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CASE SERIES

Role of Three-Dimensional/Four-Dimensional Ultrasound in Assessment of Fetal Central Nervous System Congenital Anomalies

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Abstract

Background: Using contemporary ultrasound equipment, it is possible to diagnose central nervous system (CNS) defects in prenatal life. Many CNS defects can be detected by ultrasound in the first and early second trimesters. Four-dimensional (4D) ultrasonography has a high sensitivity for detecting prenatal CNS abnormalities and is used to detect and diagnose some fetal CNS disorders.

Objective: To validate the role of 4D ultrasonography in the prenatal assessment of CNS anatomical structure and early detection of CNS congenital abnormalities.

Patients and methods: This diagnostic prospective study was carried at Radiodiagnosis Department, Al-Azhar University. The study included 30 pregnant women who were suspected to have CNS congenital anomalies during routine prenatal obstetric examination, and then, they were referred to undergo 4D detailed ultrasonography after an initial detailed diagnostic 2D ultrasonography to confirm the diagnosis.

Results: A total of 30 cases of suspected congenital malformations were assessed by 2D ultrasonography throughout the research period, and the incidence of congenital CNS abnormalities was 83.3% (25 cases) by 4D ultrasonography. In most cases, congenital CNS abnormalities were isolated, but in 36.7% (11 cases), there was a connection with other congenital CNS malformations. The most common abnormality was anencephaly, which was followed by holoprosencephaly, hydrocephalus, occipital encephalocele, and Dandy-Walker syndrome. Regarding other malformations, a lower absolute frequency was observed.

Conclusion: 2D ultrasound remains the gold standard in fetal anomaly assessment, and 4D ultrasonography is thus not a screening procedure but rather an addition to 2D ultrasonography for fetuses with known or suspected abnormalities.

Keywords: Central nervous system, Congenital anomalies, Four-dimensional ultrasound

1. Introduction

Congenital defects cause 20–25% of perinatal fatalities. Anomalies of the central nervous system (CNS) are strongly linked to spontaneous abortions.¹

After congenital heart disease, CNS abnormalities are the most common type of congenital defect.² Approximately 21% of congenital malformations involve the CNS, making it one of the most frequent congenital defects. It can occur alone or in

conjunction with other deformities of the CNS or other systems.³

From 3 to 20 weeks of intrauterine life, the CNS develops. Almost all CNS abnormalities are caused by an injury during embryogenesis at some point throughout development. Using contemporary ultrasound equipment, it is possible to diagnose CNS defects in prenatal life. Many CNS defects can be detected by ultrasound in the first and early second trimesters. Some appear or develop in late pregnancy.⁴

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The sooner the detection, the more time the clinician and parents have to arrange the pregnancy's prognosis. Minor defects help everyone be prepared for postnatal management, whereas lethal and seriously life-limiting illnesses demand early termination of pregnancy.

Prenatal diagnostics employs a variety of noninvasive and invasive techniques to detect the fetus's health or any abnormalities in an unborn fetus. Fetal visualization techniques include the following.

Noninvasive methods include ultrasound, fetal echocardiography, and MRI.

Invasive techniques include embryoscopy, fetoscopy, and amniocentesis.

For prenatal diagnosis of these abnormalities, ultrasound examination is a useful method. It is a noninvasive procedure that patients find more acceptable. Several studies have shown that the ultrasound detects CNS anatomic defects with an accuracy of 92–99%.²

Following an initial diagnostic two-dimensional (2D) ultrasonography, the current study recommends doing a CNS focused 3D/4D ultrasonography.

There is now 5D ultrasound technology, which uses new software to improve facial characteristics and depth awareness, as well as a virtual lighting source. However, it is essentially the same as 4D ultrasound.

2. Aim

The purpose of this study was to confirm the role of 3D and 4D ultrasonography in prenatal assessment of CNS anatomical structure and early diagnosis of CNS congenital abnormalities.

3. Patients and methods

This diagnostic prospective study was carried at Radiodiagnosis Department, Al-Azhar University. The study was ethically approved by the Radiodiagnosis Department. The study included 30 pregnant women aged from 18 to 40 years old, who were suspected to have CNS congenital anomalies during routine prenatal obstetric examination. They were then referred to undergo 3D/4D detailed ultrasonography after an initial detailed 2D ultrasonography to confirm the diagnosis.

Patients were enrolled in the study according to the following criteria: age of 18–40 years, pregnant women, and suspected pregnant women to have CNS congenital anomalies during routine prenatal obstetric evaluation. However, no women with suspected fetal neurological anomalies were excluded from the study.

Following a thorough obstetric medical history, all pregnant women had detailed 2D diagnostic ultrasonography, followed by 3D/4D ultrasonography.

The primary goal of the study was to determine the role of 3D/4D ultrasound in the assessment of fetal CNS congenital abnormalities.

3.1. Statistical analysis

Data were gathered, edited, coded, and entered into the Statistical Package for the Social Sciences, version 20 (IBM SPSS, Cairo, Egypt). Numbers and percentages were used to represent qualitative data, whereas mean, SDs, and ranges were used to represent quantitative data. Following that, proper statistical analyses were performed. The confidence interval was set at 95%, whereas the acceptable margin of error was set at 5%.

4. Results

A total of 30 cases of suspected congenital malformations were assessed by 2D ultrasound throughout the research period, and the incidence of congenital CNS abnormalities was 83.3% (25 cases) by 3D/4D ultrasound. The mean gestational age for diagnosis of CNS anomalies was 23.33 ± 1.54 weeks. Overall, 23.3% (seven patients) of the patients had previous history of miscarriages, and 6.7% (two patients) of the patients had previous birth with CNS malformation. The 25 newborns with congenital CNS abnormalities had an average birth weight of 2694.6 ± 872.8 g, ranging from 1000 to 4760 g. The mothers' average age was 29.53 ± 5.81 years, with a range of 18–40 years, whereas the fathers' average age was 33.3 ± 5.36 years, with a range of 20–45 years. Most of ultrasonographic examinations (28 cases) (93.33%) were done at the second trimester. The detailed demographics data are presented in [Table 1](#).

In the majority of cases, congenital CNS abnormalities were isolated, but in 36.7% (11 cases), there was a connection with other congenital malformations. The most common abnormality was anencephaly, which was followed by holoprosencephaly, hydrocephalus, occipital encephalocele, and Dandy-Walker syndrome. Regarding other malformations, a lower absolute frequency was observed, as shown in [Table 2](#) and [Figs. 1–4](#).

5. Discussion

Many studies have been conducted to evaluate the role of 3D ultrasonography in the evaluation of prenatal CNS congenital anomalies. Previous

Table 1. Demographic data.

	Presence of CNS congenital anomalies (25)	No CNS congenital anomalies (5)	P value
Mothers' age	30.12 ± 5.71	26.6 ± 5.98	0.22
Fathers' age	33.84 ± 5.15	30.6 ± 6.23	0.22
Gestational age at time of diagnosis	23.44 ± 1.53	22.8 ± 1.64	0.41
History of miscarriage	1	1	0.31
History of previous CNS congenital anomalies	6	1	0.67
Age groups [n (%)]			
Older than 30 years (15)	14 (93.33)	1 (6.66)	0.33
Younger than 30 years (15)	11 (73.33)	4 (26.67)	
Trimester			
2nd	23	5	1.0
3rd	2	0	

CNS, central nervous system.

studies have shown that 3D ultrasonography is effective in detecting fetal CNS abnormalities; however, in our current study, CNS defects were detected on ultrasound at a rate of 90%, which is higher than in previous studies (Table 3).

Congenital CNS malformations were found in 31.8% of all malformations studied, a figure comparable to that reported by certain authors but higher than that found by Noronha et al.⁵ and Pitkin⁶, who found 13 and 21% of cases, respectively. In contrast to the findings of Pitkin⁶ and Victora et al.,⁷ congenital CNS abnormalities, orthopedic deformities, and craniofacial anomalies were the most common. Another study found that the most prevalent abnormalities are craniofacial and limb malformations.⁸

Despite the fact that our study only included 30 pregnant women, the rising prevalence of CNS congenital anomalies may be explained by increased awareness among treating doctors as well as the improvement and evolution of 3D ultrasound technology in recent years.

A case–control research found that neurological factors may play a role in the etiology of some

congenital orthopedic abnormalities by producing changes in the spinal cord or nerves.⁹ Although cardiovascular malformations are reported to be the most common malformations in certain studies, there are substantial differences in research populations, as well as in the criteria and diagnostic procedures used, which may contribute to underdiagnosis of minor disorders. A higher prevalence of such congenital anomalies has been discovered as a result of regular echocardiography, which is a rare procedure in most health facilities.¹⁰

Early detection of anomalies, especially in the first and early second trimesters, helps with intervention and management planning. In this study, the average gestational age for ultrasonography diagnosis of CNS anomalies was 23.44 ± 1.53 weeks. It was most likely related to the patients in our study who lacked adequate health knowledge of early pregnancy monitoring. Following a thorough 2D assessment, all women who were found to have fetal CNS anomalies were given a 3D detailed ultrasound.

Despite extensive epidemiological research, the majority of cases of CNS disorders remain unknown. Women who have previously had a child with a neural tube abnormality have a higher risk of recurrence, according to our findings. A reliable prognosis is possible because CNS abnormalities are detected early. Ultrasound screening has been demonstrated to be helpful in detecting NTDs during pregnancy. Ultrasound technology innovation is a significant component of a higher detection rate for CNS disorders. The development of tools for prenatal identification of congenital defects has caused substantial ethical and practical concerns as a result of elective terminations.¹¹

A retrospective study found that hydrocephalus was the most prevalent congenital CNS anomaly, followed by myelomeningocele. In contrast to Moore et al.,¹² anencephaly, corpus callosum

Table 2. Frequency of central nervous system congenital anomalies.

	Frequency	Percent
No anomalies	5	16.7
Holoprosencephaly	5	16.7
Hydrocephalus	3	10.0
Occipital encephalocele	2	6.7
Corpus callosum agenesis	1	3.3
Anencephaly	7	23.3
Dandy-Walker	2	6.7
Arnold-Chiari	1	3.3
Spina bifida	1	3.3
Congenital scoliosis	1	3.3
Microcephaly	1	3.3
Schizencephaly	1	3.3
Total	30	100.0

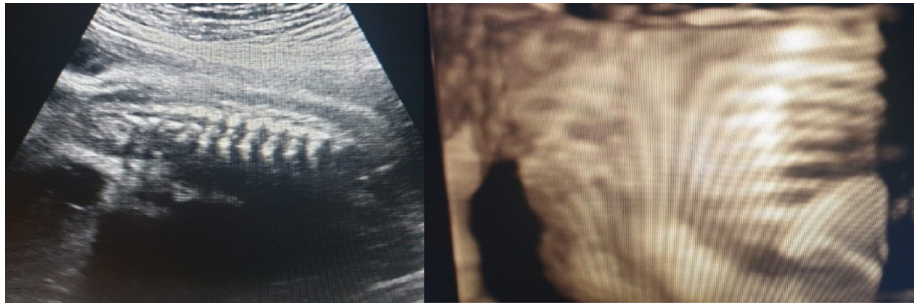


Fig. 1. A 30-year-old woman nondiabetic nonhypertensive gravida 1 with no previous abortions, no history of drug intake or radiation exposure during the first trimester, and no history of previous CNS fetal anomalies. Because her doctor suspected a vertebral column abnormalities, the mother was referred for a comprehensive anomaly scan. A 2D scan suspected hemivertebra, and a comprehensive 4D anomaly scan revealed a normal vertebral column. 2D, two dimensional; 4D, four dimensional; CNS, central nervous system.

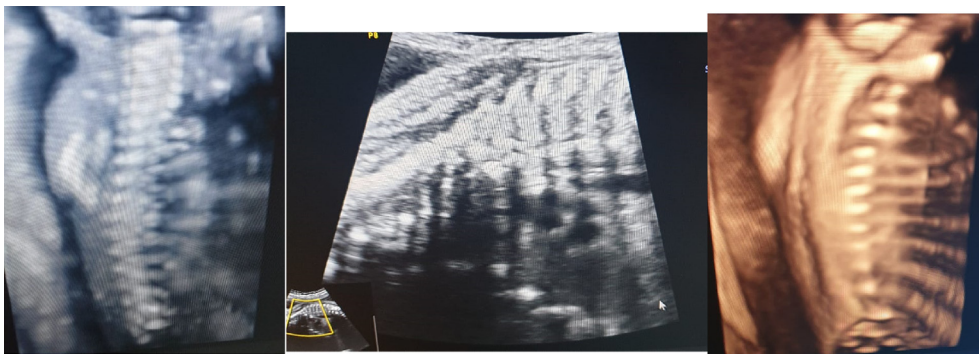


Fig. 2. A 31-year-old women nondiabetic nonhypertensive gravida 2 with no previous abortions, no history of drug intake or radiation exposure during the first trimester, and no history of previous CNS fetal anomalies. We performed a 4D scan after the mother had a standard 2D scan and suspected scoliosis with hemivertebra. Scoliosis and hemivertebra were suspected in a 2D scan, and a detailed 4D anomaly scan revealed a normal vertebral column. 2D, two dimensional; 4D, four dimensional; CNS, central nervous system.

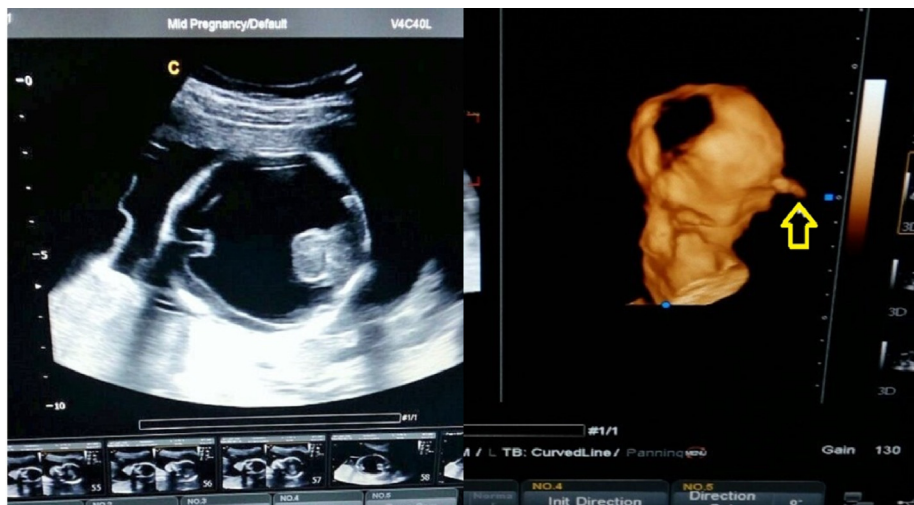


Fig. 3. A 28-year-old woman nondiabetic nonhypertensive gravida 3 with no history previous abortions, no history of drug intake or radiation exposure during the first trimester, and no history of previous CNS fetal anomalies. The woman was referred for a comprehensive anomaly scan after her doctor discovered a family history of CNS congenital abnormalities. Detailed 3D anomaly scan was done showing 23-week 4-day old fetus with alobar holoprosencephaly (monoventricle, absent corpus callosum, absent interhemispheric fissure, absent cavum septi pellucidum, and absence of third ventricle); note also the facial abnormalities (flat midface with absence of normal nose and prominent proboscis which is appendage-like structure is seen projecting from the midline fetal face (yellow arrow)). 3D, three dimensional; CNS, central nervous system.

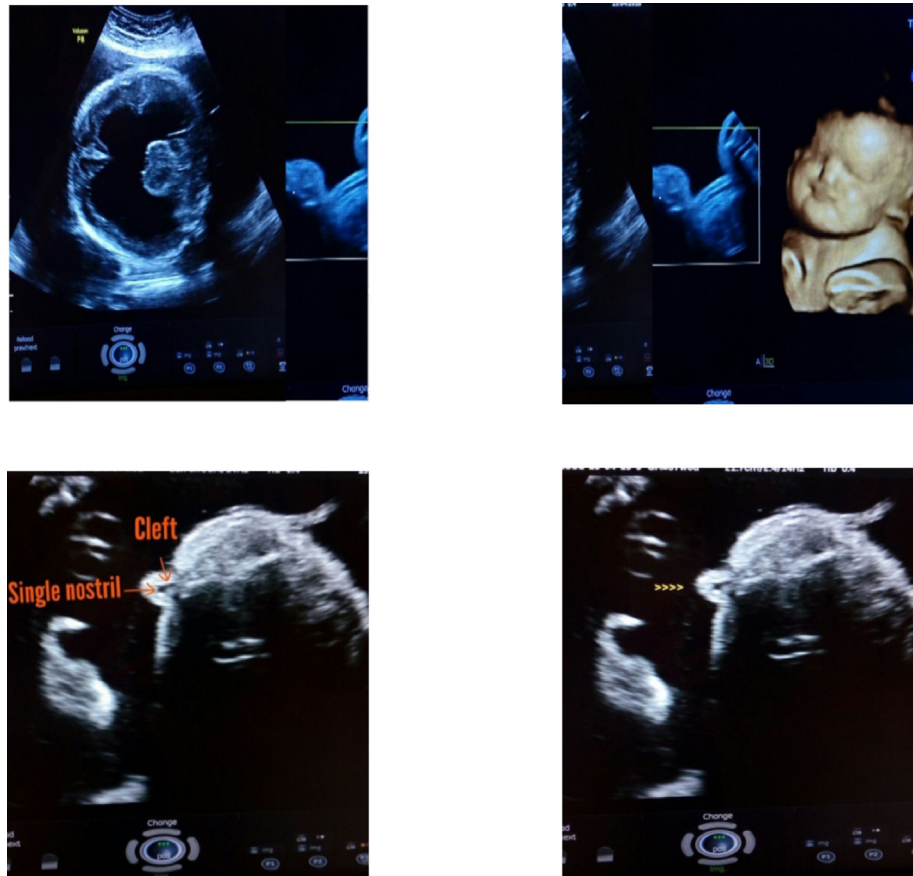


Fig. 4. A 35-year-old woman nondiabetic nonhypertensive gravida 6 with history of two previous abortions, no history of drug intake or radiation exposure during the first trimester, and no history of previous CNS fetal anomaly. As a result of her doctor's report of a previous CNS fetal abnormalities and positive relative relation, the woman was referred for a comprehensive anomaly scan. A detailed 3D abnormality scan was performed to demonstrate this 37-week-old fetus with alobar holoprosencephaly, with fascial anomalies (hypotelorism, single nostril with cleft lip, and low set ears, as shown in the 3D image). 3D, three dimensional; CNS, central nervous system.

agenesis, and encephalocele were shown to be more prevalent in succession. Among neural tube abnormalities, myelomeningocele, anencephaly, and encephalocele were discovered in that order. A similar pattern has been documented by several writers. According to Dávila-Gutiérrez,¹³ hydrocephalus and corpus callosum agenesis are connected. Other authors, on the contrary, described occurrences of coexistence of hydrocephalus with myelomeningocele.¹⁴

In our analysis, the majority of cases had isolated congenital CNS abnormalities, whereas 36.7% (11 cases) had a relationship with other congenital CNS

anomalies. Anencephaly was the most prevalent anomaly, followed by holoprosencephaly, hydrocephalus, occipital encephalocele, and Dandy-Walker syndrome. The absolute frequency of the other abnormalities was found to be lower.

It is well understood that factors such as apparatus quality, sound wave interaction with tissues, examination techniques such as appropriate gain waveform adjustment, dedicated scan time, the experience and knowledge of the medical sonographer, as well as factors such as high maternal BMI, fetal statics, advanced gestational age, and decreased amniotic fluid index, can all affect the results. Constant refining and in-depth knowledge of specialized procedures like as Doppler¹⁵ and volumetric (3D/4D) ultrasonography, together with developments in technology and cutting-edge equipment, have contributed to the method's high sensitivity in detecting abnormalities, with prognostic consequences. False-negative results accounted for 20.5% of all findings. One of the

Table 3. Detection of central nervous system anomalies by three-dimensional detailed anomaly scan.

Study	Year	Percentage
Carroll et al. ¹⁸	2000	77
Yeo et al. ¹⁹	2002	80
Szigeti et al. ²⁰	2007	65
Present study	2020	90

observable limits is oligohydramnios, which is present in 25% of false-negative sonographic tests. Unlike ultrasonography, MRI improves diagnostic accuracy with gestational age and is unaffected by decreased amniotic fluid levels, maternal obesity, or fetal statics.¹⁶

Additionally, one of the most valuable contributions of MRI is to support ultrasonography's function in the investigation of fetal CNS disorders, which is limited by advanced cranial bone ossification during late pregnancy. Nonetheless, owing to constraints such as the technology's high cost, fetal motion artifacts, and claustrophobia, suggestions not to do the method in the first gestational trimester, and its restricted availability, MRI should only be used to support ultrasonography. As a result, ultrasonography allows for early identification with excellent sensitivity, which, combined with the method's ease of use and availability, has served to cement its place as the modality of choice in routine screening for fetal CNS disorders.¹⁷

Ultrasonography is being used as a routine procedure for identifying prenatal anomalies, with a detailed 3D model. Pregnant women use it because of its efficiency, availability, low cost, and real-time capabilities. Prenatal CNS abnormalities were discovered in 90% of the people in our study. Prenatal diagnosis of brain disorders has improved with the introduction of 3D ultrasound imaging technologies. The findings have substantial implications for parental counseling in terms of the type of abnormalities as well as the prognosis for neurological and developmental difficulties.¹⁴

Hydrocephalus can be adequately treated with a ventriculoperitoneal shunt or other shunting operations. Even in such cases, neurodevelopmental issues may persist after successful surgery due to broad brain abnormalities in addition to hydrocephalus.⁸

Despite initial efforts, few trials on CNS congenital malformations have been conducted in our nation. Other minor connected anomalies were found in two of our research cases (Alobar holoprosencephaly associated with facial abnormalities). Minor anomalies must be identified because they may be associated with a syndrome. False-positive diagnoses are extremely rare with today's ultrasound technology.

6. Conclusion

2D ultrasound remains the cornerstone in fetal anomaly assessment, and 4D ultrasonography is therefore not a screening procedure but rather an addition to 2D ultrasonography for fetuses with known or suspected abnormalities.

Conflict of interest

None declared.

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