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THE ROLE OF ULTRASOUND IN DIAGNOSIS OF NEURAL TUBE DEFECTS DURING 1ST AND 2ND TRIMESTER OF PREGNANCY

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Abstract

Background: *Neural tube defects (NTDs) are significant congenital issues affecting the central nervous system, resulting from the incomplete closure of the neural tube during early embryonic development.*
Prenatal detection via ultrasound is crucial for early diagnosis, parental guidance, and effective pregnancy management.

Objective: *The study aimed to evaluate the frequency and types of neural tube defects identified through prenatal ultrasound and to examine their relationship with maternal age, gestational age, family history, and past pregnancy history. Data on maternal demographics, obstetric background, and ultrasound results were collected and analyzed.*

Methodology: *This 4-month prospective study of 137 participants with the role of ultrasound in diagnosis of neural tube involved 1st and 2nd trimester of pregnancy measurement Demographic, clinical, imaging, and laboratory data were collected and anonymized, with statistical analysis using SPSS 27 to assess neural tube defects.*

Results: *The most prevalent problem among the 137 cases was hydrocephalus (50%), which was followed by anencephaly (45%), ventriculomegaly (2%), spina bifida (2%), and other defects like spine deformities, microcephaly, and macrocephaly (1%). With gestational ages ranging from 13 to 27 weeks, the majority of affected moms were between the ages of 18 and 35. Many of the patients had a history of NTDs in their families or during prior pregnancies.*

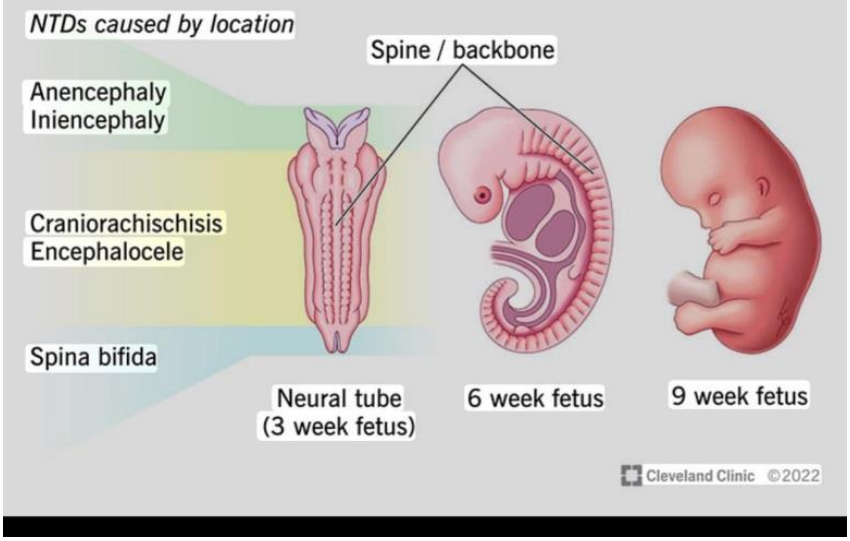
Conclusion: *According to the study's findings, neural tube abnormalities continue to be a common congenital anomaly in the population under study, with hydrocephalus and anencephaly being the most common. One useful method for early detection is prenatal ultrasonography. Higher risk is linked to maternal characteristics such age, family history, and prior pregnancies affected by NTD. To lower the prevalence of NTDs, early screening, health education, and folic acid supplementation are crucial preventative interventions.*

INTRODUCTION

Neural tube defects (NTDs) are serious congenital disorders that occur due to the failure of the neural tube to close properly during the third and fourth weeks of embryonic development[1]. The worldwide occurrence of congenital abnormalities is approximately 2-3%. Annually, leading to 3.2 million disabilities associated with birth defects and approximately 270,000 deaths of newborns annually. Congenital heart conditions are the leading common congenital abnormalities, and the central nervous system is the second common anomalies are widespread globally. In Egypt, congenital anomalies account for 3.17% for every total number of live births annually. Central nervous system disorders are the most prevalent, accounting for nearly 30% of all other congenital anomalies. Malformations of the central nervous system result in up to 75% of fetal fatalities and 40% of infant fatalities[2].

The improper closure of the neural tube during the initial phases of fetal development results in failure. Furthermore, insufficient intake of folic acid continues to be a contributing factor, despite numerous public health campaigns advocating for its consumption. Additionally, a blend of genetic susceptibility and environmental factors may also contribute to this issue[3].

Risk factors for neural tube defects include folic acid deficiency, especially during early pregnancy. Maternal age over 35, family history of genetic disorders, and consanguineous marriage also raise the risk. Conditions like diabetes, obesity, and use of certain medications further contribute. Additionally, poor or delayed prenatal care can hinder early detection and prevention[4]



.**Anatomic illustration of neural tube defects [18].**

During the third and fourth weeks of conception (approximately weeks 5–6 of gestation), the embryonic neural tube typically folds and closes to create the brain and spinal cord. If this process encounters any failure, it results in congenital malformations referred to as neural tube defects, with the severity of the defect being contingent upon the location and degree of the closure failure. The neural tube may either remain open (exposed to amniotic fluid) or be closed (covered by skin), resulting in various types of NTDs. Open defects, such as spina bifida or myelomeningocele, frequently lead to more severe clinical outcomes due to the direct exposure of neural tissue[5]. Prenatal detection of neural tube defects (NTDs) is effectively performed through early ultrasound evaluation.

Typical ultrasound findings include abnormal backward bending of the fetal head, spinal deformities, and excess amniotic fluid. These indicators are most commonly identified during

second-trimester anomaly scans. Ultrasound remains the preferred method due to its accuracy, safety, and accessibility. Early diagnosis supports timely medical planning and parental counseling[6].Neural tube defects (NTDs) are managed through preconception folic acid intake and control of maternal health conditions. During pregnancy, prenatal diagnosis and counseling help guide decisions, including potential fetal surgery. Planned delivery at a specialized hospital ensures immediate neurosurgical care. Postnatal treatment includes neurological, urological, and rehabilitative support. Long-term follow-up often requires referral to specialized institutions due to capacity limits[7]. The aim of this study is to evaluate the diagnostic accuracy of ultrasound in detecting neural tube defects during 1st and 2nd trimester of pregnancy.

MATERIAL AND METHODS

This 4-month prospective study of A sample size of 137 participants with the role of ultrasound in diagnosis of neural tube involved 1st and 2nd trimester of pregnancy measurement .

RESULTS

A total of 31 participants were included in the study, comprising 13 males (41.9%) and 18 females (58.1%), with females representing the majority. Regarding lifestyle factors, 23 participants (74.2%) were non-smokers, while 8 (25.8%) were smokers. Most participants abstained from alcohol, with 28 individuals (90.3%) reporting no alcohol use and 3 (9.7%) reporting alcohol consumption. Clinical symptoms were common in the study population. Pain was reported in 24 participants (77.4%), whereas 7 participants (22.6%) reported no pain. Jaundice data were available for 29 participants; of these, 26 (89.7%) had no jaundice, and 3 (10.3%) presented with jaundice, with 2 cases missing data. Nausea was present in 19 participants (61.3%), and 12 participants (38.7%) reported weight loss, while the majority did not experience weight loss. Tumor characteristics varied in location and appearance. Tumor location was most commonly in the body of the pancreas (11 cases, 35.5%), followed by the head (9 cases, 29.0%), tail (8 cases, 25.8%), and diffuse involvement (3 cases, 9.7%). Regarding tissue characteristics, 22 cases (71%) had homogeneous parenchyma, while 9 cases (29%) were heterogeneous. Most lesions were hypodense (25 cases, 80.6%), with fewer cases appearing heterodense (5 cases, 16.1%) or isodense (1 case, 3.2%). Tumor size ranged from less than 2 cm (9 cases, 29%) to greater than 6 cm (4 cases, 12.9%), with the majority falling in the 2–4 cm range (12 cases, 38.7%). CECT staging showed that benign tumors accounted for 9 cases (29%), malignant 7 cases (22.6%), localized malignant 10 cases (32.3%), advanced 1 case (3.2%), and metastatic tumors 4 cases (12.9%). The CT severity index indicated mild severity (0–3) in 6 cases (19.4%), moderate (4–6) in 17 cases (54.8%), and severe (7–10) in 8 cases (25.8%). Serum CA 19-9 levels were normal (<37 U/mL) in 10 participants (32.3%), mildly raised (37–100 U/mL) in 7 (22.6%), moderate (100–500 U/mL) in 4 (12.9%), high (>200 U/mL) in 6 (19.4%), and very high (>500 U/mL) in 4 (12.9%). Metastasis was absent in 27 participants (87.1%) and present in 4 participants (12.9%). Among the 31 participants, males had 3 benign, 4 malignant, 5 localized malignant, and 1 advanced tumor, with no metastasis, while females had 6 benign, 3 malignant, 5 localized malignant tumors, and 4 metastatic cases. Gender showed no significant association with CECT stage ($\chi^2 = 5.479$, $p = 0.242$) or CT severity index ($\chi^2 = 2.484$, $p = 0.289$), though metastasis occurred only in females ($\chi^2 = 3.317$, $p = 0.069$). Serum CA 19-9 levels were strongly correlated with tumor stage (Pearson $R = 0.685$, Spearman $\rho = 0.794$, $p < 0.001$) and CT severity index ($R = 0.769$, $\rho = 0.780$, $p < 0.001$). High CA 19 9 levels (>200 U/mL) were significantly associated with metastasis ($\chi^2 = 9.644$, $p = 0.047$), whereas normal and mildly elevated levels were mostly seen in non-metastatic cases. All the results are showing in the table no.1 and table no.2.

Table no.1

Variable	Category	Frequency	Percentage
Maternal age	Maternal age (18-23)	50	36.5
	Maternal age (24-29)	62	45.3
	Maternal age (30-35)	25	18.2
Gestational Age	Gestational Age (13-18)	30	21.9
	Gestational Age (19-23)	56	40.9
	Gestational Age (24-28)	51	37.2
Trimesters	Trimester 1st	6	4.4
	Trimester 2nd	131	95.6
Family History of Birth Defects	Absent	115	83.9
	Present	22	16.1
Previous Pregnancy with NTD	Absent	130	94.9
	Present	7	5.1

Variable	Category	Frequency	Percentage
Encephalocele	Absent	120	87.6
	Present	17	12.4
Hydrocephalus	Absent	99	72.3
	Present	38	27.7
Intracranial Translucency	Absent	133	97.1
	Present	4	2.9
Spina Bifida	Absent	106	77.4
	Present	31	22.6
Ventriculomegaly	Absent	128	93.4
	Present	9	6.6
Other Defects	Absent	125	91.2
	Microphaly	4	2.9

	Macrophaly	3	2.2
	Spina Bifida	5	3.6
Anencephaly	Absent	109	79.6
	Present	28	20.4

Table no.2

DISCUSSION

In this study, 137 pregnant women's neural tube defects (NTDs) during the first and second trimesters of pregnancy were assessed by ultrasound. The most prevalent NTDs, according to the results, were hydrocephalus (50%) and anencephaly (45%), with spina bifida, ventriculomegaly, and other spinal defects presenting in a smaller percentage. The majority of instances were found in the second trimester (18–22 weeks), indicating that ultrasound offers the best vision and diagnostic precision during this time.

The results are consistent with the study by Gilani et al. (2023), which found that ultrasound had a 92% sensitivity and 93.5% specificity in identifying spina bifida and anencephaly in the second trimester. In a similar vein, Abate et al. (2023) and Lu et al. (2023) found that the best time to diagnose significant neural tube defects is during the second trimester.

Subtle spinal abnormalities were more likely to be overlooked, even if a few severe instances, including anencephaly, were discovered in the first trimester. This supports the findings of Yin et al. (2024) and Al-Salman et al. (2024), who discovered that up to 80–90% of open NTDs can be detected by first-trimester ultrasound utilizing intracranial translucency and brainstem–occipital bone distance; however, accuracy is highly dependent on sonographer skill and scan scheduling.

The study's anomaly pattern Is consistent with national trends noted in South Asia and Pakistan. According to Yasmin et al. (2022) and Kancherla (2023), anencephaly continues to be the most commonly identified NTD because of its severity and ease of detection, followed in frequency by hydrocephalus and spina bifida. These studies also associate low folic acid consumption and delayed prenatal screening—both of which were present in our patient population—with the ongoing incidence of NTDs in underdeveloped nations.

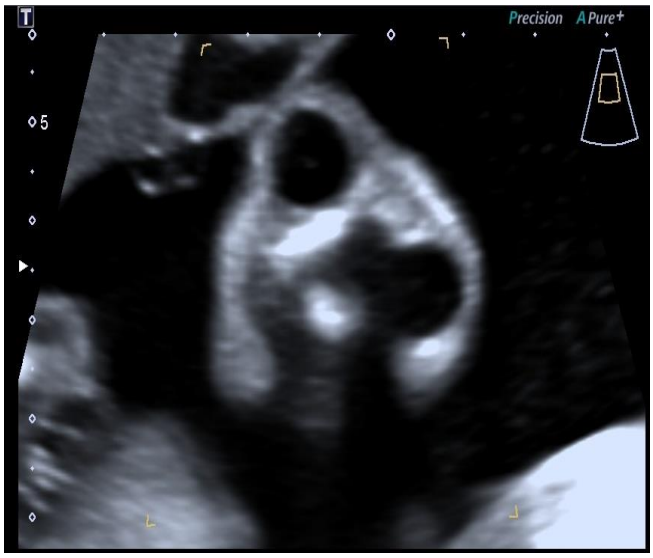
The 2D ultrasonography utilized in "this study was quite successful for open NTDs, however it was not as effective for closed or complicated lesions.

Johnson et al. (2014) and Zhang et al. (2023) came to similar results, noting that fetal MRI and 3D/4D ultrasound can help with precise counseling and surgical planning.

All things considered, the current study validates that ultrasound is a dependable, secure, and non-invasive diagnostic method for identifying NTDs. While first-trimester ultrasound is a new screening method for early diagnosis of serious abnormalities, the second-trimester scan is still the gold standard. The findings support the significance of early prenatal screening, maternal education, and folic acid supplementation in lowering the prevalence of neural tube abnormalities and are consistent with recent regional and worldwide research (2022–2024).

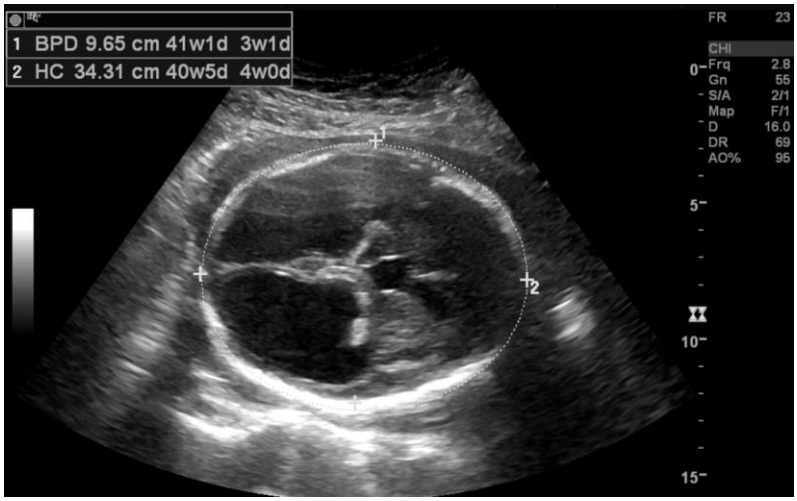
Case 1

A 20-year-old pregnant female underwent a 17-week obstetrical ultrasound showing a single live intrauterine fetus. Biometry corresponded to 17+4 weeks, but morphology revealed absent cranial vault, floating brain tissue, and prominent orbits, confirming anencephaly. Amniotic fluid was increased with an AFI of 13, indicating polyhydramnios. Placenta was fundal and right lateral with no retroplacental bleed. This case supports the importance of second-trimester ultrasound in early detection of major neural tube defects.



Case 2

27-year-old female, of 17 weeks A single, alive intrauterine fetus was seen in longitudinal lie with breech presentation.Fetal biometry showed BPD 81.8 mm, FL 56.9 mm, and AC 253.6 mm.The estimated fetal weight was 1499 g.The placenta was located along the posterior uterine wall with no retroplacental bleed.Amniotic fluid was adequate.Fetal movements and cardiac activity were present.The lateral ventricles were dilated, measuring 17 mm.No additional structural abnormalities were noted.Findings are most consistent with fetal ventriculomegaly. Follow-up ultrasound and detailed neurosonography are recommended.



CONCLUSION

According to the study, second-trimester ultrasounds (18–22 weeks) are the most accurate in identifying neural tube anomalies such anencephaly and hydrocephalus during pregnancy. For the early identification of notable abnormalities, first-trimester scans (11–13 weeks) are a useful tool. Increasing folate consumption, strengthening sonographer training, and boosting maternal education could all help reduce the incidence of neural tube abnormalities in Pakistan. Nonetheless, inadequate folic acid supplementation and delayed prenatal care remain problems in underdeveloped nations.

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