Paediatrics Today 2011;7(2):140-144
DOI 10.5457/p2005-114.27

ARNOLD CHIARI MALFORMATION ASSOCIATED WITH MULTIPLE CONGENITAL ANOMALIES

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Received: February 26, 2011
Accepted: July 20, 2011

Arnold Chiari malformation is nearly always associated with myelomeningocele, but it is rarely associated with other spine and spinal cord anomalies such as diastematomyelia and hemimyelomeningocele, as well as osseus and visceral anomalies. We report the case of a male newborn with Arnold Chiari malformation associated with diastematomyelia, syringohydromyelia, hemimyelomeningocele, multiple osseus anomalies and ren arcuatus. This article highlights the importance of detailed diagnostic examination of newborns with Arnold Chiari malformation, primarily magnetic resonance imaging, in order to identify the multiple anomalies associated with this malformation.

Key words: Arnold Chiari malformation ▪ Multiple anomalies

Introduction

In 1891, Hans Chiari described three types of rhombencephalon anomalies that were associated with hydrocephalus. One of them is Chiari II malformation, also known as Arnold Chiari (AC) malformation (1).

This malformation implies caudal displacement of the structures of the rhombencephalon through an enlarged foramen magnum into the spinal canal, wedge shaped cerebellum and kneed medulla oblongata. AC malformation is the result of the normal development of the cerebellum in abnormally small posterior fossa with low binding of the tentorium (2, 3).

Children with AC malformation usually have associated supratentorial brain anomalies, and almost all have myelomeningocele. Some authors even define AC malformation as rhombencephalon anomalies in individuals with myelo-

meningocele (4). In contrast, diastematomyelia, hemimyelomeningocele, bone and visceral anomalies are rarely seen in children with AC malformation (5). In this case report, AC malformation in the newborn is associated with multiple anomalies of the spine and spinal cord as well with other visceral anomalies. This article emphasises prompt and adequate diagnostics, including magnetic resonance (MR) with the aim of evaluating the multiple anomalies associated with AC malformation.

Case report

Multiple anomalies were observed in a full-term male newborn from the second, regularly controlled pregnancy, immediately after birth, as follows: facial dysmorphia, pes equinovarus and cystic lesions in the thoracolumbar spine segment which suggested myelomeningocele. In neurological status, the newborn was hypertonic with evident crossing of the lower extremities. Cranial ultrasound revealed hydrocephalus while magnetic resonance of the brain revealed a small posterior fossa with caudal displacement of the fourth ventricle, medulla oblongata, vermis and the cerebellar tonsils through an enlarged foramen occipitale magnum into the spinal canal at the level of the 1st thoracic vertebra, with consequent supratentorial hydrocephalus, indicating Arnold Chiari II malformation (Figure 1). MR imaging of the thoracolumbar spine segment revealed multiple anomalies of the spine and the spinal cord in the form of open spinal dysraphism: the split of the thoracolumbar segment of the spinal cord into two hemicords (diastematomyelia) with syringohydromyelia of the left hemicord; thoracolumbar spina bifida cystica with the sac filled with cerebrospinal fluid, neural placode of the "normal" hemicord and second hemicord with syringohydromyelia which corresponded to hemimyelomeningocele (Figure 2). The lumbar

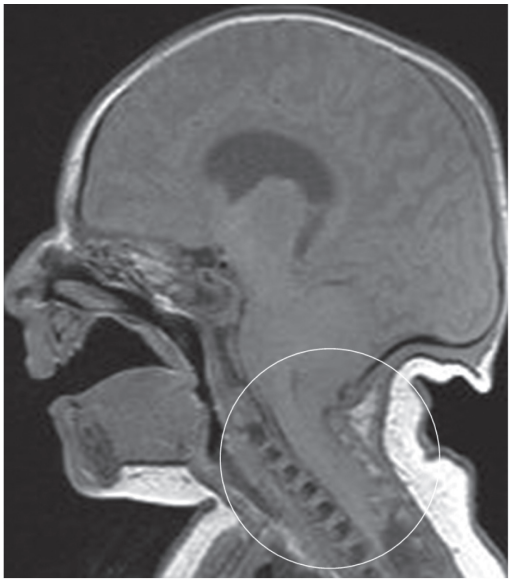

Figure 1 *The sagital unenhanced T1-weighted image of the brain shows a small posterior fossa with caudal displacement of the fourth ventricle, medulla oblongata, vermis and the cerebellar tonsils through an enlarged foramen occipitale magnum into the spinal canal at the level of the 1st thoracic vertebra.*

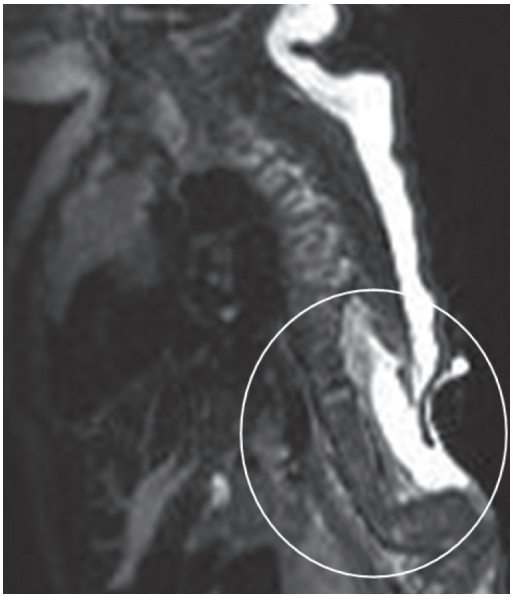

Figure 2 *The sagital unenhanced T1-weighted image of the spine shows thoracolumbar spina bifida cystica with the sac filled with cerebrospinal fluid and neural placode of the "normal" hemicord which correspond to hemimyelomeningocele.*

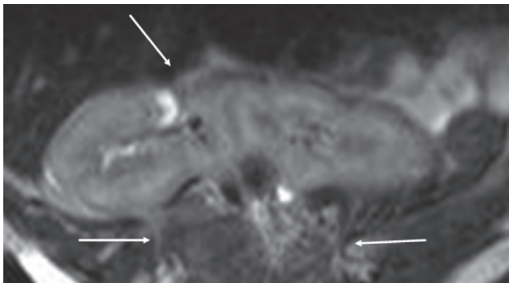

Figure 3 *The axial T2-weighted image of the spine shows a lobulated mass of more smaller formations which correspond to the supernumerary vertebras. The image also shows a ren arcuatus.*

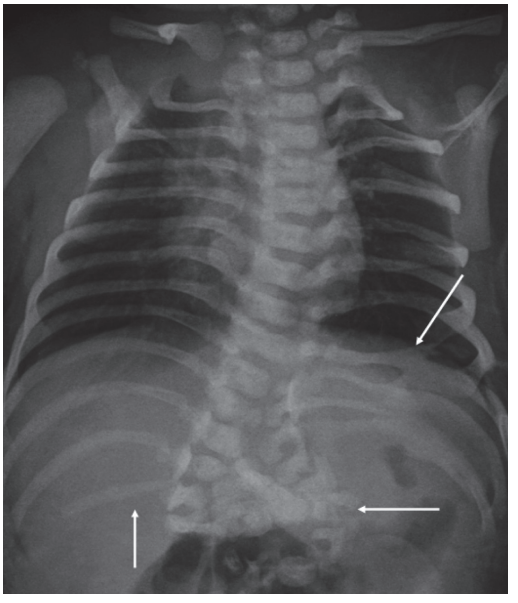

Figure 4 *Standard X-ray examination shows supernumerary lumbal vertebras with failure of formation as hemivertebras and butterfly vertebras, forming two columns. The image also shows butterfly vertebras of the lower thoracic segment of the spine, synostosis of the lower ribs on the left as well as a supernumerary 13th rib on the right.*

spine segment consisted of a lobulated mass of several smaller formations, which corresponded to the supernumerary vertebras (Figure 3).

Standard X-ray examination confirmed supernumerary lumbal vertebras with failure of formation, as hemivertebras and butterfly vertebras, forming two columns. X-ray also showed butterfly vertebras of the lower thoracic segment of the spine, synostosis of the lower ribs on the left as well as a supernumerary 13th rib on the right (Figure 4). MR imaging also revealed a ren arcuatus (Figure 4). Abdominal ultrasound did not reveal other congenital malformations. Enzyme-linked immunosorbent assay was positive for Toxoplasma, Citomegalovirus and Herpes simplex IgG antibodies. The karyotype analysis showed the normal male 46 XY karyotype. Due to the multiple anomalies, surgical treatment was not indicated. The newborn died on the thirtieth day of life.

Discussion

Chiari malformations are graded as types I, II, III and IV. Chiari II malformation is the most common and is usually associated with myelomeningocele (1, 2.) The most common supratentorial brain anomalies that are associated with AC malformation are: hypoplasia or absence of the corpus callosum, that appears in 70-90% of affected patients, enlargement of the caudate heads, hypoplasia of the falx and tentorium, and obstructive hydrocephalus (2, 6). In 95% of cases, AC malformation is associated with myelomeningocele, and in 50-90% with syringohydromyelia (2). This malformation is rarely seen with other spinal cord anomalies such as diastematomyelia and hemimyelomeningocele and with bone and visceral anomalies (5).

Besides the diastematomyelia and hemimyelomeningocele, the newborn, in our case, had multiple bone anomalies in a form of the failure of formation of the thoracal and lumbale vertebras, synostosis of the ribs, a supernumerary 13th rib, pes equinovarus, and a ren arcuatus of visceral anomalies. Due to the possible association of AC malformation with multiple anomalies, detail

diagnostic examination of newborns with myelomeningocele is necessary. In children with myelomeningocele and suspicion of AC malformation, MRI is recommended as the gold standard and obligatory diagnostic examination with the aim of full disclosure of the pathomorphological manifestation of this anomaly and its association with other anomalies of the brain, spine and spinal cord (7). Furthermore, MRI has a major role in the evaluation of the obstructive hydrocephalus which is present in more than 80% of children with AC malformation, because of the aqueduct dysfunction after initial surgical treatment of the myelomeningocele, mostly in the first 48 hours of life (8).

Approximately 33% of the children with AC malformation that are not surgically treated by five years of age, develop symptoms of damaged brainstem with swallowing disorders, stridor, weak cry, adynamic arms and apnoea during sleeping. One third of these children die, mostly because of respiratory insufficiency (9). Children with brainstem dysfunction have a significantly higher mortality rate than those without, even after neurosurgical brainstem decompression (10).

However, by early recognition of brainstem compression, based on developed symptoms and MRI of the brain, with prompt surgical intervention, it is possible to decrease the high mortality of children with AC malformation (7). On the other hand, by adequate and well-timed antenatal ultrasound examination, alpha-fetoprotein analysis and amniocentesis during pregnancy, it is necessary to diagnose myelomeningocele and other anomalies. Also, AC malformation may be revealed on the basis of ultrasound findings. With the help of timely diagnosis, parents should learn about the risks of pregnancy where they have a possibility of termination or otherwise, adequate counselling and psychological preparation if they decide that the mother will carry the pregnancy to the end (4, 11, 12).

Conclusion

Arnold Chiari II malformation is usually associated with myelomeningocele, but it is rarely associated with other spine and spinal cord anomalies or other bones and visceral anomalies. Magnetic resonance imaging has to be performed in all newborns where this anomaly is suspected in order to obtain a complete insight into the association of anomalies which would provide for appropriate decisions about the surgical treatment of the child. MRI has great importance in the evaluation of brainstem compression, and evaluation of the obstructive hydrocephalus.

Authors' contributions:
Conception and design: SM; Acquisition, analysis and interpretation of data: SM and AB; Drafting the article SM; Revising it critically for important intellectual content: SM and SS.

Conflict of Interest: The authors declare that they have no conflict of interest. This article was not sponsored by any external organisation.

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Citation: Mujagić S, Sarihodžić S, Babović A. Arnold chiari malformation associated with multiple congenital anomalies. Paediatrics Today. 2011;7(2):140-44.