancy from non-consanguineous parents.

At birth, the newborn examination revealed a lumbar mass covered with dystrophic skin, club feet, and dysmorphic facial features. Subsequently, an MRI was performed, demonstrating the following findings:

- There is an occipital bone defect measuring 14.8 mm in diameter, through which protrudes a sac of meningeal layers containing cerebellar tissue, measuring 15 x 18 x 15 mm (TxAxP) indicative of an occipital encephalocele. Additionally, there's a protrusion of cerebellar tonsils measuring 11 mm, a gyration abnormality like nodular heterotopia adjacent to the left occipital horn, with an enlarged ventricular system and a more pronounced bi-parietal cephalohematoma on the right side.
- At the spinal level, there is a defect in the posterior vertebral arch corresponding to L3-S3, giving rise to a meningeal collection with isosignal T1 and hypersignal T2, containing fibers from the cauda equina without fatty components. This measures 55 x 57 x 30 mm (HxWxAP), indicative of an open lumbosacral myelomeningocele associated with a syringomyelia cavity at the level of the terminal filum, with the spinal cord abnormally attached at L3.

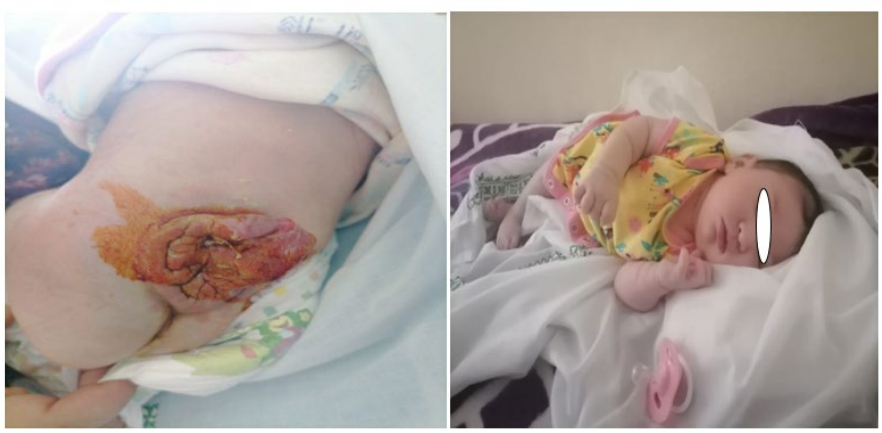
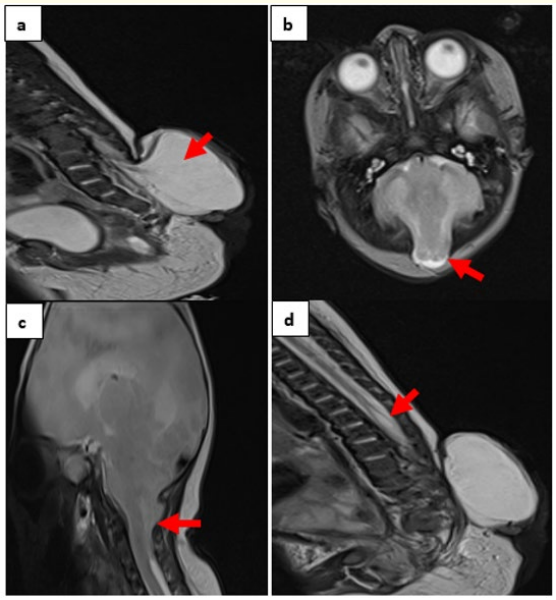


Figure 1: *A 15-day-old newborn with a lumbar mass covered by dystrophic skin, displaying club feet and dysmorphic facial features.*



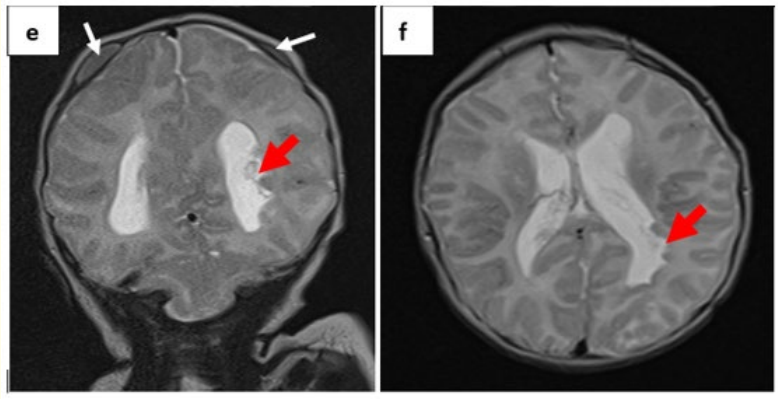


Figure 2: *MRI appearance suggestive of an open lumbosacral myelomeningocele (a) associated with an occipital encephalocele (b), cerebellar tonsil protrusion (c), syringomyelia cavity at the terminal filum level with a low-lying spinal cord at L3 (d), nodular heterotopia gyration abnormality (e, f).*

Note: More pronounced bi-parietal cephalohematoma on the right side (white arrows (e)).

Discussion

Chiari malformation (CM) type 3 is an extremely rare anomaly [7]. The incidence of encephalocele is 1 in 5,000 live births [7]. It is characterized by the herniation of the contents of the posterior fossa, including the cerebellum, brainstem, and fourth ventricle, and in some cases, the upper cervical spinal cord through an occipital and/or upper cervical bony defect [7]. CM type III is the most severe variant of ACM [2], associated with a poor prognosis leading to early death or severe disability in survivors [2].

The etiology of CM III is largely unknown. However, there is a theory suggesting that incorrect neurulation during ventricular extension leads to imperfect formation of the occipital area. Later on, there is observed prolapse of the cerebellum and brainstem [8]. Another theory is that incomplete descent of the ventricular system due to abnormal neurulation results in hypoplasia of the posterior fossa. Failure of enchondral bone ossification is also a key factor in this pathology [8].

Presentations vary, ranging from asymptomatic newborns to children presenting with seizures, severe neurological deficits, respiratory insufficiency, dysphagia, or developmental delay. Hydrocephalus is the most commonly associated anomaly [6,8].

The presence of an occipital and/or cervical anomaly alongside neural tissue herniation is crucial for diagnosis and may be associated with other anomalies [4]. Apart from encephalocele, similar anomalies to those in Chiari type II malformation are present. These include deformed midbrain, partially or completely dysgenetic corpus callosum (77%), scalloped petrous temporal bone and clivus (56%), herniated tonsils (43%), and hydrocephalus and hydromyelia in 22% and 30% of patients, respectively [3]. The content of the encephalocele varies in size and degree of dysplasia, with a dysmorphic ventricle potentially trapped within the encephalocele [3].

Syringomyelia is a rare progressive chronic condition of diverse etiologies, characterized by the presence of intramedullary cavities filled with cerebrospinal fluid. The most frequent etiology is Chiari malformation [8].

Our patient presents syringomyelia at the level of the terminal filum with a low-lying spinal cord at L3 and a nodular heterotopia gyration anomaly.

Muzumdar., *et al*. reported a case of a newborn with two distinct masses: an encephalocele and a myeloencephalocele [8], as observed in our patient as well.

Prenatal morphological ultrasound can reveal a cystic neck mass, ventriculomegaly, or microencephaly before birth. While ultrasound is the preferred method for evaluating fetal anomalies, it can be limited by the maternal body habitus, fetal position, oligohydramnios, and bone structure ossification. Recent studies have shown that prenatal MRI is useful for assessing specific brain anomalies, particularly those involving the posterior fossa [8].

Brain MRI is the gold standard for precise anatomical diagnosis of CM and for detecting associated abnormalities such as spinal cord folding, breaking of the tectum's quadrigeminal plate, spinal cord syrinx, and shallow posterior fossa. Some neurological symptoms like cranial nerve paralysis, hypotonia, and respiratory compromise can be attributed to these anomalies [4]. Preoperative angio-MRI is additionally important for localizing venous sinuses and detecting their patency; instances of double and triple division of the superior sagittal sinus have been reported. This information could prevent massive bleeding [4,5].

CT scanning complements the assessment, better suited for analyzing bone defects and all other associated bone malformations [8].

The primary treatment for meningoencephalocele in CM III patients is surgery, aiming to prevent meningitis or rupture of the encephalocele. Shunting for hydrocephalus may sometimes be performed before sac removal surgery. Vigilant observation is crucial in all cases to determine the presence of hydrocephalus [5]. Rehabilitation methods for poor head control and trunk ataxia due to CM III generally rely on neurodevelopmental treatment [5].

Conclusion

Chiari malformation type III is a rare anomaly. With its distinct clinical and radiological features, it can be easily differentiated from other Chiari types. Diagnosis can be readily achieved through MRI. Type III is associated with a grim prognosis, characterized by a high mortality rate and severe neurological and developmental deficits in surviving patients.

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