International Journal of Research in Medical Sciences
Goswami P et al. Int J Res Med Sci. 2015 Jul;3(7):1707-1711
www.msjonline.org
pISSN 2320-6071 | eISSN 2320-6012
DOI: http://dx.doi.org/10.18203/2320-6012.ijrms20150256

Research Article

Frequency and variation of neural tube defects at Liaquat University Hospital Jamshoro, Sindh, Pakistan

Pushpa Goswami[1]*, Samreen Memon[1], Vashdev Khimani[2], Farhana Rajpar[4]

[1]Department of Anatomy, Liaquat University of Medical and Health Sciences, Jamshoro, Sindh, Pakistan
[2]Department of Neurosurgery, Liaquat University Hospital Jamshoro, Sindh, Pakistan

Received: 15 May 2015
Accepted: 05 June 2015

***Correspondence:**
Dr. Pushpa Goswami,
E-mail: pushparamesh1998@gmail.com

ABSTRACT

Background: Neural tube defects (NTDs) are a group of congenital malformations of the neural tube. The incidence of NTDs in Pakistan is 13.90 per 1000 deliveries. Such type of study in our setup is conducted for the first time which shows that different varieties of (NTDs) are seen commonly in our setup also.
Methods: This descriptive study was conducted in department of Neurosurgery of Liaquat University Hospital Jamshoro, Sindh Pakistan with collaboration of department of Anatomy of Liaquat University of Medical and Health Sciences Jamshoro to determine the frequency and variations of neural tube defects seen in this institution. 45 patients with (NTDs) were included in the study. Out of 45 patients 18 were males and 27 were females. All were admitted through the outpatient department. The type of anomaly was recorded with other bio data.
Results: 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%) in addition 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.
Conclusions: The high occurrence of NTDs observed in this study calls for a special attention. A nationwide surveillance can recognize the disease burden in pre & post-natal period and related risk factors to plan future strategies for prevention, early diagnosis and timely management.

Keywords: Folic acid, Hydrocephalus, Meningocele, Neural tube defects

INTRODUCTION

Neural tube defects (NTDs) are the second most common severe disabling congenital defects. They occur because of defect in the neurulation process which begins in fourth week of intrauterine life. The neuroectoderm forms neural plate then neural folds which meet in mid line to form neural tube. The ends of the tube i.e. anterior and posterior neuropores close in the last on day 25 and day 27, so they are the most vulnerable to defects. As a result majority of NTDs arise in these areas i.e anencephaly and encephalocele in anterior and spina bifida posterior neuropore's abnormal closure respectively. Anencephaly and spina bifida so account for up to 95% of all NTDs.[1,2]

Etiology of NTDs reflect a combination of genetic predisposition, various nutritional and environmental factors like female baby, family history of NTDs, obesity, pregestational diabetes, gestational diabetes, low dietary folate intake, lack of folic acid supplementation, use of anticonvulsant drugs (sodium valporate, carbamazepine), use of folic acid antagonists (e.g. methotrexate) or genetic factors causing abnormal folate metabolism.

NTDs can be classified on embryological basis and presence or absence of exposed neural tissue, as open or closed types.[3]

Open NTDs: frequently involve the entire central nervous system; neural tissue is exposed with associated leakage of cerebrospinal fluid (CSF) due to failure of primary neurulation. It include associated hydrocephalus and Arnold- Chiari malformation.

Closed NTDs: Confined to the spine with the brain rarely involved result from a defect in secondary neurulation. Neural tissue is not exposed.[4,5]

Cranial presentations include the following

1. *Anencephaly* (Cranial vault is absent)
2. *Encephalocele* (meningocele or meningomyelocele) Meninges or brain matter herniates through a defect in skull.
3. *Craniorachischisis totalis*: congenital fissure of the skull and vertebral column.
4. *Congenital dermal sinus*: Often found in the occipital and lumbosacral areas and can connect the skin surface to the dura or to an intradural dermoid cyst.

Spinal presentations include the following

1. Spina bifida.

- *Spina bifida occulta*: Isolated laminar defects the only clinical sign is a tuft of hair or dimple at the site of defect.
- *Myelomeningocele*: Protrusion of the spinal cord with meninges outside the spinal canal.
- *Meningocele*: Protrusion of the meninges outside the spinal canal.
- *Myeloschisis*: Open neural plate covered secondarily by epithelium.

2. *Lipomatous malformations* (lipomyelomeningoceles) is lipoma or lipofibroma attached to the spinal cord.

3. *Split-cord malformations*: Sagittal cleft dividing the spinal cord into two halves, each surrounded by its pia mater.

- Diastematomyelia
- Diplomyelia

4. *Congenital dermal sinus*.

5. *Caudal agenesis* is a rare congenital anomaly resulting from an insult to the structures of the caudal eminence may be associated with anomalies of hind gut and urogenital system as they are derived from the caudal eminence.[6]

Hydrocephalus also termed as hydrodynamic Cerebrospinal fluid (CSF) disorder. It is characterized by increased CSF accumulation in central nervous system (CNS) because of imbalance between formation and absorption. This condition may be seen alone or in association with other open NTDs. Hydrocephalus may result from inherited genetic abnormalities like aqueductal stenosis or developmental disorders associated with neural tube defects like spina bifida and encephalocele. It may results from intraventricular hemorrhage, meningitis, tumors, traumatic head injury, or subarachnoid hemorrhage which block the exit of CSF from the ventricles to the cisterns.[7]

Due to lack of balanced diet in our setup the incidence of NTDs is quite high. Therefore a periconceptional folate supplementation for women who are planning to become pregnant should take 400 micrograms of folic acid daily before conception and during the first 12 weeks of pregnancy, fortification of food with the addition of folate, avoidance of anti folates has a strong protective effect against NTDs.

Affected children will require treatment by expert team of surgeons, physiotherapist and pediatrician for associated physical, developmental and learning difficulties which frequently occur in association with NTDs. The newborn with an open NTD kept warm and in prone position to prevent pressure effects with defect covered by a sterile saline dressing. Open NTDs should be closed promptly. In hydrocephalus ventriculoperitoneal shunt placed at the time of myelomeningocele closure. [8, 9]

Due to poverty, unawareness, myths commonly seen in our setup avoiding prenatal routine screening by alpha feto protein (AFP) and /or ultrasound, lack of facilities in diagnosing birth defects in second trimester leading to majority of cases diagnosed at the time of delivery or after birth. Alpha-fetoprotein in maternal serum is best detected at 16-18 weeks of pregnancy. Ultrasound is an effective technique for detecting NTDs than serum alpha-fetoprotein. It can detect anencephaly from the 12th week and spina bifida from 16-20 weeks. Second-trimester ultrasound examination increases detection rate of spina bifida to 92-95% and detection of anencephaly to 100%.[10,11]

Health care system in our setup is still needs lot of improvements in diagnosing and treating NTDS in order to provide maximum cure with minimum disabilities. In highly developed setups of USA in utero surgical repair has been practiced since many years.[12]

METHODS

This descriptive study was conducted in department of neurosurgery at Liaquat University hospital Jamshoro from September 2013 to December 2013. A total 45 patients were studied in this study. All are admitted through out 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 after confirmation of internal defect by

investigations. MRI for imaging neural tissue and for identifying contents of the defect in the new born, CT scan for direct visualization of the bony defect and anatomy. Ultrasound reports of antenatal check-ups if available were considered significant.[13]

RESULTS

During study period total 45 patients were examined, 18 males and 27 females of different age group between 01 day to 05 years, type of anomaly was noted with other details as shown in Table 1 frequencies of each anomaly is calculated and shown in Table 2.

Table 1: Showing age, gender and anomaly.

S. No	Age	Gender	Defect
1	18months	Male	Hydrocephalus
2	2 months	Male	Meningocele.
3	3 day	Female	Meningocele.
4	2 year	Female	Hydrocephalus,
5	6 month	Female	Encephalocele & hydrocephalus
6	2 month	Male	Hydrocephalus
7	19 month	Female	Hydrocephalus
8	7 days	Female	Meningocele
9	5 month	Male	Hydrocephalus
10	5 year	Male	Hydrocephalus
11	2 month	Female	Hydrocephalus & Meningocele
12	2 month	Female	Hydrocephalus & Meningocele
13	5 month	Female	Hydrocephalus
14	8 days	Female	Mylomengiocele
15	3 month	Male	Hydrocephalus
16	3 month	Female	Hydrocephalus
17	1 year	Male	Hydrocephalus
18	3 month	Male	Hydrocephalus
19	3 month	Female	Hydrocephalus
20	16 days	Female	Myelomengiocele
21	5 month	Female	Hydrocephalus
22	1 year	Female	Hydrocephalus
23	1 day	Male	Encephalocele & microcephaly
24	3 year	Male	Hydrocephalus
25	6 month	Female	Hydrocephalus
26	1.5 year	Female	Encephalocele
27	2 month	Male	Meningocele
28	8 month	Male	Hydrocephalus
29	2 month	Female	Meningocele
30	2 month	Female	Hydrocephalus & mengiocele
31	16 days	Female	myelocele
32	9 day	Male	Hydrocephalus
33	1 month	Male	Hydrocephalus & mengiocele
34	4 year	Male	Dandy Walker syndrome
35	18 months	Female	Dermoid
36	1 year	Male	Hydrocephalus
37	9 day	Female	Hydrocephalus
38	18months	Male	Lipomyelocele & mengiocele
39	1 month	Female	Hydrocephalus
40	3 month	Male	Hydrocephalus
41	13 day	Female	Mengiocele
42	1 month	Female	Myelomengiocele
43	4 month	Female	Hydrocephalus
44	9 month	Female	Hydrocephalus
45	7 days	Female	Hydrocephalus

Table 2: Showing no. Of cases and frequency of NTDs.

Type of Neural tube defect	NO. of patients affected	Frequency
Myelomeningocele	04	8.8 %
Dermoid/ congenital dermal sinus	01	2.2 %
Meningocele.	06	13.3%
Hydrocephalus	25	55.5 %
Lipomyelocele & mengiocele	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

The birth of an infant with congenital malformations particularly if not diagnosed in antenatal or even diagnosed causing mental distress to parents because of poor outcomes. Around the world 2-3 per 100 children are born with birth defects out of which 2.5/1000 babies are born with neural tube defects. NTDs are major morbid and mortal birth defects.[14]

Although different studies have been undertaken in different parts of world as well as in Pakistan, but no such study has been undertaken in Liaquat University Hospital to best of our knowledge. Thus this study was intended to to get an idea about the frequency and variation of different NTDs in our set up and its most cost effective investigations and management in this region for public awareness.

The higher incidence of NTDs in this study is probably because of being LUH is a big public sector tertiary care hospital providing health care facilities to all over Sindh province as a referral unit, dietary deficiency of folate, multi-parity, low socio economical status and cousin marriages are the major predictors in our setup.

The incidence of different NTDs varies according the geographic conditions, race, sex of baby and certain maternal factors. It is more common in white than black and twice as common in females seen in this study also which shows 27 (60%) female in 45 cases.[15]

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%) in addition 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 supported by a study conducted in Pakistan by Perveen F et al., as they have 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 (maximum) were hydrocephalic and 15.8% were 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.[16,17]

The etiology of NTD may consist of genetic and environmental factors. Prevalence of syndromic NTDs like Dandy walker syndrome which is characterized by enlargement of the fourth ventricle, complete absence of the cerebellar vermis, cyst formation near the internal base of the skull and hydrocephalus also varies in different parts of the world. In our study only 1 patient (5.3%) had dandy walker syndrome as compared to 2-17% published in data.[18]

Folic acid supplementation reduces the risk of NTD by 35-70% but a non-declining birth prevalence of NTD is a concern to our country as well as many other countries world-wide. Failure shows insufficient recommendations regarding folic acid fortification of food.[19]

Prenatal diagnosis has improved during the last 30 years and various methods of pre natal diagnosis are available. But due to lack of awarness and eonomy still not practiced in most of our surroudings. There is a high chance of recurrence of NTDs in successive pregnancies and proper preconception counseling is needed which is lacking in our setup. The diagnosis of NTDs relies on detailed ultrasound examination of the fetus along with biochemical examination of amniotic fluid.[20]

Emotional stress for parents needs counseling for early surgical intervention for better outcome. Termination of pregnancy on medical grounds for anomalies incompatible with life needs appropriate law and order.

Funding: Nil
Conflict of interest: No
Ethical approval: Not required

REFERENCES

1. Moore KL, Persaud TVN, Mark G. "Development of Nervous system"The Developing Human: clinically oriented embryology 9th ed Philadelphia, Saunders, 2011; 223-229.
2. Dias MS, Li V. Pediatric neurosurgical disease. Pediatr Clin North Am. 1998;45(6):1539-78.
3. Agopian AJ, Tinker SC, Lupo PJ, Canfield MA, Mitchell LE. Proportion of neural tube defects attributable to known risk factors. Birth Defects Res A Clin Mol Teratol. 2013;97(1):42-6.
4. Harris LW, Oakes WJ. Open neural tube defects. In: Tindall GT,Cooper PR, Barrow DL, eds. The Practice of Neurosurgery. Baltimore: Williams & Wilkins; 1996; 2779-89.
5. McComb JG, Chen TC. Closed spinal neural tube defects. In: Tindall GT, Cooper PR, Barrow DL, eds. The Practice of Neurosurgery. Baltimore: Williams & Wilkins; 1996: 2754-77.
6. McComb JG. Spinal and cranial neural tube defects. Semin Pediatr Neurol. 1997;4(3):156-66.
7. Rekate HL. A contemporary definition and classification of hydrocephalus. Semin Pediatr Neurol. 2009;16(1):9-15.
8. Lumley J, Watson L, Watson M, Bower C; Periconceptional supplementation with folate and/or multivitamins for preventing neural tube defects [Cochrane Database Syst Rev. 2011];(3):CD001056.
9. Hamilton MG. Treatment of hydrocephalus in adults. Semin Pediatr Neurol. 2009;16(1):34-41.
10. Norem CT, Schoen EJ, Walton DL, et al; Routine ultrasonography compared with maternal serum alpha-fetoprotein for neural tube defect screening. Obstet Gynecol. 2005;106(4):747-52.
11. Cameron M, Moran P; Prenatal screening and diagnosis of neural tube defects. Prenat Diagn. 2009;29(4):402-11.
12. Adzick NS, Thom EA, Spong CY, et al; A randomized trial of prenatal versus postnatal repair of myelomeningocele. N Engl J Med. 2011;364(11):993-1004.
13. Islam M, N, Siddika M, Bhuiyan M. K. J., Chowdhury A. M. Pattern of Congenital Anomalies in Newborns in a Tertiary Level Hospital in Bangladesh. Journal of Surgery Pakistan. 2013;18(1):32-36.
14. Shamnas M, Arya PS, Thottumkal VA, Deepak MG. Congenital anomalies: a major public health issue in India. IJPCBS. 2013;3(3):577-85.
15. Khattak ST, Naheed T, Akhter S, Jamal T. Incidence and management of neural tube defect in Peshawar. Gomal J Med Sci. 2008;6(1):41–4.
16. Perveen F, Tayyab S. Frequency and pattern of distribution of congenital malformations in the new

born and associated maternal risk factors. J Coll Physcians Surg Pak. 2007;17:340-3.
17. Sadaf Moin, Rubina Mushtaq, Shabbir Hussain, Moin uddin Sabir. Frequency if Neural Tube Defects Among Low Risk Pregnancies on Folic Acid Supplimentation in Military Hospital. Ann Pak Inst Med Sci. 2013;9(4):176-9.
18. Lynch SA. Non multifactorial neural tube defects. Am J Med Genet. 2005;135:69-76.
19. Eichholzer M, Tonz O, Zimmermann R. Folicacid: a public health challenge. Lancet. 2006;367:1352-61.
20. Norem CT, Schoen EJ, Walton DL, Kreiger RC, O Keefe J, ATo TT, et al. Routine USG compared with SAFP for NTD screening. Obst Gynecol. 2005;106:747-52.

Cite this article as: Goswami P, Memon S, Khimani V, Rajpar F. Frequency and Variation of Neural tube defects at Liaquat University Hospital Jamshoro, Sindh, Pakistan. Int J Res Med Sci 2015;3:1707-11.