



Possible decline in Neural Tube Defects by Controlling avoidable risk Factors

¹Pushpa Goswami,²Samreen Memon, ³Mazhar Abro

¹Assistant Professor, Department of Anatomy, Liaquat University of Medical and Health Sciences, Jamshoro Sindh Pakistan

²Associate Professor and Chairperson, Department of Anatomy, Liaquat University of Medical and Health Sciences, Jamshoro Sindh Pakistan

³Senior Lecturer, Department of Anatomy, Liaquat University of Medical and Health Sciences, Jamshoro Sindh Pakistan

¹Corresponding Author's Email: Pushparamesh1998@gmail.com

ABSTRACT

Neural tube defects (NTDs) are common and major birth defects with incidence of different NTDs vary according to the geographic locale, race, family history and gender of the baby. The current study was conducted in order to determine the risk factors associated with NTD in patient admitted at Liaquat university hospital. Due to lack of balanced diet and late/ no diagnosis in antenatal period in our setup the burden of NTDs is quite high with no reliable statistics. . A total 45 patients were studied in this study. All are admitted throughout patient department. A detailed history for any risk factor, family history of NTDs along with biodata was taken and recorded on a proforma. A thorough physical examination was performed, type of neural tube defect noted. During study period total 45 patients with varying age groups ranging between 01 day old and 05 years old were examined, out of which 17(38%) were male and 28(62%) were female patients. Mothers of the 45 patients were aged between 19 and 35 years of age and only 12 (27%) had used folic acid supplements during pregnancy while 33(73%) never used. Thirty five mothers (78%) were exposed to passive smoking and 10 (22%) had a history of active smoking. There was no history of consanguinity in 14 (31%) cases, while 16 (35%) had 1st cousin and 15(33%) had 2nd cousin marriage. Only 09 out of 45 cases of (20%) NTDs were diagnosed before birth while rests were (80%) diagnosed after birth.

Key words: Consanguinity, Folic acid, NTDs, Smoking, Ultrasound

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INTRODUCTION

Neural tube defects (NTDs) are foremost birth defects with incidence of different NTDs vary according to the geographic locale, race, family history and gender of the baby. Whites are affected more than blacks; females are twice affected than males. The precise reason behind these defects remains unidentified. Mostly NTDs results from improper closure of neural tube during early period of development. Nutritional factors like reduced level of folic acid impact grossly on its prevalence. There is significant association of low social class and cousin marriages with NTDs which is also common in our population. [1, 2]

Some NTDs like Mylomengiocele is twice common in first cousin marriage as compared with general population. One pregnancy with NTDs carries increased risk of NTDs in subsequent pregnancies. It is reported in previous studies that the ratio of couples having one previous baby with NTDs is 1 in 25 while with two past pregnancies this ratio increased to 1 in 10. [3, 4]

Many studies have reported a declining rate of NTDs at birth due to better health care facilities. Proper use of folic acid during pregnancy is reducing NTDs about seven times and medical termination of pregnancy after ultrasound screening in the prenatal diagnosis of severe and lethal types of NTDs are proved to be key elements in reduction of NTDs. [5]

During antenatal checkups mother having not only have routine checkups but health education is also given on various maternal and child care issues like proper nutrition, avoidance of possible teratogens like drugs, smoking and radiation. Multi vitamins containing folic acid are also given free in such clinic in

order to achieve better outcomes of pregnancy. Fortification of a basic food ingredient with folic acid is ideal for ensuring adequate blood levels in the mother in peri-conceptional period as most pregnancies are unplanned and NTDs arise in the fetus in early period of development in which most women are unaware that they are pregnant. When she comes to a health care worker for antenatal care, prevention of the anomalies by folic acid supplementation is not possible. Currently fortification of folic acid is 240 micrograms per 100 gram of flour is recommended. [6, 7]

Ultrasound is the diagnostic tool of choice in the detection of NTDs as it is easily available, cheaper, cost effective and safe during pregnancy with main and only drawback is its dependency on observer. Recent advances in Ultrasound such as 3D/4D techniques will improve its diagnostic efficacy even in minor congenital anomalies. [8]

Consanguinity or cousin marriage or inter-familial marriages between close relatives or same grandparents is deeply rooted social trend which is common in our Pakistani culture as favored by religious and legal grounds also. Pakistan is one of the country which has a high rate of cousin marriages and 50% of all marriages occur between first cousins which are lower in urban areas (38%) compared to rural areas (54%) and about 53% in Sindh province. Studies suggest that cousin marriages may have genetically detrimental effects so carries the highest prevalence. [9, 10, 11]

This study is conducted to outline the risk factors associated with NTD in patient admitted at Liaquat university hospital which is a public sector hospital with flow of low resource and uneducated patients heavily. Due to lack of balanced diet and late/ no diagnosis in antenatal period in our setup the burden of NTDs is quite high with no reliable statistics. Results from this study will help in refining the health care system for managing and rehabilitation of patients with NTDs in this particular locale. Knowledge of associated factors may high light their roles as risk factors for the occurrence of NTDs so providing baseline data for upcoming studies.

METHODOLOGY

This descriptive study was conducted in Department of neurosurgery at Liaquat University hospital Jamshoro with collaboration of Department of Anatomy of Liaquat University of Medical and Health Sciences from September 2013 to December 2013. A total 45 patients were studied in this study. All are admitted throughout patient department. A detailed history for any risk factor, family history of NTDs along with biodata was taken after informed consent from mother or attendant of patient. A thorough physical examination was performed, type of neural tube defect noted. Ultrasound reports of antenatal checkups which were available were thoroughly evaluated. Data was recorded on proforma and analyzed. [12]

RESULTS

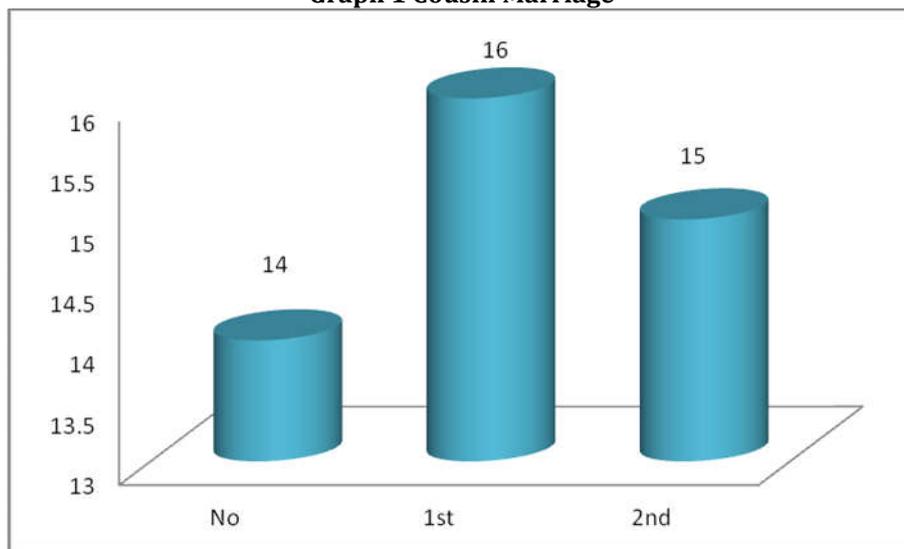
During study period total 45 patients with varying age groups ranging between 01 day old and 05 years old were examined, out of which 17(38%) were male and 28(62%) were female patients as shown in table 1. Parental demographics showed that age of mothers of the 45 patients were between 19 and 35 years of age and only 12 (27%) had used folic acid supplements during pregnancy while 33(73%) never used. Thirty five mothers (78%) were exposed to passive smoking and 10 (22%) had a history of active smoking (table 2). There was no history of consanguinity in 14 (31%) cases, while 16 (35%) had 1st cousin and 15(33%) had 2nd cousin marriage seen in graph 1. The results of this study demonstrate that only 09 out of 45 cases of (20%) NTDs were diagnosed before birth while rests were (80%) diagnosed after birth. In diagnosed cases 02 were diagnosed at rural and 07 at urban, while in undiagnosed cases 30 at rural and 06 at urban health care centers as seen in table 3 and table 4 is showing no. of ultrasound patient underwent in diagnosed and not diagnosed cases. Different types on NTDs are shown in table 5 with the commonest most is hydrocephalus (55%) followed by meningocele (13%).

Table 1 Age and Gender of Patients n=45

Age	Gender	
	Male	Female
Neonate	02	08
Infant	10	13
>1 year	05	07
	17 (37%)	28(62%)

Table 2 Maternal Demographics

Age of mothers		
Group-1	17-23 years	16
Group-2	24-30 years	24
Group-3	31-37 years	05
	Total	45
Smoking	Active smokers	Passive smokers
	10	35
Use of folic acid	Yes	No
	12	33

Graph 1 Cousin Marriage**Table 3 No. of cases diagnosed on ultrasound before and after birth in different demographic locations**

Diagnosis on antenatal u/sound before birth	
Rural	Urban
02	07
Total = 09(20%)	
Diagnosis on antenatal u/sound after birth	
Rural	Urban
30	06
Total = 36(80%)	

Table 4 No. of ultrasound in patients

No. of ultrasound in patients of NTDs							
Diagnosed after birth	Nil	01	02	03	04	05	Total No.
	08	09	04	08	05	02	36
Diagnosed before birth	00	03	01	02	03	00	09

Table 5 Types of NTDs

Type of Neural tube defect	NO. of patients	Frequency
Myelomeningocele	04	8.8 %
Dermoid/ congenital dermal sinus	01	2.2 %
Meningocele.	06	13.3%
Hydrocephalus	25	55.5 %
Lipomyelocele & meningocele	01	2.2 %
Hydrocephalus & meningocele	04	8.8 %
Dandy Walker syndrome	01	2.2 %
Encephalocele & microcephaly	01	2.2 %
Encephalocele & hydrocephalus	02	4.4 %
	45	

DISCUSSION

Birth defects are main cause of disease and death in new born with mental distress and economic burden on parents and society. Approximately 2-3 per 100 children are born with some kind of birth defects around the globe every year. Out of all anomalies 2.5/1000 babies are born with NTDs. [13, 14]

There is plenty of data available on risk factors and prenatal diagnosis of NTDs throughout world including Pakistan but this is the first study conducted at LUMHS to evaluate the possible risk factors in still worrisome situation of NTDs in rural areas of Sindh Pakistan. In particular this study was intended to get an idea about major predisposing factors of different NTDs.

The results demonstrate the higher incidence of non-compliance in use of folic acid, cousin marriage and failure of NTDs diagnosed in antenatal period as major predisposing factors. Majority of patients are rural dwellers where education and literacy rate is very low, females are treated unfairly compared to males not properly provided with better diet and basic health facilities. Moreover, the population in these rural areas belongs to low socioeconomic status, cousin marriages are commonly practiced in Muslims community which covers most of the population which may be the possible factors in causation.

The incidence of different NTDs varies according the geographic conditions, race and gender of baby. It is twice common in females than males, in this study a 28 (62%) female in 45 cases compared with 17 (37%) males almost same as seen in other studies. The age of mothers ranges between 19 and 35 years with no specific association in occurrence of NTDs which were usually seen in mothers older than 35 years which is only one seen in this study. [10, 15]

Regarding use of folic acid which is supposed to be vital in development of neural tube was taken by only 12 (27%) mothers during pregnancy while 33(73%) never used. Folic acid supplementation decreases the risk of NTD by 35-70% but a non-declining birth incidence of NTDs in our setup need evaluation. Possible causes of failures may be insufficient recommendations, non-compliance of mothers, and folic acid fortification of food moreover due to poverty there is lack of balanced diet and health care facilities. Study conducted by Farzana in KPK also showing no use of folic acid in all cases of NTDs. Authors find folic acid much better in prevention of spina bifida than anencephaly and encephalocele. [16, 17]

Thirty five mothers (78%) were exposed to passive smoking and 10 (22%) had a history of active smoking which in similar in studies conducted by others which shows passive smokers are similarly effected or even more prone to congenital defects than active smokers. [18, 19]

Consanguinity of the parents is one the significant risk factors for CNS anomalies. In our study no history of consanguinity in 14 (31%) cases, while 16 (35%) had 1st cousin and 15(33%) had 2nd cousin marriage in total (59%) which is significantly high than study conducted by Murshid (26%) and other authors (12%) and (10-45%) probably due to 99% population is Muslim. [20, 21, 22]

Ultrasound offers the possibility of early and correct diagnosis of neural tube defects. Detection of fetal abnormalities depends on type of abnormality, gestational age and sophistication of equipment and experience of operator. In this study only 09 out of 45 cases of (20%) NTDs were diagnosed before birth while rests were (80%) diagnosed after birth. In diagnosed cases 02 were diagnosed at rural and 07 at urban, while in undiagnosed cases 30 at rural and 06 at urban health care centers. These results show medical negligence contrary to medical ethics but it may be the result of deprived health facilities, furthermore one reason is the patient's awareness for seeking medical care as shown in table 4 in which 08 cases never had ultrasound or antenatal checkup therefore NTD in them diagnosed after birth. . [23]

The most common NTDs in Pakistan are the hydrocephalus and anencephaly. In this study Hydrocephalous accounts for 25 cases (55.5%) hydrocephalus with meningocele in 01 case (2.2%) and hydrocephalus with encephalocele in 02 cases (4.4%) is seen. Myelomeningocele seen in 04 cases (8.8%), Meningocele in 06 cases (13.3%), congenital dermal sinus and dandy walker syndrome 01 (2.2%) cases of each, also seen in this study 01 cases of Lipomyelocele with mengiocele and 01 case of Encephalocele with microcephaly which accounts for 2.2% of all neural tube defects.

Our findings are similar with study conducted by Perveen F et al., they also reported hydrocephalus and anencephaly most common NTDs in Pakistan. Another study conducted in military hospital Rawalpindi by Sadaf Moin et al., showed that 42.1% fetuses were hydrocephalic and 15.8% had spina bifida with meningocele. As this study was conducted on patients admitted in neurosurgery no anencephalic baby was seen as they are stillborn or die soon after birth. In our study only 1 patient (5.3%) had dandy walker syndrome which is very low when compared to 2-17% published in data. [24, 25, 26]

CONCLUSION

In nut shell three main risk factors were observed in this study; no use of folic acid, improper diagnosis in antenatal period on ultrasound and cousin marriages and all these seemed to be based on poverty and unawareness. For raising public awareness on possible preventive measures for congenital disorders pre-pregnancy and counseling on consanguinity, to ensure folic acid consumption public education to the

target audience by various communication media, provision of folic acid supplement to women of reproductive age with the help of health care professionals and proper convincing and referral of patient for at least a single fetal anomaly scan in early pregnancy by expert sonologist may be helpful in future.

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