

# Prevalence and CT-Scan Presentations of Brain Malformations in Children at a University-Affiliated Mother and Child Hospital (Cameroon)

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## Abstract

**Background:** Brain malformations (BMs) are congenital abnormalities of the shape or structure of the brain, with corpus callosum agenesis known as the most frequent. Diagnosis of most BMs can be prenatal with ultrasound and fetal magnetic resonance imaging (MRI); post-natal diagnosis is based on transfontanelar ultrasound, CT-scan, and head MRI which is the imaging gold standard technique. MRI has been recently introduced and the CT-scan was previously the reference technique for the diagnosis of BMs in our context. Almost no publication has been made in sub-Saharan Africa on the clinical and CT scan characteristics of Brain malformations in children. **Objective:** The aim of this study was, in the absence of MRI, to describe the clinical and CT-scan presentations of brain malformations in children at the Yaounde Gynaeco-Obstetric and Pediatric Hospital (YGOPH). **Patients and method:** This was a cross-sectional descriptive study conducted from February to May 2016 at the YGOPH, including all children of 15-year-old and less with BM diagnosed on CT-scan at YGOPH between April 2006 and March 2016. The studied variables were clinical (age at diagnosis, sex, clinical manifestations) and CT findings (type of cerebral malformation). The data was analyzed using the SPSS 20.0 software with the estimation of hospital prevalence of BMs, frequencies and associations. The chi-square test was used to seek for an association between variables. The threshold of significance was  $p < 0.05$ . **Results:** The prevalence of BMs was 0.52%, with 29 cases of BMs identified out of 5590 patients followed up at the pediatric neurology outpatient unit over a period

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of 10 years. The mean age at diagnosis was 37.2 months (3.1 years) and the most represented age groups were 0 - 1 year (37.9%) and 1 - 5 years (37.9%). The sex ratio was 0.81 (55.2% girls). The clinical presentation was represented by neurological signs (93.1%) with convulsions (65.5%) and psychomotor retardation (58.8%) associated with skin lesions (34.5%) and/or facial dysmorphic features (27.6%). BM was suspected on antenatal ultrasound in 14.3% of cases (4/28). Abnormalities of cortical development accounted for 65.5% of BM followed by abnormalities of brain separation (31%). Tuberous sclerosis complex was the most common BM (31%) followed by agenesis of the corpus callosum (27.6%). The presence of dysmorphic facial features was associated ( $p = 0.007$ ) with disorders of brain separation (DBS) while the presence of cutaneous lesions was associated ( $p = 0.013$ ) with anomalies of the cortical development (ACD) especially tuberous sclerosis complex. **Conclusion:** BMs are infrequent, dominated by tuberous sclerosis complex and agenesis of the corpus callosum. They are mainly revealed by convulsions and psychomotor retardation. Efforts are needed to improve antenatal diagnosis and facilitate access to cerebral MRI.

## Keywords

Cerebral Malformations, Psychomotor Retardation, Convulsions, Tuberous Sclerosis Complex, Corpus Callosum Agenesis, Cerebral CT Scan, Sub-Saharan Africa

## 1. Introduction

Brain malformations (BMs) are congenital abnormalities of the shape or structure of the brain. They represent 9% of isolated malformations and are found in 16% of polymalformative syndromes [1]. The most frequent BM is agenesis of the corpus callosum (CCA) [2] [3]. These BMs are due to a halt or a modification of brain development during intrauterine life, regardless of the etiology. BM classification incorporates morphological and genetic criteria [4] [5] [6]. The neural tube closure defects (NTCD), rhombencephalon development anomalies (RDA), disorders of brain separation (DBS), cortical development anomalies (CDA), and growth anomalies (GA) are distinguished [7] [8].

Diagnosis of most BMs can be prenatal with ultrasound and fetal magnetic resonance imaging (MRI) [9] [10]. Post-natal diagnosis is based on transfontanelar ultrasound, CT scan, and head MRI which is the imaging gold standard technique [11].

The clinical manifestations of BM are variable, ranging from refractory epilepsy to neurological and cognitive disorders of various severity, in relation to the stage of onset of disorders and the extent of malformation [12].

In Africa, most studies of BM are performed in North Africa where cortical development anomalies are the most frequent [7]. In Cameroon, BMs are often discovered during investigation for epilepsy or psychomotor retardation [13]

[14]. MRI has been recently introduced and the CT scan was previously the reference technique for the diagnosis of BM in our context. To the best of our knowledge, no publication has been made in sub-Saharan Africa on the clinical and CT scan characteristics of BMs in children. The purpose of this study was to describe the clinical and CT-scan aspects of child malformations in the Yaounde Gynaeco-Obstetric and Pediatric Hospital (YGOPH), where there is a specialized pediatric neurology unit.

## 2. Patients and Methods

This was a cross-sectional descriptive study conducted in YGOPH's Radiology and Pediatric Neurology units from February to May 2016.

### 2.1. Study Population and Procedure

It included all children aged 0 - 15 years with brain malformation (BM) confirmed by CT scan between April 2006 and March 2016 at HGOPY.

As a first step, we identified all the medical records of BMs among the files of pediatric neurology outpatients from April 2006 to March 2016. Then we contacted their parents by phone to set an appointment at the outpatient consultation in order to investigate the past history and conduct a physical examination of the child. We then retrieved from the medical records of these children data on presenting complaints, initial clinical examination, CT reports and images, and the evolution since diagnosis. All CT scans were reviewed by a pediatric radiologist and a neuropsychiatrist of at least ten years' experience. All information was recorded on a pre-conceived data collection sheet.

### 2.2. Studied Variables

The clinical data included: age of child at diagnosis of BM, sex, circumstances of diagnosis, prenatal and postnatal history, psychomotor development, convulsions, skin lesions, facial dysmorphic features and other clinical signs. CT-scan data were grouped into neural tube closure defects (NTCD), rhombencephalon development anomalies (RDA), disorders of brain separation (DBS), cortical development anomalies (CDA), growth anomalies (GA), and associated anomalies. Data on other paraclinical examinations, treatment and progression were also noted.

### 2.3. Statistical Analysis

The collected data was recorded using the CSpPro 6 software and analyzed in the SPSS 20.0 software. We used descriptive statistics tools with the calculation of frequencies and associations. The chi-square test was used to find an association between the variables; the significance threshold was  $p < 0.05$ .

### 2.4. Ethical Considerations

The study was approved by the Ethics Committee of the Faculty of Medicine and

Biomedical Sciences and the Institutional Ethics and Research Committee of the YGOPH. The confidentiality of the information and the informed consent (verbal) of the patients' careers have been assured.

### 3. Results

#### 3.1. Prevalence of Brain Malformations and Characteristics of the Study Population

Of the 5590 children in Pediatric Neurology outpatient unit at YGOPH between April 2006 and March 2016, 29 had a brain malformation, giving an in-hospital prevalence of 0.52%. Girls accounted for 55.2% of the population (16/29), giving a sex ratio of 0.81. Children's ages at diagnosis varied from 2 months to 156 months (13 years) with an average age at diagnosis of 37.2 months (3.1 years). The age distribution was 37.9% for children aged less than one year, 37.9% for those aged 1 to 5, and 24.2% for those over 5 years of age. Antenatal suspicion of BM was reported in 4 fetuses (14.3%) after discovery of hydrocephalus (2 cases) and microcephaly (2 cases).

#### 3.2. Clinical Aspects

Psychomotor retardation (PMR) was the main reason for consultation leading to the discovery of BM, followed by convulsions (31%). Concerning clinical examination, 14 patients (48.3%) had microcephaly and eight (27.58%) had signs of facial dysmorphism (hypotelorism, hypertelorism, low set ears). The cutaneous signs (achromic nevus, shagreen patch) were present in 10 patients that is 34.5%. Twenty-seven patients (93.1%) had neurological signs (**Table 1**) dominated by convulsions (65.5%) and psychomotor retardation (58.8%).

#### 3.3. CT Scanning

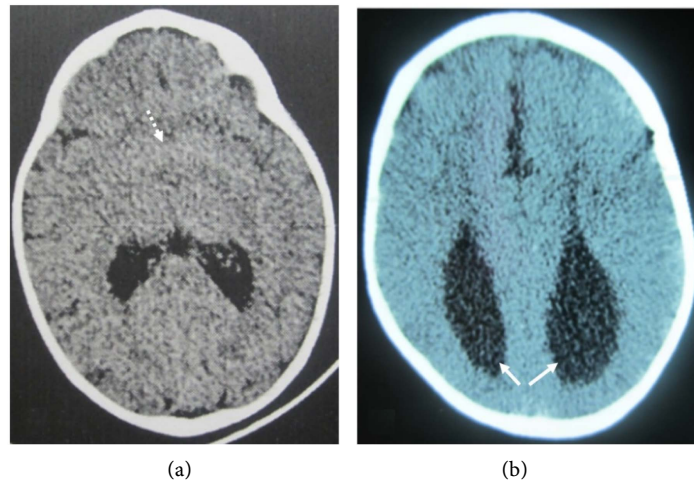
Cortical development abnormalities (CDA) were the most common BM group (**Table 2**) with 19 cases (65.5%).

The main BMs (**Table 2** and **Figures 1-3**) were tuberous sclerosis complex (TSC) with 31%, followed by agenesis of the corpus callosum (ACC) with 27.6% and pachygyria (20.7%). **Figure 4** shows the Dandy-Walker malformation.

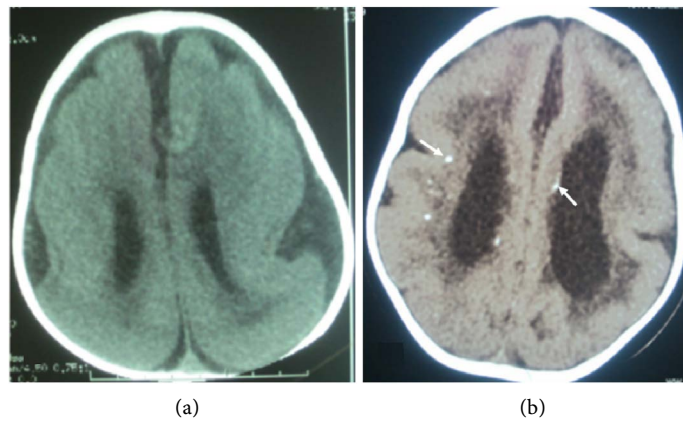
**Table 1.** Neurological signs.

Neurological signs	Number	Percentage (%)
Convulsions	19	65.5
Psychomotor retardation	17	58.8
Spastic tetraparesis	9	31.0
Axial hypotonia	4	13.8
Peripheral hypotonia	3	10.3
School delay	3	10.3
Other*	3	10.3

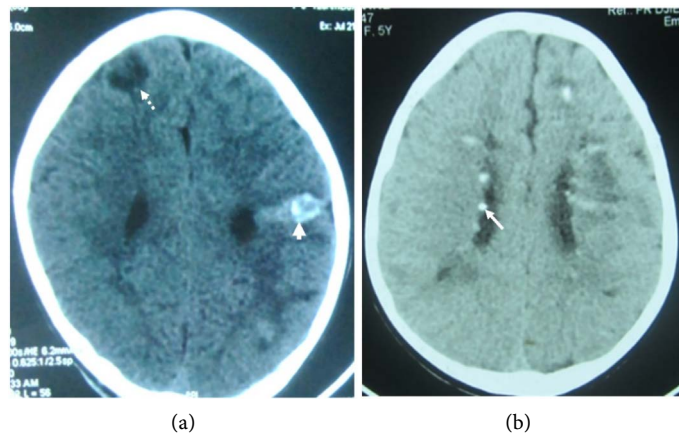
\*Other: absence from school (2 cases) walking disorders (one case).



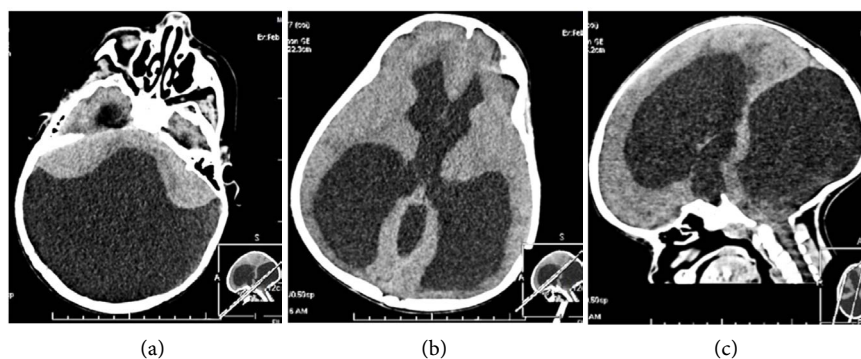
**Figure 1.** Cerebral CT scan showing brain separation abnormalities. (a) lobar holoprosencephaly with fusion of the frontal lobes (dot arrow); (b) colpocephaly (white arrows) in corpus callosum agenesis (CCA).



**Figure 2.** Cerebral scan; (a) lissencephaly (smooth brain with figure eight appearance); (b) pachygyria (thick cortex) with associated sub-ependymal and parenchymal calcifications (proved CMV infection).



**Figure 3.** Cerebral scan showing the lesions characteristic of Tuberous Sclerosis Complex: calcified sub-ependymal nodules (arrow), non-calcified cortical tubers (dot arrow) and calcified cortical tubers (arrow head).



**Figure 4.** Cerebral CT scan showing a Dandy-Walker malformation: (a) agenesis of the vermis; (b) triventricular hydrocephalus; (c) cystic expansion of V4, and ascent of the tentorium cerebelli.

**Table 2.** Distribution of different types of cerebral malformations.

Group of BM	Type of brain malformations	Number	%
CDA (n = 19)	Tuberous sclerosis complex	9	31
	Pachygyria	6	20.7
	Lissencephaly	3	10.3
	Schizencephaly	1	3.4
DBS (n = 9)	Agenesis of the corpus callosum (ACC)	8	27.6
	Holoprosencephaly	1	3.4
RDA (n = 3)	Agenesis of the vermis	2	6.9
	Dandy walker	1	3.4
GA (n = 1)	Hydrocephalus	1	3.4

Cortical Development Anomalies (CDA); Disorders of Brain Separation (DBS); Rhombencephalon Development Anomalies (RDA); Growth Anomalies (GA).

### 3.4. Associations

A statistically significant relationship was found between the group of patients with disorders of brain separation (DBS) and the presence of dysmorphic features ( $p = 0.007$ ). A statistically significant relation was found ( $p = 0.013$ ) between the anomalies of the cortical development (ACD) and the presence of cutaneous lesions, more particularly with tuberous sclerosis complex.

### 3.5. Evolution Aspects

Antiepileptic treatment was instituted in 65.5% of patients (19/29). However, epileptic seizure control was not yet effective in 13 of 19 patients (68.4% of patients on treatment). The mortality rate was 20.7% that is 6 patients (3 cases of lissencephaly, 1 case of TSC, 1 case of ACC and 1 case of holoprosencephaly).

## 4. Discussion

Over 10 years period (2006-2016) we have check off 29 cases of brain malforma-

tions with a hospital prevalence of 0.52% in neuropsychiatric consultations. Our result remains coherent with the frequency of malformations in the nervous system at birth which is 5 - 10 per 1000 births [4]. This group of infrequent affections has been the subject of several studies in particular in Morocco and Tunisia [8] [11].

The age of the patients in our study varied from 2 months to 13 years, similar to that found in Tunisia by Hammou *et al.* in 2002, which was 1 month to 16 years [8]. The mean age at diagnosis of BM in our study was 3.1 years. This result differs from that found by Houmada *et al.* [11] in 2015 in Morocco with patients of 7 days to 4 years old.

The female sex was predominant in our study. Houmada and Bennouna in Morocco in 2015 [7] [11] found the same observation. On the other hand, Hammou *et al.* [8] found a male predominance. The sex ratio in BMs varies according to the type of study and BM. Thus, Chateil *et al.* in France found a male predominance in agyria-pachygyria [15].

The main presenting complaints leading to discovery of BM were psychomotor retardation and convulsions as in the studies of Bennouna *et al.* [7] and Hammou *et al.* [8]. Indeed, in these studies the most frequent BMs were cortical development anomalies, and these BMs are known causes of refractory epilepsy [16] [17].

BMs were suspected in antenatal period in 14.3% of the fetuses with hydrocephalus or microcephaly. This low rate raises questions concerning antenatal screening for BM, which are more often post-natal discoveries following clinical manifestations. It is recommended that at least three obstetric ultrasounds be performed, one in each trimester of pregnancy at a specified time, in order to evaluate, among other things, the development and morphology of CNS structures [18]. And in some cases fetal MRI and genetic analysis are recommended [19] in addition of fetal ultrasound.

Microcephaly was found in 48.3% of our patients. This microcephaly may result either from decreased neuronal and glial proliferation in germinal areas or from increased apoptosis [20]. Its antenatal discovery must draw our attention and prompt investigation of an embryofetopathy.

Neurological signs dominated the clinical picture in our study. It was essentially convulsions, psychomotor retardation and tone disorders. Convulsions and psychomotor retardation are the most commonly reported clinical signs in the series of brain malformations [8] [11] [20] [21]. These signs were sometimes associated with cutaneous abnormalities and dysmorphic signs. Indeed, facial dysmorphism can be found in certain brain malformations, especially those of the median line [19] [22]. Similarly, BMs such as tuberous sclerosis complex are associated with cutaneous abnormalities such as the appearance of shagreen patches, hypomelanotic spots [23] [24] [25].

Anomalies of cortical development were most frequent same as in Bennouna *et al.* [7] study. Hammou *et al.* [8] found a clear predominance of growth anomalies followed by anomalies of cortical development. In addition, TSC was the

most common BM in our study followed by ACC. This predominance of TSC compared to ACC in our study can be explained by the fact that ACC can remain asymptomatic for a long time and may be accidentally discovered [2] unlike the TSC, where seizures are often forefront features [23] [24].

As limitations: some BMs such as heterotopia or focal dysplasia are difficult to diagnose with CT-scan and can be missed; retrospective review of patient files is source of missing or incomplete data.

## 5. Conclusion

Brain malformations are uncommon in our setting with an in-hospital prevalence of 0.52%. They are dominated by the TSC and ACC. The main clinical manifestations are psychomotor retardation and seizures associated or not with dysmorphic and cutaneous signs. The rate of antenatal suspicion of BM is low.

## Conflict of Interest

The authors state that they have no conflict of interest in relation to this study.

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