

Sonographic Assessment of Neural Tube Defects in 2nd and 3rd Trimester

Dr. Sidra Saeed^{1*}, Dr. Hafiza Noor Fatima¹, Dr. Maleeha Khan¹, Dr. Rabia Ghulam Nabi¹, Dr. Arfa Tareen¹, Dr. Raham Bacha², Dr. Syed Amir Gilani³, Dr. Sajid Shaheen⁷, S. Muhammad Yousaf Farooq⁴, Dr. Faiza Afzal⁵, Mam Mehreen Fatima⁶

¹MBBS, DMRD, University of Lahore, Pakistan

²Ph.D, M.Phil. Ultrasound, University of Lahore Pakistan

³Professor, PhD Ultrasound, University of Lahore, Pakistan

⁴M.Phil. Ultrasound, University of Lahore, Pakistan

⁵MBBS, King Edward Medical University FCPS, Sharif hospital, Neela Gumbad Anarkali, Lahore, Punjab 54000, Pakistan

⁶Biostatistician, University of Lahore, Pakistan

⁷MBBS, DMRD, University of Lahore, Pakistan

*Corresponding author: Dr. Sidra Saeed

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Abstract

Ultrasonography is a widely used imaging technique that has many clinical applications. Most hospitals will offer women at least 2 ultrasound scans during their pregnancy. The first is usually at around 8 to 14 weeks and is sometimes called the 'dating scan' because it can help to determine when the baby is due. Anomaly scan is an ultrasound scan that is carried out around weeks 19 to 20 of your pregnancy. Neural tube defects are abnormalities that occur in the development of the spinal cord and brain in the fetus. This scan aims to identify any physical problems with the baby. During the anomaly scan, we can identify the neural tube defects. Exorbitant choice sonography has improved it practical to picture the developing embryo and analyze fetal anomalies previously the twentieth week of pregnancy. Contingent upon the kind of neural tube defects distinguished, early end of the pregnancy might be thought about. The sonographer, just as the whole obstetric gathering, should be cognizant that there's a range of conditions with different discoveries related to a neural tube defect. Learning and awareness of these conditions will make an examination and control less demanding.

Keywords: Grey Scale Ultrasonography, Neural tube defects, 2nd and 3rd Trimester, Diagnostic Ultrasound.

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INTRODUCTION

Central nervous system (CNS) malformations are the second most frequent category of congenital anomalies, after congenital heart disease. Ultrasound examination is an effective modality for the prenatal diagnosis of these anomalies. An accurate fetal diagnosis depends upon a precise description of the sonographic appearance of the CNS and careful evaluation for associated malformations, which are often present [1]. Neural tube defect (NTD) is a simple term for a congenital malformation of the central nervous system (CNS) existing secondary to lack of closure of the neural tube [2]. They occur in the first month of pregnancy. A baby's neural tube normally develops into the brain and spinal cord. It starts out as a tiny, flat ribbon that turns into a tube by the end of the first month of pregnancy. NTDs happen if the tube doesn't close completely. NTDs can cause serious problems for babies, including death [3, 4]. Folic acid deficiency is thought to be the cause of Neural tube

defects by some authors. If women of childbearing age take 400 micrograms of folic acid every day before and during early pregnancy, it can help reduce their baby's risk for NTDs. Folic acid is a vitamin-B that every cell in the body needs for normal growth and development. Neural tube formation is divided into primary (closure) and secondary (canalization) phases. Primary closure is initiated at several discrete points along the rostrocaudal axis. First, at the boundary between future hindbrain and cervical spine (termed Closure 1), then around 12 hours later at the boundary between future forebrain and midbrain (Closure 2) and soon afterwards at the rostral extremity of the future forebrain (Closure 3) [5, 6]. NTDs are one of those types of abnormalities which can occur in the early development of a baby. The pathophysiology of the neural tube defects is still not known [7, 8]. There are many types of NTDs such as spina bifida which affects the spine and babies with spina bifida may have paralysis of the legs, lack of bladder and bowel control and curvature of the spine. It affects about 1,500 babies a year. Neural tube defects

(NTD) range from the lethal (anencephaly) to the potentially asymptomatic (some closed spina bifida) [9]. Spina bifida outcomes appear to be highly variable and difficult to predict. Anencephaly is one of the most severe NTDs and affects about 1,000 babies each year. It is Failure of closure of the rostral neuropore causes failed cranial vault development and the condition of exencephaly, the precursor of anencephaly. Ossification of the skull vault is not always apparent until 12 weeks' gestation and anencephaly should not be diagnosed before this time [Error! Reference source not found.]. This should be an easy diagnosis to make. Girls are three times more likely than boys to have anencephaly. Encephalocele is a rare NTD that affects the brain and skull. About 375 babies are born with this NTD in the United States each year. Hydrocephalus is a condition in which fluid accumulates in the brain. Globally it is estimated that approx. 300,000 fetuses are born each year with NTD's resulting in approximately 88000 deaths & 8.6% Disability Adjusted Life Years (DELAYS) [11]. An accurate estimate of the prevalence of NTD's in most countries is still unknown due to insufficient data. There seems to be some effect from prenatal diagnosis, genetic causes and possibly nutritional supplementations. NTDs can be classified on an embryological basis and presence or absence of exposed neural tissue, as open or closed types [4]. Open NTDs frequently involve the entire central nervous system; neural tissue is exposed to associated leakage of cerebrospinal fluid due to the failure of primary neurulation. It includes 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 but the direct contact of the meninges with the amniotic fluid and mechanical irritation could cause neural tissue to be exposed and lead to open neural tube defect anencephaly [11-13]. A few of the most common anomalies are discussed. The most common NTD is spina bifida which occurs on the base of the fetal spinal column. Detection rates by routine ultrasound screening should approach 100% for open spina bifida due to the presence of the easily recognizable cranial signs; the "lemon" and "banana" signs [14]. Another anencephaly occurs on the upper part of the neural tube. Which appear during the process of neurulation, a well-defined period between the 17th and 30th day after ovulation. Spina bifida is also a broad term that encompasses several subgroups of defects including myelomeningocele, meningoceles and the lipomeningocele. Encephalocele which occurs seldom. Found when part of the brain and membranes protrude through the skull. Symptoms of NTD's include paralysis, development delays and also seizures. Ultrasound has an important role in the diagnosis of fetal NTD's. Three-dimensional ultrasound can provide excellent views of a spinal lesion. An encephalocele is diagnosed by ultrasound identifying that small amount of extruded brain tissue through the cranial defect. In the US the incidence is approximately 1-2 cases per

1000 live births. Whereas the incidence in the UK is about 4 times greater. Detection of Anencephaly is reported to be 100% at mid-gestation in all cases of NTD's. Long term surveillance of NTDs in countries that have successfully implemented fortification, such as the United States, Canada, Costa Rica, South Africa and Chile and data from a supplementation program in China suggest that folic acid interventions can reduce NTD prevalence to as low as 5-6 per 10,000 pregnancies. The incidence of different NTDs varies according to the geographic conditions, race, sex of the baby and certain maternal conditions [9]. An Arnold Chiari malformation can be appreciated, defined as the displacement of the cerebellar vermis, fourth ventricle and medulla through the foramen magnum. Bananas sign defines the elongated shape of the cerebellum. Lemon sign defines in utero frontal bossing [15]. Ultrasound can play a major role in early detection. Ultrasound is the most effective, non-invasive, specific, low cost and useful technique for confirmation of NTD's and to classify its different types.

MATERIALS AND METHODS

The study design was cross-sectional descriptive study and the calculated sample size was 30. The sample size was collected from the accessible population for 6 months using a consecutive sampling technique. The study was conducted at the Diagnostic Ultrasound department of Indus hospitals.

Sampling Technique

Inclusion Criteria

All pregnant ladies during their pregnancy in the second and third trimester are included (Inclusion Criteria).

Machine

The ultrasound was performed on the Samsung HS40 machine by using a convex probe with the frequency of the 5-12 MHz transducer.

Data Collection and Static Analysis

Data were entered in Microsoft Excel and analyzed by using SPSS version 24. Consent forms were collected from the patients. Data collection sheets were collected during this study. Patients were scanned in the supine position for the transabdominal scan.

RESULTS

Total number of patients included in this study was 30. Out of these 10 patients diagnosed as Anencephaly (33.3%), 8 as hydrocephalus (26.6%), 4 as spina bifida (13.3%), 3 as meningoceles (10%), 2 as lumbar meningoceles (6.6%), 1 as hydranencephaly (3.3%), 1 as holoprosencephaly (3.3%) and 1 as encephalocele (3.3%). According to this, we found anencephaly is more common than any type of other neural tube defects. Figure 1 shows single intrauterine pregnancy corresponding to a sonographic gestational

age of 29 weeks this figure indicated the lumbar Meningocele. Figure-2 shows hydrocephalus with hypoplastic cerebellum corresponding to the gestational age of 27 weeks and 3 days. And in figure-3 shows the holoprosencephaly corresponding to the sonographic gestational age of 28 weeks and 5 days. And secondly, we can see that the disease has mostly been affected by people from 20 to 29 years old. Only 1 out of 30 patients have a genetic disorder. The table shows the occurrence of neural tube defects.

DISCUSSION

Ultrasonography is an important, non-invasive, safe, easily available and cost-effective technique for diagnosis neural tube defects in pregnancy. Secondly, it gives chance to parents to prepare them psychologically, socially, financially and medically for a baby with a health problem. Thirdly it gives chance to

parents for early termination [16]. Numbers of cases with NTDs were so small that we could not do any statically analysis. Many cases of NTDs were diagnosed in the second & third trimester. In our study, 30 patients were selected who were sent to the radiology department for routine ultrasonography regardless of their race, religion and socioeconomic status. Brief clinical history was taken about gestational age, previous pregnancies and expected date of delivery from the patient. The ultrasound was done after informed consent. Gestational age between 18 to 28 weeks is considered the ideal time for anomaly scan because high-quality sonographic images are available at this time [17]. It has been reported by Hegge FN *et al.*, that in 2/3 of prenatally diagnosed fetal anomalies clinical sign and symptoms may develop at 23 to 24 weeks of gestation [18]. In



Fig-1: Lumbar Meningocele (Figure shows single alive intrauterine pregnancy corresponding to the sonographic gestational age of 29weeks)



Fig-2: Hydrocephalus (figure shows hydrocephalus and hypoplastic cerebellum corresponding to the gestational age of 27 weeks and 3 days)

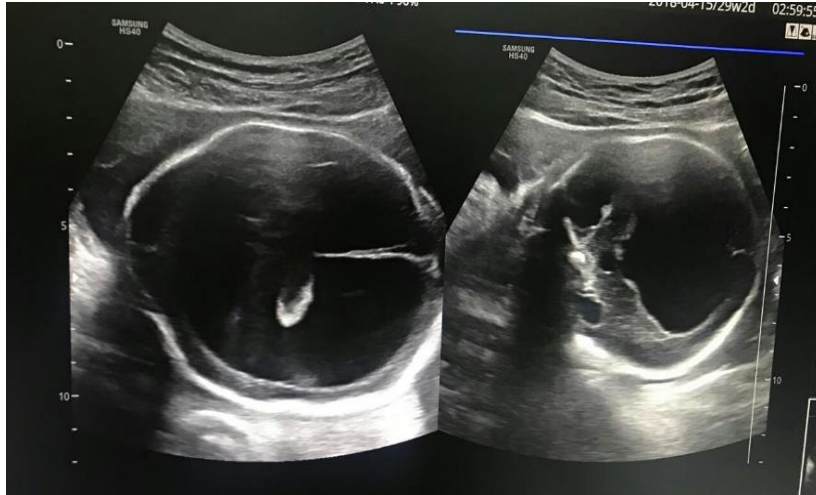


Fig-3: Figure shows holoprosencephaly corresponding to the sonographic gestational age of 28weeks and 5 days

Table-1:

Serial number:	Neural Tube defects	Numbers	Percentage
1	Anencephaly	10	33.3
2	Hydrocephalus	8	26.6
3	Spina Bifida	4	13.3
4	Meningocele	3	10
5	Encephalocele	1	3.3
6	Holoprosencephaly	1	3.3
7	Hydrancephaly	1	3.3
8	Lumbar Meningocele	2	6.6

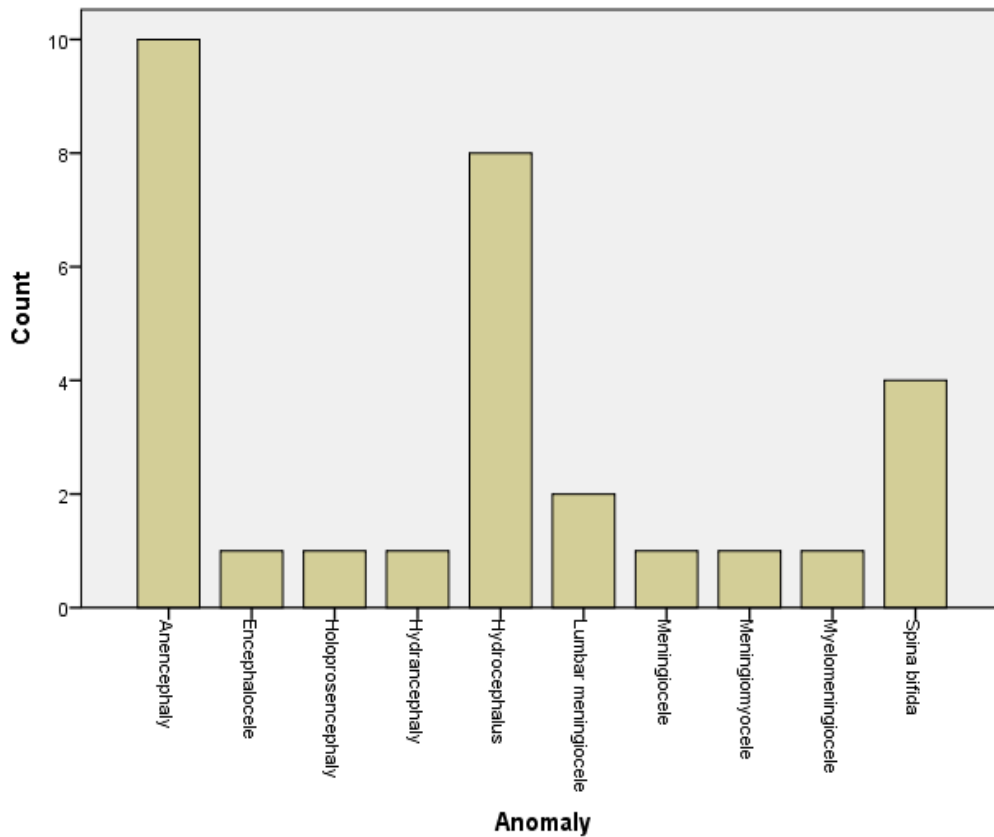


Fig-4: Bar graph shows the ratio between all types of neural tube defects that which occurs the most

This study according to our data which we were collected anencephaly is more common than any other type of anomaly in the fetus. NTD is most common in the low socioeconomic group. This is due to dietary factors and lack of awareness in the lower class. Innovation in clinical management has come from the demonstration that closure of open spina bifida lesions in utero can diminish neurological dysfunction in children [19]. Primary prevention by folic acid has been enhanced through introducing Morris JK et al and Kadir RA *et al.*, reported that the incidence of live births, stillbirths and pregnancies terminated because of a fetal neural tube defect has fallen steadily from 1972 to 1992 due to folic acid therapy of mandatory food fortification in some countries [20]. In another study, Filly RA *et al.*, reported that spina bifida among NTD is most difficult to detect by ultrasound. The transverse plane has been reported as the most useful plane for scanning these defects [21]. Another study was done in order to determine the effect of the absence of a brain on intrauterine and placenta growth, and throughout gestation, 147 pregnancies were analyzed with no brain and compared with the control group. There appears to be a low rate of intrauterine and placenta growth in the anencephalic group. So, there is another cause of anencephalic fetus [22].

CONCLUSION

Exorbitant choice sonography has improved it practical to picture the developing embryo and analyze fetal anomalies previously the twentieth week of pregnancy. Contingent upon the kind of neural tube defects distinguished, early end of the pregnancy might be thought about. The sonographer, just as the whole obstetric gathering, should be cognizant that there's a range of conditions with different discoveries related to a neural tube defect. Learning and awareness of these conditions will make an examination and control less demanding.

REFERENCES

1. Stevenson, R. E., Seaver, L. H., Collins, J. S., & Dean, J. H. (2004). Neural tube defects and associated anomalies in South Carolina. *Birth Defects Research Part A: Clinical and Molecular Teratology*, 70(9), 554-558.
2. Lemire, R. J. (1988). Neural tube defects. *JAMA*, 259(4):558-62.
3. Wallingford, J. B., Niswander, L. A., Shaw, G. M., & Finnell, R. H. (2013). The continuing challenge of understanding, preventing, and treating neural tube defects. *Science*, 339(6123), 1222002.
4. Norwitz, E. R., Arulkumaran, S., & Symonds, I. (2007). *Oxford American handbook of obstetrics and gynecology*. Oxford University Press.
5. Copp, A. J., Greene, N. D., & Murdoch, J. N. (2003). The genetic basis of mammalian neurulation. *Nature Reviews Genetics*, 4(10), 784-793.
6. Copp, A. J. (2005). Neurulation in the cranial region—normal and abnormal. *Journal of anatomy*, 207(5), 623-635.
7. Derbyshire, E. (2011). Nutrition in the childbearing years: John Wiley & Sons.
8. Blom, H. J., Shaw, G. M., den Heijer, M., & Finnell, R. H. (2006). Neural tube defects and folate: case far from closed. *Nature Reviews Neuroscience*, 7(9), 724.
9. Goswami, P., Memon, S., Khimani, V., & Rajpar, F. (2015). Frequency and variation of neural tube defects at Liaquat University Hospital Jamshoro, Sindh, Pakistan.
10. Northrup, H., & Volcik, K. A. (2000). Spina bifida and other neural tube defects. *Current problems in pediatrics*, 30(10), 317-332.
11. Zaganjor, I., Sekkarie, A., Tsang, B. L., Williams, J., Razzaghi, H., Mulinare, J., ... & Rosenthal, J. (2016). Describing the prevalence of neural tube defects worldwide: a systematic literature review. *PloS one*, 11(4), e0151586.
12. Blomberg, M. I., & Källén, B. (2010). Maternal obesity and morbid obesity: the risk for birth defects in the offspring. *Birth Defects Research Part A: Clinical and Molecular Teratology*, 88(1), 35-40.
13. Agopian, A. J., Tinker, S. C., Lupo, P. J., Canfield, M. A., Mitchell, L. E., & National Birth Defects Prevention Study. (2013). Proportion of neural tube defects attributable to known risk factors. *Birth Defects Research Part A: Clinical and Molecular Teratology*, 97(1), 42-46.
14. Copp, A. J., & Greene, N. D. (2013). Neural tube defects—disorders of neurulation and related embryonic processes. *Wiley Interdisciplinary Reviews: Developmental Biology*, 2(2), 213-227.
15. Cameron, M., & Moran, P. (2009). Prenatal screening and diagnosis of neural tube defects. *Prenatal diagnosis*, 29(4), 402-411.
16. King, J. S. (2010). And Genetic Testing for All—The Coming Revolution in Non-Invasive Prenatal Genetic Testing. *Rutgers LJ*, 42, 599.
17. Reddy, U. M., Abuhamad, A. Z., Levine, D., Saade, G. R., & Participants, F. I. W. I. (2014). Fetal imaging: executive summary of a joint Eunice Kennedy Shriver National Institute of Child Health and Human Development, society for maternal-fetal medicine, American Institute of ultrasound in medicine, American College of Obstetricians and gynecologists, American College of radiology, society for pediatric radiology, and society of radiologists in ultrasound fetal imaging workshop. *American journal of obstetrics and gynecology*, 210(5), 387-397.
18. Pilu, G., Perolo, A., Falco, P., Visentin, A., Gabrielli, S., & Bovicelli, L. (2000). Ultrasound of the fetal central nervous system. *Current Opinion in Obstetrics and Gynecology*, 12(2), 93-103.
19. Volpe, J. J., Inder, T. E., Darras, B. T., de Vries, L. S., du Plessis, A. J., Neil, J., & Perlman, J. M.

- (2017). *Volpe's Neurology of the Newborn E-Book*. Elsevier Health Sciences.
20. Greenberg, J. A., Bell, S. J., Guan, Y., & Yu, Y. H. (2011). Folic acid supplementation and pregnancy: more than just neural tube defect prevention. *Reviews in Obstetrics and Gynecology*, 4(2), 52.
 21. Terrone, D. A., & Perry Jr, K. G. (1998). Ultrasound evaluation of the fetal central nervous system. *Obstetrics and gynecology clinics of North America*, 25(3), 479-497.
 22. Honnebier, W. J., & Swaab, D. F. (1973). The influence of anencephaly upon intrauterine growth of fetus and placenta and upon gestation length. *BJOG: An International Journal of Obstetrics & Gynaecology*, 80(7), 577-588.