Prevalence of Neural tube defects at PUMHS Hospital.

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Abstract:
Introduction: Spina bifida, anencephaly, and encephalocele are neural tube defects (NTD) and result from failure of closure of the neural tube between 21and 28 days following conception. Encephaloceles are possibly post-closure defects. NTDs are an important cause of mortality and morbidity globally. NTD encompasses about one-tenth of the burden of all congenital conditions and make up the third largest congenital burden after congenital heart disease and Down’s syndrome. In low income countries, NTDs may account for 29% of neonatal deaths in observable birth defects

Objective: To know the prevalence of neural tube defects at PUMHS Benazirabad.

Methodology: All pregnant ladies when diagnosed to have fetus with neural tube defect through ultrasound examination were counselled for termination of pregnancy. Data collected through proforma with details of demography, gynecological history, and type of neural tube defect evaluated by senior gynecologist and neonatologist. NTDs were defined as cases of anencephaly, spina bifida, encephalocele and meningocoele among infants of any gestational age.

Results: A total of 7152 deliveries took place in the obstetrics department of PUMHS Hospital from January 2018 to September 2019. 38 (thirty-eight) pregnancies were terminated at different gestational ages, diagnosed as cases of neural tube defects, making a prevalence of 5.313 per 1000 births. Among the NT defects, 22/38 (57.89%) were anencephaly and 12/38 (31.57%) were spina bifida with hydrocephalus.

Conclusion: The most common neural tube defect is anencephaly followed by spina bifida with hydrocephalus. Overall prevalence of NTD is comparable published literature from South East Asia.

Keywords: Neural tube defects, Anencephaly, Spina bifida, Hydrocephalus.

Introduction:
Spina bifida, anencephaly, and encephalocele are neural tube defects (NTD) and result from failure of closure of the neural tube between 21and 28 days following conception.1 Encephaloceles are possibly post-closure defects.2 NTDs are an important cause of mortality and morbidity globally and it is estimated that approximately 300,000 babies are born each year with NTDs 3 resulting in approximately 88,000 deaths and 8.6 million disability-adjusted life years (DALYs) 4,5. This encompasses about one-tenth of the burden of all congenital conditions and make up the third largest congenital burden after congenital heart disease and Down’s syndrome.6 In low income countries, NTDs may account for 29% of neonatal deaths in observable birth defects.7 As morbidity and mortality from infectious diseases are decreasing worldwide, the contribution of birth defects to under-5 morbidity and mortality will continue to increase proportionally.7 Over the past decades many countries have reported a reduction in prevalence at birth of NTDs.8 In one series an overall 93% decrease in prevalence at birth was accounted for by a combination of second-trimester screening and termination of affected pregnancies (34%) and an underlying decrease in the prevalence of affected conceptions (59%), in part explained by folic acid supplementation,9 which led to many high-income countries adopting policies recommending supplementation for women planning pregnancy.10 Therefore it is important to know

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the local incidence of Neural tube defects so that appropriate measures to decrease the incidence of NTDs can be recommended.

**Objective:**
To know the prevalence and pattern of neural tube defects at PUMHS Benazirabad.

**Methodology:**
This cross-sectional descriptive study was conducted at the department of Obstetrics and Gynecology, Peoples University of Medical & Health Sciences, Benazirabad from January 2018 to September 2019 for a period of 21 months. Ethical approval was taken from institutional ethical committee.

**Inclusion criteria:** All pregnant ladies who presented antenatally and diagnosed to have neural tube defect fetuses in their wombs by ultrasound examination, counselled for termination of pregnancy and consented were included in the study.

**Exclusion criteria:** Those pregnant ladies who had normal fetuses and those who did not consent for study were excluded.

A proforma with details of demographics of patient, gynaecological history, and type of neural tube defect in newborn babies were all recorded. Data was collected by trained house officers and postgraduate trainees working in the department. NTDs were defined as cases of anencephaly, spina bifida, encephalocele and meningocele among infants of any gestational age. Radiologists used ultrasonography to perform prenatal diagnosis of NTD and these were ascertained by senior gynecologist and postnatal by neonatologist. Data was analyzed using SSP version 22. For numerical data independent t-test was performed, which gives mean ± standard deviation. While for categorical data, association was carried out by using Chi-square test.

**Results:**
A total of 7152 deliveries took place in the obstetrics department of PUMHS Hospital during the study period. 38 (thirty-eight) pregnancies were terminated at different gestational ages which were diagnosed as cases of neural tube defects by ultrasound examination, making a prevalence of 5.313 per 1000 births. Average age of women who underwent termination of pregnancy was 27.34 years and average gestational age was 27.36 weeks with minimum 18 Weeks and maximum 36 Weeks. Among the NT defects, 22/38 (57.89%) were anencephaly, 12/38 (31.57%) were spina bifida with hydrocephalus, 3/38 (7.89%) were encephalocele and 1/38 (2.63%) was hydrocephalus with meningocele.

**Discussion:**
In a systemic review regarding prevalence of NTDs; it has been reported that 74 members of WHO do not have data available for NTDs.\textsuperscript{11} The reported prevalence of NTDs varied greatly between different regions of the world but also highly variable between countries of the same region. For example the median prevalence of NTDs in South East Asian countries is 15.8 per 10,000 births; it varies between 1.9-66.2; per 10,000 births. Lowest figure of 1.9/10,000 is from Thailand\textsuperscript{12} while highest figure of 66.2/10,000 is from India.\textsuperscript{13} Five studies from Pakistan reported estimates between 38.6 and 124.1 per 10,000 births. This may be in part due to variation in data collection methodology among individual studies, or it could also be the true differences, resulting from other factors including nutritional factors, genetics, routine folic acid supplementation, and the presence of folic acid fortification programs. High NTD prevalence estimates throughout the literature indicate that NTDs remain an important preventable public health problem.\textsuperscript{11}

The prevalence of NTDs in our study is 5.313 per 1000 births or 53.13 per 10,000 births which is well in the range mentioned above and is less than the study by Khattak ST, et.al. who reported the prevalence at 12.14 pe 1000 births.\textsuperscript{14} Another study conducted at Jinnah Postgraduate Medical Centre (JPMC), Karachi for the year 2002, reported in 2004 by Jooma R,\textsuperscript{15} observed 9892 deliveries and amongst these, there were 34 births with various NTDs making a prevalence of 3.437 per 1000 births and is comparable with our study. Whereas another study by Cotter AM, et.al\textsuperscript{16} published in the European Journal of obs/Gyne showed prevalence of 3.047 per 1000 births from 1975 to 1999, which significantly decreased later due to Folic acid fortification. However, recently the prevalence in developed countries is reported much less than our part of the world. A study from South Africa by Syed AR et al\textsuperscript{17} showed a decline in the prevalence of NTDs from 1.41to 0.98 per 1000 births after folic acid fortification. Similarly, another study from Canada, by De Wals P et al.\textsuperscript{18} showed the decrease in prevalence from 1.89 to 1.28 per 1000 total births with folic acid fortification. This decrease in prevalence is attributed to Folic acid fortification and screening. Anencephaly was the most frequent NTD anomaly in our study with prevalence of 57.89% followed by Spina bifida with hydrocephalus, 31.57%. In study by Abbottabad,\textsuperscript{14} Anencephaly was the commonest anomaly consisting of 63 cases out of 69 followed by Spina bifida 4 cases only. Similarly, Study conducted at JPMC, reported by Jooma R, 13 anencephaly was the commonest anomaly consisting upon 19/34 cases. However, in a study from India by Mahadevan B et al\textsuperscript{19} spina bifida (54.8%) was most common defect followed by anencephaly (31.6%), and encephalocele (11.6%). Similarly, in worldwide study by Zaqanjor I, et.al.\textsuperscript{11} spina bifida was highest in percentage to total NTD prevalence, followed by anencephaly and encephalocele. This variability is like the one in the prevalence of NTDs in different parts of the world.

**Conclusion:**
The Prevalence of Neural tube defects in our study are comparable to local studies and the studies from south east Asia. Anencephaly is the most common anomaly followed by Spina bifida with hydrocephalus in our Study.
References: