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Research Article

# Role of Mri in Characterization of Congenital Malformations of Brain in A Tertiary Care Centre-A Retrospective Study

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## Abstract

**Background:** Congenital brain anomalies are rare among the congenital anomalies of various organ systems. It is crucial to recognise these disorders as soon as possible because of the extensive neurological deficiency and negative consequences they can cause. Neuroimaging is a reliable way to accurately diagnose the majority of congenital brain abnormalities. There aren't enough statistics available in developing countries like India about the frequency and types of brain abnormalities. The purpose of this study is to examine the prevalence and range of frequently occurring congenital brain abnormalities.

**Objective:** To study the whole spectrum of all substantial and frequently occurring congenital abnormalities as well as the proportional distribution of the various brain malformations in a tertiary care centre.

**Methods and materials:** The study was conducted at Department of Radio-Diagnosis, Medical College Kolkata from 1st September 2022 to 31st August 2023. 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 24 patients. The total number of brain malformations identified in these patients was 33. The age of patients at first MRI imaging ranged from 5 days to 16 years. There were 14 males and 10 females. The most common malformations were corpus callosum dysgenesis (24%), Heterotropia (15%) and Lissencephaly (12%). Seven patients (29.1%) had more than one congenital brain malformation.

**Conclusion:** The variety and complexity of congenital brain abnormalities is remarkable. The correct diagnosis is essential for the best care of these unfortunate conditions, and MRI plays a significant role in this process. Identification of distinctive neuroimaging findings of various congenital brain anomalies are immensely helpful in diagnosing the anomalies and further management.

**Key words:** obesity; dopamine; insulin; food behavior; reward; leptin; ghrelin; african american; black; afro-american

## Introduction

Congenital brain anomalies are rare among the congenital anomalies of various organ systems. There are more than 2000 different forms of congenital malformations (CM) of the brain that have been described in the literature. Congenital deformities have a wide range of etiologic causes, which can be broadly categorised into genetic, environmental, exposure to teratogens, and specific deficiencies, including folate and iodine, with some studies attributing a multifactorial origin. Known risk factors for congenital abnormalities include maternal age, drug usage, teratogen exposure, radiation exposure, maternal diseases, smoking, and alcohol consumption. Congenital brain malformations have been diagnosed using a variety of imaging modalities, including computed tomography (CT), MRI, and ultrasound. MRI is the modality of choice because it is not only perfect, especially for young patients, but also because it is multiplanar and multiaxial, has great soft-tissue delineation,

and has no known biohazard effects. There aren't enough statistics available in developing countries like India about the frequency and types of brain abnormalities. The purpose of this study is to examine the prevalence and range of frequently occurring congenital brain abnormalities.

### Congenital brain anomalies can be classified as:

1. Disorders of primary neurulation: These are mostly neural tube closure defects and early CNS anomalies occurring during 3rd and 4th gestational weeks. These include Chiari malformations, cephaloceles and myelomeningoceles.
2. Disorders of diverticulation, cleavage, sulcation and cellular migration. These include:
    a. Holoprosencephaly
    b. Lissencephaly
    c. Cortical dysplasia
    d. Heterotropia
    e. Schizencephaly.
3. Posterior fossa malformations include:
    a. Dandy-Walker malformations
    b. Joubert syndrome
    c. Rhombencephalosynapsis.
4. Disorder of histiogenesis

## Materials and Methods

### Patients:

The study was conducted at Department of Radio-Diagnosis, Medical College Kolkata. 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. The study was conducted from 1st September 2022 to 31st August 2023.

Inclusion and exclusion criteria:

All paediatric patients (<18 years) who had MRI conducted in our institute or those with congenital brain anomalies were included. Patients over 18 years of age were excluded from the study.

### Imaging technique:

In our institution, all brain MRI scans were performed in accordance with the standard protocols and positioning for brain MRI. The patients were lying supine on the MRI table with the median sagittal plane equally spaced from the table edges. An orthogonal 3-plane localizer was then obtained, consisting of axial, sagittal, and coronal views. Sagittal images were obtained on a coronal or axial localizer image. The corpus callosum, the Sylvian aqueduct, the fourth ventricle, and the cervical spinal cord were used as landmarks to identify the midsagittal plan. Axial images were obtained on a sagittal localizer and were positioned parallel to the bicommissural line or parallel to a line linking the floor of the sella turcica to the fastigium of the fourth ventricle. Coronal images were obtained on a sagittal localizer positioned parallel to the posterior surface of the brainstem.

### Imaging Sequences:

- T1-weighted sequences
- T2-weighted sequences
- Fluid-attenuated inversion recovery sequences
- Diffusion weighted sequences
- Gradient echo sequences

A slice thickness of 4 mm with 1 mm was used for all sequences done at our institute. Flip angles of 90° and 180° were used for T1- and T2-weighted sequences, respectively.

### Image analysis:

The interhemispheric fissure, cerebral cortex, ventricles, white matter, basal ganglia, internal and external capsule, corpus callosum, brain stem, cerebellum, intracranial vessels, pituitary and sella, petrous pyramids, paranasal sinuses, and orbit were evaluated for size, shape, position, signal characteristics, and potential space-occupying lesions.

### Ethical Consideration:

The study was conducted after getting approval from Institutional Ethics Committee and other authority.

## Results

This study included 24 patients. The total number of brain malformations identified in these patients was 33. The age of patients at first MRI imaging ranged from 5 days to 16 years. There were 14 males and 10 females.

| **Age(years)** | **No.** | **%** |
|---|---|---|
| <1 | 7 | 29.2 |
| 1-10 | 11 | 45.8 |
| 10-20 | 6 | 25 |
| Min-Max | 5d-16 yrs. | |
| Mean+- SD | 6 +-1.2 | |
| Median | 7 | |

**Table 1**: Distribution of patients according to age.

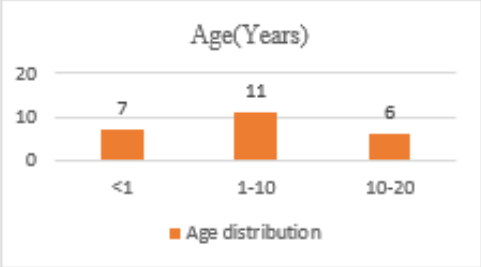


**Figure 1: Distribution of patients according to age.**

| Sex | No. | % |
|---|---|---|
| Male | 14 | 58.3 |
| Female | 10 | 41.6 |

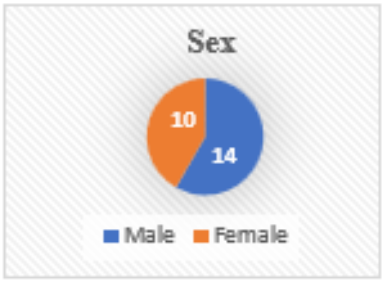



**Table 2 and Figure 2:** Distribution of patients according to sex.

Delayed developmental milestones was the most common presenting symptom in 10(41.6%) of cases, followed by seizures 5(20.8%), macrocephaly 4(16.6%), vomiting 3(12.5%) and mental impairment in 2(8.3%) of cases.

| Symptoms | No. | % |
|---|---|---|
| Delayed developmental milestones | 10 | 41.6 |
| Macrocephaly | 4 | 16.6 |
| | | |
| Seizures | 5 | 20.8 |
| Mental impairment | 2 | 8.3 |
| Vomiting | 3 | 12.5 |

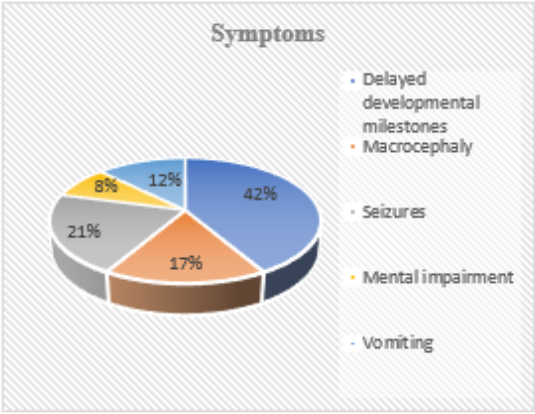



**Table 3 and Figure 3:** Distribution of Patients according to symptoms.

The total number of brain malformations identified in these patients was 33. Seven patients (29.1%) had more than one congenital brain malformation. The most common malformations were corpus callosum dysgenesis (24%), Heterotropia (15%) and Lissencephaly (12%).

| Diagnosis | No. | % |
|---|---|---|
| Corpus callosum dysgenesis | 8 | 24 |
| Heterotropia | 5 | 15 |
| Lissencephaly | 4 | 12 |
| Schizencephaly | 2 | 6 |
| Porencephalic cyst | 1 | 3 |
| Pachygyria | 3 | 9 |
| Chiari 2 Malformation | 2 | 6 |
| Dandy walker Spectrum | 2 | 6 |
| Holoprosencephaly | 2 | 6 |
| Arachnoid Cyst | 3 | 9 |
| Aqueductal stenosis | 1 | 3 |

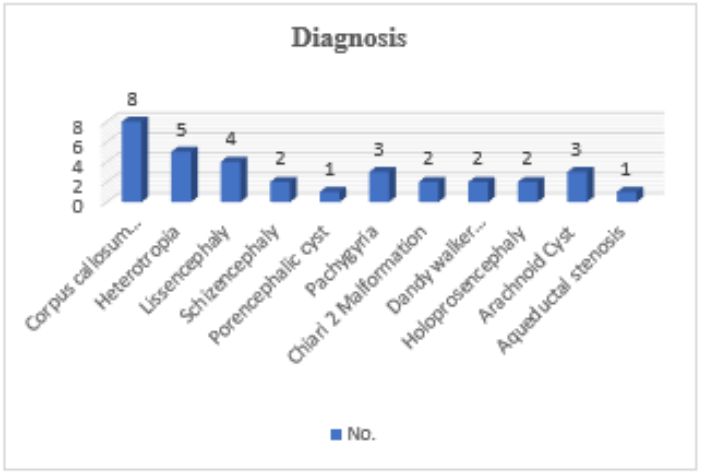
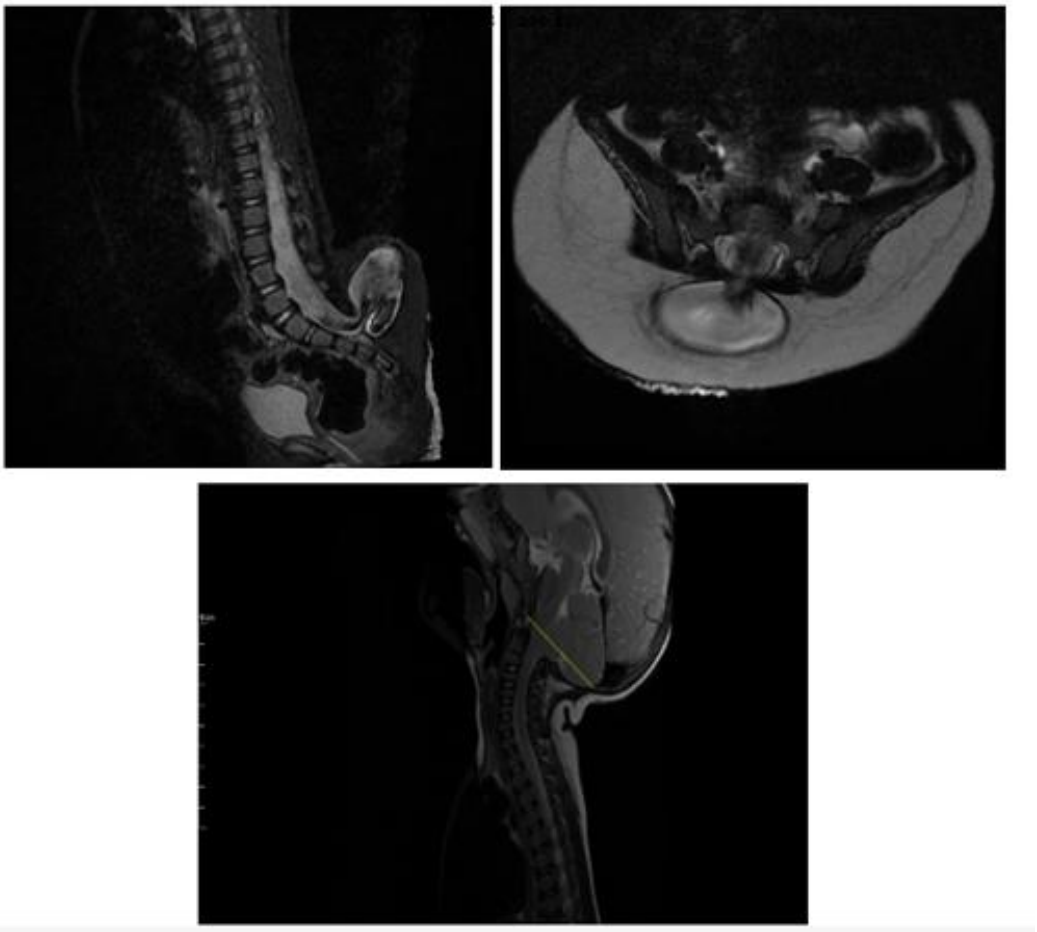



**Table 4 and Figure 4:** showing frequency distribution of congenital malformations.

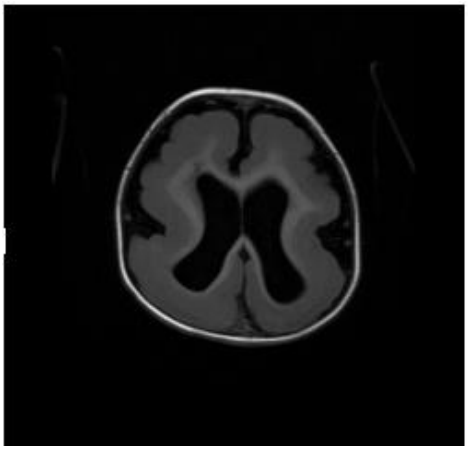
**Images**

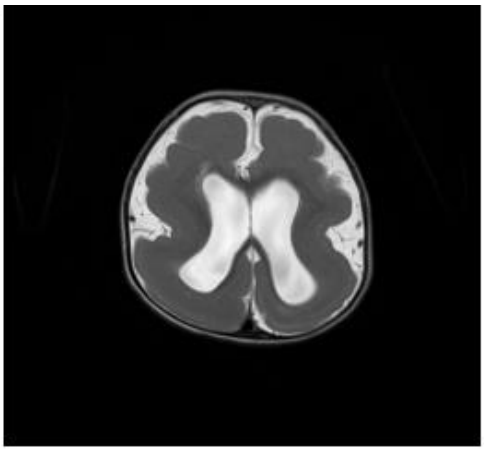
## Case 1:



A 1-year-old female showing myelomeningocele and inferior displacement of cerebellar vermis consistent with Chiari 2 Malformation.

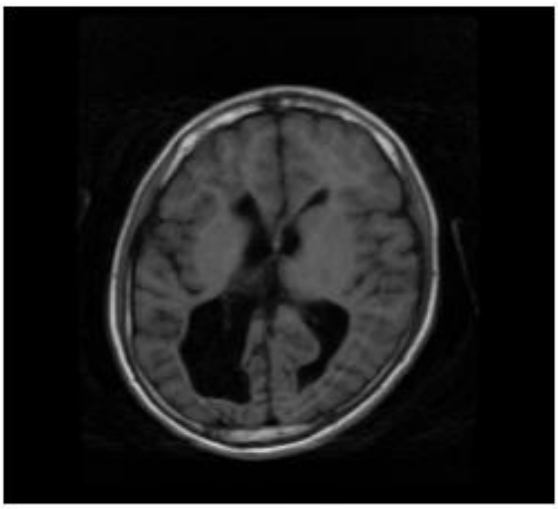
## Case 2:

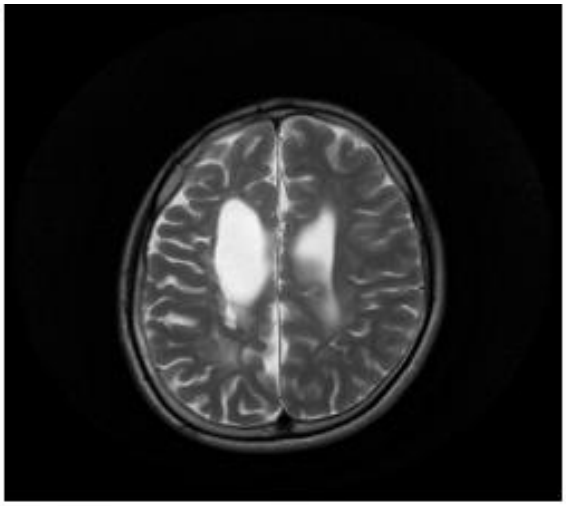


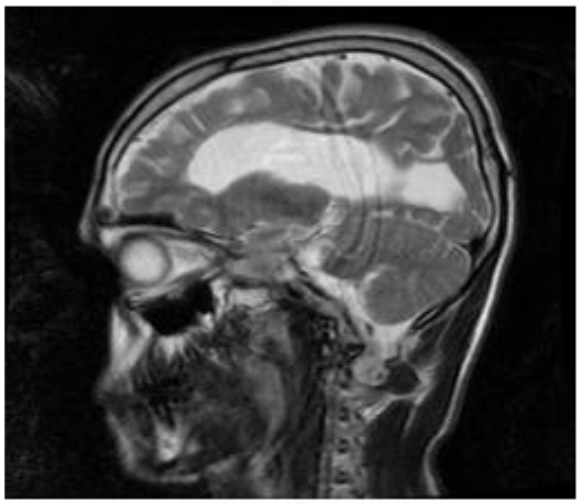


An 18 months old female with developmental delay. Axial T1W and T2W images show marked and diffuse thickening of the cerebral cortex bilaterally with a few poorly formed gyri and a smooth outer surface of both cerebral hemispheres giving figure of 8 appearance consistent with Classic Type 1 Lissencephaly with pachygyria. There is also dilatation of ventricular system.

## Case 3:

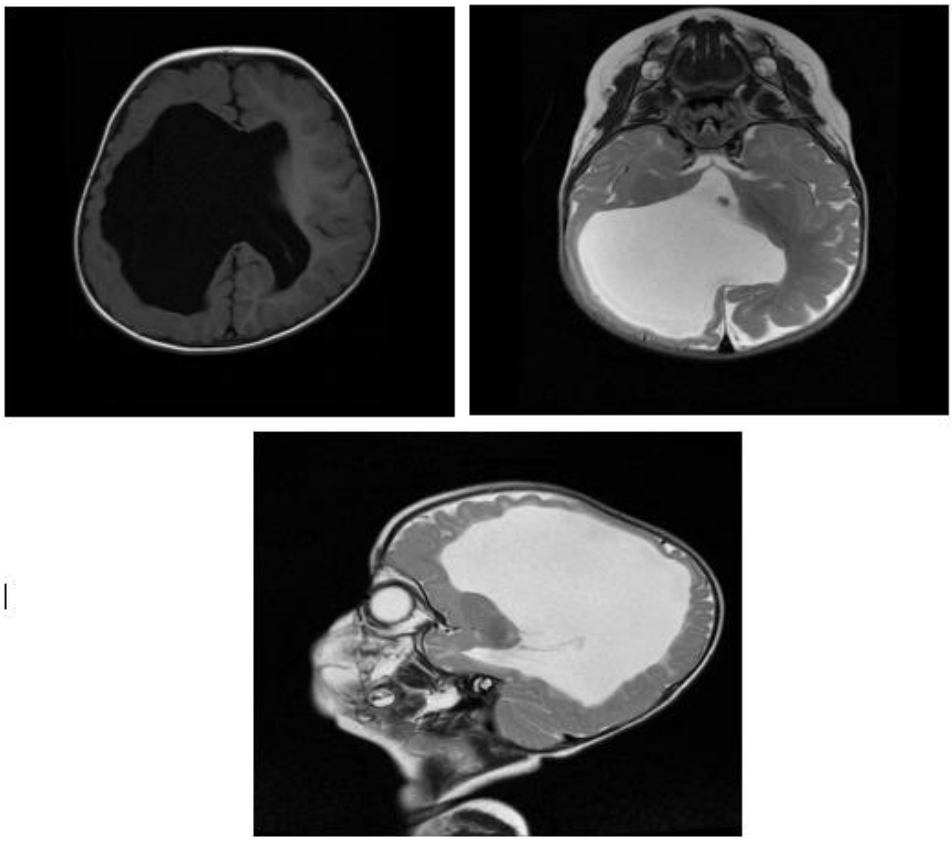






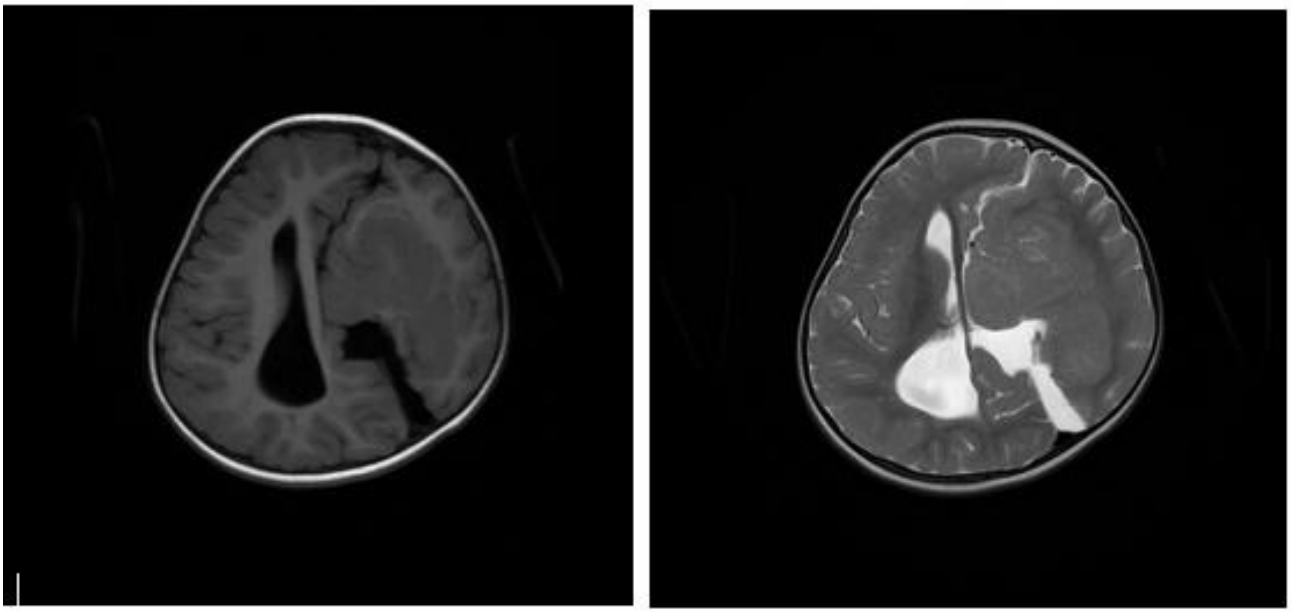
A 5-year-old male child with developmental delay. Axial T1W, T2W and Sagittal T2W images show complete absence of corpus callosum, parallel non converging lateral ventricles and colpocephaly consistent with corpus callosum agenesis.

## Case 4:



A 6-month-old female with seizures. Axial T1W, Sagittal and coronal T2W images show a large cystic cavity in right fronto-parieto-temporal lobes communicating with right lateral ventricle consistent with porencephalic cyst.

## Case 5:



A 2-year-old female with seizures. Axial T1W and T2W images show open lip schizencephaly, nodular grey matter heterotropia and pachygyria on left side.

## Discussion

Congenital brain abnormalities are structural or functional defects that develop in utero and can be detected during pregnancy, at birth, or as symptoms and signs later in life. Some are never symptomatic and are later recognised as incidental discoveries. MRI has had an important impact on the study and understanding of congenital cerebral malformations. MRI allows for study of the entire spectrum of such malformations, from mild to severe. Moreover, MRI permits multiple cuts in multiple planes at multiple occasions, providing a better understanding

of the temporal evolution of these diseases. Since most brain structures develop at roughly the same time during foetal life, it is common to see multiple anomalies in association. As a result, one case with multiple anomalies may fit into many classes of cerebral malformations.

Embryologically, the brain develops in 4 stages. Stage 1 involves dorsal induction with formation and closure of the neural tube, and this takes place between 3rd and 5th week of embryological life. Failure of this stage results in neural tube defects. Stage 2 involves ventral induction with formation of the brain segments and face at 5–10 weeks. Failure of this results in holoprosencephaly, Dandy–Walker malformation, and facial anomalies to mention a few. Stage 3 involves migration and histogenesis at 2–5 months of gestation with the failure of migration resulting in heterotopias, agyria-pachygyria, polymicrogyria, schizencephaly, corpus callosal agenesis, while the failure of histogenesis results in aqueductal stenosis, arachnoid cysts, megalencephaly, phakomatoses, congenital vascular malformations, and neoplasms. Cerebral atrophy and hydrocephalus occur at the stage of myelination (Stage 4).

In our study of 24 patients, the total number of brain malformations identified were 33 in number. The age of patients at first MRI imaging ranged from 5 days to 16 years. There were 14(58%) males and 10(42%) females. The most common malformations were corpus callosum dysgenesis (24%), Heterotropia (15%) and Lissencephaly (12%). Seven patients (29.1%) had more than one congenital brain malformation. Neural tube defect was seen only in 2(6%) patients. Cortical migrational defects were found in 9 (27.27%) patients, 5 of whom had lissencephaly and the remaining 4 gray matter heterotopia. Three patients were found to have disorders of cortical organization in the form of pachygyria. In only one of these patients was there also schizencephaly in association with cortical migrational defects (periventricular nodular heterotopia).

In our studies, a predominance of cortical migrational abnormalities and corpus callosum anomalies has been demonstrated and the results are similar to what has been observed in other parts of the world. Although most congenital cerebral malformations followed the usual and commonly described pattern and appearance, some did not.

## Conclusion

The variety and complexity of congenital brain abnormalities is remarkable. The correct diagnosis is essential for the best care of these unfortunate conditions, and MRI plays a significant role in this process. MRI plays a significant role in delineating these anomalies, with a significant impact on the study and understanding of the malformations and their evolution. Identification of distinctive neuroimaging findings of various congenital brain anomalies are immensely helpful in diagnosing the anomalies and further management.

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