

Open Neural Tube Defects at the National Hospital, Abuja : an Analysis of Clinical Patterns and Neonatal Outcome

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Summary

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Background: Neural tube defect is the most common congenital abnormality of the central nervous system.

Objectives: To document the clinical patterns and neonatal outcome of babies with open neural tube defects at the National Hospital, Abuja.

Design: Retrospective review.

Patients: All babies admitted into the Newborn Unit of the hospital with a diagnosis of open neural tube defect between July 2000 and June 2003.

Methods: Case notes were retrieved and relevant information was extracted for analysis.

Results: Eighteen babies (13 males, five females) presented with open neural tube defects of which 16 (88.9 percent) were outborn. Although 14 of the mothers received routine antenatal care, only eight of them took folic acid regularly. Prenatal diagnosis was made in one of five mothers who had antenatal abdominal ultrasound. The predominant clinical type was lumbosacral myelomeningocele (61.1 percent). Six (33.3 percent) babies had hydrocephalus at presentation, while three of the babies with encephalocele were microcephalic. Vaginal delivery was associated with an increased risk of ruptured myelocele sac. Limb paralysis and sphincteric dysfunction occurred in eight (44.4 percent) of the babies. The defects were repaired at a mean age of 7.8 ± 3.1 days in five patients, while three babies died (anencephaly=1, ruptured sac with meningitis=2), resulting in a mortality rate of 16.7 percent.

Conclusion: We suggest that efforts be intensified to diagnose these lesions antenatally, in order to allow for prenatal counseling, planned cautious delivery and prompt multidisciplinary postnatal management, as these would most likely result in improved neonatal outcome.

Introduction

NEURAL tube defects (NTD) constitute a significant proportion of congenital abnormalities of the central nervous system.¹ while spina bifida occulta may be of little clinical significance,² spina bifida cystica or cranium bifidum is often associated with significant mortality and long-term morbidity.³ although there is an association between NTD and periconceptual folate deficiency,⁴ maternal use of valproic acid^{5,6} and genetic predisposition,² no risk factor is identifiable in a large number of cases of NTD as in other major congenital malformations.⁷ Antenatal diagnosis can be made early in pregnancy by abdominal ultrasound and by estimation of maternal serum and amniotic fluid alpha-feto protein.⁸ Even where the option of pregnancy termi-

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nation is not culturally acceptable, prenatal diagnosis allows room for anticipatory management including deciding on the best mode of delivery, while parents are psychologically prepared for the birth of an abnormal child. Immediate and long term outcome are related to the size and site of the defects as well as the presence of complications or associated malformations in other organs such as the heart. The management of this condition requires a multidisciplinary approach.⁹

Although early surgical intervention on all babies with meningomyelocele was carried out in the past to minimize injury to nervous tissue and the attendant worsening of neurological dysfunction, long-term follow-up of such patients has demonstrated unpleasant morbidity among survivors necessitating the need for pre-operative selection criteria³ to optimize the benefits of operative intervention. In resource poor countries such as Nigeria, where access to comprehensive medical care is limited to a negligible proportion of the population,¹ neural tube defects are expectedly associated with high morbidity and mortality. Shehu *et al* had earlier documented functional disability in 38 percent of cases of spina bifida cystica who underwent surgery in Zaria and attributed this to inadequate facilities for postoperative rehabilitation.¹¹ Neural tube defects therefore constitute a burden on parents as well as the community. We undertook a review of all cases of open neural tube defects seen at the Neonatology Unit of our hospital over a period of three years (July 2000 - June 2003) to document their clinical patterns and assess their neonatal outcome. The National Hospital, Abuja (NHA) is a tertiary institution, which is a referral centre for some other hospitals in the north central part of Nigeria.

Subjects and Methods

Babies admitted to the Neonatal Unit of the National Hospital, Abuja between July 2000 and June 2003 with myelomeningocele, encephalocele or anencephaly

formed the subjects of this analysis. One case of meningocele had incomplete records and was therefore, excluded from the analysis, as were cases of spina bifida occulta as the study was restricted to open neural tube defects. A list of babies who satisfied these diagnostic criteria was compiled from the admission register kept in the special care baby unit. This list was then used to retrieve the case notes from the records department. Relevant demographic and clinical information were extracted from the case notes and transcribed to a pre-designed proforma to ensure uniformity and ease of analysis of information thus obtained. Lesions were classified on the basis of location along the cranio-vertebral axis, while associated neurological deficits as well as other significant malformations were noted. Information on management including neonatal outcome was also extracted. For those who had surgery, the age at surgery was noted.

Results were analyzed for clinical presentation, risk factors associated with increased morbidity and mortality, as well as the neonatal outcome.

Results

A total of 18 cases of NTD were seen over the three-year period, accounting for 0.8 percent of the 2250 neonatal cases admitted during the period. Thirteen (72.2 percent) were males, giving a male to female ratio of 2.6:1. Sixteen (88.9 percent) babies were outborn and two (11.1 percent) inborn. The total number of deliveries at the National Hospital over the period was 3,632, giving an incidence of 0.5 per thousand inborns. Nine (56.3 percent) of the outborn babies were referred from government hospitals within and outside Abuja, five (31.2 percent) were delivered in private hospitals and two (2.5 percent) were products of home delivery. Fourteen (77.8 percent) mothers attended antenatal clinic, with the mean gestational age at booking of 20.4 ± 5.6 (SD) weeks, while eight (44.4 percent) of them took folic acid regularly. Abdominal ul-

Table I

Types of Neural Tube Defects in 18 Newborns

Type of Defect	No. of Cases (% of Total)
Anencephaly	1(5.6)
Fronto-nasal encephalocele	2(11.1)
Occipital encephalocele	2(11.1)
Thoracolumbar myelomeningocele	1(5.6)
Lumbo-sacral myelomeningocele	11(61.0)
Sacral myelomeningocele	1(5.6)
Total	18(100.0)

trasound was performed on five (27.8 percent) mothers; one of these revealed anencephaly. The ultrasound did not suggest the presence of congenital malformation in the other four mothers.

The mean age of the mothers was 29 ± 9.9 (SD) years. None gave a history of having a previous child with similar lesion. Also, there was no case of parental consanguinity. Sixteen (88.9 percent) babies were born at term while two (11.1 percent) were preterm. Spontaneous vertex delivery accounted for 13 (72.2 percent), two (11.1 percent) were delivered by elective caesarian section (C/S) and three (16.7 percent) were breech deliveries. The elective C/S was carried out on account of pregnancy-induced hypertension in one and pro-

longed labour with foetal distress in the other. There were no signs of hydrocephalus in these babies at birth.

Classification of NTDS

Thirteen (72.2 percent) of the cases of NTD were myelomeningoceles, four (22.2 percent) were encephaloceles while one (5.6 percent) was an anencephaly. Lumbo-sacral myelomeningocele was the most frequently encountered in this series, accounting for 11 (61.1 percent) cases. Table I shows a detailed classification and relative frequency of each type, while Figure 1 shows a term baby with lumbo-sacral myelomeningocele, hydrocephalus and spastic limbs.

Associated complications

Ruptured myelomeningocele sac with cerebrospinal fluid (CSF) leak was seen in nine (50 percent) cases (Table II). Seven (77.8 percent) of these were delivered spontaneously per vagina while two (22.2 percent) were breech deliveries. None of the babies delivered by elective C/S had CSF leak. Of the six cases with hydrocephalus, five (83.3 percent) were observed at presentation while one (16.7 percent) was diagnosed post-operatively. All babies with NTDs seen in our newborn unit routinely undergo transfontanelle ultrasonography to detect hydrocephalus. Hydrocephalus occurred only in babies with thoraco-lumbar (3) and lumbo-sacral (3) lesions while microcephaly was seen in babies with encephaloceles. Sphincteric dysfunction was predominantly associated with lumbo-sacral myelomeningocele (six of eight cases). Two babies were moderately asphyxiated; one had the cord round his neck at birth, while there was no identifiable risk factor in the other.



Fig. 1 Term Infant with a lumbo-sacral myelomeningocele.

Table II

Complications and Other Malformations Associated with the NTDs

Complications/Malformations	No of Cases (%)
Rupture of sac with CSF leak	9(50.0)
Talipes equinovarus	8(44.4)
Sphincteric dysfunction (bladder/rectum)	8(44.4)
Spastic paraplegia/quadruplegia	8(44.4)
Hydrocephalus	6(33.3)
Microcephaly	3(16.3)
Meningitis	2(11.1)
Asphyxia	2(11.1)
Congenital hip dislocation	1(5.6)
Cephalhaematoma	1(5.6)

CSF = Cerebrospinal fluid
NTDs = Neural tube defects

Neonatal outcome

Five babies (27.8 percent) had surgical closure within the neonatal period, three of them by a visiting neurosurgeon and two by resident general surgeons. The mean age at surgery was 7.8 ± 3.1 (SD) days. Five (27.8 percent) were referred to the neurosurgeon at the Usman Danfodio Teaching Hospital, Sokoto, two (11.1 percent) left the hospital because they could not afford surgery while three (16.7 percent) were taken away from the hospital following parental counseling. Three died before surgery; one of the three had anencephaly and the remaining two had ruptured myelomeningocele with meningitis. The mortality rate was thus 16.7 percent. Deaths from NTD accounted for 1.9 percent of all deaths in the newborn unit over the study period. The overall mortality rate for all neonatal admissions during the period under review was 7.3 percent.

Discussion

We have presented 18 cases of neural tube defects seen in the newborn unit of the National Hospital, Abuja (NHA) over a period of three years, representing 0.8 percent of all neonatal admissions. A significant proportion (88.8 percent) of them was outborn, and referred to our hospital for management. This finding, although similar to reports from two other tertiary health institutions in the southern (15 cases in two years)¹⁰ and northern (96 in 12 years)¹¹ parts of the country, may not be an accurate representation of the prevalence of these malformations in the region. This is because there are two other tertiary health institutions serving the same geographical area: Jos University Teaching Hospital Jos, Plateau state and Ahmadu Bello University Teaching Hospital Zaria, Kaduna state, where such major congenital abnormalities could also be referred for management.

The computed incidence of NTD using the total number of deliveries in our hospital (0.5 per 1000), is lower than the 1-2 per 1000 often quoted for the general population.¹¹ An incidence of 1.6 per 1000 was reported in Zaria in 1989.¹⁰ The relatively high cost of medical care at the National Hospital limits the patients' population to the high social class. Only two of 18 cases were delivered at the NHA. Additionally, nutritional deficiency, which is said to increase the risk of NTDs, is unlikely to be prevalent in the mothers who patronize our hospital. Furthermore, our figures excluded minor forms of spina bifida (spina bifida occulta), which are often included in the estimation of the population incidence.

The utilization rate of abdominal ultrasound in pregnancy as seen in this study was low; only a third of the

mothers who had antenatal care (ANC) had abdominal ultrasound done. It is worthy of note that the diagnosis of congenital abnormality was made in only one of them despite the fact that this is a fairly common investigative tool that has the capacity of recognizing open NTDs *in utero*.^{8,9} It should however be noted that most of the mothers attended private and secondary public health institutions where they may not have been privileged to access the services of qualified obstetricians. This may explain the apparently low diagnostic yield of this procedure. Late booking (20 weeks) for ANC as shown in this study precludes early prenatal diagnosis even where efficient ultrasound services are routinely available.

Only eight (44 percent) mothers had taken folic acid regularly beginning from the second trimester of pregnancy. Although this does not necessarily imply a causal relationship between inadequate folate intake and NTD in our study, it can be postulated that the mothers of our patients were probably not exposed to adequate amounts of folic acid in early pregnancy and that this probably contributed to the development of NTD in their babies. Furthermore, it was not possible to extract adequate information about other known risk factors for NTD such as the use of anti-epileptic drugs like valproic acid and maternal pre-conceptional nutritional status, this being one of the limitations of a retrospective study.

Lumbo-sacral myelomeningocele as shown in this study, is reportedly the most prevalent of open neural tube defects.^{2,10-13} The rarity of anencephaly⁴ as demonstrated in the present study where only one case was seen does not necessarily reflect the true incidence since a large proportion are aborted spontaneously. Rupture of myelomeningocele sac was seen in the babies delivered per vagina (SVD/Breech). This complication is associated with increased risk of meningitis and ventriculitis and also delays surgical intervention. None of the babies delivered by caesarean section had a cerebrospinal fluid (CSF) leak. Although the number of babies involved in this study may be too small to enable reasonable determination of the relative risk of CSF leak associated with vaginal delivery, a cautiously supervised labour and delivery of an antenatally diagnosed foetal NTD will minimize the risk of trauma to the lesion. Where facilities are available and affordable, an elective C/S is reportedly associated with a better long-term neurological function in such patients.¹⁵ This may therefore be a better delivery option.

The association of NTD with hydrocephalus is related to the presence of Arnold Chiari malformations in babies with NTD.¹⁴ Hydrocephalus is said to be rare in upper thoracic and sacral lesions.¹⁶ The finding in our study is in keeping with this. However, the preva-

lence of hydrocephalus is widely variable (30-80 percent).^{10,17} The relatively low incidence in our study (33.3 percent) may be partly attributed to the fact that most of our patients could not be managed in our hospital. A number of them could have developed clinically obvious hydrocephalus after leaving the hospital. Microcephaly was seen in the three babies with encephaloceles. This suggests the presence of a significant amount of brain tissue within the protruding sac. The herniating brain tissue often contains dysgenetic neural tissue resulting in various degree of cerebral dysfunction among survivors, the resulting developmental aberrations becoming obvious on long-term follow-up. Although two babies were asphyxiated, this perinatal complication was not directly related to the presence of neural tube defects in them. Anencephaly, a severe form of NTD¹⁷ is not compatible with extra-uterine life.¹⁴ The only case seen in our study died within one week of delivery. The presence of limb paralysis and sphincter dysfunction poses additional challenges in the management of NTD beyond the neonatal period.

Surgical intervention was delayed in those who underwent surgery before discharge. This delay was due to several factors among which were late presentation to the hospital and the absence of a neurosurgical unit in our institution. While delayed surgical closure is not necessarily associated with long-term morbidity, early closure is known to reduce the risk of meningitis and ventriculitis.¹⁸ The period before surgery could be utilized for consultations, discussions and decision making while babies at risk of meningitis are covered with appropriate antibiotics.

The mortality rate of 16.7 percent in our study was twice as high as the overall mortality rate for all neonatal admissions (7.3 percent). Major congenital abnormalities contribute significantly to neonatal deaths.¹⁹ The mortality rate in the present study was much higher than the three percent reported from Zaria.¹¹ The difference could be accounted for, by the predominance of meningoceles in the Zaria series. Meningocele are milder forms of NTDs and are less likely to be associated with complications.

Based on our experience, we make the following recommendations. All pregnant women should have two abdominal ultrasound examinations (1st and 2nd trimesters) during the period of antenatal care. While early diagnosis offers the opportunity for termination of a grossly abnormal conceptus, late antenatal diagnosis will enable prenatal counseling, planned cautious delivery and prompt and timely surgical intervention when indicated. Mothers who have had previous abnormal babies should be closely monitored in subsequent pregnancies. Since most pregnant women regis-

ter very late for antenatal care and neural tube closure occurs at a time conception is hardly recognized, routine use of folic acid by women of childbearing age beginning from the period of wedlock may be useful as a long term measure. Meanwhile, public awareness of the need for early booking for ANC should be heightened. A large scale, multicentre, prospective study may be necessary to identify the various risk factors associated with NTDs and other major congenital malformations in our community. This will enable the evolution of a comprehensive package of preventive strategies for major congenital malformations. Some designated tertiary institutions should have human resources adequately trained to provide the immediate care required by these babies and others with major congenital malformations. All prenatally diagnosed cases of neural tube defect should be referred to such centres for delivery and subsequent baby care.

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