# Arnold-Chiari Malformation: Anatomical Variations and Latest Embryological Perspective. Review of Literature

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**ABSTRACT**

One of The most common anomaly of craniovertebral junction is Chiari malformation. These malformations involve both skeletal and neural structures. Earlier it was estimated that the occurrence of Chiari malformation is 1 in 1000 births but with the increased use of imaging techniques such as CT scans and MR imaging it is suggested that this condition is much more common. However it is very difficult to estimate the exact rate of occurrence as some of cases as asymptomatic or do not develop symptoms till adulthood. Chiari malformations are more common in women than in men and type II Chiari malformation is more prevalent in certain ethnic groups like the people of Celtic descent. In the present case report a new born was presented with myelomeningocele and MR imaging revealed the herniation of cerebellar tonsils and vermis along with Brain Stem through foramen magnum hence confirming it to be a case of type II Chiari malformation.

**Keywords:** Hind brain, Pons, Meningomyelocele, Malformation, Chiari malformation, Arnold-Chiari malformation, Myelomeningocele.

## INTRODUCTION

Chiari malformations include a large spectrum of anomalies of hindbrain formation which appear at different stages of development of the central nervous system. 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 in 1891 and 1896 and gave a detailed description of hindbrain malformations.[1] 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.[2] In his initial description, Chiari classified the hindbrain malformations into type I, II and III and then latter added type IV malformation.[3]

The exact embryological period of the occurrence of Chiari malformation is unclear. The anatomical variations attributing to the Chiari malformation is due to the failure of pontine flexure to form normally from 28-29$^{th}$ day of gestation which leads to elongation of brainstem. Normally growth of the cerebellum during the third month of intra uterine life causes the caudal vermis and choroid plexus to come under the tonsils. However if there is failure of this process, the vermis and choroid plexus remain in extraventricular position. These structural deformities lead to blockage in the flow of cerebrospinal fluid, producing an embryological hydrocephalus. The secondary hydrocephalus causes further herniation of cerebellar tonsils into spinal canal and drags tentorium along with it, thus reducing the dimensions of posterior cranial fossa.[4] A theory was given by Daniel and Strich, which stated the developmental arrest, especially in the progression of pontine flexure during 28$^{th}$ and 29$^{th}$ day of gestation as a cause of Chiari malformation.[5]

The theory of overgrowth suggest that the overgrowth of neural plate before neurulation prevents fusion of neural folds. Barry et al reported two cases of human foetuses of 17 and 18 weeks of development with increased volume of cerebellum and brain stem having Chiari malformation.[6] Contradictory to this it has been observed that cerebellum weighs less in patients with Chiari malformation than in normal people, at all ages.[7,8]

According to hydrodynamic theory, imbalance between pulsating choroid plexus of forth and lateral ventricles result in Chiari malformation.[6] According to Jenning et al, Chiari malformation occurs because the normal zone of fusion at third and fourth somites is displaced caudally below the third to fifth somite pairs thus causing the displacement of the area of formation of cervicomedullary junction.[9]

Chiari malformation is not as rare as would be expected from the small number of reported cases but with the increased use of CT Scans and M.R.I's it is suggested to be much more common. The defect is almost always, but not invariably, associated with meningomyelocele or spina bifida occulta in lumbosacral region. Hydrocephalus is present in most cases. Other associated defects of development include creniolacunia, hydromyelia, sryngomelia, double cord, basillar impression.[2]

## CLASSIFICATION OF CHIARI MALFORMATION

Chiari malformations were described to be of four types:

**Type I**

It is the most commonly observed Chiari malformation. In this type, there is tonsillar herniation through foramen magnum. It is often associated with syringomyelia but not hydrocephalus. This type of Chiari malformation is congenital as well as acquired. Radiologically, Type I is described as tonsillar decent of 5 mm below foramen magnum. Patients with Type I Chiari malformation may be asymptomatic or present with mixture of cerebellar and pyramidal tract signs associated with dysfunctioning of lower cranial nerves.[10]

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**Type II**

It is also called as classic Chiari malformation or Arnold-Chiari malformation. It is less common. In type II Chiari malformation there is caudal descent of cerebellar tonsils and the vermis into the spinal canal along with brain stem and fourth ventricle. Type II Chiari malformation is usually accompanied by myelomeningocele. Hydrocelaphus is seen in 90% of cases. Symptoms arise from dysfunctioning of brain cells and lower cranial nerves. Myelomeningocele results in the partial or complete paralysis of area below the spinal opening. Due to the severity, Type II patients become symptomatic in infancy or early childhood.[10]

**Type III**

It is the most serious form of Chiari malformation. There is occipital or cervical encephalocele alongwith intra cranial abnormalities seen in type II Chiari malformation and a wide foramen magnum. This defect is readily visible and palpable. Plain radiographs help to identify the scull or cranial defects while MR imagining identifies the herniated brain tissue.[10]

**Type IV**

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 includes 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.

## DISCUSSION

Number of studies have been carried out on Chiari malformation. It has been noted that the prevalence of Chiari I malformation is one per thousand in general population. With the improvement in the imaging modalities, the diagnostic abilities have also improved.

According to Stevenson KL, approximately 1/3rd of the patients with Chiari II malformation develop signs and symptoms of brain stem compression.[11] Curnese JT carried out a study on 33 patients with Chiari II malformation and found out that 36% of patients were symptomatic while 64% were asymptomatic.[12] Niels Geerdinll et al carried out MR imaging study on 79 children and concluded that the reliable morphological features leading to diagnosis of Chiari II malformation on MR imaging are downward herniation of the cerebellum, downward displacement of the medulla, pons and fourth ventricle, medullary kinking, abnormally shaped fourth ventricle, hypoplastic tentorium and breaking mesoncephalic tectum.[13] Most of these morphological features are seen on the MR imaging of the present case (figure-1,2,3).

Gammal T et al stated that myelomengocele is present with Chiari II malformation almost in all cases. However, the reverse is not true all the time.[14] According to Rauzzino M et al, Hydrocephalus is seen in 90% of the cases and ventricles are seen asymmetrically.[15] These observations are in correlation with the present case.

Emery JK et al stated that approximately 6% of cases show spinal bifida.[16] Mclendon RE et al in their study found out

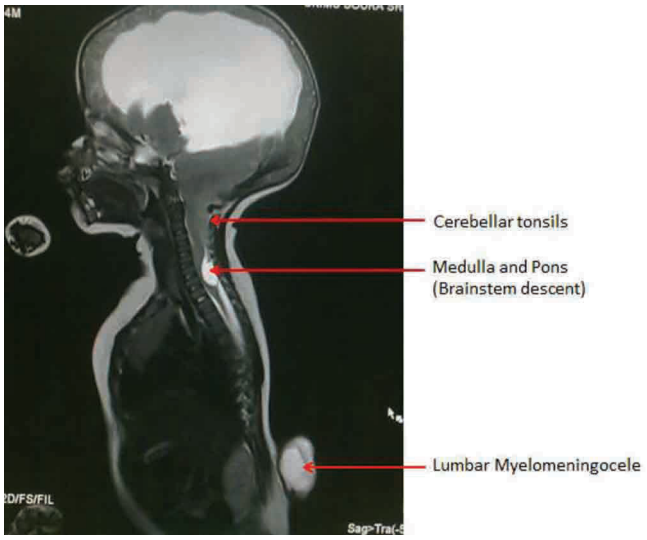


**Figure-1:** Demonstrating Arnold Chiari Malformation with Lumbar Myelomeningocele

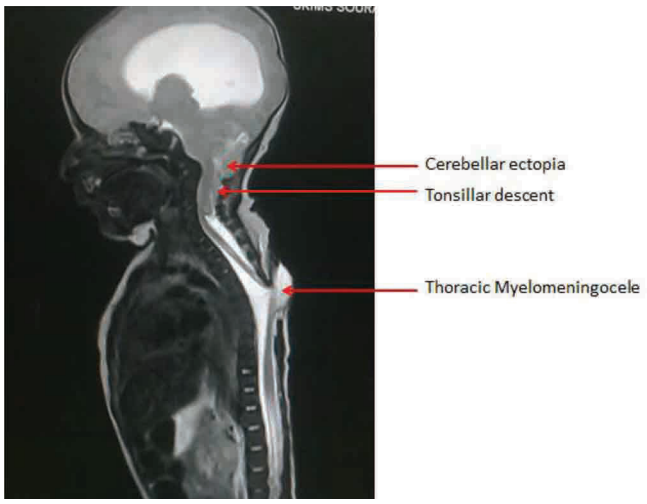


**Figure-2:** Demonstrating Arnold Chiari Malformation with Thoracic Meningomyelocele

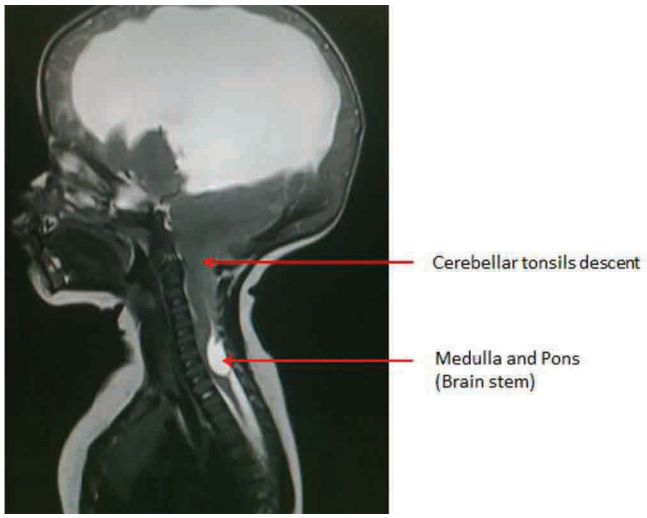


**Figure-3:** Demonstrating Arnold Chiari Malformation with Prominent Tonsillar Descent

that partial or complete agenesis of corpus Callosum with absence of septum pellucidum, polygyria, prominent anterior commissure, obliterated longitudinal fissure between parietal

and occipital lobes and absent cingulate gyrus are frequently seen to be associated with Chiari II malformation.[17] According to Cunnes JT et al in symptomatic children, the typical krinking of medulla is often seen. It was also reported that the upper cervical spine shows Klippel–Fiel anomaly with hypoplastic posterior arch of first cervical vertebra and scalloped dense.[13]

Tsai T et al did a biometric analysis of 25 patients with myelomeningocele and Chiari II malformation and concluded that degree of vermion herniation and cervicomedullary junction herniation are independent variables in Chiari II malformation while the size of posterior cranial fossa is an important factor in explaining the variability of vermian herniation.[18] Wolpert SM et al carried out a study to see the relation between the amount of brain stem herniation and neurological status of the children with Chiari II malformation and found out that the neurological status was not effected by either the amount of herniation of the characteristics of cervicomedullary and hence concluded that the breathing and swallowing difficulties experienced by children with Chiari II malformation is due to other factors like disorganization of brain stem nucli.[19]

## CONCLUSION

To conclude it appears that the phenomenon of cervicomedullary junction is independent although different theories have been suggested resulting in it. The most common embryological cause is found to be due to the failure of pontine flexure to form normally from 28$^{th}$ -29$^{th}$ day of gestation which leads to elongation of brainstem. Recognition of the vermis, medullary kink, cervical cord, C1 arch, fourth ventricle, and myelomeningocele are important radiological features to confirm the diagnosis of Chiari II malformation.

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