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ORIGINAL ARTICLE

Prevalence of neurological fetal anomalies diagnosed by ultrasonography in Sohag Governorate

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ABSTRACT

Keywords:Ultrasonography, neurological, anomalies .

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Background: Congenital anomalies affecting the central nervous system are among the most prevalent .Fetal neurological malformation are second only to cardiac malformations in their frequency of occurrence.The most common central nervous system anomalies are neural tube defects, which affect roughly 1-2 of every 1000 newborns. **The aim of this study:** The study detected the accuracy of ultrasound to diagnose CNS abnormalities **. Methods:** Descriptive cross-sectional study on 567 pregnant women patients coming for antenatal care at Sohag Governorate undergoing mid anatomical scan between September 2021 and December 2022. After getting approval from the ethical committee at Sohag University and written consent from each woman. **Results:** Mid anatomical scans were performed on 567 pregnant women who presented for antenatal care in the Sohag Governorate. It was noticed that (90% n=510) females in the current study had not neurological congenital anomalies in her baby and (10% n=57) had neurological congenital anomalies in her baby. **Conclusions:** Based on these findings, we can say that ultrasonography is useful for detecting and diagnosing some defects of the fetal central nervous system.

INTRODUCTION

The central nervous system (CNS) is composed of the brain and the spinal cord. They both develop from the embryonic ectoderm alongside other structures like the skin. Their development begins as early as the 3rd and 4th weeks of embryonic life, starting with the process of neurulation, which is the development of the neural tube. The neural tube closes spontaneously rostrally and caudally. In the fifth to sixth week, the first appearance of the brain, the prosencephalic development ensues. The primitive brain is comprised of prosencephalon, mesencephalon, and rhombencephalon. The prosencephalon divides further into telencephalon and diencephalon through a series of developmental stages, namely: formation, cleavage, and development of the midline [(1,3)].

Any form of developmental alteration in these leads to the malformation of the developing brain [4].

Fetal central nervous system (CNS) abnormalities are second only to cardiac malformations in their frequency of occurrence. Early and accurate diagnosis at prenatal US is therefore essential, allowing improved prenatal counseling and facilitating appropriate referral [5].

Central nervous system (CNS) malformations are some of the most common of all congenital abnormalities. Neural tube defects are the most frequent CNS malformations to about 1–2 cases per 1000 births [6].

Prenatal detection and accurate definition of CNS malformations are important because these anomalies frequently have a severe prognosis and are often associated with genetic syndromes [7].

The development of the brain and spinal cord is an extremely complicated process which continues into the second decade before final maturity is achieved. Abnormality in the development of CNS is common, up to 75% of fetal deaths and 40% of deaths in infancy are due to CNS malformations [7].

Four standard recommended views-transventricular, falx, cavum, and posterior fossa or transcerebellar views-provide an overview of fetal intracranial anatomy during the second trimester anatomy scan [5].

CNS abnormalities can be organized into six main categories at prenatal US. Developmental anomalies include neural tube defects and neuronal migration disorders. Posterior fossa disorders include Dandy-Walker malformation variants and Chiari II malformation. Ventricular anomalies include aqueductal stenosis. Midline disorders include those on the spectrum of holoprosencephaly, agenesis of the corpus callosum, and septo-optic dysplasia. Vascular anomalies include veins of Galen malformations. Miscellaneous disorders include hydranencephaly, porencephaly, tumors, and intracranial hemorrhage [5].

Aim of the Work

The work is to detect the Prevalence of neurological fetal anomalies diagnosed by ultrasonography in Sohag Governorate.

Patients and Methods

This descriptive cross-sectional study was carried out on 567 patients coming for antenatal care at Sohag Governorate undergoing mid anatomical scan from September 2021 to December 2022 after approval from the ethical committee at Sohag University and obtaining a written consent from each woman.

Criteria of patient selection:

Inclusion criteria: Pregnant women have mid-anatomical scan at 20-22 weeks.

Methods

All patients were subjected to the followings:

- Complete history was taken from patients age body mass index (BMI), consanguinity, maternal history, and family history.
- General examination.
- Radiological investigations:
- Two-dimensional Ultrasound (US) were performed to detect the presence of antenatal anomalies.

Two-dimensional ultrasound technique:

Antenatal examination of the anomalies was done for all pregnant women using two-dimensional US (The Voluson E8 color Doppler US diagnostic instrument) during the second trimester. First, the abdominal convex array probe C1-5-D was adopted, and the probe frequency was set to 2.0 MHz. The lower abdomen was scanned for the presence of any anomalies [8].

Four standard recommended views-trans ventricular, falx, cavum, and posterior fossa or trans cerebellar views-provide an overview of fetal intracranial anatomy during the second trimester anatomy scan.

Results

This study was a descriptive cross-sectional study conducted on 567 pregnant women coming for antenatal care at Sohag Governorate undergo mid anatomical scan.

Table1: Demographic data of the studied mothers

		N=567
Age (years)	**Mean ± SD**	29.7 ± 5.97
	Range	16 – 48
BMI (kg/m^2)	**Mean ± SD**	28.9 ± 4.06
	Range	19 – 41
Consanguinity	**Yes**	44 (7.76%)
	No	523 (92.24%)

The age ranged from 16 to 48 years with a mean value (± SD) of 29.72 (±5.97) years. BMI ranged from 19 to 41 kg/m2 with a mean value (± SD) of 28.92 (±4.06) kg/m2. Consanguinity was present in44 (7.76%) patients and not in 523 (92.24%) patients. **(Table)**

Table2: Maternal medical history of the studied patients with neurological anomalies

		N=57
Maternal medical history	**Negative**	50 (87.72%)
	Positive	7(12.28%)
	Positive TOURCH infection	5 (8.77%)
	Positive teratogenic drugs	1 (1.75%)
	Brain atrophy	1 (1.75%)

Maternal medical history was negative in 50 (87.72%) patients and positive in 7(12.28%) patients. Positive maternal medical history was Positive TOURCH infection in 5 (8.77%) patients, teratogenic drugs in 1 (1.75%) patient and brain atrophy in1 (1.75%) patient. **(Table 2)**

Table 3: Family history of the studied patients .

		N=57
Family history	**Negative**	37 (64.91%)
	Positive	20(35.09%)
	Positive congenital in other siblings	9 (15.79%)
	Microcephaly	1 (1.75%)
	Brain atrophy	6 (10.53%)
	Epilepsy	1 (1.75%)
	Hydrocephalus	1 (1.75%)
	Deafness, CHD	1 (1.75%)
	Hydrocephalus	1 (1.75%)

CHD: Congenital heart disease.

Prevalence of neurological congenital anomalies in Sohag Governorate

Mid anatomical scans were performed on 567 pregnant women who presented for antenatal scan in the Sohag Governorate.

It was noticed that (90% n=510) females in the current study had not neurological congenital anomalies in her baby and (10% n=57) had neurological congenital anomalies in her baby findings as shown in **figure1:** so the prevalence of neurological congenital anomalies detected by ultrasound in Sohag Governorate was 10%.

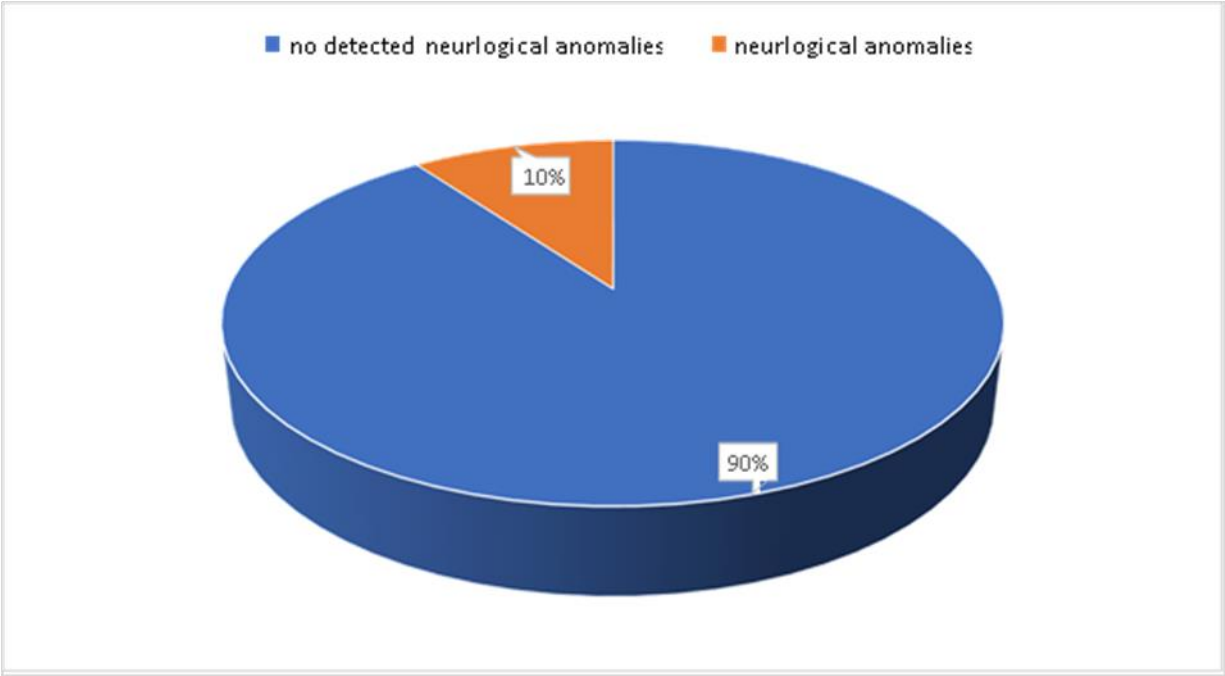

Figure 1 :Prevalence of neurological congenital anomalies.

Table 4: Antenatal diagnosis of the studied patients

		N=57
Antenatal diagnosis	**Microcephaly**	8 (14.04%)
	Microcephaly, hepatosplenomegaly	2 (3.51%)
	Dandy-walker syndrome	7 (12.28%)
	Joubert syndrome	2 (3.51%)
	Hydrocephalus	16 (28.07%)
	Hydrocephalous, CHD	1 (1.75%)
	Biventricular dilation hydrocephalus	1 (1.75%)
	Holoprosencephaly, hydrocephalus	1 (1.75%)
	Hydrocephalus aqueductal stenosis	1 (1.75%)
	Holoprosencephaly, cleft lip and palate	1 (1.75%)
	Intracranial calcification IUGR	1 (1.75%)
	Occipital encephalocele meningomyelocele	1 (1.75%)
	Left cerebral hemisphere atrophy	1 (1.75%)
	Left ventriculomegaly agenesis of corpus callosum	1 (1.75%)
	Retrocerebellar dermoid cyst	1 (1.75%)
	Corpus callosum agenesis	4 (7.02%)
	Spina bifida	1 (1.75%)
	Anencephaly	3 (5.26%)
	Vein of Galen aneurysm	2 (3.51%)
	Middle cerebral artery aneurysm	1 (1.75%)
	Intracranial hemorrhage	1 (1.75%)

CHD: Congenital heart disease.

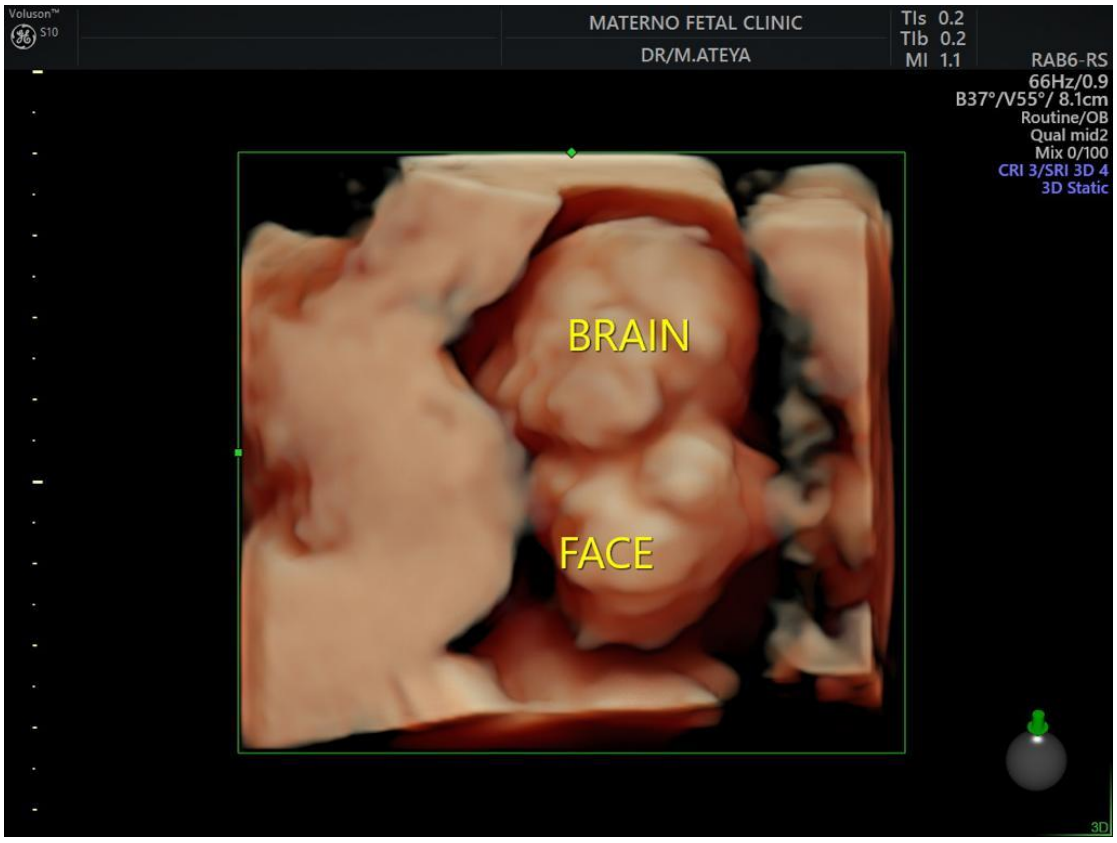

figure 2 : Three-dimensional ultrasound showing exencephaly of the studied patients.

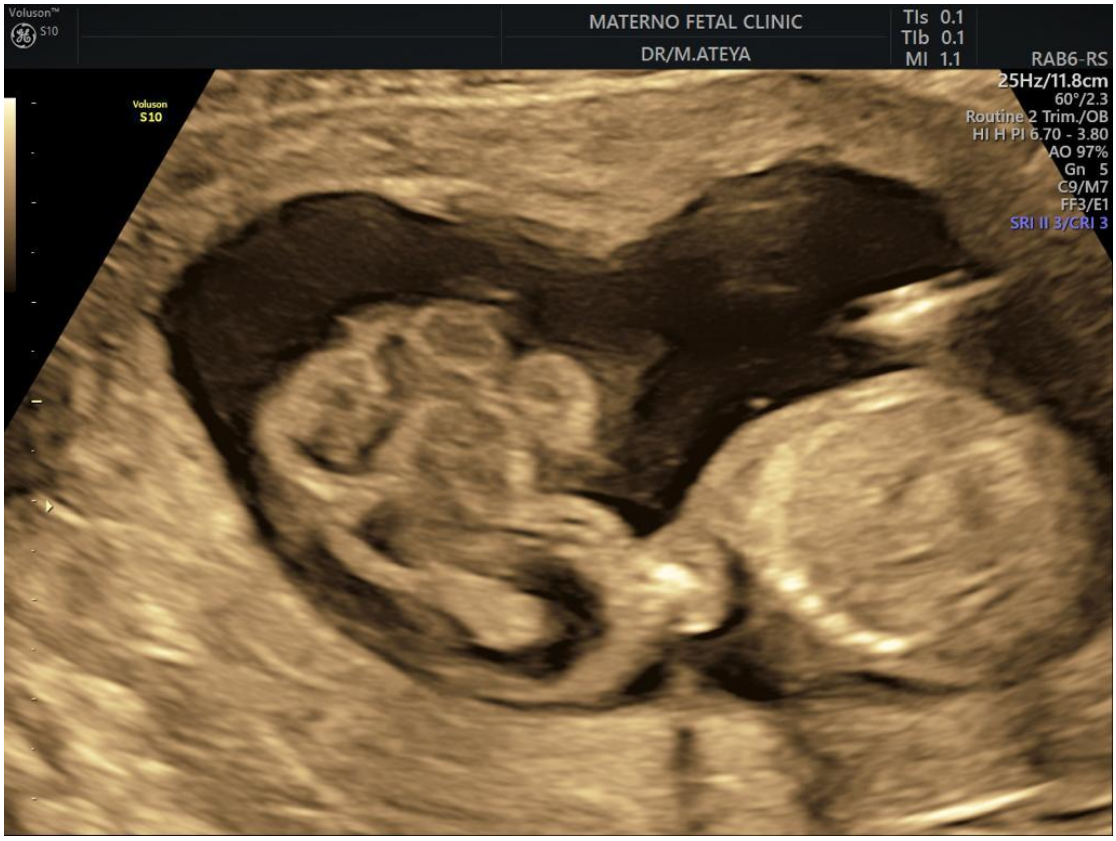

Figure 3 :Two-dimensional ultrasound showing exencephaly of the studied patients.

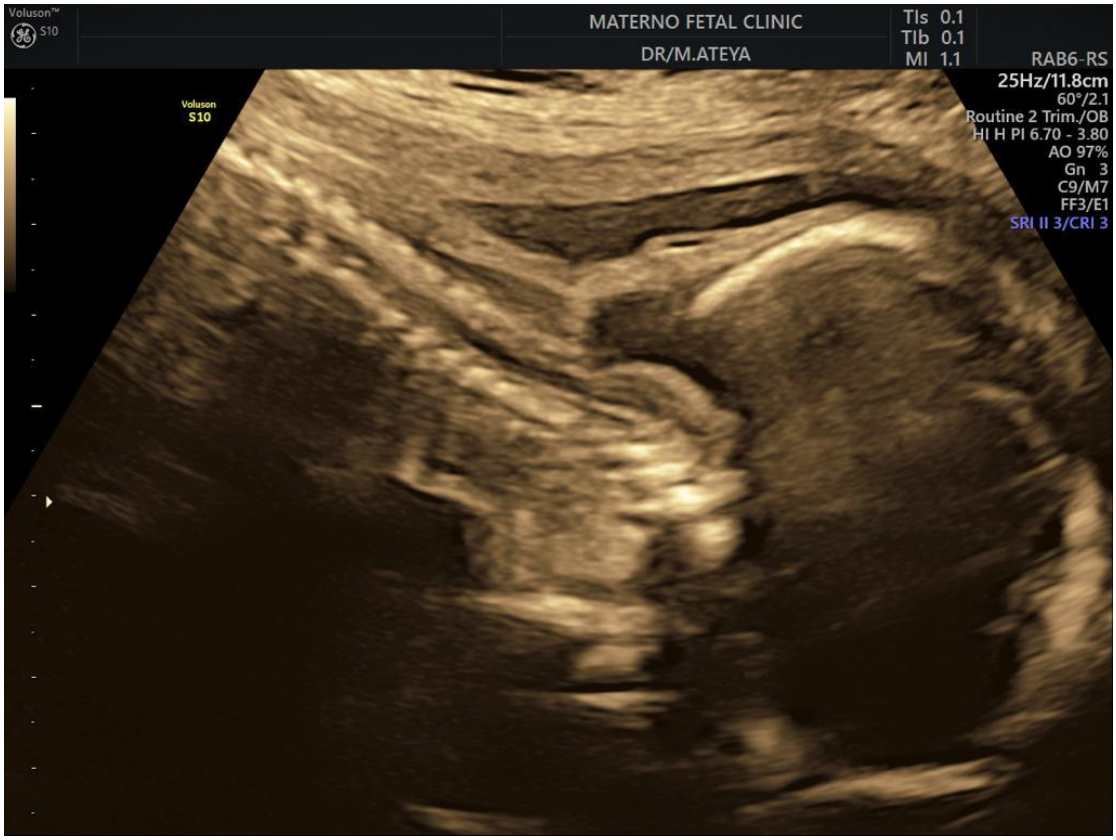

Figure 4: Two-dimensional ultrasound showing a case of cervical spina bifida.

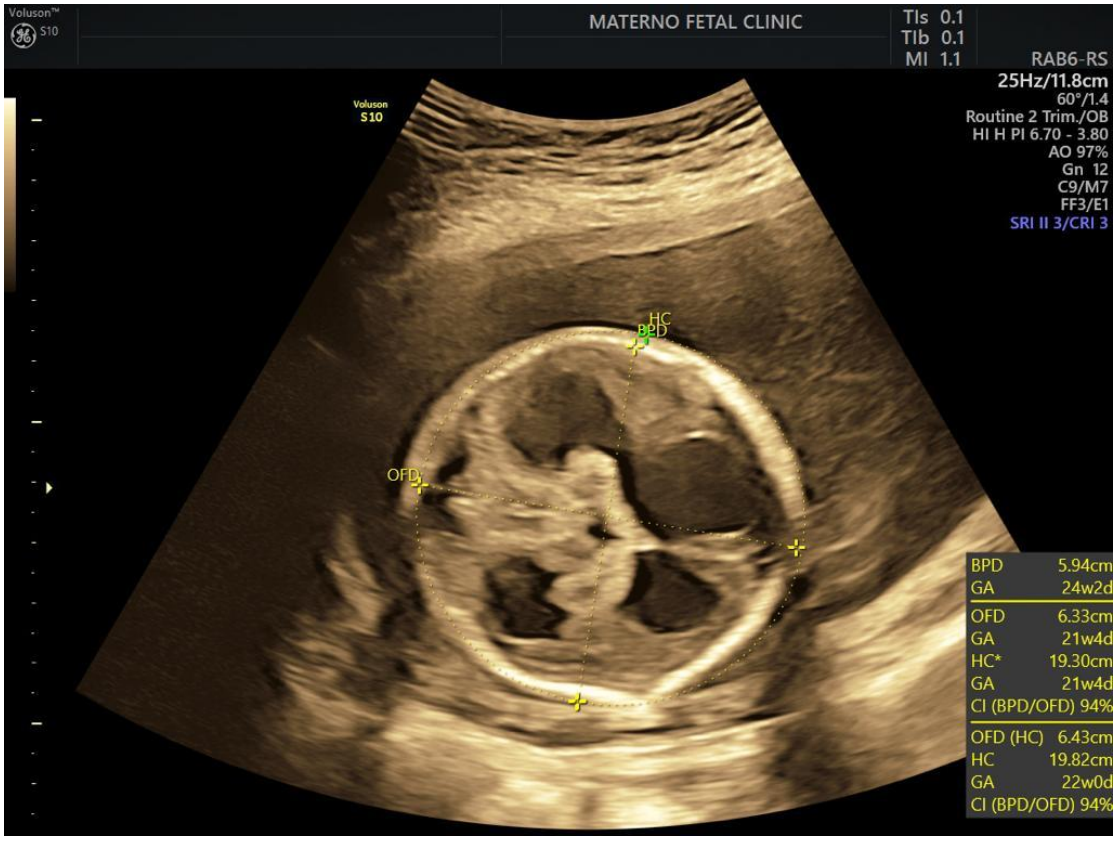

Figure 5 : Two-dimensional ultrasound showing ventriculomegaly of thestudied patients

Discussion

Antenatal diagnosis showed microcephaly in 8 (14.04%) patients, Microcephaly, hepatosplenomegaly in2 (3.51%) patients, Dandy-walker syndrome in 7 (12.28%) patients, Joubert syndrome in 2 (3.51%) patients, Hydrocephalus in 16 (28.07%) patients, Hydrocephalous, CHD, Biventricular dilation hydrocephalus, Holoprosencephaly, hydrocephalus, Hydrocephalus aqueductal stenosis, Holoprosencephaly, cleft lip and palate, Intracranial calcification IUGR, Occipital encephalocele meningomyelocele, Left cerebral hemisphere atrophy, Left ventriculomegaly agenesis of corpus callosum, Retrocerebellar dermoid cyst, Spina bifida, middle cerebral artery aneurysm and intracranial hemorrhage in 1 (1.75%) patient, corpus callosum agenesis in 4 (7.02%) patients, anencephaly in 3 (5.26%) patients and vein of Galen aneurysm in 2 (3.51%) patients. (Table 4)

The evaluation and diagnosis of CNS malformation during the prenatal period can be performed by the US at any gestational age. The ultrasonography evaluation includes the study of the brain and spinal cord. It is important to determine whether the CNS structures present complex embryology and anatomy because the CNS undergoes most of its changes during gestation. CNS changes are associated with changes in the US aspects of the CNS during gestation [(9)].

Therefore, every professional involved in the fetal evaluation should be aware of the embryology and anatomy of CNS as well as of its ultrasound characteristics of different gestational ages to avoid diagnostic errors [(9)].

Ultrasonography evaluation of the fetal CNS in the first trimester is usually performed in the axial, sagittal, and coronal planes using abdominal and vaginal approaches. Most efforts to diagnose CNS malformations occur during the second trimester in the examination of fetal

morphology conducted at 20 to 24 weeks of gestations **(ISUOG, 2007)**. Brain development features are susceptible to changes throughout gestation mainly secondary to the effect of external agents such as infection, trauma, and hemorrhage. So, it is important to emphasize that abnormal CNS assessment in the second-trimester morphology scan does not exclude the emergence of fetal alterations during pregnancy, hence it's necessary to re-evaluate the fetal brain morphology throughout pregnancy **(10)**. Conventionally, the ultrasound evaluation of brain development during pregnancy is performed in the axial planes of the fetal skull; however, that type of evaluation has some limitations **(9)**.

For instance, the attenuation of the sound beam by the skull can impair the evaluation of the cerebral hemisphere proximal to the transducer, and because the brain is a three-dimensional organ with a complex anatomy, the midline structures such as the corpus callosum, the brain stem, the cerebellar vermis and the cerebral cortex are not probably evaluated if the scan of the fetal skull is performed only in the axial planes **(10)**.

Timor Tritsch et al., (1996) (11) described a fetal neurosonography technique that involves multiplaner analysis of the fetal brain structures incorporating sagittal and coronal views of the fetal skull. The international society of ultrasound in obstetrics and gynecology (ISUOG) has issued guidelines for the ultrasonographic study of the brain and spine in fetuses; they are divided into two categories: basic CNS assessment and neurosonographic evaluation**(12)**.

The aim of our work was to detect the prevalence of neurological fetal anomalies diagnosed by ultrasonography in Sohag Governorate. This was a descriptive cross-sectional study conducted on 567 pregnant women coming for antenatal care at Sohag Governorate undergo mid anatomical scan.

In our study, the age ranged from 16 to 48 years with a mean value (± SD) of 29.72 (±5.97) years. BMI ranged from 19 to 41 kg/m^2 with a mean value (± SD) of 28.92 (±4.06) kg/m^2.

In our study, maternal medical history was negative in 50 (87.72%) patients and positive 7(12.28%) patients. Positive maternal medical history was Positive TOURCH infection in 5 (8.77%) patients, teratogenic drugs in 1 (1.75%) patient and brain atrophy in1 (1.75%) patient.

According to **Munim et al. (2006)** (13) who aimed to describe the trends of congenital abnormalities seen at a tertiary care facility in Karachi. They reported that the mean age of the women in this study was 27.3 years with SD ± 5.3. Among the study subjects 11.6% were women above the age of 35 years. Only 8.8% of them had a previous history of congenital malformations.

According to **Hassan et al. (2022**) (9) who aimed to detect the proportion of C.N.S malformations in patients attending the fetomaternal unit at Minia University. The study reported that 25% of cases had a previous history of congenital fetal malformations, and 6% of cases had a previous history of C.N.S fetal malformations.

Hassan et al. (2022) (9) reported that, as regarding ultrasound detected CNS anomalies in 79 fetuses. The most common anomaly detected wasVentriculomegaly that found in 22%. Ventriculomegaly should be interpreted with care as brain formation is still ongoing and mild ventriculomegaly may still be a variation of normal. Nevertheless, first-trimester enlargement of the lateral ventricle has been described in fetuses with aneuploidy; this explains the high percentage of Ventriculomegaly in our sample as 42% of cases had a history of aneuploidy. Ultrasound examination is operator dependent with entails trial and error needs careful and skillful examination. Regarding out of 79 fetuses with CNS anomalies, 60% cases were treated by conservative management while 19% cases were treated by therapeutic termination of pregnancy, after delivery, 64% cases confirmed the diagnosis of anomalies after delivery.

Fatma et al.,(2019) (14) reported that 3D ultra-sonography is effective in the assessment of fetal CNS anomalies with detection of CNS anomalies on ultrasound was 90%. They concluded that CNS malformations have been detected prenatally in 90 % of patients.

Conclusion

Based on these findings, we can say that ultrasonography is useful for detecting and diagnosing some defects of the fetal central nervous system. The prevalence of neurological congenital anomalies detected by ultrasound in Sohag Governorate was 10%. However, because of the positioning of the fetus, it may be difficult to identify certain defects using two-dimensional ultrasonography.

Recommendations

- Our results need to be confirmed by more multicenter clinical trials.
- Ultrasound examinations at different stages of pregnancy are highly recommended for all pregnant women.

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