Case Report

A case report of a female child with Chiari II malformation with associated heterotopias.

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Abstract

Several brain malformations have been linked with Chiari II malformation. The presentation of heterotopias in classic Chiari malformation is however often uncommon, compared to other known associations, such as callosal dysgenesis, myelomeningocele, and absent septum pellucidum. This is a case of a child who presented at infancy with a myelomeningocele and a limb deformity. She had brain and spine MRI at presentation, and a follow up brain MRI. The initial neuroimaging established the diagnosis of Arnold-Chiari (Chiari II) malformation with other common associations, which included myelomeningocele, poorly defined corpus callosum, absent septum pellucidum and tiny subependymal nodules. These pathological associations were however well established in the 5 years follow up study, as they became more prominent. This publication is thus aimed at emphasising the importance of follow up studies in pediatric age group, especially new-born and infants, and to present the association between Chiari II malformation and brain heterotopias.

Keywords: Chiari II, Arnold-chiari, Brain heterotopia, Paediatric

Introduction

Heterotopia is a subgroup of migration abnormalities, identified by arrest of radial migration of neuroblasts. Being a well-known reason for formative delay, mental retardation, and epilepsy, association of heterotopia with some other circumstances has also been incuiuded. There are many case reports about the concurrence of heterotopia and encephalocele; however, no such association has been announced with cervical meningocele. Herein, we present the first case of periventricular nodular heterotopia included with cervical meningocele. Our case additionally emphasizes the role of brain MRI in the evaluation for those with neural tube defects. Identification of possible included heterotopia is crucial for planning proper treatment and genetic counseling [1-3]. Several malformations of brain development, such as Chiari II malformation have been claimed to be an outcome of severe inter-linked time-dependent failures in the development of the ventricular system. It had been proposed that the cerebrospinal fluid leakage from the neural tube defect is a significant factor responsible for small posterior fossa. More so, non-distention of the developing telencephalic ventricular system invariably leads to various cerebral abnormalities, such as callosal dysgenesis, heterotopias, and polymicrogyria. Chiari II malformation generally comprises small posterior fossa, cerebellar tonsillar and/or vermis herniation, and inferiorly pooled brain stem with subsequent elongation and low-lying of the fourth ventricle. There is also angulation of the aqueduct with subsequent aqueductal stenosis and hydrocephalus, as well as spinal myelomeningocele and cord tethering [4-6]. Other associated central nervous system abnormalities may include: Spinal (syringohydramyelia, scoliosis, diastematomyelia, segmentational anomalies), cerebral (corpus callosum dysgenesis, absent septum pellucidum, fenestration of falx, stenogyria, tectal beaking), cranial vault (scalloping of petrous temporal bone, enlarged foramen magnum, luckenschadel skull) and skeletal (club foot) abnormalities. Associated neuronal migration malformations (heterotopia) have been shown to be of high proportion in patients with Chiari II malformation. There has also been a report of periventricular heterotopia in childhood Chiari I malformation. Clinical presentations of Chiari II malformation usually vary with age. Neonates generally present with myelomeningocele, brainstem dysfunction or neurogenic bladder. While childhood presentations could be due to hydrocephalus or musculoskeletal abnormalities, young adults usually present with syrinx and/or scoliosis. Heterotopias, however is known to be associated with seizures as the most prominent clinical presentation [7-9].

Case Presentation

This is case of a 6year old girl who was being followed up since birth. She was a product of full-term pregnancy and delivered through caesarean section. At birth, she presented with talipes equinovarus and lumbosacral myelomeningocele. There was a positive family history of talipes equinovarus in her aunt. She had corrective surgery for the Talipes at age 6 months. She also had a Ventriculo-Peritoneal (V-P) shunt for her hydrocephalus [10]. Patient had diagnostic brain and spine Magnetic Resonance Imaging (MRI) at infancy, and a follow up study after about 5 years when she presented with headache and vomiting. The initial MRI at infancy shows evidence of Chiari II malformation (small posterior fossa, cerebellar tonsillar herniation, inferiorly displaced medulla, stretching of

4th ventricle), hydrocephalus and posterior lumbosacral myelomeningocoele and some cord tethering. Other associated findings were thin/partially visualised and poorly delineated corpus callosum, absent septum pellucidum, as well as some tiny subependymal soft tissue nodules (subependymal heterotopias) (Figure 1) [11].

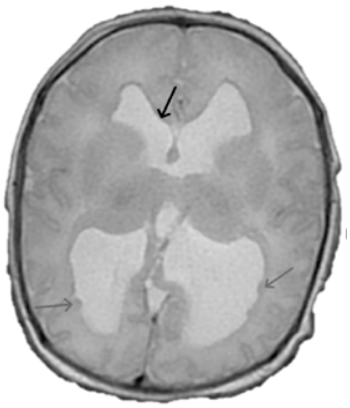

Figure 1. *Brain MRI (T2 axial and sagittal) showing tiny subependymal nodules (heterotopias) in the occipital horns of the IateraJ ventricle (blue arrows). Partially visualised corpus callosum is seen as a thin line structure lining the frontal horns of anterior horns of the lateral ventricles (black arrow).*

The patient's follow up brain MRI showed prominence and confirmation of the previously noted findings which now included multifocal periventricular and subependymal heterotopias, tonsillar herniation of up to 7.0 mm below the foramen magnum and inferiorly displaced medulla. Dysgenesis is now better seen as absence of its posterior half. There was also obstructive hydrocephalus despite an established association between heterotopias and seizure, There was however no clinical history of seizures at the time of this publication (Figure 2) [12].

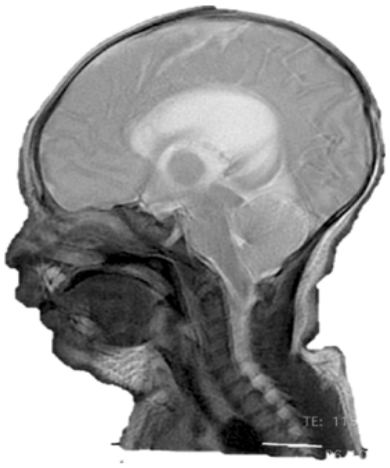

Figure 2. *There is some herniation of the cerebellar tonsil and brain (sagittal image) in image B. Other findings include dilated lateral and third ventricles.*

Results and Discussion

Abnormality of neuronal migration, especially heterotopias had been reported to co-exist with Chiari-II malformation as in the index case, as well as in childhood Chiari-I malformation. Presence of other structural brain pathologies (such as corpus callosum agenesis, ventriculomegaly, brainstem abnormalities, and pachygyria etc.) in more than half of the children with band, subependymal or periventricular heterotopias.

A similar pattern of periventricular and subependymal heterotopias, is in association with pachygyria. Associated pachygyria, aside other brain abnormalities, in children with grey matter heterotopias (Figure 3) [13].

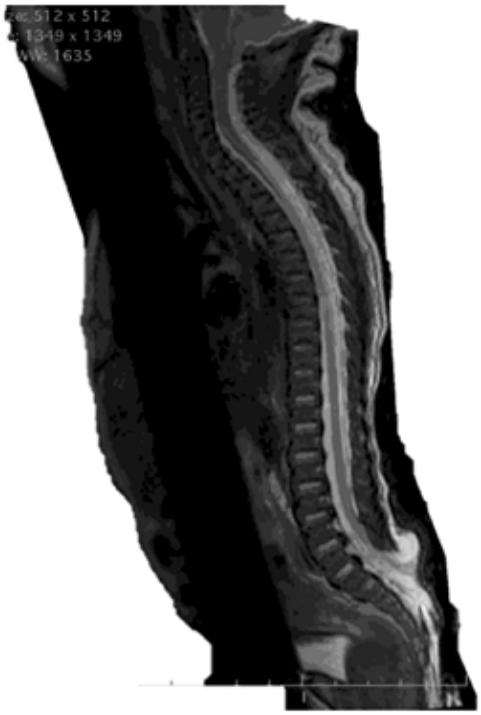

Figure 3. *Spine MRI (T2 sagittal and axial) showing lumbosacral myelomeningocoele with some cord tethering.*

The relevance of a follow up neuro-imaging cannot be over-emphasised, in which the previously missed or unclear heterotopias, were better visualised or confirmed at the follow up study.

Their publication further stressed the unavoidable limitation of neuro-imaging during foetal life or early neonatal period (Figures 4-7) [14,15].

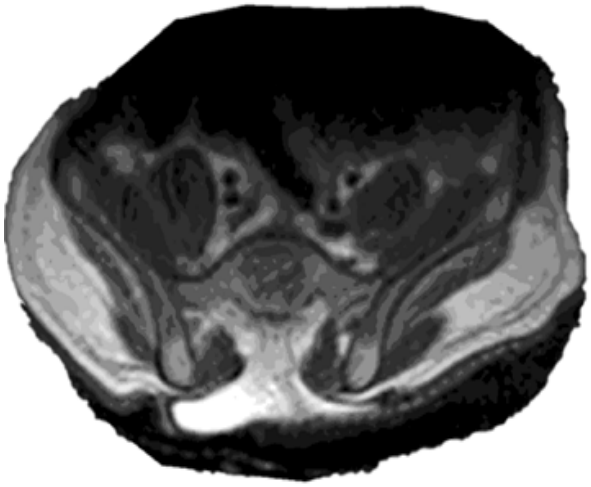

Figure 4. *Neuro-imaging during foetal life.*

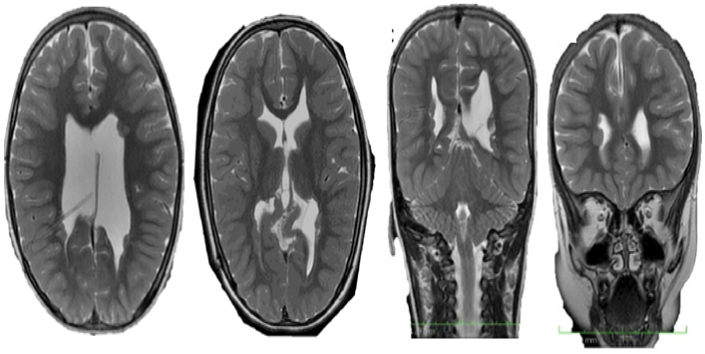

Figure 5. *Brain MRI (T2 axial and coronal) showing multiple periventricular and subependymal nodules (heterotopias). VP shunt is seen within the body of right lateral ventricle.*

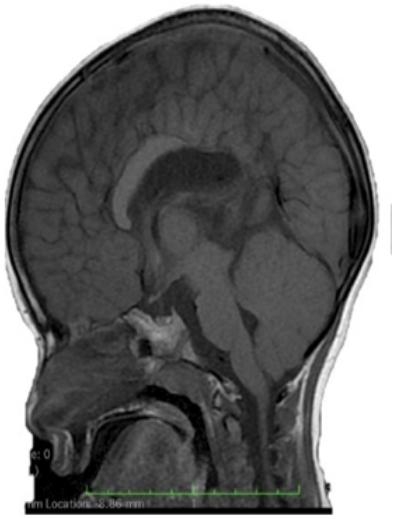

Figure 6. *MRI brain (TI and T2 sagittal) showing hypoplastic corpus callosum, and cerebellar tonsillar and brainstem herniations.*

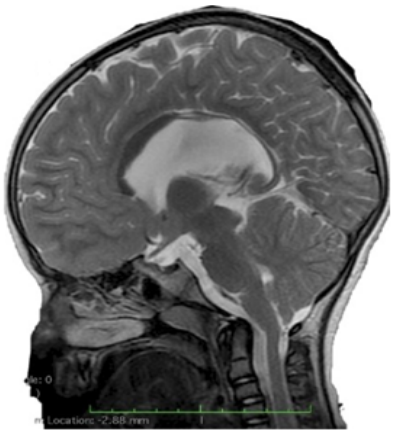

Figure 7. *MRI brain (TI and T2 sagittal) showing brainstem herniations.*

Conclusion

This study aims to document other possible associations (especially heterotopias) with Chiari-II malformation and to stress the need for follow up imaging, to further confirm a previous radiological diagnosis and seek other initially missed findings. Periventricular nodular heterotopia was seen in a relatively high proportion of children with Chiari II malformation, suggesting that it may be associated with severe hindbrain deformity. Knowledge of the occurrence of cortical malformation development and relationship to hindbrain in Chari-II malformation.

Consent

A verbal consent was obtained from the parents, having assured them of maintaining strict patient's anonymity in all writings and images involved in this publication.

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