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Israël Maoneo,
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# Brain abnormalities associated with myelomeningocele at Kinshasa University Teaching Hospital in the Democratic Republic of the Congo

**Dmien Kamate Kirauli[1], Stéphane Yanda Tongo[2], Blaise Sumbu Matondo[3], Pierre Akilimali Zalagile[4], Aimé Lumaka Zola[5], Prosper Lukusa Tshilobo P.[5], Gilbert Lelo Mananga[6], Larrey Kasereka Kamabu[7], Israël Maoneo[8], Glennie Ntsambi Eba G.[8]**

[1] Paediatric Surgery Department, Kinshasa University Hospital, University of Kinshasa, DRC
[2] Radiology and Medical Imaging Department, Kinshasa University Hospital, University of Kinshasa, DRC
[3] Department of Medical Biology, Kinshasa University Hospital, University of Kinshasa, DRC
[4] Public Health School, University of Kinshasa, DRC
[5] Genetic Unity, Kinshasa University Hospital, University of Kinshasa, DRC
[6] Neurology Department, Neuro-Psycho-Pathology Centre, University of Kinshasa, DRC
[7] Neurosurgery Department, Faculty of Medicine, Catholic University of Graben, DRC
[8] Neurosurgery Department, Kinshasa University Hospital, University of Kinshasa, DRC THAILAND

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Corresponding author:
**Israël Maoneo**

Kinshasa University Hospital, University of Kinshasa, DRC

maoneo@yahoo.fr

## ABSTRACT

**Background**: Myelomeningocele (MMC) is the most prevalent neural tube defect, with significant implications for both health and development. The presence of associated brain abnormalities is crucial in determining the prognosis for affected children, making early detection and accurate diagnosis essential.

**Objective**: This study aimed to evaluate the prevalence of brain abnormalities in children with MMC, assess the accessibility of cranial imaging, and identify the most common associated brain malformations.

**Methods**: A retrospective study was conducted, focusing on children diagnosed with myelomeningocele and admitted to Kinshasa University Teaching Hospital from January 2014 to December 2021. The study reviewed clinical data, including the use of cranial tomography, and identified the frequency and types of brain abnormalities.

**Results**: Thirty-five children with MMC underwent cranial tomography. The average



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age of these patients was 43 days, and the male-to-female ratio was 1.5. Notably, 34.3% of mothers did not undergo any obstetric ultrasound, and only 5.7% of mothers who had an ultrasound received an antenatal diagnosis of MMC. Brain abnormalities were observed in 91.4% of the cases. Hydrocephalus was the most prevalent anomaly (93.7%), with aqueductal stenosis identified as the primary cause (60%). The most common surgical treatment was the simultaneous treatment of MMC with ventriculoperitoneal shunt (44.4%). Postoperative recovery was uneventful in 59.2% of patients.

**Conclusion**: The study highlights the high prevalence of brain abnormalities in children with myelomeningocele, particularly hydrocephalus caused by aqueductal stenosis at Kinshasa University Teaching Hospital. The findings underscore the critical need for improved access to prenatal care and diagnostic imaging to detect congenital malformations early, especially in resource-limited settings like Kinshasa.

## INTRODUCTION

Myelomeningocele (MMC) is the most prevalent form of spinal dysraphism, accounting for approximately 80% of all cases [1,2]. MMC is one of the neural tube defects (NTDs) that develops during the fourth week of embryonic life [3]. The prevalence of MMC varies by region: 0.2 to 0.4 per 1000 live births in the USA [2], 0.6 in China [4], 0.86 in Europe [5], and 1.87 in Malaysia [6]. In Africa, the prevalence is reported to be 1.3, with higher rates observed in Algeria, Ethiopia, Tanzania, and Cameroon [7]. The prognosis of MMC is influenced by the site of the lesion and associated brain abnormalities [8,9]. The condition typically results in a reduction or loss of motor and sensory function in the limbs, as well as loss of control over rectal and bladder function [10,11].

Approximately 75% of MMC cases are associated with hydrocephalus, which remains the leading cause of morbidity and mortality [12-13]. In addition to hydrocephalus, sub- and supratentorial abnormalities can result in difficulties with learning, mathematical calculations, concentration, memory, perception, motor coordination, organization, and reasoning [14-16].

Given the severity of the prognosis, medical termination of pregnancy is often considered in many countries [17-18]. In Kinshasa, MMC is the most common NTD, and its diagnosis is frequently made at birth due to the lack of antenatal diagnosis [19].

While brain abnormalities associated with myelomeningocele have been described globally, no scientific publications exist on these anomalies in the Kinshasa hospital setting. This gap in knowledge justifies the present study. The objectives were to assess the accessibility to cranial imaging (cranioencephalic tomography), determine the proportion of children born with MMC who have brain abnormalities, and inventory the brain anomalies associated with MMC.

## METHODS

### Study design

This is a retrospective study.

### Setting and study period

This study was conducted at the pediatric surgery department of Kinshasa University Hospital from January 2014 to December 2021.

### Study Population and sample

One hundred and two children were admitted for MMC. Out of these, 35 performed the cranioencephalic CT-scan and constituted the study sample.

### Inclusion and exclusion criteria

Children diagnosed with MMC and who had undergone cranioencephalic scanning during the study period were included in the analysis. Exclusion criteria involved children who did not undergo cranioencephalic imaging as part of their diagnostic workup.

### Data Collection and variables

Data were retrospectively extracted from medical records and patient charts available in the pediatric surgery department at Kinshasa University Hospital. Variables collected included demographic information (such as age, sex), clinical presentation, antenatal history, type of myelomeningocele, associated brain abnormalities identified on cranioencephalic imaging, and surgical interventions performed.

The imaging technique used to detect brain abnormalities was the tomography (CT) scans. CT scans were protocoled by radiologists of the University teaching hospital of Kinshasa

### Data Management and Analysis

After collection, data were entered, coded, and stored using Microsoft Excel 2016 software.

Descriptive statistics were employed to summarize and present the collected data. For categorical variables, frequencies and proportions were calculated, and results were presented in tabular form. Continuous variables were summarized by their means, standard deviations, as well as quartiles, minimum, and maximum values.

The statistical analysis was performed using R software version 3.6.3, "Holding the Windsock" (2020-02-29). The Chi-square test was used to compare the proportions of categorical variables, while the student's t-test was applied to compare the means of two continuous variables. A p-value of ≤ 0.05 was considered statistically significant for all tests.

### Ethical Considerations

This study was conducted in compliance with ethical standards and was approved by the Ethics Committee of the School of Public Health, University of Kinshasa (ESP/CE/O21/2024).

## RESULTS

Thirty-five patients were included. A male predominance was observed, with a sex ratio of 1.5:1. Thirty-four percent of the patients were transferred to our hospital between one week and two years after birth.

The average age of fathers was significantly higher than that of mothers ($t = 6.1$; $p < 0.001$). Among 35 fathers, 25 (71.4%) were over 35 years old, compared to 5 of 35 mothers (14.3% ) in the same age group. In terms of occupation, 2 fathers (5.7%) were bureaucrats, 11 held liberal professions (31.4%), and 22 were unemployed (62.9%). Regarding mothers, 28 were housewives (80 %), while 7 (20%) had professional jobs. The distribution of sociodemographic and anthropometric characteristics in the study population is summarized in Table 1.

**Table 1.** Distribution of Sociodemographic and Anthropometric Parameters in the Study Population

| Variables | | Infants | | | Parents age (years) | |
|---|---|---|---|---|---|---|
| | | Age (days) | Birth weight (grams) | Sibling Rank | Father | mother |
| Mean | | 43.23 | 3159.29 | 2.86 | 38.23 | 29.43 |
| Median | | 3.00 | 3000.00 | 2.00 | 39.00 | 29.00 |
| Mode | | 1 | 3000 | 2 | 39 | 32 |
| Standard deviation | | 144.457 | 620.427 | 1.734 | 6.098 | 5.987 |
| Minimum | | 1 | 2000 | 1 | 25 | 21 |
| Maximum | | 730 | 4800 | 8 | 55 | 45 |
| Centiles | 25 | - | 2700.00 | - | - | - |
| | 50 | - | 3000.00 | - | - | - |
| | 75 | - | 3500.00 | - | - | - |

In the medical history of the mothers, we identified one case of pregnancy-induced hypertension, one family history of diabetes mellitus, and one case of a urinary infection during the third trimester. Two mothers reported spontaneous abortions at 3 and 5 months of pregnancy, respectively. No stillbirths were recorded in our series. Additionally, there was no history of neural tube defects or other congenital malformations. Alcohol consumption was reported by 27.7% of fathers and 17.1% of mothers, while 14.3% of mothers reported passive smoking. None of the mothers took folic acid during the periconceptional period, and no consanguineous marriages were reported. Details regarding antenatal diagnosis, birth weight, and birth resuscitation are provided in Table 2.

**Table 2.** Antenatal Diagnosis, Birth Weight, and Birth Resuscitation

| Variables | Frequency | % |
|---|---|---|
| ▪ Obstetric sonography | | |
| Yes | 23 | 65.7 |
| No | 12 | 34.3 |
| ▪ MMC antenatal diagnosis | | |
| Yes | 1 | 2.9 |
| No | 34 | 97.1 |
| ▪ Hydrocephalus antenatal diagnosis | | |
| Yes | 1 | 2.9 |
| No | 34 | 97.1 |
| ▪ Birth weight (grams) | | |
| <2500 | 3 | 8.6 |
| 2500 – 3500 | 24 | 68.6 |
| 3501-4000 | 5 | 14.3 |
| >4000 | 3 | 8.6 |
| ▪ Resuscitation at birth | | |
| Yes | 1 | 2.9 |
| No | 34 | 97.1 |

Among the 35 children in our series, 19 (54.3%) did not show any other apparent malformations apart from MMC. Fifteen children (42.9%) presented with clubfeet, with 14 of them having bilateral clubfeet (2 cases of talus clubfoot and 12 cases of equinovarus clubfoot). The unilateral clubfoot was of the left equinus varus type. Genu valgum, genu varum, and bilateral second and third toe syndactyly were noted separately in three neonates with clubfeet. One child had low-set ears.

Regarding brain abnormalities, three out of the 35 children (8.6%) had a normal cranioencephalic CT scan. The brain anomalies detected in the 32 children are presented in Table 3.

**Table 3.** Brain Abnormalities Associated with Myelomeningocele

| Brain abnormality | n =32 (%) |
|---|---|
| Left univentricular hydrocephalus | 1(3.1) |
| Biventricular hydrocephalus | 2(6.3) |
| Triventricular hydrocephalus due to Sylvius aqueduct stenosis | 18(56.2) |
| Triventricular hydrocephalus due to Sylvius aqueduct stenosis +callosum corpus agenesis | 1 (3.1) |
| Triventricular hydrocephalus due to fourth ventricular mass | 1(3.1) |
| Quadriventricular hydrocephalus due to Chiari I | 2(6.3) |
| Quadriventricular hydrocephalus due to Chiari II | 3(9.3) |
| Quadriventricular hydrocephalus due to Dandy Walker | 2(6.3) |
| Callosum corpus agenesis | 1(3.1) |
| Intermediate holoprosencephaly between lobar and alobar pattern | 1(3.1) |

No surgical procedure was performed on 8 (22.9%) of our patients. Two infants died from respiratory distress before surgery, and 6 were discharged at their parents' request, who wished to prepare financially. The various procedures performed on the patients are presented in Table 4. In 4 patients (11.4%), a ventriculoperitoneal shunt (VPS) was placed 3 to 6 months after the treatment of myelomeningocele (MMC).

**Table 4.** Procedures Performed on Patients [MMC=Myelomeningocele; VPS=Ventriculo-Peritoneal Shunt]

| Perfomed procedures | n=27 (%) |
|---|---|
| Only MMC treatment | 10 (37.1) |
| MMC treatment followed by VPS | 4 (14.8) |
| Simultaneous MMC treatment + VPS | 12 (44.4) |
| VPS followed by interval MMC treatment | 1 (3.7) |

**Intra-hospital post-therapeutic evolution**

Of the 27 patients who underwent surgery, 16 (59.2%) had an uneventful postoperative recovery. Simpler outcomes were more commonly observed in patients who received both myelomeningocele (MMC) treatment and ventriculoperitoneal shunt placement under the same anesthesia. Hydrocephalus, meningitis, and surgical wound dehiscence were more frequently observed in patients who underwent MMC treatment without the addition of a ventriculoperitoneal shunt. Two patients died: one from meningitis and the other from a combination of meningitis, hydrocephalus, and surgical wound dehiscence. Postoperative complications are summarized in Table 5 below.

**Table 5.** Postoperative Events

| Postoperative event | Only MMC treatment n=10 | MMC treatment followed by VPS n=4 | Simultaneous MMC Treatment +VPS n=12 | VPS followed by interval MMC treatment n=1 | TOTAL |
|---|---|---|---|---|---|
| Uneventful postoperative | 3 | 3 | 9 | 1 | 16 |
| Hydrocephalus | 3 | 0 | 1 | 0 | 4 |
| Surgical wound dehiscence | 0 | 0 | 1 | 0 | 1 |
| Surgical wound dehiscence +hydrocephalus | 2 | 0 | 0 | 0 | 2 |
| Meningitis | 0 | 1 | 0 | 0 | 1 |
| Meningitis+hydrocephalus | 1 | 0 | 0 | 0 | 1 |
| Death | 1 | 0 | 1 | 0 | 2 |

The average time from admission to scan diagnosis and the average time from scan diagnosis to surgery were significantly correlated with the average length of hospital stay ($t = 7.46$; $p < 0.001$, $t = 2.78$; $p < 0.001$ respectively). As these delays increased, the length of hospital stay also lengthened. The distribution of scan diagnosis time, surgical time, and hospital stay is summarized in Table 6.

**Table 6.** Distribution of Scan Diagnosis Time, Surgical Time, and Hospital Stay in the Study Population

| Variables | Scan diagnosis delay (days) | Operating delay (days) | Hospital stay (days) |
|---|---|---|---|
| Mean | 40.000 | 33.6571 | 90.029 |
| Median | 30.000 | 34.0000 | 90.000 |
| Mode | 30.00 | 34.00 | 90.000 |
| Standard deviation | 42.609 | 20.11576 | 49.249 |
| Minimum | 2.000 | 2.00 | 29.000 |
| Maximum | 220.000 | 80.00 | 266.000 |

**Out-of-hospital post-therapeutic evolution**

The phone calls concerned 31 children (88.6%). The remaining four children (11.4%) had already died: two awaiting surgery and two others postoperatively. Among the 31 children to whom we made the phone calls, 7 calls (22.6%) were conclusive, of which one child walked alone without anal incontinence but with urinary incontinence, and six children were declared dead. Twenty-four appeals (77.4%) were inconclusive. Indeed, for 16 calls the phones did not go through while for 8 calls, the parents did not recognize the children.

Based on various socio-demographic, clinical, paraclinical, and therapeutic parameters, we have categorized our patients according to vital outcomes in Table 7.

**Table 7.** Distribution of the Study Population According to Vital Outcome

| Variables | Modality | Dependant: vital infant outcome | |
|---|---|---|---|
| | | Alive | Dead |
| Age (days) | Mean (SD) | 57.5 (169.6) | 7.5 (11.6) |
| Sibling rank | Mean (SD) | 2.8 (1.7) | 3.1 (1.8) |
| Sex | Male | 15 (71.4) | 6 (28.6) |
| | Female | 10 (71.4) | 4 (28.6) |
| Father's age (years) | Mean (SD) | 38.1 (6.1) | 38.6 (6.5) |
| Mother's age (years) | Mean (SD) | 29.2 (5.8) | 29.9 (6.6) |
| Father's alcohol use | No | 19 (73.1) | 7 (26.9) |
| | Yes | 6 (66.7) | 3 (33.3) |
| Mother's alcohol use | No | 20 (69.0) | 9 (31.0) |
| | Yes | 5 (83.3) | 1 (16.7) |
| Father's tobacco history | No | 21 (70.0) | 9 (30.0) |
| | Yes | 4 (80.0) | 1 (20.0) |
| Spontaneous abortion | No | 23 (69.7) | 10 (30.3) |
| | Yes | 2 (100.0) | 0 (0.0) |
| Surgical procedure | Yes | 20 (74.1) | 8 (25.9) |
| | No | 5 (62.5) | 2 (37.5) |
| Hospital stay (days) | Mean(SD) | 90,3 (41.3) | 89,3 (68.0) |
| Operating delay (days) | Mean(SD) | 34,4 (20.5) | 31,8 (20.2) |
| Scan diagnosis delay (days) | Mean(SD) | 38,4 (31.3) | 44,0 (64.9) |

## DISCUSSION

This study involved 35 newborns with myelomeningocele. Among the mothers, 34.3% did not have access to obstetric ultrasound. Among those who had at least one obstetric ultrasound, only two (5.7%) had an antenatal diagnosis of myelomeningocele or hydrocephalus. Cranioencephalic CT scans revealed brain malformations in 91.4% of children with hydrocephalus, primarily due to stenosis of the Sylvian aqueduct.

### 1. Sociodemographic data

At admission, our patients had an average age of 43 days while surgical treatment of myelomeningocele must be performed within 48 to 72 hours after [20]. The mystical consideration of congenital malformations, the absence of a reference system and the difficulties for parents to pay for the care of their children may be the main causes behind this late arrival. The mothers of our patients had an average age of 29 years. This average age is the same found by Komolafe in Nigeria [21]. It aligns with the average age of motherhood reported in the USA by Martin [22] and in Europe by Fertility Statistics [23], though it is higher than the 26.9 years reported by Gedefaw [27] in Ethiopia. Almost all the mothers (91.4%) were aged between 20 and 35 years, similar to the results of Gedefaw [24] and Talebian [25], who reported 87.7% and 82.0%, respectively, in this age group. Mothers aged 40 or older represented 8.5% of the sample. A meta-analysis by Vieira [26] showed that maternal age ≥40 years is associated with an increased risk of myelomeningocele.

### 2. Clinical Data

No family history of neural tube defects was reported in our study. Most cases of myelomeningoceles are considered sporadic [2,27-28], although some authors have documented familial occurrences, including Gedefaw [24] (2.7%), Al-Wassia [29] (5.4%), Omer [30] (10.7%), and Forci [31] (6.8%). A follow-up study by Dupépé [32] found 17.7% of cases with a family history of neural tube defects. Our study recorded 6% of mothers with a history of spontaneous abortion, which is considered a risk factor for neural tube defects in subsequent pregnancies [33-35]. However, Lu [36] argued that abortion alone does not constitute a risk unless associated with a short interpregnancy interval (less than 6 months). A significant proportion of the mothers in our study (34.3%) did not have access to obstetric ultrasound, despite being in the capital city.

The rate of antenatal diagnosis of myelomeningocele in our study was 2.8% (1 in 35), which is much lower than the rates reported by other studies (63–100%) in Africa, Asia and Europe and [27,31,37-38]. Several factors may explain this low

rate, including the absence of obstetric ultrasound for 34.3% of mothers in our study, as well as the poor quality of ultrasound equipment and limited operator experience.

### 3. Paraclinical Data

Brain abnormalities were present in 91.4% of our patients. Paschereit [39] and Schneider [40] reported brain abnormalities in 95% and 100% of patients, respectively. Hydrocephalus was the most common brain abnormality (90.6%) in our study. It was also the most frequently observed abnormality in several studies [39,4-43]. Stenosis of the Sylvian aqueduct was the leading cause of hydrocephalus (63.3%) in our study. According to Kaur [44], stenosis of the Sylvian aqueduct is the most common cause of congenital hydrocephalus. Warf [45] reported a high frequency of this stenosis, which was associated with myelomeningocele in 82.7% of cases. On the other hand, Arnold Chiari Type II malformation was found in 9.3% of our patients. This rate is significantly lower than those reported by several authors: 36% for Paschereit [39], 67% for Alexiou [46], 89% for Morais [42], 90% for Spoor [43], and 95% for Kuhn [47]. All the authors we referenced used magnetic resonance imaging (MRI) to identify brain abnormalities. In the absence of MRI, we relied on cranioencephalic scanning to detect brain abnormalities. MRI has been available in the city of Kinshasa for about ten years, but it remains inaccessible to many parents of children who require proper sedation for the exam. This justifies our use of cranioencephalic scanning, which was performed more than four weeks after admission for more than half of our patients.

### 4. Therapeutic Data

The rate of CSF shunting in our study (56.7%) is significantly lower than that reported in several studies [40, 48-50]. Many families of our patients still hold a negative cultural perception of surgical interventions, particularly those involving the newborn's central nervous system. Additionally, since our hospital was not subsidized during the study period, parents of children born with myelomeningocele and hydrocephalus were unable to afford emergency care. This led to delayed diagnoses, prolonged surgery delays, and extended hospital stays.

While some authors recommend delaying CSF shunting as much as possible to ensure its sterility [27, 45], most myelomeningoceles are ulcerated. These authors are able to monitor their patients clinically and paraclinically, ensuring prompt CSF drainage once the operative indication is established. In our study, twelve patients (44.4%) had simultaneous repair of myelomeningocele with ventriculoperitoneal shunt, with a simple postoperative follow-up for 75% of these patients. Postoperative complications, including hydrocephalus, meningitis, wound dehiscence, and death, were more common in the group of children who underwent myelomeningocele repair without CSF shunting. In our setting, simultaneous treatment of hydrocephalus and myelomeningocele seems to be the most appropriate approach.

Two years after the discharge of the last patient in our series, we recorded 10 deaths (28.5%), including preoperative and postoperative deaths. This mortality rate may be underestimated for many reasons. Most parents (45.7 %) were not reached during telephone follow-up. Some parents (22.9%) did not recognize the children for whom they were contacted. The reassignment of phone numbers by telecommunications companies may explain why we reached someone other than the child's parent. Additionally, many parents leave the hospital with the understanding that they will pay their bills at subsequent appointments, which they do not attend. Some parents may also pretend to have lost contact with their child, particularly if the child has passed away.

Since October 2023, the national government has covered the costs of prenatal consultations, related paraclinical exams, and childbirth under its Universal Health Coverage (UHC) program. Child care is also covered under the UHC. There should be a particular emphasis on acquiring adequate equipment and providing training or retraining for staff responsible for pregnant women and newborns. This will increase the rate of antenatal diagnosis, allowing parents to be psychologically prepared for the birth of a malformed child. It will also enable medical staff to better prepare for the newborn's care and perform surgery within a reasonable timeframe.

## Limitations

This was a retrospective and single- center study. Other limitations were the use of CT scan instead of

MRI, the small sample including only children with MMC who underwent CT-scan and the large proportion of children whose parental phone calls were inconclusive.

## Conclusion

This study highlighted that MMC occurs in children who lack access to basic healthcare, especially access to antenatal medical imaging for detecting congenital malformations. Brain abnormalities were found in almost all of the children, with hydrocephalus due to aqueductal stenosis being the most frequent malformation.

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