MRI EVALUATION OF CONGENITAL MALFORMATIONS OF BRAIN

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ABSTRACT

BACKGROUND

Central nervous system (CNS) malformations constitute an important spectrum of disorders and more than 2,000 malformations have been described. Approximately 75% of fetal deaths, 40% of infant deaths and 0.5% of deaths in children under one year of age are caused by CNS malformations. In developing countries like India, there are no adequate statistics regarding the incidence and type of brain malformations. This study has been undertaken to analyse the incidence and spectrum of routinely encountered congenital malformations of brain.

OBJECTIVE

To analyse the entire spectrum of all important and routinely encountered congenital malformations and the proportional distribution of the various malformations of the brain in a tertiary care institute in South India

METHODS AND MATERIALS

All MRI brain examinations at our hospital over a period of one year were retrospectively reviewed and analysed by sex, age at presentation, type of congenital cerebral malformation, and other associated congenital cerebral malformations.

RESULTS

This study included 52 patients. The total number of brain malformations identified in these patients was 60. The age of patients at first MRI imaging ranged from 3 days to 60 years. There were 28 males and 24 females. The most common malformations were corpus callosum dysgenesis (18%), posterior fossa malformations (17%) and neural tube closure defects (15%). Eight patients (15.3%) had more than one congenital brain malformation.

CONCLUSION

Congenital malformations of the brain are both complex and multiple. MRI plays an important role in reaching the correct diagnosis necessary for optimum management of these unfortunate conditions.

KEYWORDS

Nervous system malformations, congenital, magnetic resonance imaging, brain anomalies.

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INTRODUCTION: Central nervous system (CNS) malformations constitute an important spectrum of disorders and more than 2,000 malformations have been described. They occur in almost one percent of all births and represent at least l0% of all systemic malformations. Approximately 75% of fetal deaths, 40% of infant deaths and 0.5% of deaths in children under one year of age are caused by these malformations. The severe forms of malformations are incompatible with life and are usually aborted. The ones that are clinically encountered are relatively mild in severity.[1] Abnormal development, leading to dysplasia or malformation, is a common finding in the neuroimaging studies of children with developmental delay, mental retardation, or epilepsy.[2,3,4] It is important to identify these conditions at the earliest due to its far reaching neurological deficit and detrimental outcome.[5]

In developing countries like India, there are no adequate statistics regarding the incidence and type of brain malformations. This study has been undertaken to analyse the incidence and spectrum of routinely encountered congenital malformations of brain.

OBJECTIVE: To analyse the entire spectrum of all important and routinely encountered congenital malformations and the proportional distribution of the various malformations of the brain in a tertiary care institute in South India.

MATERIALS AND METHODS: The study was carried out in the Department of Radio-diagnosis in Victoria Hospital attached to Bangalore Medical College and Research Institute, Bangalore. Ethical clearance for the study was obtained from the institution.

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All MRI studies of the brain for over a period of 12 months from Jan 2014 to December 2014 were retrospectively reviewed. All MR imaging examinations were performed on a Siemens Avanto 1.5-T magnet MR system. MRI was performed in all patients in the axial, coronal and sagittal plane with T1 & T2-weighting, along with Inversion recovery and Diffusion Weighted Imaging sequences. Contrast agents were utilized in specific conditions if required.

Cases with inadequate or technically suboptimal examination were excluded from this study. Congenital malformations were analysed by sex, age at presentation, type of congenital cerebral malformation, and other associated congenital cerebral malformation.

OBSERVATION AND RESULTS: Congenital brain malformations were detected in 52 patients. The total number of cerebral malformations identified in these patients was 60. The age of patients at first MRI imaging ranged from 3 days to 60 years. Most of the malformations were detected in the first decade of life (n=33, 55%). Around 21.7% (n=13) were detected in the second decade life. Table 1 shows the age wise distribution of the various malformations.

Age group in years	Number of malformations	Percentage
0-10 years	33	55.0
11-20 years	13	21.7
21-30 years	10	16.7
31-40 years	3	5.0
41-50 years	0	0
51-60 years	1	1.6

Table 1: Age wise distribution of different malformations in the study group

Of the 52 patients, there were 28 males and 24 females. There was slight male preponderance (M: F-1.16:1). (Table 2)

Gender	No. of cases	Percentage
Male	28	53.8
Female	24	46.2
Total	**52**	**100.00**

Table 2: Gender wise distribution of cases

The most common malformations were corpus callosum dysgenesis (18%), posterior fossa malformations (17%) and neural tube closure defects (15%). Eight patients (15.3%) had more than one congenital brain malformation. Table 3 shows the distribution of the sixty malformations in the fifty two patients.

There was no significant difference in the incidence of various malformations in both the sexes. Table 4 shows the gender distribution of the various malformations.

Corpus Callosum Dysgenesis: Corpus callosum dysgenesis was seen in 11(18%) patients. In 6(54%) patients the entire corpus callosum was absent (agenetic). In 5(46%) patients the callosal agenesis partially affected the splenium and posterior half of the body. Of the 11 patients with corpus callosum dysgenesis, 4(36%) patients had the anomaly with no other associated cerebral malformations, while 7(64%) patients had other associated malformations. In these 7 patients, the encountered cerebral malformations were Chiari II malformation (n=2), Dandy Walker spectrum (n=1), Schizencephaly (n=1) heterotopia (n=1), and polymicrogyria (n=1).

Type of malformation	Number	Percentage
Corpus callosal dysgenesis Partial Complete	11 06 05	18.3
Posterior fossa malformations Dandy-Walker spectrum Cerebellar malformation Joubert syndrome	10 06 03 01	16.7
Neural tube closure defects Chiari I Chiari II Chiari III Occipital encephalocoele	09 04 03 01 01	15.0
Cortical organisational defects Focal cortical dysplasia Polymicrogyria Schizencephaly	08 02 03 03	13.3
Cortical migration defects Lissencephaly complex Heterotopia	06 05 01	10.0
Neurocutaneous syndromes Neurofibromatosis Sturge-Weber syndrome Tuberous sclerosis Von Hippel Lindau disease	07 01 01 04 01	11.7
Vascular malformations Arteriovenous malformation Developmental venous anomaly Cavernous malformation	05 02 02 01	8.3
Others	**04**	**6.7**

Table 3: Distribution of the different malformations in the study group

Type of malformation	Males	Females
Corpus callosal dysgenesis	6	5
Posterior fossa malformations	5	5
Neural tube closure defects	5	4
Cortical organisational defects	4	4
Cortical migration defects	3	3
Neurocutaneous syndromes	4	3
Vascular malformations	2	3
Others	2	2

Table 4: Gender distribution of different malformations in the study group

Posterior Fossa Malformations: Posterior fossa malformations constituted the second most common malformation (n=10, 17%). Of the ten patients, Dandy

walker spectrum was noted in six patients, three patients had cerebellar hypoplasia and one patient with Joubert syndrome. One case of Dandy walker malformation had associated corpus callosal dysgenesis.

Neural Tube Closure Defects: Neural tube closure defects was seen in 9(15%) patients. This included eight cases of Chiari malformation and one case of occipital encephalocoele. Chiari I malformation was seen in four patients, of which two patients had syringohydromyelia. Chiari II malformation was seen in three patients, all of which were associated with meningomyelocoele. Two of these patients had also dysgenetic corpus callosum. One patient with Chiari II malformation had associated polymicrogyria. Only one patient with Chiari III was identified.

Cortical Organizational Defects: Eight (14%) patients were found to have disorders of cortical organization. This included three cases of schizencephaly, three cases of polymicrogyria and two cases of focal cortical dysplasia. One case of schizencephaly and one case of polymicrogyria was associated corpus callosal dysgenesis. One case of polymicrogyria was associated with Chiari II malformation.

Cortical Migrational Defects: Cortical migrational defects were found in 6(10%) patients, 5 of whom had lissencephaly-pachygyria spectrum and the remaining 1 grey matter heterotopia which had associated corpus callosal dysgenesis. One patient had agyria and four patients had varying degrees of pachygyria.

Neurocutaneous Syndromes: The Neurocutaneous syndromes constituted 12% (n=7) of the cases. This included 4 cases of tuberous sclerosis and one each in neurofibromatosis type 1, Von Hippel Lindau and Sturge Weber syndrome. Of the four cases in tuberous sclerosis, one case was associated with sub ependymal giant cell tumour.

Vascular Malformations: Vascular malformations constituted 8% (n=5) of the cases, of which two were developmental venous anomaly, two cases of arteriovenous malformation and one case of cavernous malformation. Developmental venous anomaly was incidentally detected.

Others: Other malformations detected include semilobar holoprosencephaly (n=1), septo-optic dysplasia (n=1), hypothalamic hamartoma (n=1) and intracranial lipoma (n=1).

DISCUSSION: Congenital brain malformations are extremely diverse and often more than one malformation exists in the same patient. Earlier, these malformations were studied and classified based on pathologic examinations. However, the development of magnetic resonance imaging (MRI) has led to a much better understanding of the entire spectrum of congenital brain malformations. Furthermore, MRI has allowed us to observe the temporal evolution of these malformations as the brain undergoes maturation.[6]

Because most neural structures develop at about the same time during foetal life, it is common to see multiple associated anomalies.[7] Hence, one case with multiple anomalies may fit into many classes of brain malformations. Corpus callosal dysgenesis is of two types: complete or partial (Figure 1). It can be associated with Chiari II malformation, migration disorders (heterotopias, lissencephaly, and schizencephaly), Dandy-Walker malformations, and holoprosencephaly and corpus callosal lipoma.[8,9]

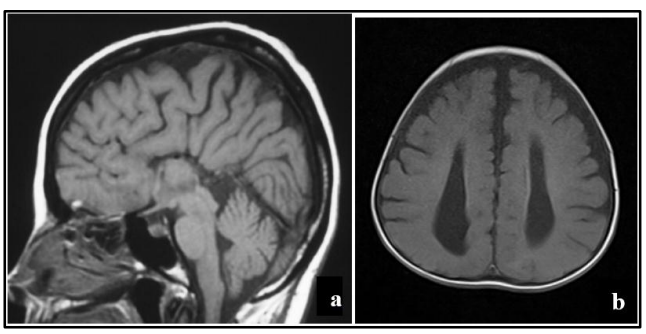

Fig. 1: Complete corpus callosal agenesis. (a) Sagittal T1 weighted image shows medial hemispheric sulci radiating into the third ventricle because of lack of inversion of the cingulate gyrus. (b) Axial T1 weighted image shows that bodies of lateral ventricles are parallel

Posterior fossa malformations include a spectrum of entities such as Dandy-Walker malformation, Dandy Walker variants, Joubert syndrome (Figure 2), cerebellar hypoplasia and developmental variants such as mega cisterna magna, arachnoid cyst and Blake's pouch cyst.[10] Dandy-Walker malformation is diagnosed on the basis of a characteristic triad of cystic dilatation of the fourth ventricle, complete or partial agenesis of the vermis and an enlarged posterior fossa. The diagnosis of Dandy-Walker variants comprises those cases that are commonly linked by varying degrees of vermian cerebellar hypoplasia with normal size of posterior fossa.[11]

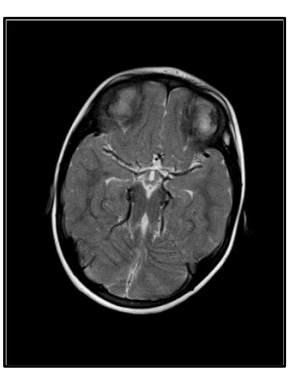

Fig. 2: Joubert syndrome. Axial T2W image at level of mid brain shows characteristic "molar tooth" appearance of the midbrain secondary to the narrow isthmus and elongated superior cerebellar peduncles

Neural tube closure disorders identified in this study include Chiari malformations and occipital encephalocoele. The characteristic feature of Chiari I malformation (Figure 3) is the cerebellar tonsillar herniation below foramen magnum. The hallmark of Chiari II is the small posterior fossa due to a low tentorial insertion leading to herniation of cerebellum inferiorly below the foramen magnum and almost always associated with lumbar myelomeningocele, and frequently other cerebral malformations.[12]

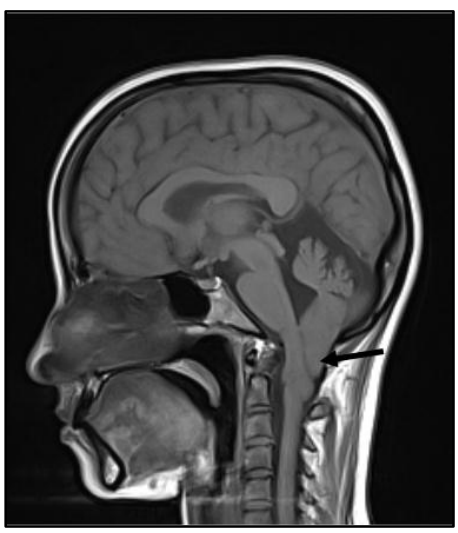

Fig. 3: Chiari 1 malformation. A. Sagittal T1W MR image of the brain and upper cervical spine shows inferior displacement of the cerebellar tonsils into the upper part of cervical spine (black arrow)

Chiari III malformation is an extremely rare disorder and only one case was identified in this study. In this malformation there is, in addition to the manifestations of Chiari II malformation, there is posterior herniation of the cerebellum and sometimes of the brainstem by spina bifida at the C1 or C2 level.[13]

When neurons reach the cortex area but fail to develop into normal gyri the condition is referred to as a disorder of cortical organization and it can be focal or diffuse. There is a lack of normal gyral formation with a thickened cortex.[14] The most common malformations include focal cortical dysplasia, polymicrogyria and schizencephaly (open or closed lip) (Figure 4).

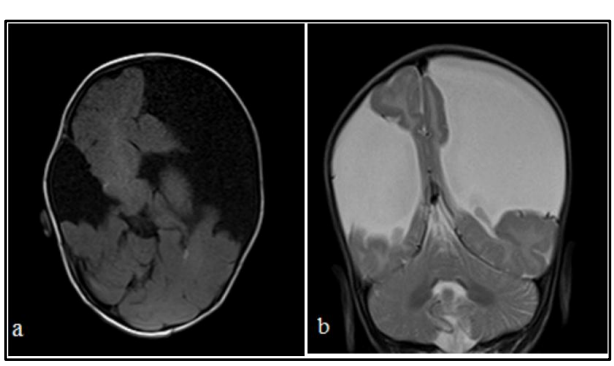

Fig. 4: (a) Axial T1W image and (b) Coronal T2W image showing bilateral opening lip schizencephaly

Migrational abnormalities occur due to arrest of neurons at a distance short of the normal location of the cortex. In type 1 (classical) lissencephaly, the brain surface is completely smooth called agyria (Figure 5) or has broad, flat gyri separated by a few, shallow sulci (pachygyria). On the other hand, the previously termed type 2 lissencephaly forms part of the cobblestone complex that is often seen in association with muscular dystrophy.[15]

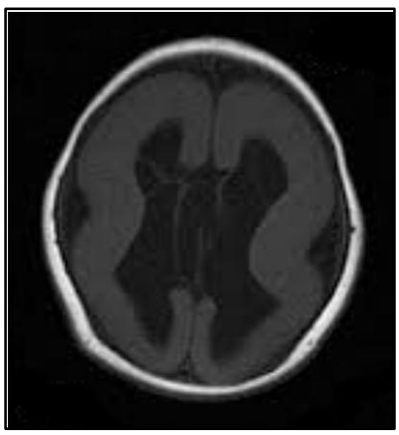

Fig. 5: Lissencephaly. Axial T1W image shows a complete smooth brain with thickened cortex and shallow sylvian fissures giving the brain characteristic figure of eight appearance

Grey matter heterotopias refer to the presence of normal neurons at abnormal locations are due to arrest of the normal neuronal migration along radial glial fibres. It can be divided into three types, periventricular, subcortical and band heterotopias.

Neurocutaneous syndromes" or "Phakomatoses" is a group of congenital malformations characterized by cutaneous lesions associated with CNS anomalies. This includes tuberous sclerosis (Figure 6), Neurofibromatosis type 1 and type 2, Sturge Weber syndrome and Von Hippel lindau syndrome.[16]

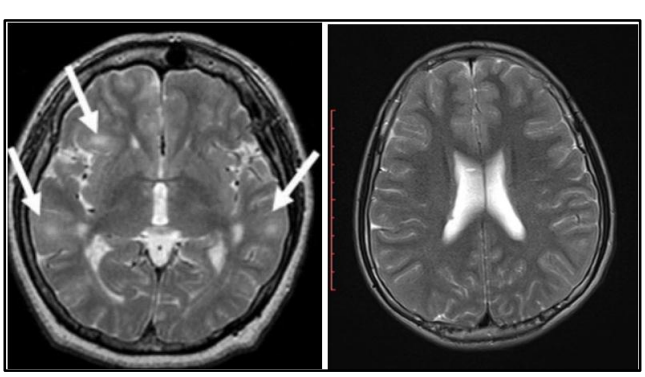

Fig. 6: Tuberous sclerosis. Axial T2W images showing multiple hyperintense cortical tubers (arrows) and hypo intense sub ependymal nodules

CONCLUSION: In conclusion, a predominance of corpus callosal dysgenesis, posterior fossa malformations and neural tube defects has been demonstrated in this study. There was no sex predilection in most of these malformations and most of the malformations were detected

in the first two decades of life. MRI is the imaging modality of choice in the evaluation of congenital malformations. It is essential to know the imaging findings of these malformations and to have knowledge about the various abnormalities associated with them to provide an accurate diagnosis which is very important for predicting the prognosis and planning further management.

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