

Case Series

ARNOLD CHIARI MALFORMATION

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ABSTRACT

Chiari malformations (CM) are named for Hans Chiari, an Austrian pathologist, who first identified type I-III in 1891. This study was conducted on 400 fetuses obtained from department of Obstetrics and Gynecology, Government Medical College and Hospital, Chandigarh. 4 cases were diagnosed with Chiari malformation. All cases were associated with meningocele. In our study we found that 3 cases were of type 2 ACM (ELSTER AND CHEN) which included herniation along with meningocele and 1 case of type 3 (Small brain, parietal and occipital bones are not properly formed, cerebellum seen protruding through foramen magnum, leading to herniation). Arnold Chiari Malformations can be prevented by preconceptional folic acid and Vitamin B 12 supplementation

KEY WORDS: Chiari malformations, Meningocele, Herniation, Preconception supplementation.

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INTRODUCTION

Arnold-Chiari malformation (ACM) is a group of complex brain abnormalities that affect the area in lower posterior skull where the brain and spinal cord meet. It occurs in about 0.4:1000 live births and has formed 3% of all abortions [1]. Chiari malformations (CM) are named for Hans Chiari, an Austrian pathologist, who first identified type I-III in 1891. Ulius Arnold further expanded the definition of Chiari malformation type II and some medical sources began using the name Arnold-Chiari malformation. Arnold chiari malformation (ACM) are of four types [2-4].

Diagnosis of Arnold chiari malformation is made with U/S or 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 [5].

Child born with ACM requires frequent examinations and diagnostic testing to monitor the development of head as the child grows.

MATERIALS AND METHODS

This study was conducted on 400 fetuses obtained from department of Obstetrics and Gynecology, Government Medical College and Hospital, Chandigarh, India. Out of 400 fetuses 5 were diagnosed with Arnold Chiari syndrome. Autopsy was done on all fetuses. 1 fetus with normal anatomy of head and neck and CNS was also dissected for comparison. These fetuses were between 18 - 24 wks. Out of 5 fetuses one fetus of 16 wks age (5th fetus) could not be dissected because of advanced stage of maceration. The indication of termination of pregnancy in cases 1,2 and 3 was diagnosis of open neural tube defect and Arnold Chiari malformation on U/S. In case 4 and 5 U/S

diagnosed defect before spontaneous expulsion of fetus took place. Relevant history from the parents was taken. The details of which are given in Table 1. The fetuses were examined externally and were radio graphed. Routine autopsy procedure was followed for each autopsy. For observing the posterior cranial

fossa sagittal section of the head in each case was done. The depth of the posterior cranial fossa was measured from internal occipital protuberance to upper margin of foramen magnum. The findings were noted and described in the light of the available literature.

Table 1: Case histories.

	Case I	Case II	Case III	Case IV	Case V
Maternal history	21 Years, PGR	25 Years, G ₃ P ₂ L ₂	26 years, PGR	22 Years, PGR	21 yrs, PGR
Gestational age, Sex of the baby	24 wks , male baby	18 + 4 wks Male baby	21+2 wks, female baby	21+3 wks, Male baby	16 +5 wks, Male baby
History of previous pregnancies	NA	Normal, Elder male baby , 3 Years, G2 10 mths female baby	NA	NA	NA
Past history	Not contributory	Not contributory	Not contributory	Not contributory	Not contributory
Family history	Not contributory	Not contributory	Not contributory	Not contributory	Not contributory
Antenatal history of present pregnancy	Not contributory	Not contributory	Not contributory	Not contributory	Not contributory
Investigations	U/S shows Arnold Chiari malformation type II with meningocele, kyphosis Normal karyotype	US done at17 wks show small posterior cranial fossa, banana sign + Hydrocephalus., meningocele at cervicodorsal region, thoracolumbar spine dysplastic showing severe, kyphoscoliosis, mild polyhydroamnios	Rh-ve, U/S Finding at 19 wks shows Arnold Chiari malformation, spina bifida, myelomeningocele	US/ showed Arnold Chiari malformation, Small thorax, kidneys absent bilaterally	U/S AT 16 wks showed echogenic mass
Termination of pregnancy	MTP	MTP	MTP	Spontaneous expulsion	Spontaneous expulsion

OBSERVATIONS

Total number of cases 4 out 400(1%)

Table 2: Showing the findings on External examination.

Feature	Case I	Case II	Case III	Case IV	Case V
Head, curvature of spine	Head normal, kyphosis, Meninomyelocele in thoracolumbar region	Anterior posterior flattening of head due to soft posterior cranium, (meningoencephalocele) wide nasal bridge, Mild kyphoscoliosis	Head normal, Meninomyelocele in lumbosacral region (4 x 3 cm)	Normal , Small thorax Meningocele in thoracolumbar region	Side to side Flattening of the head, Open neural tube defect in lumbar region
Limbs	Normal	Normal	Normal	Right club foot	Normal
Thorax	Normal	Normal	Normal	Normal	Normal
Abdomen	Normal	Normal	Normal	Normal	Normal

Table 3: Showing the findings on Internal examination.

Feature	Case I	Case II	Case III	CASE IV
Skeletal system	Posterior cranial fossa 2 cm 9 (fig 1)	Posterior cranial fossa 1.5 cm (fig 2)	Posterior cranial fossa 1.1 cm (fig3)	Right club foot Posterior cranial fossa small (1.2 cms) Small thoracic cavity (fig 4)
CNS	Small brain, meningocele (thoracolumbar region)	Small brain, parietal and occipital bones are not properly formed, cerebellum protruding foramen magnum, leading to herniation	Myelocele in thoracolumbar region, spinal cord degenerated at thoracolumbar region	Herniation of the cerebellum in to foramen magnum Open neural tube defect in thoracolumbar region
GIT	Normal	Normal	Normal	Normal
CVS	Normal	Normal	Normal	Normal
Respiratory system				Normal
Urinary system and supra renals	Normal	Normal	Normal	Kidney absent bilaterally, Small mass of tissue was seen open in into bladder on right side, Right adrenal present, left adrenal absent (figure-5)
Genital system	Normal	Normal	normal	Normal

Fig. 1: Posterior cranial fossa 2 cm. **Fig. 2:** Posterior cranial fossa 1.5cm. **Fig. 3:** Posterior cranial fossa 1.1cm.

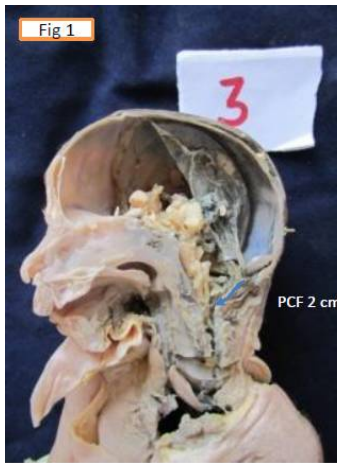


Fig. 4: Right club foot Posterior cranial fossa small (1.2 cms) Small thoracic cavity.

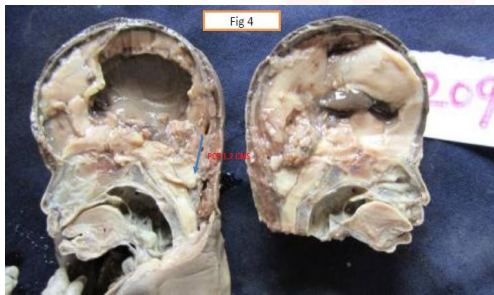


Fig. 5: Showing Bilateral absence of Kidney.



DISCUSSION

Chiari malformations are abnormalities of the posterior fossa that affect the cerebellum, brainstem, and the spinal cord with prevalence rates of 0.1%–0.5% [6,7]. In our study we came across 4 fetus with ACM out of 400 cases, which accounts for about 1% of cases. Advanced studies on ACM had reported that 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) [8-10]. It should also be noted 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 [11].

A small posterior fossa is frequently demonstrated by necropsy and radiographic studies [12].

In our study we measured the depth of posterior cranial fossa from internal occipital protuberance to upper margin of foramen magnum. We found that the normal depth of posterior cranial fossa in normal fetuses (gestational age of 21 wks) was 2.5 cm, that of fetuses of same gestational period with ACM was ranging from 1.1- 2 cm. So we noted that there was a decrease in the depth of PCF in fetuses with ACM making the posterior cranial fossa of affected fetus small than the normal fetus.

ACM can be classified into 4 types

Type 1- Refers to herniation of cerebellar tonsils alone, radiologically as simple tonsillar herniation 5mm or greater, below the foramen magnum

Type 2- Herniation of both cerebellum and lower brain stem with spina bifida

Type 3- Rare type of brain herniation in association with cervical or occipital encephalocoele

Type 4- Extreme cerebellar hypoplasia and caudal displacement of posterior cranial fossa contents [13].

Tubbs et al described two additional type of chiari malformation

Chiari type 0- Syringohydromyelia with distortion of contents in posterior fossa but without cerebellar tonsillar herniation

Chiari type 1.5- Caudal migration of brainstem and cerebellar tonsils often associated with syringomyelia [14].

Table 4: Classification of Arnold Chiari malformation.

Type	Signs	Symptoms
Arnold Chiari type I (most common form)	Herniation of cerebellar tonsil, flocculi and medial aspect of the caudal medulla in the upper cervical canal. Presents with headache and cerebellar symptoms May not appear until adulthood	Pain in the occipital or cervical region and arms. Double vision, Blurred vision, oroscillopsia. Seizures and developmental delay.
Arnold Chiari type II	Same as type I but caudal portion of the 4 th ventricle positioned in the upper cervical canal. Towering cerebellum, tectal beaking, hydrocephalus, hypo/ agenesis of corpus callosum, lükenschadel skull, colpocephaly Accompanied by lumbar myelomeningocele May not present until early adult hood	Same as type I but more severe
Arnold Chiari type III	Small cerebellum from physical appearance, lower brainstem & 4 th ventricle pushed in to cervical canal, encephalocele Associated with syrinx present at birth	Rare severe neurological defects
Arnold Chiari type IV	Underdeveloped cerebellum and brainstem located within the posterior cranial fossa. Present at birth	rare

In our study we found that 3 cases were of type 2 ACM(ELSTER AND CHEN) which included herniation along with meningomyelocele and 1 case of type 3 (Small brain, parietal and occipital bones are not properly formed, cerebellum seen protruding through foramen magnum, leading to herniation).

Je. G. Chi and Weon Seo 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 ,which was somewhat equaling with our findings. In case 4, we found ACM with bilaterally absent Kidney, and adrenal gland. Apart from this they also noted skeletal anomalies such as kyphoscoliosis,club foot, pes equinovarus along with ACM .We ,in our study also came across ACM with kyphosis in case 1, and club foot in case 4 [15].

Arnold-Chiari malformation usually is seen as

an associated with spina bifida, while most authors focused on the relationship of meningomyelocele with ACM. One theory proposed in this association suggest that the spinal cord fixed at the meningomyelocele creates downward traction on the hindbrain as the fetus grows. This hypothesis has been disproven experimentally [16]. It has been shown that traction is dissipated within four segments of the nonmobile defect. In addition, this theory cannot explain the Arnold-Chiari malformation without spina bifida.

Cameron (1957) proposed a myelomeningocele hydrodynamic theory. He proposed that all the abnormalities found in the Arnold-Chiari malformation are the consequence of an escape of cerebrospinal fluid from the spina bifida into the amniotic cavity during the first half of intrauterine life. So an increase in intraamniotic pressure would be transmitted to the fetal skull, and this downward pressure from the skull was thought to force the hindbrain into the upper cervical cord. However, the hydrodynamic theory fails to explain the Arnold-Chiari malformation cases without hydrocephalus and those without spina bifida [17].

Another study proposed that the development of the cerebral malformation precedes the development of the skull in fetal life and the abnormalities of the skull and dura are the consequence of the hindbrain abnormality. He proposed that the primary disturbance is a failure of formation of the pontine flexure. Lack of flexion would cause the hindbrain elongation typical of this anomaly [18].

Skull malformations in ACM may include calvarial defects (lacunar skull), small posterior fossa with low lying transverse sinuses, wide foramen magnum. Brain anomalies include inferiorly displaced vermis, medullary kink and spur. Corpus callosal hypoplasia or complete agenesis may also be present. Myelomeningocele is present in all cases. We speculate that hind brain herniation is secondary to meningomyelocele. This results as a consequence of failed posterior neural pole closure. The posterior neural pole usually closes at 6 wk of menstrual age [19]. In our study also almost all cases of ACM showed meningomyelocele. From an embryological perspective, the foramen magnum consists of cells

of 2 different origins. One is the chondrocranium that constitutes the bones at the base of the cranium by endochondral ossification and the other constitutes the parachordal cartilage around the cranial end of the notochord. They fuse into the cartilaginous mass that is derived from the sclerotomal regions of the occipital somites. These cartilaginous masses grow extensively around the cranial end of the spinal cord and contribute to the base of the occipital bone. Finally, these masses form the boundaries of the foramen magnum. After birth, growth of the cranial base continues at the sphenooccipital synchondrosis until adolescence [20]. Studies has also suggested that premature stenosis of the sphenooccipital synchondrosis causes premature closure of the sphenooccipital synchondrosis, similar to craniosynostosis of the calvaria. The shape of the posterior cranial fossa eventually changes into a narrow funnel shape, reflecting the slab-sided and narrow posterior cranial fossa, such as inverted oxycephaly of coronal suture craniosynostosis [21]. Routine anatomic ultrasound performed in the second trimester has a detection rate of approximately 70-90% for fetal congenital abnormalities [22]. The central nervous system abnormalities are one of the most common ones detected. Chiari malformation is among the CNS abnormalities diagnosed in the fetal period [23]. The obvious sonographic findings are the frontal bone scalloping (lemon sign) and absent cerebellum or abnormal anterior curvature of the cerebellar hemispheres (banana sign) [24]. However, the lemon sign is frequently not present in later pregnancies and can be seen in healthy fetuses and in other conditions [25].

In view of the severe morbidity associated with these malformations, a daily intake of 400µgms of folic acid is recommended in the preconception period. The Methylene tetrahydrofolate dehydrogenase gene (MTHFD) is one of the key genes involved in folate pathway. A large metanalysis study strongly suggests that the MTHFD 1 G1958A gene polymorphism is strongly related to neural tube defects [26]. It is believed that 1; 00,000 years ago our ancestors ate 14-16 servings of fresh fruits and vegetables (major source of folic acid) each day [27].

The main treatment of ACM is to enlarge the

foramen magnum and expand the dura by surgical approaches, such as a simple suboccipital craniectomy, craniectomy with C1, C2 laminectomy, duroplasty [28].

Open neural tube defects, can be repaired in utero, as documented recently in the Ochsner experience [29].

CONCLUSION

In our study we found that 3 cases were of type 2 ACM (ELSTER AND CHEN) and 1 case of type 3. Incidence was 1%. Arnold Chiari Malformations can be prevented by preconceptional folic acid and Vitamin B 12 supplementation. In India, most of the pregnancies are unplanned. The rostral and caudal neural pores close at 6 wk 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. Therefore, identification of features of the Chiari II malformation virtually ensures that myelomeningocele is present. Probably, among these supratentorial findings are the so called lemon sign (inward scalloping of the frontal bones) and ventriculomegaly.

The diagnosis of myelomeningocele in a fetus is important for many reasons. It provides the parents with an opportunity to consider pregnancy termination. Among parents electing to continue the pregnancy, adequate counseling and psychological preparation can be provided.

Conflicts of Interests: None

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