Original Article

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Paediatrics Section

# Evaluation of Cerebral and Spinal Abnormalities on Magnetic Resonance Imaging in Infants with Myelomeningocele: A Retrospective Observational Study

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## ABSTRACT

**Introduction:** Spinal dysraphism includes a diverse collection of congenital spinal abnormalities that occur due to flawed closure of the neural tube and can be classified as open or closed types. The most common form of open spinal dysraphism is Myelomeningocele (MMC), which is characterised by a bony spinal defect and a sac in the back region containing dysplastic neural tissue and cerebrospinal fluid. MMC is associated with various intracranial and spinal anomalies that can lead to neurological, intellectual, and cognitive impairments. Imaging is necessary for the diagnosis of these anomalies and for surgical management planning and follow-up.

**Aim:** To evaluate the associated intracranial and spinal anomalies in infants with MMC who presented with low back swelling since birth.

**Materials and Methods:** The retrospective observational study was conducted in the Department of Radiology at Gajara Raja Medical College, Gwalior, Madhya Pradesh, India, from January 2020 to December 2022. A total of 117 subjects under one year of age, clinically diagnosed with MMC, were included in the study. Brain Magnetic Resonance Imaging (MRI) images were available for 84 patients, and spine MRI images were available for 113 patients. The MRI images were evaluated for bony and spinal anomalies. Data analysis was performed using IBM's Statistical Package for Social Sciences (SPSS) version 20.0.

**Results:** The mean age of the study participants was 1.7±2.9 months, with 70 males and 47 females (M:F=1.5:1). The most common location of spina bifida was lumbosacral 36 (31.9%), followed by lumbar 31 (27.4%), dorsolumbar 30 (26.5%), and sacral 16 (14.2%). A low-level spinal defect was observed in 39 cases (34.5%), and a high-level spinal defect was observed in 74 cases (65.5%). The spinal anomalies detected on MRI included syringomyelia 80 (70.8%), low-lying cord 59 (52.2%), bony abnormalities 26 (23.01%), diastematomyelia 16 (14.2%), and fatty filum terminale 3 (2.7%). Among the intracranial anomalies, the most common was dilated lateral ventricles 62 (73.8%). The most common infratentorial anomaly was Chiari II malformation 58 (69.1%). Corpus Callosum (CC) abnormalities were found in 40 cases (47.6%), with complete agenesis seen in three cases (3.6%), cerebellar hypoplasia in 20 cases (23.8%), absent septum in 15 cases (17.9%), and colpocephaly in 10 cases (11.9%).

**Conclusion:** Spinal dysraphism, such as MMC, and cerebral anomalies often co-exist and can have an impact on neurological, intellectual, and cognitive functions. Chiari II malformation, corpus callosal anomalies, ventriculomegaly, and syringomyelia are frequently associated with MMC. Therefore, imaging evaluation of both the spine and brain is necessary for surgical treatment planning and long-term follow-up in MMC.

**Keywords:** Chiari II malformation, Intracranial anomalies, Open neural tube defect, Spina bifida, Spinal dysraphism, Ventriculomegaly

## INTRODUCTION

Spinal dysraphisms include a diverse collection of congenital spinal abnormalities that occur due to flawed closure of the neural tube and can be open or closed types [1,2]. The closed form of spinal dysraphisms can be asymptomatic as they are covered by intact skin. The open form of spinal dysraphisms has an overlying skin breach and is frequently associated with neurological impairments depending on the extent of neural placode abnormality and bony spinal defect level [1]. The most frequent form of open spinal dysraphism is MMC [2], which occurs due to incomplete closing of the lower end of the neural tube in the third and fourth gestational week [1]. MMC is characterised by a bony defect in the posterior elements of vertebrae with bulging out of a part of the spinal cord and nerves through the bony defect, and the development of a sac in the back region consisting of dysplastic neural tissue and cerebrospinal fluid [2]. The most common sites of MMC are lumbosacral or sacral, although thoracic and cervical MMC are also observed [2,3].

The MMC is associated with motor and sensory weakness, bowel and bladder dysfunction, and talipes as the neural placode comes into communication with the outer atmosphere [1,3,4]. MMC is also related to various intracranial anomalies that cause impairment of intellectual and cognitive functions [1,2]. It has been reported that hydrocephalus is the main reason for cognitive impairment [5]. Chiari II malformation, often seen with MMC, can also contribute to multiple cognitive deficits [5]. Chiari II malformation is characterised by inferior herniation of posterior fossa structures (cerebellar vermis, brainstem, and fourth ventricle) with a diminished size of the posterior fossa [3]. Other intracranial findings associated with MMC, in addition to ventriculomegaly and Chiari II malformation, include cerebellar hypoplasia, brainstem hypoplasia, CC dysgenesis/hypoplasia, cortical malformations, colpocephaly, absent septum pellucidum, wide interhemispheric fissure, etc. [4]. Patients with spinal dysraphism can also have other spinal anomalies such as diastematomyelia, low-lying cord, syringomyelia, filum terminale thickening, vertebral bony anomalies, spinal lipoma, dermal sinus, etc. [6].

These Central Nervous System (CNS) findings can be diagnosed and evaluated using Ultrasonography (USG) and MRI and are required for surgical management planning [1-3]. Conventional X-rays can detect bony defects and vertebral deformities [1,5]. Computed Tomography (CT) is not used for imaging in the perinatal period due to radiation exposure and low contrast resolution, and is only utilised for bony anomalies [2,5]. USG is used for prenatal diagnosis of MMC and has a sensitivity of 90% [1].

However, it has limitations such as low resolution in the posterior fossa and operator dependency in the postnatal period. MRI offers advantages such as multiplanar imaging, high spatial resolution, and no radiation risk [2, 3, 5]. MMC can have a persistent progressive course, leading to an increase in symptoms over time. Therefore, patients require timely operative treatment and regular monitoring even after surgical repair, as they may develop worsening of associated abnormalities like hydrocephalus, syringomyelia, and tethered cord [1, 5, 7]. The present study aimed to investigate the associated intracranial and spinal anomalies in infants with MMC who present with low back swelling since birth at a single tertiary care centre.

## MATERIALS AND METHODS

A retrospective observational study was conducted in the Department of Radiology at Gajara Raja Medical College, Gwalior, Madhya Pradesh, India, from January 2020 to December 2022. The study was performed after receiving approval from the Institutional Ethics Committee (13/IEC-GRMC/2023). Informed written consent was obtained from the parents/guardians of all infants prior to MRI, allowing the use of their images and clinical information for academic/educational/publication purposes without revealing their identity, following departmental protocol.

**Inclusion criteria:** Patients under one year of age, clinically diagnosed with MMC, and who had undergone preoperative MRI in the Department were included in the study.

**Exclusion criteria:** Patients older than one year, presence of spinal tumours, traumatic injury, infection, abscess, artifacts in MRI images hindering proper evaluation, and patients who had undergone previous intracranial or spinal surgery were excluded from the study.

### Study Procedure

The MRI was performed using a 1.5 Tesla scanner (Ingenia, Philips Healthcare) with a head coil. MRI findings in the brain and spine of the included subjects with MMC were retrospectively evaluated. The spine MRI images were assessed for the location of the bony spinal defect, MMC sac diameter, spinal cord, spinal canal, and bony vertebral anomalies. Brain MRI images were examined for the presence of findings such as Chiari II malformation, dilated lateral ventricles, CC dysgenesis/hypoplasia, and other associated intracranial anomalies. The spina bifida defect was classified as a low-level defect when the highest level of bony defect was below the third lumbar vertebra (L3) and high-level when it was at or above the L3 level [4]. The presence of a low-lying cord was determined when the conus medullaris was observed below the level of the L2-3 intervertebral disc [3]. Dilated lateral ventricles were identified when the ventricular trigone diameter measured equal to or greater than 1 cm on the axial plane [4].

## STATISTICAL ANALYSIS

All imaging findings and demographic characteristics were recorded using a Microsoft Excel 2007 spreadsheet. The mean with Standard Deviation (SD) was calculated for numerical variables such as age, size of bony defect, and size of MMC sac. Frequency (n) with percentage (%) was determined for sex, location of MMC sac, spinal cord changes, bony anomalies, and various brain anomalies. IBM SPSS version 20.0 (Armonk, NY, USA) was used for data analysis. A t-test was performed to assess the association between mean age and gender.

## RESULTS

A retrospective study included 117 patients with a clinical diagnosis of MMC, with a mean age of 1.7±2.9 months (range: 2 days to 11 months) [Table/Fig-1]. Among them, there were 70 males (59.8%) and 47 females (40.2%), resulting in a male-to-female ratio of 1.5:1. The mean age for males was 1.9±3.3 months (range: 2 days to 11 months), and for females, it was 1.2±2.3 months (range: 2 days to 11 months), with no significant difference in age between male and female patients (p-value=0.2 using t-test). Brain MRI images were available for 84 patients, and spine MRI images were available for 113 patients.

| Age (in months) | Frequency (n) | Percentage (%) |
|---|---|---|
| <1 | 82 | 70.1 |
| 1-3 | 13 | 11.1 |
| 3-6 | 9 | 7.7 |
| 6-12 | 13 | 11.1 |

**[Table/Fig-1]:** Age distribution of Myelomeningocele (MMC) patients

The most common location of spina bifida among these patients was lumbosacral 36 (31.9%), followed by lumbar (31, 27.4%), dorso-lumbar 30 (26.5%), and sacral 16 (14.2%). A low-level spinal defect was observed in 39 cases (34.5%), while a high-level spinal defect was seen in 74 cases (65.5%). The mean size of the spina bifida defect was 23.1±10.7 mm (range: 5-54 mm), and the mean size of the MMC sac diameter was 47.5±15.8 mm (range: 10-93 mm). The spinal anomalies found on spine MRI are shown in [Table/Fig-2], with syringomyelia being the most common (70.8%).

| Spinal anomalies | Frequency (n) | Percentage (%) |
|---|---|---|
| Syringomyelia | 80 | 70.8 |
| Low lying cord | 59 | 52.2 |
| Bony vertebral anomalies | 26 | 23.01 |
| Diastematomyelia | 16 | 14.2 |
| Fatty filum terminale | 3 | 2.7 |

**[Table/Fig-2]:** Frequency of various spinal anomalies associated with Myelomeningocele (MMC).

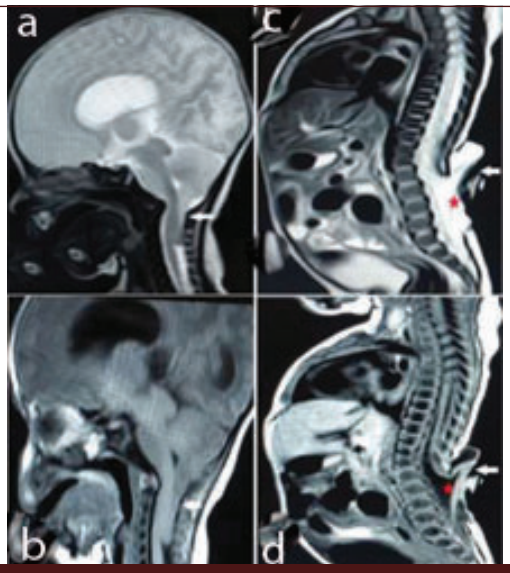
Among the intracranial anomalies, the most common was dilated lateral ventricles (73.8%), with severe dilatation observed in 12 patients (14.3%). The most common infratentorial anomaly was Chiari II malformation (69.1%). Additionally, Chiari I malformation was noted in 10.7% of cases. Two patients (2.4%) presented with less than 5 mm of cerebellar tonsillar herniation. CC abnormalities were found in 40 patients (47.6%), with complete agenesis observed in 3 patients (3.6%), partial agenesis in 21 patients (25%), hypoplasia of CC in 14 patients (16.7%), and stretching in 2 patients (2.4%). Dandy-Walker malformation and semilobar holoprosencephaly were found in one patient each. The intracranial anomalies are listed in [Table/Fig-3]. Images of two representative cases of MMC with Chiari II malformation are shown in [Table/Fig-4,5].

## DISCUSSION

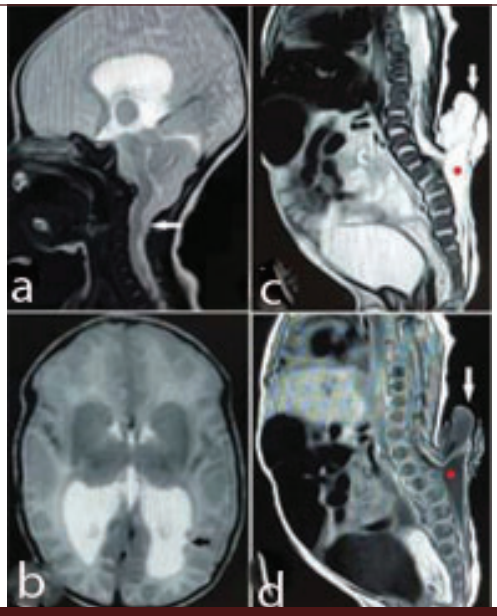
The MMC has been found to have a frequency of about 0.6-1.0 for every 1000 live births and is somewhat more frequent in females compared to males [2]. However, in the present study, there were

| Cerebral anomalies | Frequency (n) | Percentage (%) |
|---|---|---|
| Dilated lateral ventricles | 62 | 73.8 |
| Chiari II malformation | 58 | 69.1 |
| Cerebellar hypoplasia | 20 | 23.8 |
| Brainstem hypoplasia | 16 | 19.1 |
| Chiari I malformation | 9 | 10.7 |
| Corpus Callosum (CC) anomalies | 40 | 47.6 |
| Absent septum pellucidum | 15 | 17.9 |
| Colpocephaly | 10 | 11.9 |
| Wide interhemispheric fissure | 8 | 9.5 |
| Heterotopia | 5 | 5.9 |
| Cortical abnormalities | 5 | 5.9 |
| Gyral interdigitation | 4 | 4.8 |
| Abnormal white matter maturation | 2 | 2.4 |

**[Table/Fig-3]:** Frequency of various cerebral anomalies associated with Myelomeningocele (MMC).




**[Table/Fig-4]:** Sagittal MRI brain of a nine-day-old female showing Chiari II malformation (arrow) on T2W (a) and T1W (b) images and sagittal MRI spine showing spina bifida in lumbar region (red asterisk) with Myelomeningocele (MMC) sac (arrow) on T2W (c) and T1W (d) images.




**[Table/Fig-5]:** MRI brain of a 18-day-old female showing Chiari II malformation (white arrow) on sagittal T2W (a) and heterotopia (black arrow) on axial T2W (b) images and sagittal MRI spine showing spina bifida in lumbosacral region (red asterisk) with Myelomeningocele (MMC) sac (white arrow) on T2W (c) and T1W (d) images.

more males than females. The most frequent age group was less than one month since early postnatal imaging is required for timely management of MMC. The most common location of spina bifida was lumbosacral, which is consistent with previous studies [2,3,8]. Patients with open spinal dysraphism may present with varying degrees of motor, sensory, cognitive, and bowel/bladder dysfunction and are often seen concomitantly with a range of intracranial anomalies [8]. Apart from CNS anomalies, urogenital, skeletal, gastrointestinal, pulmonary, cardiac, and craniofacial anomalies have also been reported in these patients [8].

Maurice P et al., retrospectively reviewed MMC cases using USG and MRI brain to assess the presence of brain abnormalities apart from Chiari II malformation. They reported ventriculomegaly in 56%, small head size in 46%, CC abnormalities in 60%, heterotopias in 11%, and gyral abnormalities in 3% [4]. MRI findings showed good correlation with USG findings in these cases, with agreement between prenatal and postnatal imaging findings. Morais BA et al., previously reported a higher incidence of Chiari II malformation, hydrocephalus, and CC anomalies (89.1%, 94.5%, and 86.4%, respectively) in 37 cases of MMC imaged with MRI brain compared to the present study [9].

Alexiou GA et al., previously noted that supratentorial anomalies are common in MMC, similar to the present study [10]. Among them, 87.5% of MMC cases had lateral ventricular dilatation, 58.3% had CC abnormalities with complete agenesis in 8.3%, 41.6% had interhemispheric fissure widening, 12.5% had colpocephaly, and 8.3% had absent septum, periventricular heterotopia, and white matter abnormalities each. Among the infratentorial abnormalities, they observed Chiari II malformation (54.6%), small size of the posterior fossa (74%), cerebellar hypoplasia (12.5%), brain stem hypoplasia (30%), Chiari I malformation (12.5%), and cervical cord syringomyelia (4.2%). These MRI findings reported by Alexiou GA et al., are comparable to the MRI brain findings of the current study [10]. Ventriculomegaly and Chiari II malformation may be present simultaneously or unrelated to each other [8]. Apart from Chiari II malformation and ventriculomegaly, various cerebral anomalies were noted in 60.7% of patients. This high frequency of cerebral abnormalities seen in open-type spinal dysraphisms is not found in closed forms of spinal dysraphism [11]. There was no statistically significant difference in frequency of intracranial anomalies between males and females in the present study, which is analogous to the previous study by Paschereit F et al., [8].

Morais BM et al., evaluated cerebral anomalies in 37 cases of MMC on MRI [12]. They observed a higher frequency of most intracranial abnormalities associated with MMC compared to this study. They reported Chiari malformation in 94%, hydrocephalus in 93%, CC dysplasia in 69%, absent septum in 38%, colpocephaly in 96%, cortical malformations in 48%, gyral interdigitations in 81%, heterotopia in 16.2%, abnormal white matter maturation in 8%, and brainstem hypoplasia in 82% [12]. However, cerebellar hypoplasia was reported in only 8% of cases compared to 23.8% in the present study.

Open neural tube defects cause modifications in brain growth in the early gestational period, resulting in a spectrum of brain abnormalities, including Chiari II malformation (which is distinctive for MMC), decreased dimension of the cerebellum and posterior fossa, CC dysgenesis particularly affecting the splenium and rostrum, and cortical malformations occurring in varying degrees [13]. Alterations of white matter tracts in MMC can be evaluated with diffusion tensor imaging [13].

Kitov B and Kehayov I reported a case of an adolescent male with lumbosacral MMC since birth who was operated on at two months of age with partial excision of the MMC sac [14]. On long-term follow-up, he developed weakness in both lower limbs and decreased sensation in the right leg along the L4-S1 nerve root supply. MRI revealed Chiari II malformation and low-lying tethered cord with syringomyelia in the spinal cord at C6-D1 levels and ventral epidural lipoma in the lumbosacral spine. It was suggested that MMC can often cause deterioration of clinical symptoms postoperatively. Hence, postoperative clinical and imaging follow-up is required in these patients in the long run [14].

Surgical repair is performed in the early postnatal period to cover the open spinal defect in MMC, and a ventriculoperitoneal shunt is placed for ventriculomegaly [15-18]. High-level spinal defects have a poorer prognosis compared to low-level defects in MMC due to a higher possibility of urinary tract infection and the requirement of shunt revision surgery [1,15]. Operative repair of the spinal defect is also being done globally in the prenatal period in some centers in select cases with a consequent reduction in cerebellar herniation and hydrocephalus [16,17]. However, ventriculomegaly may develop in the early gestational period before fetal surgery is performed [8]. Prenatal imaging with sequential occurrence of congenital malformations, along with genotype study, will improve understanding of various types, causes, and consequences of open spina bifida [8].

### Limitation(s)

The main limitation of the present study was the inability to assess long-term follow-up and prognosis of patients treated surgically. Future prospective studies with clinical and surgical follow-up are recommended to improve understanding of the effect of associated anomalies on motor, sensory, and cognitive functions in MMC patients and to guide therapeutic strategies.

## CONCLUSION(S)

Spinal dysraphism, such as MMC, and cerebral anomalies often occur simultaneously and have an influence on neurological, intellectual, and cognitive functions. Chiari II malformation, corpus callosal anomalies, ventriculomegaly, and syringomyelia have been observed to be more commonly associated with MMC. Therefore, imaging evaluation of both the spine and brain is required for planning surgical treatment and long-term follow-up.

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