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Original Research Article

Arnold Chiari Malformation - A hospital based autopsy study

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Abstract

In Arnold Chiari II malformation the cerebellar tonsils are displaced inferiorly through the Foramen Magnum into the upper cervical spinal canal. It is a complex anomaly with skull, dura, brain, spine and cord manifestations. This study was conducted on 400 fetuses obtained from department of Obstetrics and Gynaecology, Government Medical College and Hospital, Chandigarh. Arnold Chiari malformation was noted in 10 out of 300 (3.33%) fetal autopsy cases. All cases were associated with meningomylocele. In our study we found that 7 cases were showing cerebellar tonsilla rectopia and 3case of type II .Arnold Chiari Malformations can be prevented by preconceptional folic acid and Vitamin B 12 supplementation.
Keywords: Chiari malformation, Folic acid, herniation, meningomylocele.

1. Introduction

Arnold chiarimal formation is a congenital abnormality of CNS characterized by downward displacement of the parts of cerebellum, fourth ventricle, pons and medulla oblongata into the spinal canal.[1] It is named after Hans Chiari and his professor Dr. Arnold. These authors were first to emphasize the importance of this. Classification of Arnold chiari is based on two basis i.e. anatomical and etiological basis.

1.) Anatomical classification:

I - herniation of cerebellum tonsil (most common form)
II - herniation of cerebellum and lower part of brain stem (classic Arnold chiari malformation)
III - herniation of brain stem together with cervical or occipital encephalocele (most severe form)
IV - extreme cerebellar hypoplasia with anomalous shape of the brainstem (rarest form)[2]

2.) Etiological classification:

Primary/congenital

It occurs due to change in the structure of the brain and spinal cord that take place during pregnancy. Primary defect of the paraxial mesoderm leads to underdevelopment of the occipital bone and shallow posterior cranial fossa. This may also due to the genetic problems (unexpected changes in the genes as the fetus develops).

Acquired/secondary

Occurs when large amount of CSF are drained from the spinal area and due to severe injury and exposure to harmful substances or infection.

Tubbs *et al* described two additional type of chiari malformation.

1.) Chiari type 0

Syringohydromyelia with distortion of contents in posterior fossa but without cerebellar tonsillar herniation.

2.) Chiari type 1.5

Caudal migration of brainstem and cerebellar tonsils often associated with syringomyelia.

Arnold chiari malformation is present with an incidence of 0.4:1000 live births and usually associated with hydrocephalus and myelomeningocele [2]. As a causative factor of death, it contributes to 3% of all abortions and 1-2% recurrence risk [1]. Diagnosis of Arnold chiari malformation is made with MRI evaluation of posterior cranial fossa. If scan show that the cerebral tips have

exceeded a distance of 5 mm below the foramen magnum, the diagnosis is confirmed. With routine use of MR imaging, Chiarimal formation is discovered with increasing frequency. Antenatal diagnosis of this abnormality during USG screening for congenital anomalies is an indication for fetal karyotyping followed by counselling, prognostication with/without termination of pregnancy[3].

Bony abnormalities occur during the embryonic development. In Chiari I, posterior fossa may be smaller than the normal. If too small, the effect can be overcrowding of brainstem and cerebellum, as well as herniation of tonsil through the foramen magnum. Scoliosis is a curvature of the spine. There is high rate of scoliosis associated chiari and syringomelia, especially in children.

2. Material and Methods

Current study was conducted on 300 human fetuses in the Department of Anatomy, Government Medical College & Hospital, Chandigarh. Fetuses were sent by the Department of Obstetrics & Gynaecology of the same institute for routine autopsy. Written consent was taken from the parents to perform autopsy and other relevant research work. These fetuses were between 18 - 24 wks. Routine autopsy procedure was followed for each autopsy. Relevant maternal and family histories along with antenatal USG findings were recorded.

During autopsy external findings observed were - sex of the fetus, gestational age of fetus by fetal weight and measurements and presence of other CNS and extra CNS structural anomalies.

Method used

First of all, the fetus brain was injected with formalin solution for fixation for almost 2 weeks. For observing the posterior cranial fossa sagittal section of skull was made with the help of brain knife to open the brain. After that we identified the bony landmarks of foramen magnum i.e. opisthion and basion.

Basion

The midpoint on the anterior margin of the foramen magnum.

Opisthion

The midpoint on the posterior margin of foramen magnum. Put a thread at these points and carefully view any herniation of brain matter below this line and measure it with the help of divider. The depth of the posterior cranial fossa was also measured from internal occipital protuberance to upper margin of foramen magnum (Figure 1).

The most commonly used criteria for diagnosis of chairi I malformation is cerebellar tonsillarectopia of at least 5mm herniation below the level of foramen magnum.

3. Results

Arnold Chiarimal formation was noted in 10 out of 300 (3.33%) fetal autopsy cases. In 7 cases the herniation of the lowest part of the brainstem below the line connecting opisthion and basion measured upto 5 mm (Cerebellar Tonsillar Ectopia) and in 3 case the value was more than 5mm (Type II).

The incidence of Arnold Chiari malformation in our fetal autopsy series was 3.33%. The major type of Arnold Chiari malformation observed was Type I (Figure 2). Associated anomalies were seen in all the cases. Moderate hydrocephalous has been documented in about 20% (n=2/10) cases. Meningomyelocele is seen in all 100% cases. Skeletal anomalies are found in 50% (n=5/10) of cases.

In one case Arnold Chiari Malformation was seen as part of OEIS complex. Male fetus with gestational age 18^{+4} weeks and Posterior cranial fossa measurement 1.1cm shows Spinal defect in the lumbosacral region. Cardio and respiratory system were normal. Omphalocele is present with triplication of large intestine beyond the left 1/3 part of transverse colon. There was an imperforate anus. Urinary bladder was bifurcated and each part receives one ureter. Two penile structures noted with no scrotal sacs (Figure 3). In another case fetus with gestational age of 16^{+6} weeks shows left side renal agenesis. Right renal is also enlarged with tortous ureter. Other related clinical findings are shown in table 2.

Table 1: Showing the frequency of Arnold Chiari Malformation In relation to the gestational age and sex of the fetus

Gestational age	Male	Female	% age
15^{th} -20^{th} wks	7	2	60%
20^{th} -25^{th} wks	1	O	40%
Total	8	2	100%

Table 2: Other related clinical findings associated with Arnold Chiari Malformation

S. No	Associated Anomalies		No
A	Neural Tube Defects (Spina Bifida)	Hydrocephalus	2
		Corpus Callosum Hypoplasia	1
		Thoraco-Lumbar Region	10
B	Musculoskeletal	Club foot	3
		Scoliosis	1
		Hyperextended joints	1
C	Gastrointestinal Anomaly	Omphalocele	1
D	Urinary System	Agenesis of Kidney	1
		Bladder Malformation	1
E	Genitalia	Double Penis	1
		Anal Atresia	1

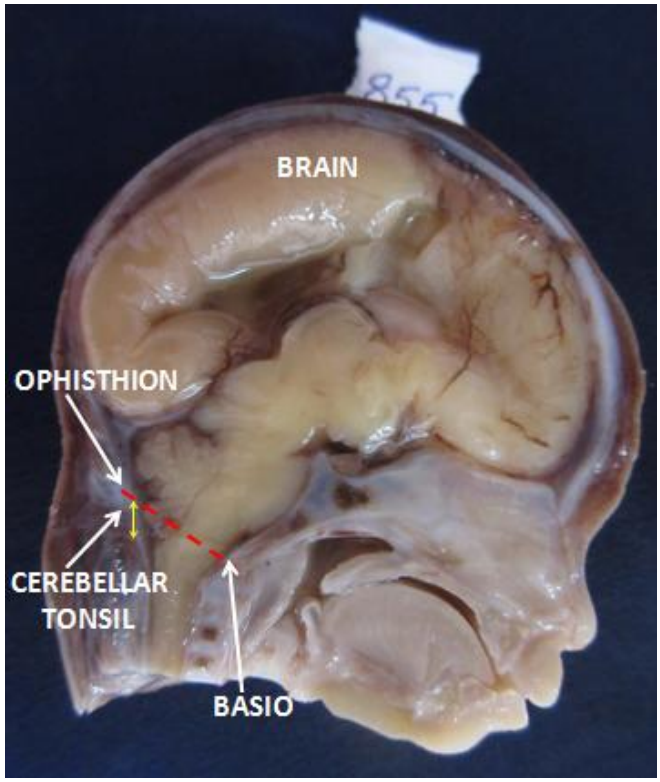

Figure 1: Showing measurement of herniation of brain matter below the line joining basion and Opisthion

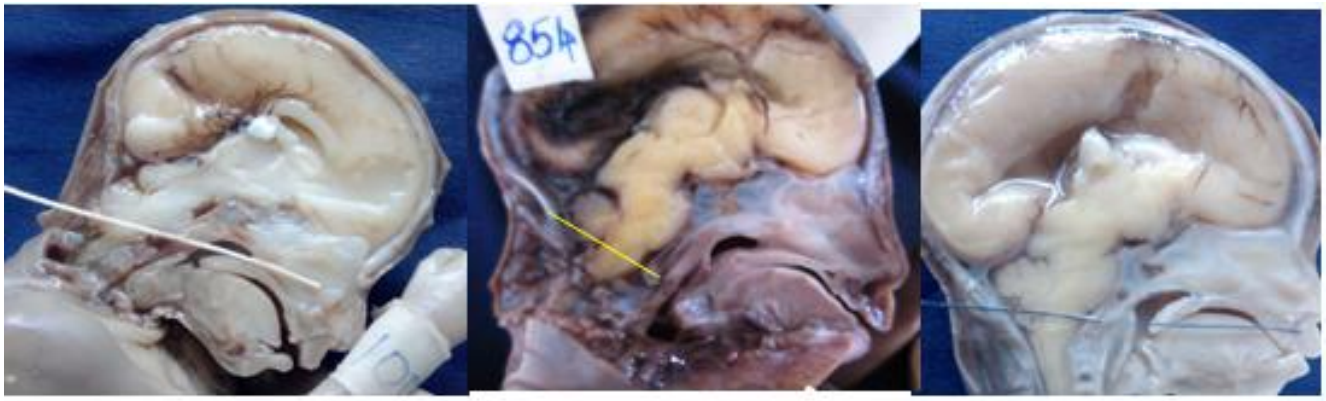

Figure 2: Showing Herniation of cerebellar tonsil

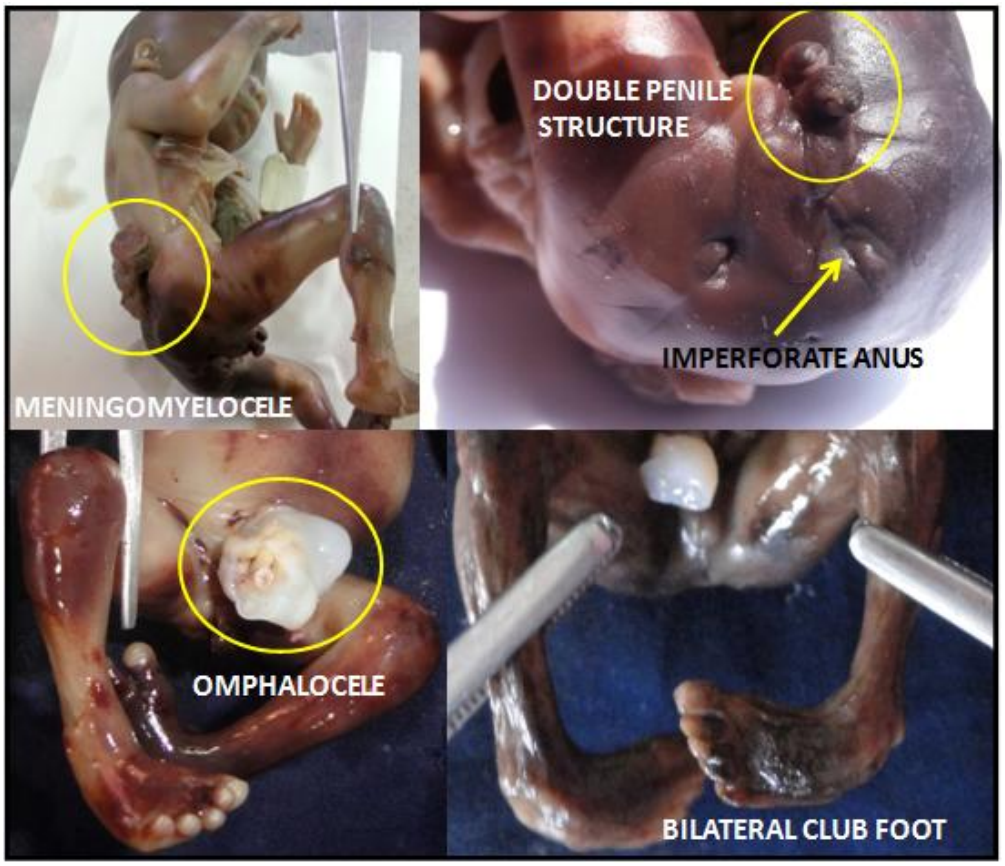

Figure 3: Showing Arnold Chiari Malformation as part of OEIS complex

4. Discussion

Chronic cerebellar tonsillar herniation (CTH) occurring in CM primarily results from a paraxial mesodermal defect .which leads to underdevelopment of the occipital bone and overcrowding of developing hindbrain within a primary, small, and shallow posterior cranial fossa (PCF).[4-6]

For Chiari I, prevalence rates of 0.1-0.5% with a slight female predominance are suggested by recent studies. We also found 8 cases of chiari I with male risk [5]. It is hypothesized that Chiari type I originates as a disorder of paraaxial mesoderm, which subsequently results in formation of a small posterior fossa. The development of the cerebellum within this small compartment results in overcrowding of the posterior fossa, herniation of the cerebellar tonsils, and impaction of the foramen magnum.

Theories regarding embryogenesis of Chiari II malformation must taken into account its invariable association with myelomeningocele. An attractive theory is the "CSF loss" theory. It is hypothesized that escape of fluid through the open placodein myelomeningocele results in an inadequate stimulus for mesenchymal condensation at the skull base. The disordered and inadequate growth of the posterior fossa results in upward herniation of vermis, downward herniation of brainstem and distortion of tectum (tectalbeaking). Furthermore, collapse of the developing ventricular system because of fluid loss results in associated abnormalities such as agenesis of corpus callosum and enlargement of massainter media [6].

The fact should also be taken in account that the "normal' position of cerebellar tonsils varies with age. In neonates the tonsils are located just below the foramen magnum and descend further during childhood, reaching their lowest point somewhere between 5-15 years of age. As the age advances the tonsils ascend and come to rest at the level of foramen magnum [7].

Kruyff, states high frequency of small posterior cranial fossa by radiographic studies [8]. We also observed that the normal depth of posterior cranial fossa in normal fetuses was 2.5cm and this depth get decreased in ACM cases (range=1-2cm).

Je. G. Chi and WeonSeo Bark reported 6 cases of ACM. In one of their case there was anal atresia as well as agenesis of kidneys, urinary bladder, rectum, urethra and prostate, Hypoplastic lungs and hiatal hernia along with ACM [9]. In the present study we also observed a case of ACM with associated omphalocele, anal imperforation, bifid urinary bladder and double penile structure. One another case of AC was also noted with agenesis of left kidney and tortous ureter and enlarged kidney on the right side. Beside these skeletal anomalies such as

kyphoscoliosis, club foot also noted by Je. G and Weonseo Bark [9]. Present study also came across three cases with club foot and single case of scoliosis.

5. Conclusion

In our study we found 10 cases were of type 2 ACM. Knowledge of the anatomy of posterior cranial fossa is essential for proper surgical management of Arnold Chiari malformation. Arnold Chiari Malformations can be prevented by preconceptional folic acid and Vitamin B 12 supplementation. The rostral and caudal neural pores close at 4th week of gestation. Rostral at 24th day and Caudal at 28th day of gestation. A delayed folic acid supplementation is bound to miss the vital period of organogenesis and neural tube closure. The cranial findings associated with the Chiari II malformation are found exclusively in fetuses with myelomeningocele. The diagnosis of myelomeningocele in a fetus is important for many reasons. It provides the parents with an opportunity to consider pregnancy termination.

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