

# Ultrasound evaluation of fetal central nervous system anomalies and its correlation with postnatal outcome

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## Abstract

**Introduction:** CNS anomalies are the most serious congenital abnormalities. Ultrasound is an effective and non invasive modality of prenatal diagnosis of these anomalies. The purpose of the current study was to determine the frequency of CNS anomalies and its postnatal outcome. **Objectives:** Present study was undertaken to find out the incidence of CNS anomalies in utero by ultrasound and to confirm them by autopsy or postnatal examination. **Materials and Methods:** Ultrasound scanning of 5761 pregnant women was performed. Pregnancies with ultrasound findings of CNS anomalies were followed up. Prenatal findings were compared with postnatal findings and confirmed by autopsy wherever possible. In case of live birth post natal findings were noted. **Results:** Ultrasound detected 50 anomalies in 42 cases. Ultrasound findings were exactly matching in 90%. 4 cases had variations 3 on autopsy & 1 on postnatal USG. **Conclusion:** The incidence of CNS malformations on ultrasound was 0.73%. Autopsy and postnatal findings showed high degree of correlation with prenatal ultrasound findings.

**Keywords:** Antenatal ultrasound, CNS anomalies, Congenital malformations, Fetal autopsy

## Introduction

Central nervous system (CNS) malformations are one of the commonest congenital anomalies encountered in pregnancy. There is significant variation in incidence of congenital CNS anomalies in different regions of world including Europe [1]. Severe and lethal CNS malformations requires termination of pregnancy, however minor defects can be managed in the earliest possible postnatal period.

Targeted antenatal Ultrasound can detect most of the CNS malformations before 22 weeks of gestation. Some anomalies such as anencephaly can be diagnosed as early as 11- 13 weeks [2]. Early diagnosis of serious fetal anomalies makes the termination process easier, reduces maternal risks and lessens the psychological trauma of the parents.

Postmortem examination of aborted embryos and fetuses and examination of live born or stillborn infants has an important role in the quality control of the work performed by ultrasonographers. Several studies have compared prenatal ultrasound examination with autopsy findings in cases with congenital anomalies However, few have specifically addressed the discrepancies between prenatal and postmortem findings [3].

**Incidence and Epidemiology-** Neural tube defects are the major malformations of the CNS. The neural tube account for 0.5-2 per 1000 pregnancies worldwide, with variations in prevalence ranging from 0.2 to 10 per 1000 in specific geographical locations. [4] Incidence of NTDs in India varies from 0.6-13/1000 births & varies in different population. Highest rates of NTDs occur among certain ethnic groups, such as Welsh, Irish, Sikhs. Incidence in U.K., U.S.A. Denmark, & Oman reported as 1.5 / 1000 births [5].

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## Objective

- The purpose of our study was to find out the incidence of various Central Nervous System anomalies in utero by ultrasound and their imaging findings on antenatal ultrasound.
- Relation of gestation age, maternal age group and gravid status with incidence of CNS anomalies.
- To correlate and compare the prenatal ultrasound findings with postnatal outcome and to evaluate the diagnostic accuracy of ultrasound as part of general quality control.

## Materials and methods:

**Study Design:** This was a hospital based prospective study, observational and non-interventional study.

**Setting:** Ultrasound was performed with Philips HD 11 XE 3D and 4D machine with 2-5 MHz broadband convex probe.

### Inclusion Criteria

- Pregnant cases with gestational age > 10 weeks.
- Pregnant cases with fetal CNS anomalies with or without other associated anomalies.

### Exclusion Criteria

- Pregnant cases with gestational age < 10 weeks.
- Other anomalies without fetal CNS anomaly.

**Participants-** Pregnant women with gestational age more than 10 weeks were evaluated by ultrasound. Informed consent was taken. Fetuses with CNS anomalies with or without associated other anomalies were included. Their family and maternal history was noted. Antenatal ultrasound findings were confirmed after the delivery of fetus with either clinical examination/Radiographs/USG /autopsy.

### Variables:

- Number of foetuses
- Fetal cardiac activity
- Gestational age
- Placenta
- Liquor status
- Fetal CNS anomalies
- Other associated anomalies

## Results

Out of 5761 cases, Ultrasound detected 50 CNS anomalies with or without associated anomalies in 42 fetuses. The incidence of CNS anomalies was 0.73% [Table/Fig 1 and 2) Age groups of mothers having fetus with CNS

Follow up of the cases were done till the final outcome of the pregnancy. Findings were confirmed after birth by: Autopsy / Direct Clinical examination / Radiograph / Imaging.

**Data Source-** The prospective study was carried out at Govt. Rajindra medical college and hospital Patiala, Punjab from Jan 2015 to Dec 2015. Institutional review board approval for conducting this study was obtained and informed consent of study patients was taken.

**Bias-** Each patient was examined for previous obstetric history, history of drug intake, diabetes, epilepsy, smoking, alcohol. Previous history of congenital anomalies, history of consanguineous marriage, no folic acid intake, poor nutritional status were bias factors.

**Study Size-** Total 5761 pregnant women with gestational age more than 10 weeks were evaluated by USG. 50 CNS anomalies were detected in 42 cases which were included in the study and followed postnatally.

### Quantitative Variables

- Incidence of cases diagnosed with CNS anomalies
- Number of CNS anomalies detected
- Distribution of various CNS anomalies in 42 positive cases
- Maternal age group
- Gestational age
- Gravid status
- Obstetric history
- Liquor status

PPV, NPV, Sensitivity, Specificity and Diagnostic accuracy were calculated.

**Statistical Methods-** The data was collected as described above, tabulated and statistically analyzed. Data was tabulated using MS Excel and was analyzed using SPSS 16 software. Data were expressed as percentages and comparisons were performed by chi square test ( $\chi^2$ ) of Pearson. P value was calculated using Chi square test and a p value of <0.05 was used as the threshold for statistical significance.

malformations varied from 21 to 40 years in our study. Majority of women having fetal CNS anomalies were between 21-25 years followed by 26-30 years. Gestational age during which CNS malformations detected was in between 19-40 weeks with mean gestational age 22.76 weeks. 2 patients had history of consanguineous marriage. 5 patients had history of previous abortion and 1 case of recurrence of CNS anomalies were detected in previous study. 17 patients were primi gravida, 12 patients were second gravida, 11 patients were 3rd gravida and 2 patients were 4th gravida [Table]. 5 patients had history of congenital CNS malformation in previous child. 25 autopsies were performed and rest of the cases were confirmed by postnatal examination. Various CNS malformations detected in our study are listed below

**Table No.-1: Incidence of CNS anomalies.**

Total no. of cases scanned	Number of cases	Incidence
5761	42	0.73%

**Table No.-2: showing incidence of CNS anomalies.**

Anomalies (n=5761)	Number	Percentage
Anencephaly	14	0.24
Spinal dysraphism	10	0.17
Encephalocele	08	0.14
Exencephaly	07	0.12
Hydrocephalus	05	0.087
Ventriculomegaly	03	0.052
Acrania	01	0.017
Holoprosencephaly	01	0.017
Occipital meningocele with dandy walker malformation	01	0.01
<b>Total</b>	<b>50</b>	<b>0.86</b>

Congenital CNS malformations are very frequently encountered among the congenital malformations, in which neural tube defects are the most common. Anencephaly and spinal deformity are the commonest among the neural tube defects. Anencephaly is the CNS malformation which can be diagnosed at 11-14 weeks of gestational age. We diagnosed one case at 11 weeks 1 day and another at 13 weeks of gestational age.

**Table No.-3: Distribution of CNS anomalies according to gravid status.**

Obstetrics History	G1	G2	G3	G4	TOTAL
Number	17	12	11	2	42
Incidence	40.48	28.57	26.19	4.76	100

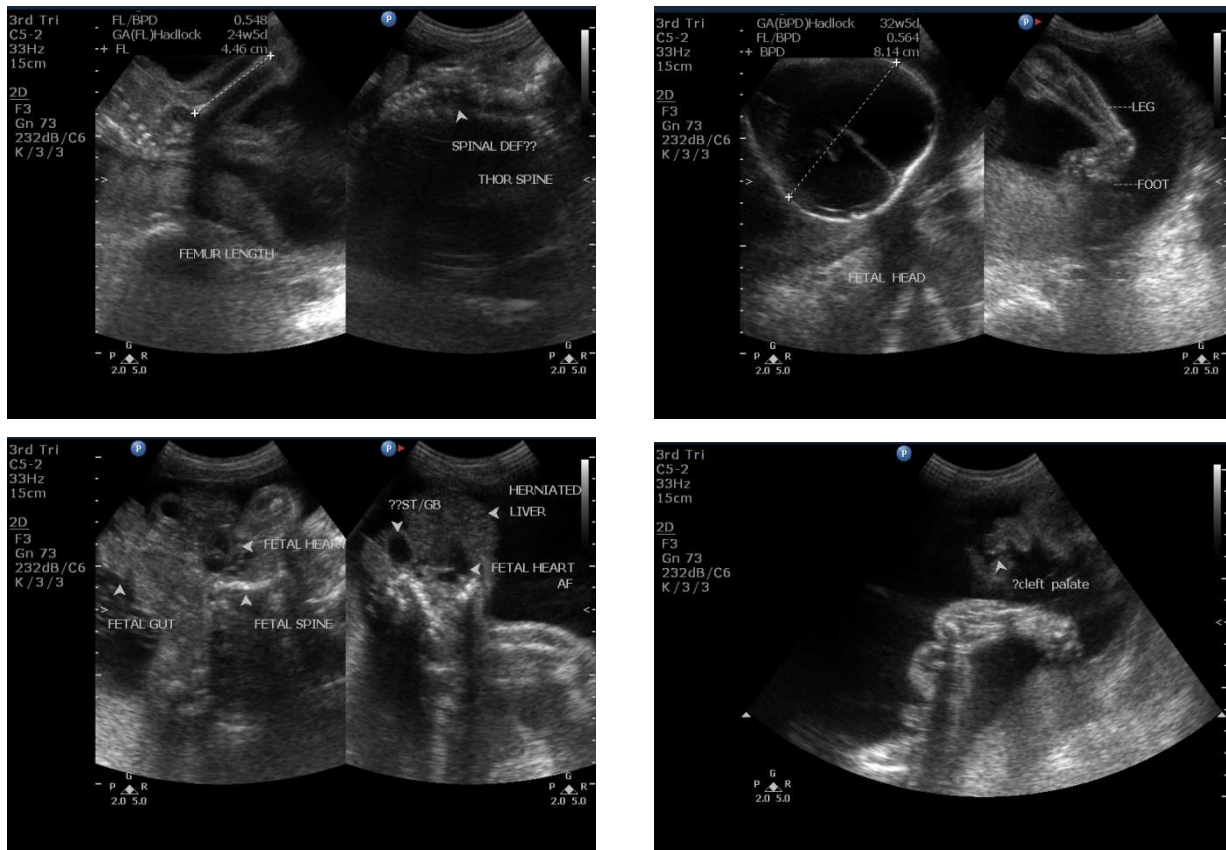
**Table No.-4: Patient screened: Patient diagnosed positive/negative for CNS anomalies.**

	Present at Birth	Absent at birth	Total
Diagnosed	41	1	42
Not Diagnosed	3	5716	5719
<b>Total</b>	<b>44</b>	<b>5717</b>	<b>5761</b>

Sensitivity of USG -93%                      PPV-97.6%  
 Specificity of USG-99.98%                  NPV-99.9%

**Table No.-5: Variation of ultrasound and postnatal findings in 4 cases.**

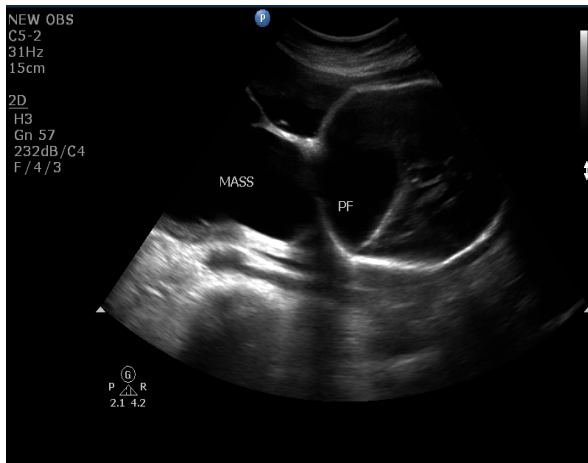
Prenatal USG findings	Postnatal confirmation by AUTOPSY/CLINICALEXAMINATION/USG/CT
Ventriculomegaly	Normal (confirmed by USG)
Anencephaly	Anencephalic fetus with Rachischisis ( confirmed by autopsy)
Exencephaly with anterior sacral MMC with thoracic spinal dysraphism	Exencephalic fetus with thoracic Spinal dysraphism (confirmed by autopsy)
Holoprosencephaly	Holoprosencephaly with facial anomalies like Proboscis (confirmed by autopsy)



**Fig 1: Antenatal USG showing hydrocephalus, ectopia cordis, omphalocele, with associated cleft palate, clubfoot and spinal deformity.**



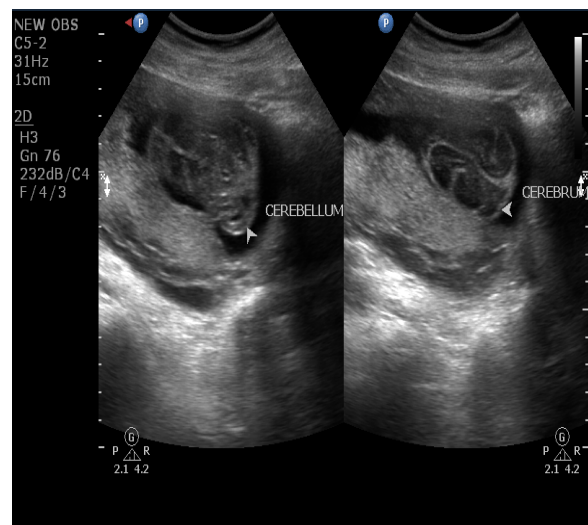
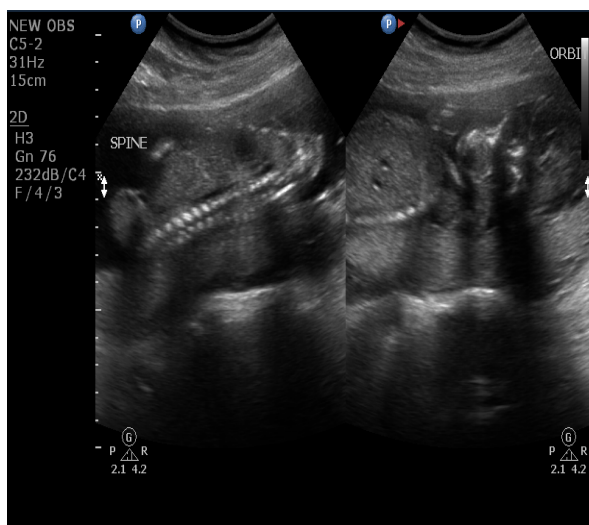
**Fig 2: Abortus shows the same findings**



**Fig 3: Antenatal USG shows posterior fossa cyst, occipital meningocele associated with Dandy walker malformation.**



**Fig 4: Alive baby CT scan demonstrates occipital meningocele associated with Dandy-walker malformation and a posterior fossa cyst.**



**Fig 5: Antenatal USG shows acrania-absence of calvarium, with cerebrum and cerebellum lying outside**



**Fig 6: Abortus shows acrania**

## Discussion

A total of 5761 pregnant women were scanned at different stages of gestation. CNS anomalies were found in 42 fetuses. Overall incidence of craniospinal anomalies in present study was 0.73%. This is comparable with the study of Rajan et al [6] 0.6%, Nuzhat amer et al [7] 0.57% and Jaiganesh Shivalingam 0.46% [8].

Overall incidence of anencephaly in this study is 0.24% while in the study conducted by Dhapate et al[11] was 0.19% and Rajan et al[6] was 0.23%. Among all CNS anomalies anencephaly forms a major part i.e. 38.89%. Similar results were found by Dhapate et al [11] 48.57%, Rajan et al [6] 33.33% and Campbell[13] 24.92%.

In the present study maximum numbers of anomalies were detected in second trimester. In this study average gestational age for diagnosis for CNS anomalies is 22.76 wks, whereas in study conducted by Onkar et al [9] average gestational age was 23.54wks and in Carrol et al[14] it was 26.4 wks. This is probably because of late visit of patient to the hospital for ultrasound examination, as the majority of population under this study was from a rural locality with low economic status, poverty and ignorance.

High degree of correlation was seen between prenatal and postnatal findings. Total number of variations in antenatal ultrasound and postnatal findings were seen in 4 cases. In present study autopsy and ultrasound were exactly matching with prenatal ultrasound in 90% whereas Onkar et al [9] in 2014 showed 85.7% and Carrol et al[14] in 2000 showed 89% concordance. The autopsy was done in 25 cases. Ultrasound findings were

exactly matching with autopsy in 22 cases (88%) comparable to the studies of Isaksen et al [3] in 1998 (89%) and Onkar et al [9] in 2014 (83%). The possible reason for increased detection rate over the years could be technological improvement in ultrasound and increased experience of ultrasonologists over the years. Total number of variations in antenatal ultrasound and postnatal findings were seen in 4 cases out of which autopsy findings varied in 3 cases [Table/Fig-5]. It did not alter the prognosis. Attention to minor defect of CNS anomalies can be difficult on ultrasound due to factors like fetal positioning [15]

Proper autopsy of delivered abnormal fetuses should be done in all the possible cases which will confirm the anomalies and help us to improve the accuracy of ultrasound diagnosis and to assess the possibility of recurrence in future pregnancy. It provides psychological benefits to some patients by confirming the fetal anomalies detected by ultrasound [9]. Unfortunately it was not possible in all cases in our study due to religious customs and the patients not willing for the procedure.

## Conclusion

Ultrasound is an effective investigation for in-utero screening for anomalies including CNS. Ultrasound imaging in antenatal period practically gives an anatomical record of the developing fetus. Confirmation of the anomalies in cases of aborted fetuses definitely helps in increasing accuracy of ultrasound diagnosis and better counseling of the patient. Additionally, it provides very useful educational information. Autopsy reveals additional anomalies and gives feedback to the

ultrasonologist. This also provides psychological benefits for some patients by confirming the reality of fetal anomalies. Early detection of anomalies especially in first and early second trimester helps in planning interventions and further management. In the present study, average gestational age for ultrasound diagnosis of CNS anomalies was 22.1 weeks. It was probably due to late visit of the pregnant women to the hospital, as majority of the patients under this study was from a rural area with low socio-economic status. All the women diagnosed with fetal CNS anomalies were undergoing ultrasound for the first time.

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**Permission of IRB: Yes**

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