ISSN: 2415-038X (Print)

Journal of Preventive and Rehabilitative Medicine

2018; 1(1): 12-18
Published Online:23/03/2018 (http://medicine.unza.zm/research/journal)
doi: 10.21617/jprm.2018.0101.2

Original Paper

Profiling Children with Neural Tube Defects at the University Teaching Hospital, Lusaka, Zambia

Micah Simpamba[1], Margaret M. Mweshi[1] and Patricia M. Struthers[2]

[1]School of Health Sciences, Department of Physiotherapy, University of Zambia, Lusaka, Zambia
[2]School of Public Health, University of Western Cape, Cape Town, Republic of South Africa

E-mail: micamutuna@yahoo.co.uk

To cite this article:
Micah Simpamba, Margaret M. Mweshi and Patricia M. Struthers. Profiling Children with Neural Tube Defects at the University Teaching Hospital, Lusaka, Zambia. *Journal of Preventive and Rehabilitative Medicine,* Vol. 1, No. 1, 2018, pp. 12-18. doi: 10.21617/jprm.2018.0101.2

Abstract

Background: Neural tube defects (NTDs) are the commonest disabling congenital defects which affect about 300,000 infants each year. In developing countries like Zambia, these defects are unrecognized and under reported. Therefore, the aim of this study was to determine the profile of children with NTDs at the University Teaching Hospital (UTH) in Lusaka.
Methodology: A retrospective review of medical records of children with NTDs who were who were admitted at the UTH between January and December, 2010 was done. Data was collected using a data extraction sheet and descriptive analysis was done using Statistical Package for Social Science (SPSS) version 20.
Results: A total of 101 medical records were identified and 50 of them were for children with NTDs giving a prevalence rate of 50%. The sample of children with NTDs had more boys (58%) than girls (42%). The majority of the children were from Lusaka province with 28%, while the minority were from North-Western and Copperbelt provinces with 2% each. The profile showed that many children (78%) presented with Spina Bifida (SB), with encephalocele (20%) and lastly one with both SB and encephalocele (2%). Myelomeningocele was the most common type of SB (44%) while the lumbar region was the commonest site (52%). Occipital encephalocele (60%) was the commonest compared with the nasal (30%) and frontal (10%).
Conclusion: The majority of the children with NTDs admitted at the UTH were from Lusaka province and SB was the most common type of NTDs. Myelomeningocele was the most common presentation while the lumbar region was the most common site. Occipital encephalocele was the commonest site compare with other regions.

Key words: Neural tube defects, spina bifida, meningocele, myelomeningocele, encephalocele, Zambia

1.0 Introduction

Neural tube defects (NTDs) are the second commonest group of serious birth defects after cardiac abnormalities which result in infant mortality and severe disability [1]. These defects result from failure of the neural fold to fuse during fetal development and occur early in pregnancy between 21 and 28 days of conception [2]. Failure of fusion of the anterior neural tube results in cranial NTDs namely anencephaly and encephaloceles, while failure of fusion of the posterior neural tube results spinal NTDs referred to as spina bifida (SB) [3]. Spina bifida is classified into spina bifida cystica and spina bifida occulta, with spina bifida cystica further classified into meningocele and myelomeningocele [2].
The worldwide incidence of NTDs is estimated to range between 1.0 and 10.0 per 1000 births [4]. The highest incidence was reported in China during the period of 2002 and 2004, at 19.94/1000 births and in 2003 at 13.87/1000 births [5,

6]. A worldwide systematic review (1990-2014) on global prevalence of NTDs reported the prevalence of 11.7/1000 births in Africa [7]. In low and middle income countries such as those in Africa, the prevalence of NTDs may even be higher [8]. In Zambia, a retrospective review of congenital anomalies at a tertiary hospital in Ndola district found CNS congenital anomalies to be the most common (40%) [9]. Hospital based prevalence and incidence rates which are common practice of reporting prevalence and incidence rates in most low and middle income countries may not reflect the actual prevalence. Neural tube defects are reported to cause approximately 88,000 deaths globally (in 2012) and 8.6 million disability adjusted life years [7]. Among all the NTDS, anencephaly is the most lethal, and therefore incompatible with life, with most affected pregnancies resulting in stillbirths and livebirths dying shortly after birth [3]. Meningocele and myelomeningocele are compatible with life and constitutes about 95% of spinal NTDs [2]. Myelomeningocele is the commonest complex form of NTDs with most survivors living with severe lifelong disabilities [8, 10].

According to Burton and colleagues [11], about 70% of myelomeningoceles are in the lumbar or lumbosacral regions while the less common sites are cervical, thoracic and sacral regions [11]. Most studies in sub-Saharan African countries have reported the lumbosacral region to be the commonest site for spinal NTDs [12, 13]. For encephaloceles, the commonest site is the occipital region and this is consistent with Burton [11] who observed that 75% of encephaloceles are on the occipital region.

The causes of NTDs are multifactorial, involving complex interactions between genetic and environmental factors, with folic acid deficiency being the major risk factor [2, 14]. Despite the complex nature of the epidemiology of NTDs, advances in research have resulted in evidence based preventive and therapeutic measures against these defects [15]. Preventive measures involve the use of periconceptional folic acid during childbearing years while therapeutic measures involve in utero repair of the spina bifida for the unborn child, resulting in significant outcomes for the child [15]. Postnatal management involves surgery to close the lesion immediately after birth, while subsequent surgeries may be required for tethered spinal cord, hydrocephalus and other orthopaedic and neurological problems [14].

Closure of the open myelomeningocele is advisable within 24 hours, or 48 hours at most, as closure beyond this period results in infection of the defect [16]. Besides reducing infection rates, other reported advantages of early surgery for myelomeningocele include low incidence of preoperative myelomeningocele rupture, low incidence of postoperative dehiscence, lower proportion of ventriculoperitoneal shunt and a lower incidence of one year neurodevelopmental delay [17].
Despite the advances made in prevention and management of NTDs, African countries still lag behind with most countries experiencing challenges related to prevention and management. Furthermore, the health care services for children with NTDs are often more costly and may not be affordable for most families from developing countries who are mainly from low socioeconomic background [18]. Besides the cost of care involved with NTDS, children in developing countries face many other challenges such as harsh social economic conditions, harmful taboos, religious beliefs, and inadequate medical personnel and facilities, which all lead to late presentation of patients for appropriate health care [13]. Additionally, surgical services in most developing countries are only found in big cities and this makes it difficult for the poor rural communities to access the services [19]. Having surgical services only in larger cities makes it difficult for the poor rural communities to access these services [19]. This may result in late presentation for surgery, high mortality rates and severe secondary complications for those who survive [8].

Despite all the challenges associated with management and prevention of NTDs in developing countries, these anomalies are unrecognised and underreported in most sub-Saharan African countries [12]. In Zambia, children who are born with NTDs from any part of the country are supposed to be transported to Lusaka if they require surgical management. This is could be because surgical repair and shunting for NTDs is only done in Lusaka at the University Teaching Hospital (UTH) and Beit Cure Hospital, a private hospital. Therefore the aim of this study was to report on the clinical profile of children with NTDs, who were managed at the UTH in Lusaka, Zambia during the period of January to December, 2010.

2.0 Methodology

Study Design

A descriptive retrospective design was used to review records of children with NTDs who were admitted to ward D01 from January to December 2010. Ward D01 is a paediatric surgical ward at UTH. The UTH is a tertiary level hospital in Lusaka, the capital city of Zambia. This is a national referral center which receives referrals from all parts of the country and also acts as the principal training institution for almost all health and allied disciplines.

Population and Sampling

The study population included all the 101 medical records of children who were admitted during the stated period. This population was identified using the admission register. Before data extraction from individual patients' medical files, a list of names and file numbers for all patients with NTDs who were admitted in the year 2010 were generated from the admission register. Using this list, a search for individual patients' files was conducted and only 50 medical files were found, as it was learnt that some parents went home with the children's medical files.

Procedure of data collection

Ethical clearance for the study was obtained through written permission from the Senate Research Ethics Committee of the University of the Western Cape and the University of Zambia

Research Ethics Committee. Permission to carry out the study at the UTH was granted by the UTH management.

Data collection was done using a data extraction sheet that was developed based on research objectives, the researcher's experience and information from literature on similar studies [20, 13]. Information recorded on the data capturing sheet was entered in SPSS (version 20.0) software for analysis. Data analysis consisted of descriptive and inferential statistics and presented in frequency tables. Associations between variables were tested by chi-square. These associations included the following variables: province of referral and type of NTD, type of NTDs and neurological status; and type of NTDs versus associated impairments. The level of significance was set at $p< 0.05$.

3.0 Results

The results of this study are presented under demographic characteristics, type of NTDs, site of lesion, neurological impairments, associated impairments, radiological investigations, surgical management, and complications of management and follow up visits.

Demographic Characteristics

Table 1 shows the distribution of the children according to province of referral. The majority of children were referred from Lusaka province (28%). The gender distribution for the study sample was 29 (58%) males and 21 (42%) females. The age distribution ranged from one day old to 48 months with the majority (80%) aged between one day to one month (n = 40). All children referred from Lusaka Province were aged between one day and one month. Chi-square tests showed a significant relationship between province of referral and age of child on admission ($p = 0.0001$).

Types of neural tube defects

The profile showed that 39 (78%) children with NTDs presented with Spina Bifida (SB), 10 (20%) with encephalocele and 1 (2%) had both SB and encephalocele. Myelomeningocele was the most common type of SB (44%) while the lumbar region was the commonest site (52%). Occipital encephalocele (60%) was the commonest compared with the nasal (30%) and frontal (10%). Chi square tests showed no association between type of NTD and sex of the child ($p = 0.232$). There was no significant association between type of NTD and province of residence ($p = 0.172$).

Site of neural tube defect

Table 2 shows the distribution of children with NTDs according to the site of the NTD. The lumbar region was the commonest site (52%) for spinal lesions while the occipital region was the common site for cranial defects.

Neurological impairments

Figure 1 shows neurological impairments of the children with SB in the study group. The most common neurological impairments recorded in the patients' files where incontinence and paraplegia. From this sample (n = 50), the distribution of neurological impairments was as follows: 23 patients had paraplegia; one patient had incontinence only; 15 patients had no neurological impairments and 11 patients had no records on neurological deficits. Out of the 23 patients who had paraplegia, 15 had also incontinence giving a total of 16 patients with incontinence out of the total sample (n = 50). Chi square tests shows a significant association between type of NTD and neurological impairment ($p= 0.0001$).

Associated impairments

Figure 2 shows the distribution of associated impairments according to type of NTDs. The most common associated impairment was hydrocephalus (56%) which was either present as the only impairment or in association with other congenital and chromosomal anomalies. Apart from two children, whose status was unknown, all children with myelomeningocele had either hydrocephalus or a combination of hydrocephalus and clubfeet. The chi-square tests did not show any significant relationship between type of NTDs and associated impairment ($p = 0.086$).

Radiological investigations

Ultrasound was the most common radiological investigation done with 80% while MRI (2%) was the least used radiological investigation as shown in table 3. The medical records for five children (10%) had no notes on radiological investigations and therefore were recorded as not done.

Surgical management

Surgical management consisted of either repair of the defect (20%), insertion of a shunt only (28 %) or both the repair of the defect and the insertion of a shunt (22%). The majority of the children (30%) did not undergo any surgical management. The type of shunt for all the children whose surgery done was the ventriculo peritoneal shunt (VPS). Figure 3 shows the distribution of surgical management among the children.

Complications

The majority of the children (52%) had some complications compared to those without (48%). Among the complications, wound infection was the most common complication (40%).

Follow up visits

Table 4 shows that the majority (66%) of the children had no follow up notes in their medical records. Furthermore, even for those children who had follow up notes, most of them did not have more than three (3) follow up visits recorded in their medical records.

Table 1. Distribution of study sample according to province of referral (n=50)

Province	Frequency (n)	Percentage (%)
Lusaka	14	28%
Southern	6	12%
Central	7	14%
Eastern	5	10%
Western	8	16%
North - Western	1	2%
Northern	5	10%
Luapula	3	6%
Copperbelt	1	2%

Table 2. Distribution of Neural Tube Defect according to site (n=50)

Site	Frequency (n)	Percentage (%)
Spina bifida		
Lumbosacral	10	25%
Lumbar	26	66%
Sacral	1	3%
Thoracolumbar	1	3%
Cervical	1	3%
Encephaloceles		
Occipital	6	60%
Nasal	3	30%
Frontal	1	10%
Both encephalocele and meningocele		
Occipital/lumbar	1	100%
Total	**50**	**100%**

Table 3: Radiological investigations (n=50)

Radiological investigation	Frequency (n)	Percentage (%)
Cranial Ultrasound	40	80%
CT Scan	4	8%
MRI	1	2%
No radiological investigation	5	10%
Total	**50**	**100**

Table 4: Number of follow up visits for children with NTDs (n=50)

Follow up visits	Frequency (n)	Percentage (%)
No follow up notes	33	66%
1 follow up visit	4	8%
2-3 follow up visits	7	14%
Above 3 follow up visits	4	8%
Referred to another hospital (BCH)	2	4%
Total	**50**	**100**

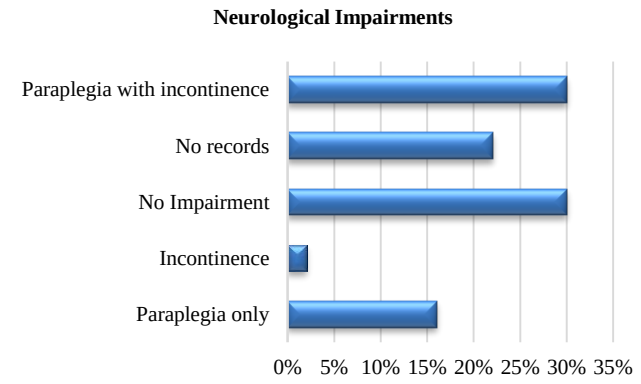

Figure 1. Neurological impairments

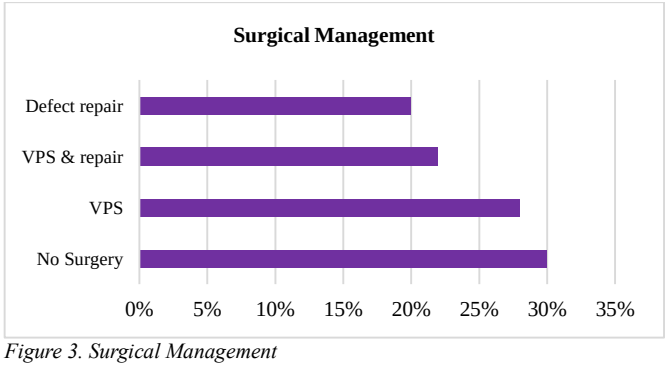

Figure 3. Surgical Management

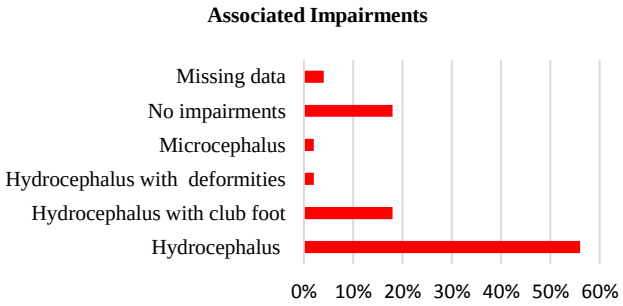

Figure 2. Associated Impairments

4.0 Discussion

The aim of this study was to determine the profile of children with NTDs at a tertiary hospital in Zambia. The profile included demographic characteristics, prevalence of NTDs, types of NTDs, site of the NTD, neurological status of the affected children, complications associated with NTDs, surgical management and follow up visits.

Data on the profile of NTDs in developing countries like Zambia is very scanty and where available, it is restricted to hospital based studies. The majority of children in this study where referred from Lusaka province. However, contrary to

this, a retrospective record review which was done at UTH and Beit Cure hospitals in Zambia between 2001 and 2010 found the majority of children with SB to have come from Southern province [20]. This could perhaps be because the earlier study covered nine years and also included children from Beit Cure Hospital that had an outreach program in the Southern Province of the country. The authors of this study attributed this to environmental predisposing factors related to the rock stratum and geological pattern in the Southern province of Zambia which is in line with assertions by Wang et al [22]. In the current study, the high prevalence of children with NTDs in Lusaka could be attributed to geographical accessibility as reported in another study which was done in Cameroon where the majority of the patients (66%) came from within the city of Yaounde where the hospital was based [21]. Geographical accessibility is one of the dimensions of access that have been reported to favour people living in urban areas compared to those living in rural areas [23].

While findings on age at presentation are similar to some studies done in some African countries, there are other studies within Africa whose findings were contrary to this study. Studies reporting similar findings include a one year prospective study conducted in a Nigerian neurosurgical unit, which reported the age at presentation to range from two days to 60 months with a mean age of between 5.8 to 11.47 months [12]. On the contrary, a retrospective medical record review that was done in Cameroon (2000-2006) reported that about 44.13% of children presented on the first day of life with only 7.25% presenting between the first week and fourth week of life [20]. The age on admission for children with NTDs is an important factor in determining the outcome of the management. Surgical closure for open NTDs should be done within 36 to 72 hours after birth to minimise the risk of CNS infection and improve the neurological outcome [24].

The male predominance in the current study is consistent with findings from another study done in Zambia [25] and other similar studies in some African countries such as Nigeria [11] and Cameroon [19]. Contrary to these findings, other studies done in Africa and other regions have reported female predominance among children with NTDs [26, 27, 28, 29]. There are many theories that are used to explain the gender variations among children with NTDs. One assertion is that female children are more likely to have cranial NTDs than spinal defects [30]. This is supported by findings from some studies done in Africa which reported higher frequency of cranial defects in females than males [26, 27]. However, the current study found more males (n = 9) than females (n = 2) among children who had encephalocele.

The high prevalence of myelomeningocele in study was also reported in another retrospective study done in Zambia between 2001 and 2010 at the University Teaching Hospital and Beit Cure Hospital where 61% of spina bifida cases where myelomeningocele [25]. Other studies done in both African and non-African countries have reported myelomeningocele as the most common type of NTD [31, 30, 20]. Although this study did not report any case of anencephaly, there are other studies which reported more cases of anencephaly than encephalocele in countries such as Iran [32], Texas [31] and Sudan [26]. All these studies which reported high incidences of anencephaly were characterised by high mortality rates among live births, foetal deaths and abortion rates because anencephaly is incompatible with life. It is for this reason that we cannot rule out the availability of cases of anencephaly in Zambia because most of these children are unlikely to reach tertiary level hospitals.

Findings from this study regarding the lumbar region being the commonest site for spinal NTDs are consistent with the previous findings in Zambia by Mweshi and colleagues [25] in which 60% of defects were in the lumbar region. Similar findings have also been reported in studies in Cameroon [20] and Kenya [31]. However, other studies in this area have reported the lumbosacral region to be the commonest site for spinal defects (34, 35, 36]. The level of the spinal NTDs is an important factor in determining neurological status and functional independence among patients with spina bifida [37]. The occipital region as a common site for encephaloceles is also consistent with findings from other studies including those done in Uganda [38], Nigeria [12] and Alabama [39].

Hydrocephalus was the commonest associated impairment reported in the current study (56%) and also was more associated with myelomeningocele. These findings are supported by Mweshi et al [40] who noted that about 80-90% of children with myelomeningocele have hydrocephalus. Furthermore, Warf [41] observed that about half to two thirds of children with myelomeningocele require treatment for hydrocephalus and about one third of those with encephalocele will require management for hydrocephalus.

The commonest radiological investigation done among children in the current study was cranial ultrasound which was done in 80% of the children. The use of cranial ultrasound has been recommended for use in developing countries because it is fast, easy to use, feasible, practical, cost-effective and an efficient technique [42]. In addition, Adeleye and colleagues [12] noted that cranial ultrasound is one of the most convenient and affordable methods for detection of foetal neural tube defects as well as early diagnosis of hydrocephalus in infants born with NTD.

Surgical management involved closure of the lesion only, shunt only, or both shunt and closure of the lesion and 70% of children underwent surgery. The other 30% did not undergo any surgical management as most of them who had infected lesions were sent home and their mothers advised to return on given dates. This study did not report on whether the children who had both shunting and closure of lesion had either staged or simultaneous surgery. Literature on surgical management of children with NTDs indicates that there are variations in the surgical management with some authors supporting simultaneous surgery while others go for staged surgery [40, 34]. According to Margaron and colleagues [43], simultaneous surgery may not be possible for most children in African countries because of the late presentation which is characterised by infected wounds and therefore poses a high risk of shunt malformation.

Wound infection was the commonest complication reported in the study. The presence of complications in the study may be an under estimation because over 66% (n = 33) of the medical records had no follow up notes after the initial discharge despite the records indicating the need for follow up. Wound infection, which is a common observation in most studies done in Africa [35, 13] is associated with delayed closure of the defect which is a consequence of the late presentation of children. The low incidence of children presenting with meningitis in the current study has also been reported in other studies [20, 44]. The low incidence of shunt malfunction could be attributed to the fact that most children who presented with wound infection were discharged for wound cleaning without any surgical intervention. This is in line with the recommendation to delay shunting for children with infected wounds by first putting them on antibiotics and wound cleaning for at least a week before surgery [43].

The lack of follow up notes in 66% of medical notes in the current study is consistent with findings by Mweshi and colleagues [25] who reported that 81% of the patients were lost to follow up. The lack of follow up on children with NTDs has been reported in studies done both in Africa and elsewhere [34, 20, 44]. This could be associated with geographical accessibility, affordability and acceptability of health care services. Follow up visits for children with NTDs have been reported to have a bearing on survival rates of these children. According to Warf and colleagues [35], it is important to note that despite providing competent and evidence based initial management of children with NTDs, their survival largely depends on adequate follow up management. It is extremely important to encourage follow-up programs in Zambia for such could help prepare children to participate in chores expected of them regardless of their physical status [46].

Study limitations

Limitations of medical record reviews include incomplete or missing data in medical notes, difficulty interpreting or verifying documented information and variability in quality of documentation among health personnel [45]. The strategies used in the development of the data capturing tool ensured that such limitations were minimised. Other limitations included the inability by the researcher to locate 51 medical records, and of the 50 that were located, about half of them had no follow up notes.

Conclusion

The profile of children with NTDs at UTH shows that the majority of children were from Lusaka province and most children were admitted between three days and one month after birth. Spina bifida was the most common type of NTDs and among children who had Spina bifida, those with myelomeningocele were the majority. Hydrocephalus was the most common associated impairment while wound infection was the most common complication. The majority of medical records had no follow up notes. The findings from this record review suggest that management of children with NTDs in Zambia is faced with challenges such as late presentation, wound infection and loss to follow up management. This is consistent with literature which indicates that developing countries have higher incidences of children with NTDs and yet are faced with many challenges related to prevention and management.

Recommendations

The high prevalence of wound infection and loss to follow up indicates the challenges with accessibility of specialty services for children with NTDs in Zambia. In order to improve the health care delivery for these children, there is need for the government to provide transport to these children during first referral as well as follow up management. There is also need to consider follow up management through community based rehabilitation programmes as this would ensure many children access to follow up management. Furthermore, the outcome of patients who are lost to follow up is also not known and therefore there is need for research in this area so as to determine the outcome of these patients.

Acknowledgements

The authors would like to thank Management at the University Teaching Hospital for allowing the study to be conducted there. Appreciation also goes to the nursing officer in-charge of Ward D01 and the records clerk for making it possible to access the patients' records.

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