

Major Malformations of the Central Nervous System as seen at The University Teaching Hospital, Ilorin

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Summary

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Background: The pattern of central nervous system (CNS) malformations seen at the University Teaching Hospital, Ilorin in the middle belt of Nigeria may be different from that seen in southern Nigeria and elsewhere.

Objectives: To present the major types of CNS malformations encountered in our centre over a five-year period.

Methods: A prospective study of all cases of CNS malformations seen on the neonatal and children's wards over a period of five years (2000-2005) at the University of Ilorin Teaching Hospital, Ilorin, was carried out. Diagnosis of the malformations was based on detailed history, clinical, as well as neurological and radiological evaluations. The data obtained were entered onto a proforma during hospitalization and follow-up, and were later analysed.

Results: One hundred and thirty five children (76 males and 59 females) were seen during the period. Spina bifida cystica (SBC) was the commonest among eight major malformations of the CNS encountered, accounting for 61 (45.2 percent) in the 135 children. The commonest (73.8 percent) form, a closed myelomeningocele occurred in isolation in 30 cases and co-existed with hydrocephalus in 15 other infants suspected to have Arnold Chiari type 2 malformation. SBC was most frequently located over the lumbosacral (74 percent) segment of the spine. The death of three children who developed meningitis or ventriculitis following rupture of myelomeningocele, accounted for the postoperative mortality rate of 6.7 percent among the infants with spina bifida. Hydrocephalus, the second commonest anomaly (50; 37 percent) was largely due to aqueduct stenosis (68 percent). Many of the affected infants presented late with grotesque head enlargement and varying degrees of visual failure. Death following shunting occurred in two children, a surgical mortality rate of 5.4 percent. Encephalocele which was predominantly occipital (91.7 percent) constituted 8.9 percent of the total, while craniosynostosis (five cases), subgaleal inclusion cyst (three), microcephaly (two), porencephaly (one) and hydranencephaly (one) constituted less common anomalies. Of the 103 (76.3 percent) children who underwent surgical intervention, six died, resulting in an overall surgical mortality rate of 5.8 percent.

Conclusions: The commonest central nervous system malformation encountered in this centre was a spina bifida cystica followed by hydrocephalus. Parental socioeconomic status militated against standard management of many children with CNS malformations in this series.

Introduction

THE complicated development of the central nervous system (CNS) involves the delicate neuro-embryological interplay between the brain and the

spinal cord as well as the associated structures such as the skull, vertebrae, meninges and the vasculature.¹ Congenital malformations of the system are therefore among the most common anomalies.² Although most aberrations of embryonic development related to the CNS are evident at birth or shortly after, the true incidences of many CNS malformations in the general population are not known¹ and the pattern of distribution appear to vary with geographical locations. The incidence of hydrocephalus occurring as a single congenital disorder in the US populations, is reported to be 0.9 to 1.5 per 1000 births and that

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of hydrocephalus occurring with spina bifida, from 1.3 to 2.9 per 1000 births.³ In most countries, the incidence of open neural tube defects had dropped remarkably; that of the United Kingdom,⁴ from 2 per 1000 to 0.6 per 1000 live births.⁵ In most indigenous African countries, the incidence of CNS malformations is unknown, because infant mortality records are scanty, live and still births are underreported, autopsy examination is grossly deficient, and published reports are based on hospital experience rather than population studies, with variation in the hospital population producing different results. For example, early documentations on CNS malformations in Nigeria revealed varying incidences of 1.37 per 1,000 live births in Lagos⁷ and 5.2 per 1000 live and still births in Ibadan⁶ with anencephaly and hydrocephalus being the respective commonest anomalies in these locations. Hospital-based studies in tertiary centres located in the southern^{8,9} and northern¹⁰ regions of Nigeria have also established geographical variation in the pattern of distribution of CNS anomalies, the most frequent malformation being hydrocephalus in the south and myelomeningocele in the north;⁸⁻¹⁰ the pattern in the middle belt has not been documented. The present study was undertaken in order to establish the major types of malformations of the CNS encountered at the University Teaching Hospital, Ilorin, a tertiary health centre that serves a large proportion of the middle belt of Nigeria.

Subjects and Methods

One hundred and thirty five consecutive children with central nervous system malformations managed by

the neurosurgical unit of this hospital between October 2000 and September 2005 were studied prospectively. Only children that were admitted into the paediatric and neonatal ward were included in the study. Children aged more than three years who had head enlargement were excluded as were those whose enlargement followed meningitis or tumours. A detailed family, antenatal, intra-partum and postnatal history was obtained. In each patient, diagnosis was based on detailed clinical examination, with particular emphasis on the central nervous system. The head size (in cm), was taken as the greatest occipito-frontal circumference. When and where indicated, radiological evaluations included trans-fontanelle ultrasound brain scan, ultrasound scans of cystic back lesions and abdomen, skull and spine radiographs, and in a few cases, cranial computed tomography, depending largely on parental economic status. In children with the 'small head syndrome', microcephaly was diagnosed in those who had abnormally small, though symmetrical heads with head circumference greater than 2 standard deviations below the mean for sex and gestational age, while craniosynostosis was diagnosed in infants with often asymmetrical small heads showing features of early closure of cranial sutures. Appropriate surgical treatment which was offered to each patient was refused by a few parents for various reasons. Children who underwent surgical management were followed up for a variable period of six months to four years. Their clinical data were entered consecutively onto a pre-designed proforma during hospitalization and follow up.

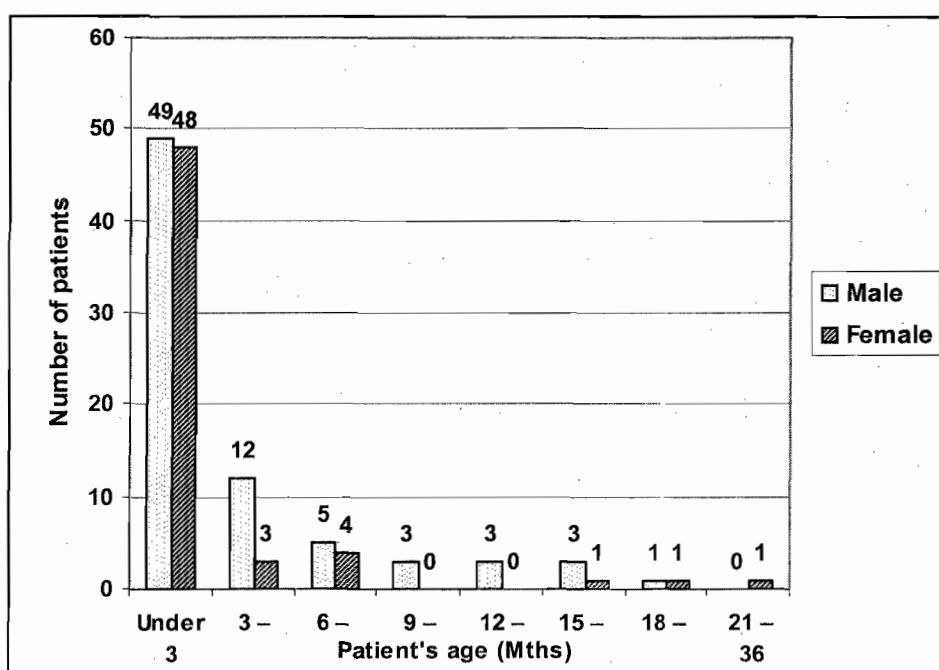


Fig. 1: Age and sex distribution of children with central nervous system malformations

Results

Frequency of malformations

Over the five-year period, eight major malformations of the central nervous system (CNS) were encountered in 135 children comprising 76 males and 59 females aged between 12 hours and three years (Table 1; Fig. 1). Over 90 percent of the children were aged six months and below (Fig. 1). Yearly admissions increased from six children in 2000 through 12 (2001), 18 (2002), 29 (2003), 32 (2004) to 38 children managed in 2005. The commonest malformations of the CNS encountered were spina bifida cystica, hydrocephalus and encephalocele. Less

common malformations were craniosynostosis, porencephaly, microcephaly and hydranencephaly. Of the 135 children, 103 (76.3 percent) underwent appropriate neurosurgical procedures, while others declined surgical intervention predominantly because of financial constraint; they sought alternative and probably less costly therapies elsewhere.

Characteristics of each malformation

Spina bifida cystica

Spina bifida cystica which was the commonest malformation encountered, occurred in 61 children consisting of 32 males and 29 females (Table I) and

Table 1

Major Central Nervous System Malformations seen at the University Teaching Hospital, Ilorin

Type of Malformation	Sex and Number		Total	Percent of Total	No. who had Surgery	Deaths (Mortality Rate %)
	Male	Female				
Spina bifida cystica	32	29	61	45.2	45	3(6.7)
Hydrocephalus	30	20	50	37.0	37	2(5.4)
Encephalocele	7	5	12	8.9	12	1(8.3)
Craniosynostosis	3	2	5	3.7	4	-
Subgaleal inclusion cyst	2	1	3	2.2	3	-
Microcephaly	1	1	2	1.5	-	-
Hydranencephaly	1	0	1	0.75	0	-
Porencephaly	0	1	0.75	1	-	-
Total	76(56.3%)	59(43.7%)	135	100.0	103(76.2%)	6(5.7)

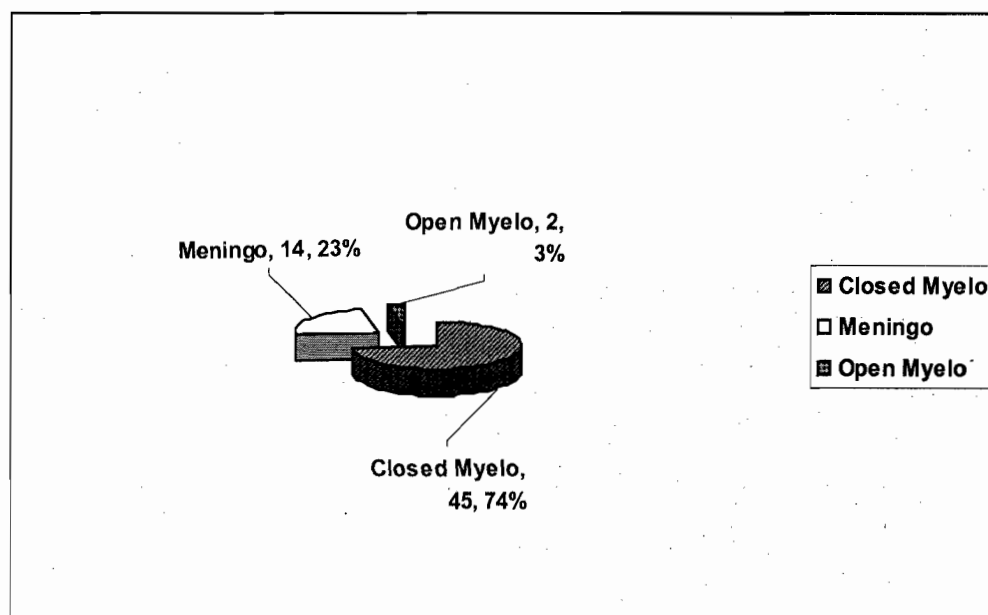


Fig. 2: Types of spina bifida cystica encountered
[Myelo = myelomeningocele, Meningo = meningocele]

constituting 45.2 percent of all malformations. Majority (74 percent) of the lesions, which were myelomeningocele (77 percent) and meningocele (23 percent) were sited over the lumbo-sacral segment of the spine, the cervical region being the least affected (Figs. 2 & 3). Myelomeningocele (77 percent) was either closed (74 percent) or open (three percent). Clinical diagnosis was easy; the child presented with a midline lesion at the back that had been present since birth, and weak lower extremities with or without incontinence of urine and faeces. Profound sensory loss in the lower limbs was complicated by deep decubitus ulcers of the gluteal region in one case (Fig.

Hydrocephalus

Isolated hydrocephalus, the second most common CNS malformation, was encountered in 30 males and 20 females, constituting 37 percent of the total (Table 1). Forty percent of the children presented either at birth, neonatal period, or between the first and third months of life, while 50 percent were seen between the 4th and 12th months of life. Their ages ranged between one day and three years (mean \pm SD of 15.00 ± 3.55 months). They presented most commonly with progressive head enlargement, sunseting eyes, vomiting, convulsion, paraparesis, wasting and varying degrees of visual loss or

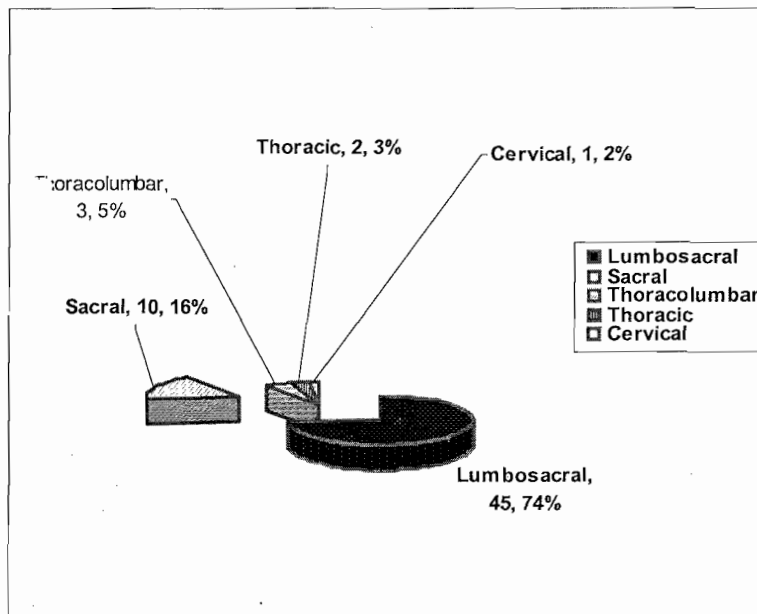


Fig. 3: Locations of spina bifida cystica along the vertebral column in 61 children

4). Myelomeningocele was associated with hydrocephalus in 15 infants with suspected Chiari type-2 malformation. The sac had ruptured before presentation in six cases, four had developed meningitis and one ventriculitis. Of the 61 infants, 45 (74 percent) comprising 14 and 31 cases of meningocele and closed myelomeningocele respectively, underwent surgical repair and closure of their lesions. In addition to repair, children with complicating hydrocephalus were shunted using Chhabra slit-in-spring ventriculo-peritoneal shunt system (*G Surgwear, India*). Complications following spina bifida repair included cerebrospinal fluid leak, wound sepsis and wound dehiscence in two infants each. The infants with open myelomeningocele were not offered surgery because their cord lesions were high, exposed and thus potentially infected and were associated with no reasonable neurological function. Three of the infants whose sacs had ruptured at presentation died, accounting for the mortality rate of 6.7 percent among children with this malformation (Table 1).

blindness. Hydrocephalus was due to aqueduct stenosis in 68 percent of the cases. Ten percent of the affected infants presented late with grotesque head enlargement and varying degrees of visual failure while four children were blind without visual fixation nor tracking at presentation. Thirty seven (74 percent) of the hydrocephalic children underwent ventriculo-peritoneal shunting (VPS) procedure employing a medium pressure, Chhabra slit-in-spring shunt system (*G. Surgwear, India*). Shunt blockage which occurred in 10.8 percent of the shunted group was the commonest complication followed by internal and external shunt infections (8.1 percent) due most commonly to the staphylococcus species. Other complications include wound sepsis, burst abdomen, and seizure disorder in two cases each, while bilateral hydrocoele, adynamic intestinal obstruction, cerebrospinal fluid (CSF) ascites (Fig. 5) and decubitus scalp ulcers occurred in one case each. The overall complication rate was 29.6 percent. The shunt was removed in two cases of shunt infection. Two

Encephalocele

Twelve children, (seven females and five males) constituting 8.9 percent of the total, presented with encephaloceles which varied in size, shape and location (Figs 6a, b, c). The lesions were occipital in 11 infants (Fig. 6a & b) and sincipital or fronto-nasal in one (Fig. 6c). A peculiar case of occipital encephalocele had a bi-lobed sac with each lobe containing a cerebral hemisphere and the half empty cranium appearing anencephalic in configuration (Fig 6b). Her planned staged management was truncated when her parents opted for a voluntary discharge. Surgical repair in children with occipital lesions consisted of simple excision and closure of the wound in layers from dura to the scalp. In addition to repair of the encephaloceles, an associated hydrocephalus was shunted in one infant. The fronto-nasal encephalocele encountered in a male neonate had ruptured at birth and brain tissue was herniating from two small frontal cranial defects separated by a narrow bony midline raphe with the rest of the head appearing anencephalic in configuration (Fig. 6c). He died after an extra-dural and extra-cranial repair, resulting in a mortality rate of 8.3 percent among infants with this anomaly.

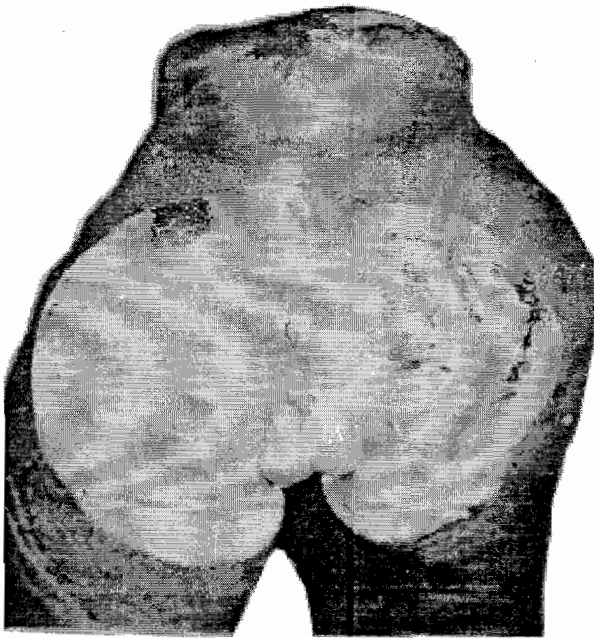


Fig 4: Lumbosacral myelomeningocele in an infant. Note extensive bi-gluteal pressure ulcerations

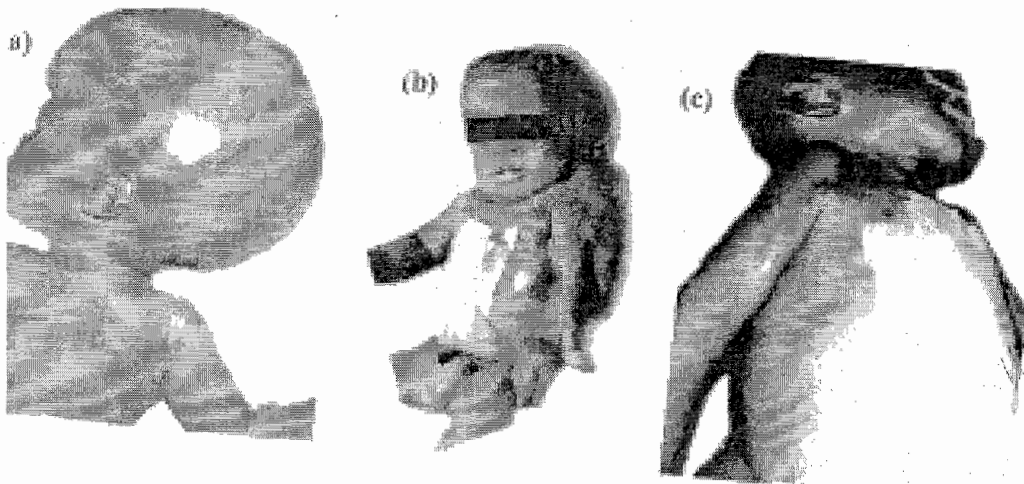


Fig 5: A shunted hydrocephalic infant showing:
(a) grotesque head enlargement
(b) bilateral hydrocoele and

children died after shunting, giving a surgical mortality rate of 5.4 percent. They were infants aged six and nine months who presented with grotesque head enlargement (occipito-frontal circumference greater than 65cm) and multiple systemic anomalies. Following VPS, one of the infants developed shunt infection and blockade while the other developed shunt infection and CSF ascites.

(c) distended abdomen resulting from CSF ascites and adynamic intestinal obstruction

Craniosynostosis

Craniosynostosis, either syndromic (two cases) or non-syndromic (three cases), was diagnosed clinically in five children (3.7 percent) who presented with microcephaly, ridging of cranial sutures and abnormal head shape. The diagnosis was confirmed on skull radiography and/or cranial CT scanning. The syndromic type manifested as Apert's and Crouzon's syndromes in two children. Children with non-

syndromic craniosynostosis underwent linear strip craniectomy of the affected sutures at between nine and 18 months of age while the case of Apert's syndrome underwent lateral canthal advancement and separation of fused digits at the age of 14 months with satisfactory cosmetic result. The child with Crouzon's syndrome was not operated upon because of financial constraint.

Sub-galeal inclusion dermoid cyst of the anterior fontanelle

A soft, non-tender and fluctuant solitary scalp swelling over the anterior fontanelle (Fig. 7), which had been present at birth, was encountered in three (2.2 percent) otherwise normal children during the first year of this study. The head circumference and fontanelle sizes were normal for age, while trans-fontanelle ultrasound revealed extra-cranial subgaleal cyst over the anterior fontanelle, without any link with apparently normal intracranial contents. Simple excision of the cysts was apparently curative since there has been no recurrence after four years of follow-up. Subsequent histology revealed a dermoid cyst.

Microcephaly

Symmetrically small heads were found in two neonates who presented with intermittent spasms, restlessness, poor appetite, weight loss, poor neurological development and irritability. Brain CT scan confirmed cerebral infarcts in the territories of the right and left middle cerebral arteries, respectively. Their parents refused treatment, opting for a voluntary discharge.

Porencephaly

A six-month-old female infant presented with head enlargement and mild left hemiparesis. A trans-fontanelle ultrasound scan revealed dilated lateral and third ventricles suggesting hydrocephalus. She was promptly shunted but developed a blockade which was found at shunt revision, to be due to intraventricular haemorrhage. A cranial CT scan revealed a cyst of the right frontal lobe communicating with the anterior horn of the right lateral ventricle. Her hemi-paresis improved remarkably following craniotomy and excision of the cyst.

Hydranencephaly

A male infant presented with grotesque head enlargement, sutural diasthesis, failure to thrive, listlessness, lethargy, paraparesis and blindness suggesting a diagnosis of hydrocephalus. Initial trans-fontanelle ultrasound revealed a large fluid filled cavity occupying the supra-tentorial portions of the anterior and middle cranial fossae without discernible

ventricular landmarks, while subsequent cranial CT scan demonstrated loss of significant portions of the cerebral hemispheres which were replaced by fluid with intact basal ganglia and brain stem; features consistent with the diagnosis of hydranencephaly. The initial plan to shunt the infant was shelved and the child was discharged to follow-up.

Discussion

This study has revealed spina bifida cystica to be the commonest malformation of the CNS encountered at the University of Ilorin Teaching Hospital, a tertiary health institution serving over 30 million inhabitants of the middle belt of Nigeria. This finding is similar to that from Jos¹⁰ northern Nigeria, where myelomeningocele predominated among CNS anomalies but slightly differs from those from Ibadan⁸ and Enugu⁹, representing south Nigeria, where hydrocephalus was the commonest anomaly, thus suggesting a geo-ethnic variation in the frequency of CNS malformations in the country. The incidence and pattern of distribution of congenital CNS malformations are known to vary with geographical, familial or ethnic factors and sex¹¹ as this series revealed. The study also lends support to the male-gender predilection of CNS malformations as documented by previous authors,^{1,2,6} although the reasons for such a predilection are not clearly understood.

Parental socioeconomic status militated against standard management of many children with CNS malformations in this series. Although our approach to the management of patients with spina bifida cystica was non-selective, this policy became naturally selective as the socioeconomic and educational levels of parents emerged as the chief determinants of acceptance or rejection of surgical intervention, leading to a denial of surgery in 36 percent of infants with this anomaly. In this institution and elsewhere in Nigeria, treatment of patients with spina bifida cystica was for many years, left in the hands of general, orthopaedic and paediatric surgeons. However, with the current establishment of neurosurgical services manned by qualified neurosurgeons in many tertiary institutions, an improvement is expected in the quality of care, outcome and survival rates of such children. In centres where a higher standard of care is linked with better facilities, early death in both treated and untreated patients is currently associated with advanced hydrocephalus and multi-system congenital anomalies, with the reported mortality rate in selected (14 percent) and unselected (15 percent) treated groups being similar¹² but these figures are even higher than the mortality rate of 6.7 percent obtained in this series.

Hydrocephalus was easily diagnosed clinically and confirmed by ultrasound, since CT scan and magnetic resonance imaging (MRI) are not routinely available. Although these imaging techniques provide superior anatomical details in the investigation of hydrocephalus, cranial ultrasound reliably demonstrates ventricular enlargement and determines with reasonable confidence, the common underlying pathology thus eliminating with some reliability, common differentials of an abnormally large head. The present series revealed hydranencephaly as the most common cause of abnormal head enlargement that was easily mistaken for hydrocephalus. Pathologically in hydranencephaly, the cerebral hemispheres are absent or significantly reduced and are represented by a sac of membranes over which remnants of the frontal, temporal and occipital lobes are scattered, the basal ganglia and brain stem being present. While these features were confirmed on cranial CT scan of the infant with hydranencephaly, cranial ultrasound was less informative. A distinction between these conditions is necessary because, while the child with hydrocephalus benefits substantially from shunting, the procedure is not recommended for hydranencephaly which is associated with psychomotor retardation.¹³

The shunt infection rate of 8.1 percent following surgery of hydrocephalus in this series is less than the reported rates which vary from > 1 to < 30 percent of operations¹⁴ depending on the patient's age and other factors. It is higher in paediatric than adult populations and even higher in shunted premature infants, with the rates approaching 15-20 percent.¹⁴ Although shunt infections carry a substantial morbidity and have the potential to cause severe disability or even death, there may be no adverse effects on long-term outcome¹⁵ as confirmed by this study in which no infant died because of shunt infection. The major determinants of mortality were the complications of grotesque head enlargement and coexisting congenital anomalies. Outcome of the shunting procedure is however best in the absence of shunt infection.¹⁵

This study has revealed a preponderance of occipital over fronto-nasal encephalocoeles, confirming previous reports.¹⁶ It also confirmed delayed presentation in a majority of patients with un-ruptured encephalocoele until after the lesions had achieved enormous sizes. In Nigeria, parents of such children may shy away from neurosurgical consultation and treatment because of the significant social stigma which an encephalocoele generates, the victims being labeled as having "occipital or frontal horns" or "two heads". However, the treatment

outcome appears not to be related to the size but to the content of each sac. Only two cases of subgaleal inclusion dermoid cyst of the anterior fontanelle were encountered and both were seen during the first of the five-year study period. Within a 10-year study period, 18 cases of this type of lesion were managed at a sister tertiary institution in Ibadan, southern Nigeria,¹¹ while the anomaly was not mentioned in a three-year study from Jos, northern Nigeria,¹⁰ suggesting a seasonal and geographic variation in its occurrence.

Our experience shows that the major morbidity factor in children with craniosynostosis, whether syndromic or non-syndromic, is cosmetic, while other factors such as elevated intracranial pressure in the primary type or neuro-developmental delay in the secondary type assume less prominence. Apart from the contentment resulting from improved cosmetic appearance in the patient with Apert's syndrome, her postoperative neurology and prognosis for intellectual and motor recovery were also satisfactory, even though surgery was delayed till 14 months due to late presentation. The result is however best, if reconstructive surgery for craniosynostosis is performed below six months of age.

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