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Original Research Article

STUDY OF CHIARI MALFORMATIONS IN A TERTIARY CARE HOSPITAL IN NORTH-EAST INDIA

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ABSTRACT

Background: Chiari malformations are herniation of part of cerebellum into the foramen magnum. Professor Hans Chiari classified chiari malformations into four types: type I to type IV. Type I chiari malformation is the most extensively studied entity. Type III and IV are very rare variety.

Aim: Magnetic Resonance Imaging (MRI) study of the different types of chiari malformations in a tertiary care hospital in North-East India.

Materials and Methods: After performing MRI of brain and spinal cord, 63 cases of chiari malformations were included in the study. The cases were classified according to the classification of Hans Chiari.

Results: 73.02% cases were type I followed by 22.22% cases of type II chiari malformations. Male predominated female and more than half of the patients were under twenty years of age. Hydrocephalus and syringomyelia were mostly associated with type I chiari malformations.

Conclusion: various aspects of such a rare type of congenital anomaly were studied in this part of the country.

KEY WORDS: Chiari malformations, MRI, Syringomyelia, Hydrocephalus.

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INTRODUCTION

Chiari malformations (CM) are a congenital heterogeneous group of disorders characterized by anatomic anomalies of the cerebellum, brain stem, and cranio-cervical junction associated with downward displacement of the cerebellum, alone or with lower medulla, into the cervical spine canal [1].

Toward the end of the nineteenth century, Professor Hans Chiari developed a classification system that is still used today [2]. He described four types of patients based on postmortem dissection studies. Type I Chiari malformations [2] are patients without neural tube defects who have the cerebellar tonsils move through the foramen magnum. Radiologically, significant

caudal displacement is considered to be more than 5 mm. The brain stem remains within the posterior fossa and there may or may not be the development of a syrinx. Type II Chiari malformations [2] are seen exclusively in patients who have a neural tube defect and have caudal displacement of the cerebellar vermis and lower brain stem. This movement is thought to occur in utero prior to the full development of the cerebellar tonsils. Type III Chiari malformations [2] are a rare and extreme form of hindbrain hernia where a portion of the cerebellum and brain stem migrate out of the craniocervical junction through a defect in the dura, skull, and soft tissue layers to present as a mass or sac on the back of the neck. Type IV Chiari malformations [2] were originally thought to be part of the hindbrain spectrum where the cerebellum was hypoplastic or absent. This group of patients is also uncommon, usually has no therapeutic options available, and is not appropriate to be considered with the hindbrains hernias [2].

Chiari type I malformation is the most extensively studied entity and its prevalence rate ranges from 0.1% to 0.5% according to epidemiological studies, with female preponderance [3].

The basic presentations of symptoms include occipital headaches, balance and coordination issues, and other associated cerebellar signs. This disease process affects both adult and pediatric patient populations, but symptoms vary by age. The presenting symptoms can be quite variable as well [4].

Chiari malformation is the commonest anomaly of the craniovertebral junction involving both the skeletal as well as the neural structures. This entity has rapidly evolved over the past decade with newer visualization techniques, thus posing new challenges to diagnosis and management [5].

MATERIALS AND METHODS

The study is a retrospective one based on cases of Chiari malformations (CM) attending the Department of Radiology, Gauhati Medical College and Hospital, Guwahati, from June 2010 to June 2015, for MRI (Magnetic Resonance Imaging) with common symptoms of headache, swallowing problems, dizziness, nystagmus, etc. In all cases MRI of the brain and spinal cord was done.

Altogether, 63 cases of Chiari malformations were included in the study. The cases were classified, according to the classification developed by Hans Chiari, into four types: type I to type IV [3]. Data obtained from the MRI studies of 63 cases were tabulated, compared and calculated. The study was approved by the Institutional Ethics Committee of Medical College and hospital.

RESULTS

Total 63 cases of Chiari Malformations were included in the study.

Table 1: Number and percentage of different types of Chiari Malformation cases.

Types of Chiari Malformations	Number of cases of Chiari Malformations	Percentage of cases of Chiari Malformations
Type I	46	73.02%
Type II	14	22.22%
Type III	2	3.17%
Type IV	1	1.59%
Total	**63**	**100%**

The type IV Chiari Malformation case was an incidental finding as the patient came for MRI with histopathologically proven adenoid cystic carcinoma of right parotid gland. There was cerebellar agenesis including vermis, hypoplastic cerebellar peduncles and hypoplasia of brain stem predominantly the pons.

Table 2: Different types of Chiari Malformation cases according to religion.

Religion of patients with Chiari Malformations	Number of Chiari Malformations Patients							
	Type I	% (out of 46)	Type II	% (out of 14)	Type III	% (out of 2)	Type IV	% (out of 1)
Hindu	37	80.43%	6	42.86%	0	0	1	100%
Muslim	9	19.57%	7	50.00%	2	100%	0	0
others	0	0	1	7.14%	0	0	0	0
Total	**46**	**100%**	**14**	**100%**	**2**	**100%**	**1**	**`100%**

Majority of patients were Type I CM (73.02%) followed by Type II (22.22%), type III CM (3.17%) and type IV (1.59%) (Table 1) (figure 1B, 2A, 3). Out of 73.02%type I patients, 80.43% were Hindu by religion whereas all Type III and half of type II CM patients were Muslim by religion Table 2).

55.56% CM patients were male and rests were female. Thus, there was male predominance with ratio of male: female is 1.25.

In contrast to the male predominance (64.29%) over female (35.71%) in type II variety, only about 4% gender differentiation was observed in type I CM (male 52.17%, female 47.83%).

Highest numbers of CM patients (25.40%) were below 5 years of age followed by patients in the age group between 5 years to less than 10 years (14.29%). Among all CM patients, lowest recorded age was 3 days and highest was 50 years.

Table 3: Number of different Chiari Malformations cases with different age groups.

Different Age Groups	Number of Chiari Malformations Patients			
	Type I	Type II	Type III	Type IV
0 to <5 yrs	2	12	2	0
5 yrs to <10 yrs	8	1	0	0
10 yrs to <15 yrs	7	0	0	0
15 yrs to <20 yrs	8	0	0	0
20 yrs to <25 yrs	6	0	0	0
25 yrs to <30 yrs	6	0	0	0
30 yrs to <35 yrs	4	0	0	0
35 yrs to <40 yrs	2	0	0	0
40 yrs to <45 yrs	1	0	0	1
45 yrs to <50 yrs	1	1	0	0
50 yrs to <55 yrs	1	0	0	0
55 yrs to <60 yrs	0	0	0	0
60 yrs to <70 yrs	0	0	0	0
Total	**46**	**14**	**2**	**1**
Mean	**3.54**	**1.08**	**0.15**	**0.077**
Standard deviation	**±3.072**	**±3.303**	**±0.555**	**±0.277**
Standard Error of mean	**±0.852**	**±0.916**	**±0.154**	**±0.077**

Type I CM was common in the age groups between 5 to 20 years and type II was mostly found under 5 years of age (Table 3).

Level of significance of age distribution between type I and type III chiari malformations is extremely significant with p value 0.0007 (<0.05). Also, p value for age distribution between type I and type IV chiari malformations is 0.0005 (<0.05) which is extremely significant.

Table 4: Number of different Chiari Malformations cases with associated anomalies.

Associated anomalies in Chiari Malformations	Number of Chiari Malformations			
	Type I	Type II	Type III	Type IV
Hydrocephalus	12	4	1	0
Syringomyelia	24	5	1	0
Corpus callosum agenesis	0	11	1	0
Occipitalisation of atlas vertebra (atlanto–occipital assimilation)	3	0	0	0
Ectopic Kidney	0	1	0	0
Total	**39**	**21**	**3**	**0**

Type I CM patients were mostly associated with hydrocephalus and syringomyelia (Figure no.1A). Out of 14 type II CM patients, 11 were associated with corpus callosum agenesis (table no.4). Atlanto-occipital assimilation was found only with type I CM. One case of ectopic right kidney located in presacral region was found with type II CM patient (Table 4).

Table 5: number of different Chiari Malformations cases with other findings.

Associated findings in Chiari Malformations	Number of Chiari Malformations Patients			
	Type I	Type II	Type III	Type IV
Small Posterior cranial fossa	3	13	1	0
Tectal beaking	0	10	1	0
Dilatation of ventricular system (Ventriculomegaly)	7	8	2	0
Basilar invagination	9	0	0	0
Lobulated Parotid gland	0	0	0	1
Total	**29**	**32**	**4**	**1**

Basilar invagination was found only in type I CM (figure no.4). In type II CM, small posterior cranial fossa was a common finding with tectal beaking (table no.5) (Figure 2A, 2C).

53.33% (16 out of 30) CM cases with syringomyelia were in the age groups between 15 to less than 30 years. Again, 52.94% (9 out of 17) CM cases with hydrocephalus were found under 15 years of age and 41.18% (7 out of 17) CM cases with ventriculomegally were under 5 years of age.

83.33% (10 out of 12) CM cases with corpus callosum agenesis and 70.59% (12 out of 17) CM cases with small posterior cranial fossa were observed under 5 years of age. On the other hand, 77.78% (7 out of 9) CM cases with basilar invagination were found between 10 years to less than 25 years.

Table 6: Distribution of length of herniated cerebellar tonsil in type I Chiari Malformations.

Range of length of herniated cerebellar tonsil	Number of type I Chiari Malformations patients with herniated cerebellar tonsil
5 to <10 mm	16
10 to <15mm	18
15 to <20 mm	8
20 to <25 mm	2
25 to <30 mm	2
30 to <35 mm	0
Total	**46**

Length of herniated cerebeller tonsil in 73.91% (34 out of 46 cases) cases of type 1 CM were in between five mm to less than 15 mm (table no.6). The smallest length recorded of herniated cerebeller tonsil in type I CM was 5.2 mm and longest being 28 mm.

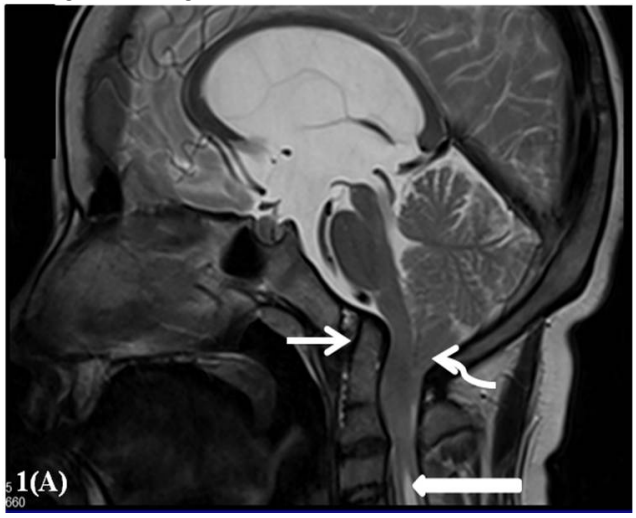


Fig. 1(A): Sagittal T2 weighted MR image of patient with type I chiari malformations showing tonsillar ectopia with pointed appearnce (curved arrow) with normal tectum & 4th ventricle. The dens is retroflexed (straight thin arrow). There is syringomyelia in upper cervical canal starting from the opposite of C3 vertebra (thick arrow).

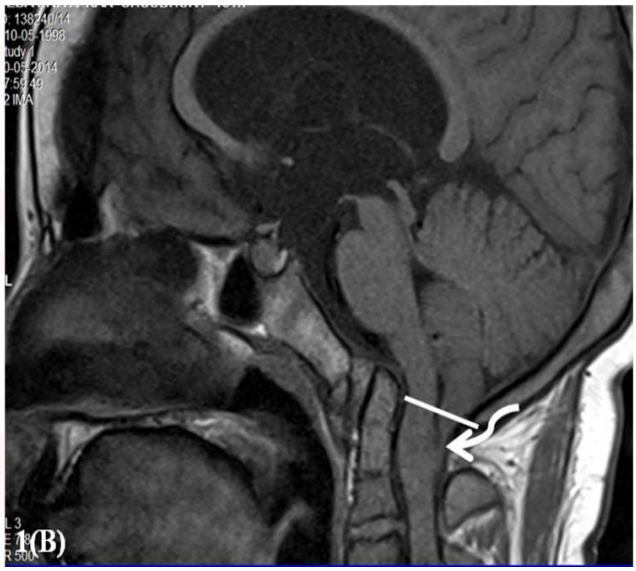
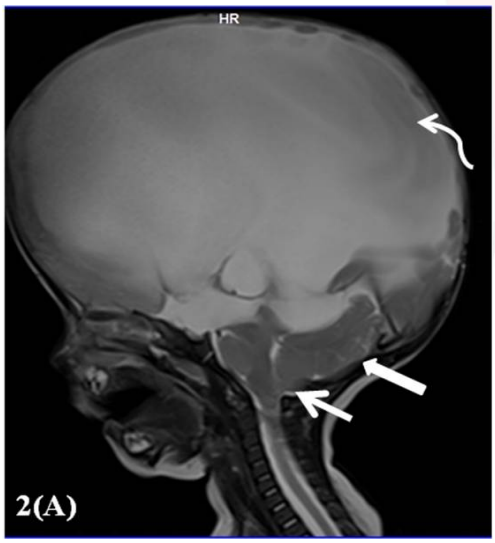


Fig. 1(B): Sagittal T1 MR image of the same type I chiari malformation patient clearly showing tosillar herniation (Curved arrow) below the Basion-Opisthion line (white straight arrow).

Fig. 2: **(A)** Sagittal T2 weighted scan of 3 months old female patient with type II chiari malformation showing small posterior fossa (thick arrow) with gross supratentorial hydrocephalus (curved arrow). There is herniation of medulla and cerebellar vermis (thin arrow). **(B)** Sagittal T2 weighted scan of the same patient of type II chiari malformations showing tethered cord (thin arrow) with syringomyelia in the lower cord (thick arrow) with lumbar myelomeningocele (curved arrow).



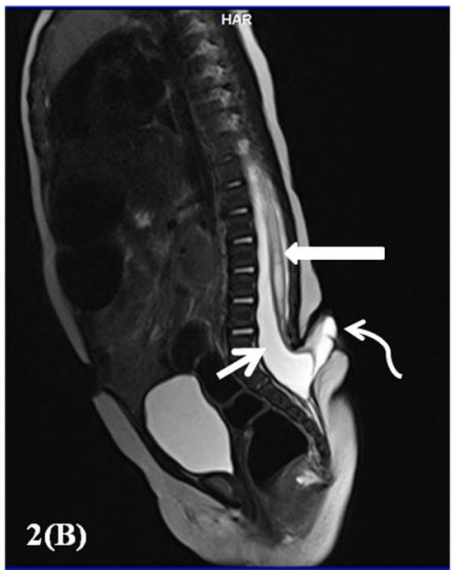


Fig. 2(C): Axial T2 MR image in patient with type II Chiari malformation showing dilated occipital horn of lateral ventricle (thick arrow) with beaked tectum (thin arrow).

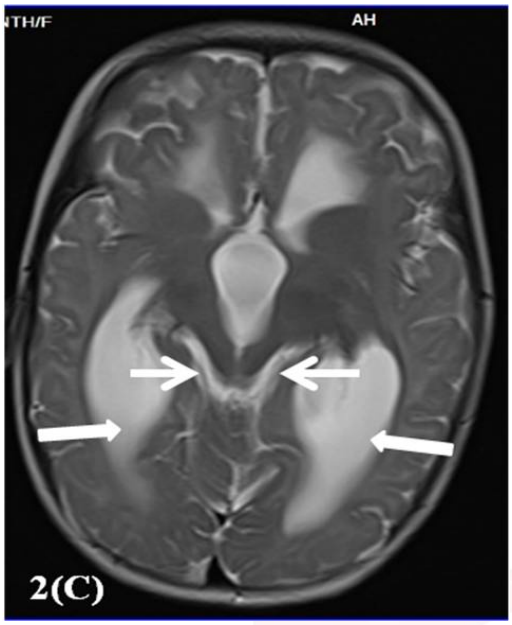
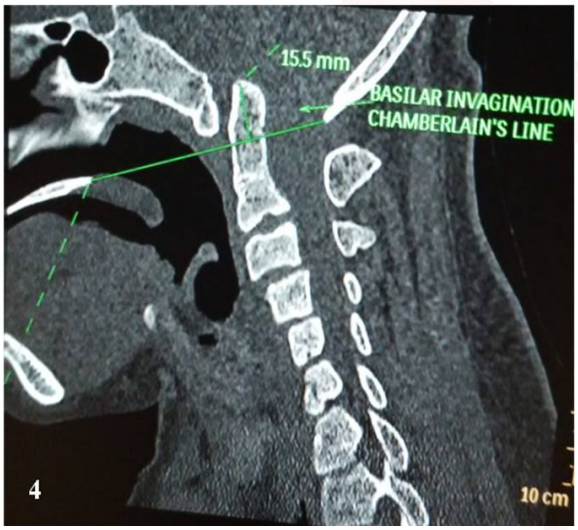


Fig. 4: Sagittal CT scan image of type I chiari malformation patient showing that the tip of dens is 15.5mm above the Chamberlain's line suggestive of basilar invagination (Chamberlain's line or the Palato-occipital line joins the posterior pole of hard palate to opisthion. Basilar invagination was diagnosed when the tip of the odontoid process was at least 2 mm above Chamberlain's line [24]).

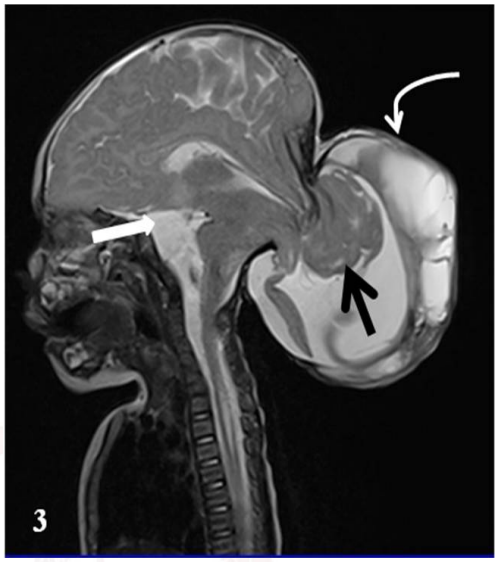


DISCUSSION

The term Chiari malformation, as currently defined, embraces a heterogeneous group of disorders with different pathogenetic origins [6]. It is generally congenital but, rarely can be of acquired origin [7].

Syringomyelia is the term given to define a fluid filled cavity laying within the spinal cord, most commonly occupying space between the

Fig. 3: Sagittal T2 weighted image of a 3 month old male patient with type III chiari malformation showing occipital cephalocele (curved arrow) that contains herniated dysplastic appearing brain (black thin arrow). The 4th ventricle (white thick arrow) is enlarged, elongated and tugged downwards towards the cephalocele.



cervicothoracic vertebral levels [8] (Figure 1A, 2B) . It is commonly associated with a Chiari I malformation and may be an incidental finding [9].

In the present study 73.02% cases were type I CM, followed by 22.22% of type II CM cases (table no.1) (figure no.1B, 2A). In Choudhury PR et al. [10] study, 82.22% were type I and 13.33% were type II CM cases. Moreover, in the present study, 55.56% CM patients were male which was almost similar to 53.33% male CM patients observed in Choudhury PR et al. [10] study. According to Fernandez AA et al. [9], chiari I and II malformation tend to be more frequent in women but male predominance in both type I and II CM was found in the present study.

In Choudhury PR et al. [10] study maximum numbers of cases of type I CM were in the age group between 15 years to less than 20 years and type II CM cases were under 5 years of age. In the present study, most of type I CM cases were in the age groups between 5 to 20 years and type II CM patients were in age group between 0 to less than 5 years (table no.3). The usual age of onset of Type I CM ranged from the second to the fourth decade as mentioned by Verma R et al [3].

Lee S et al. [11] observed that ventriculomegaly was common (but no syrinx or scoliosis) was

found in less than 3 years of age of type I CM patients and scoliosis was most common presentation of type I CM patients between 3 years to 5 years of age. In the present study, 41.18% (7 out of 17) CM cases with ventriculomegally were under 5 years of age.

In one-third to half of the cases of Chiari type I malformation was associated with syringomyelia[12] and 30% was associated with hydrocephalus [13]. Among 46 type I CM patients in the present study, 52.17% were associated with syringomyelia (figure no.1A) and 26.09% were associated with hydrocephalus (table no.4). According to Fernandez AA et al.[9], hydrocephalus only occurs in 3-10% of patients with Chiari type I malformation. In contrast, practically always occur in Chiari type II malformation [9]. Fernandez AA et al. [9] also mentioned that 40-75% of Chiari type I malformations have associated syringomyelia.

In type II CM, moderate hydrocephalous has been documented in about 25% cases. Syringomyelia is present in 60% of established cases [14]. Among 14 type II CM patients in the present study, 28.57% were associated with hydrocephalus and 35.71% were associated with syringomyelia (Figure 2A, 2B).

Partial or complete agenesis of the corpus callosum with absent septum pellucidum, polygyria, prominent anterior commissure, obliterated longitudinal fissure between parietal and occipital lobes, dysgenesis of olfactory tract/bulb, absent cingulate gyrus and heterotopic gray matter are frequently seen with type II CM [15]. In the present study, 78.57% (11 out of 14 cases) of type II CM were associated with corpus callosum agenesis (Table 4).

In a report of 364 cases of symptomatic Chiari I malformations, anomaly of the occipital bone—including occipitalization of the atlas, condylar hypoplasia, and other atypia of the occipital bone itself—did not exceed 5.2% [12]. In the present study, 6.52% cases of type I CM were associated with occipitalization of the atlas vertebra (Table 4).

De Barros et al., reported 22/66 individuals with Chiari I showing basilar invagination [16].

In the present study, out of 46 type I CM cases, nine were associated with basilar invagination (table no.5) (figure no.4). Basilar invaginations are found in 50% of cases of type II CM [14]. In the present study, basilar invagination was absent in type II CM (Table 5).

In chiari type I, craniovertebral junction abnormalities such as basilar invagination, platybasia, small posterior fossa, concavity of the clivus, occipitalization of the atlas and spina bifida in upper cervical region can be encountered at nearly 20-30 percent rate[7]. In the present study, 6.52% type I CM cases were associated with small posterior cranial fossa. On the other hand, 13 out of 14 type II CM cases were associated with small posterior cranial fossa (Table 5) (Figure 2A).

Many supratentorial abnormalities have been described with type II CM which includes callosal dysgenesis, a small third ventricle, enlarged interthalamic adhesions, a beaked tectum, polymicrogyria, heterotopias, skull deformities (the “lemon sign”), colpocephaly, and ventriculomegaly [17]. In the present study, 71.43% type II CM cases were associated with tectal beaking (Table 5) (Figure 2C).

Length of herniated cerebel-ler tonsil in 56.76% (21 out of 37 cases) cases of type 1 Chiari malformations are in between 10 and less than 20 mm[10]. In the present study, length of herniated cerebeller tonsil in 73.91% (34 out of 46 cases) cases of type 1 CM were in between five mm to less than 15 mm (Table 6).

Aboulez et al. reported that on T1 weighted images in the sagittal plane, the average distance from the tonsillar tips to the foramen magnum ranged from 2.9 mm above to 3.4 mm below [18]. They indicated that extension of the tonsils to up to 3 mm below the foramen magnum can be considered normal and that extension is clearly pathological only when it exceeds 5 mm [18].

Detailed evaluations using current imaging methods have introduced more subtle forms of the chiari malformations currently named as Chiari 0 and Chiari 1.5. Also introduced are terms like “asymptomatic” and “incidental” chiari malformations depending upon the extent of cerebellar ectopia and clinical findings [5]. Moreover, Chiari IV malformation, characterized by cerebellar hypoplasia or aplasia and

abnormalities in the pons [19], is now recognized within the cerebellar hypoplasia group [20].

CM type II and CM type III are gross defects of neuroectodermal origin (figure no.2A, 3), there is accumulating evidence that CM type I is a disorder of the paraaxial mesoderm that results in underdevelopment of the posterior cranial fossa (PCF) and overcrowding of the hindbrain [21] (Figure 1B).

Chiari malformations can be the result of a known etiology or can occur without any known causative factors. The chiari that occurs from an unknown etiology is likely the result of a genetically determined small posterior fossa volume [4].

In a recent study mentioned that the combination of patient age and BMI (body mass index) plays a significant role in the development of CM type 1 signs and symptoms [22]. Older obese patients presented with significantly larger chiari malformations, more cerebellar symptoms, and fewer headaches [22].

Currently, the only form of treatment to alleviate symptoms is to perform a posterior fossa (PF) decompression surgery. The goal of this surgery is to expand the cranial base to provide additional room for the cerebellum while also restoring normal cerebrospinal fluid flow [23]. Another recent study suggested that Arnold Chiari Malformations can be prevented by pre-conceptional folic acid and Vitamin B 12 supplementation [14].

CONCLUSION

With data obtained from the MRI, an effort has been made to study 63 numbers of different types of chiari malformations in this part of India. Unlike the established literature, male predominates female and type I chiari malformations being the commonest type among the four. More than half of the patients are under twenty years of age. Type I chiari malformation is mostly associated with syringomyelia and hydrocephalus, whereas type II is associated with corpus callosum agenesis. Small posterior cranial fossa is present in almost all the cases of type II variety.

Conflicts of Interests: **None**

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