

Evaluation of Fetal Central Nervous System Anomalies by Ultrasound and Its Anatomical Co-relation

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ABSTRACT

Introduction: Central nervous system anomalies are often severe and are the most common indications for therapeutic abortions. Ultrasound examination helps to identify and evaluate them well before birth.

Objectives: Present study was undertaken to evaluate the incidence of central nervous system anomalies in utero by ultrasound and to confirm them by autopsy or postnatal examination.

Method: Ultrasound screening of 7485 pregnant women was performed. Pregnancy with ultrasound findings of central nervous system anomalies were followed up. Prenatal ultrasound findings

were confirmed by autopsy in cases of therapeutic abortions and fetal losses. In case of live birth postnatal findings were noted.

Results: Ultrasound detected central nervous system malformations in 24 fetuses. Ultrasound findings were matching in 83% on autopsy. Autopsy and postnatal findings together were matching with ultrasound in 85.7%. Two cases had additional minor findings on autopsy.

Conclusion: The incidence of central nervous system malformations on ultrasound was 0.31%. Autopsy and postnatal examination showed high degree of correlation with ultrasound findings.

Keywords: Antenatal ultrasound, Central nervous system anomaly, Congenital malformations, Fetal autopsy, Neural tube defects, Preconceptual counseling

INTRODUCTION

Central nervous system (CNS) anomalies are common and most devastating. They occur in frequency of about 1.4 to 1.6 per 1000 live births but are seen in about 3-6% of stillbirths. CNS develops from 3 to 20 weeks of intrauterine life. Almost all CNS anomalies are result of the insult in embryogenesis at some point of development. Ultrasound can diagnose many CNS anomalies in first and early second trimester. Some develop or become apparent in late pregnancy [1]. Earlier is the detection more is the time available for the clinician and parents to plan about the outcome of pregnancy. Lethal and severely life limiting disorders warrant early termination of pregnancy, whereas detection of minor anomalies helps everybody to be prepared for postnatal management. In a cross sectional study, 92.8% of the CNS anomalies were detected prenatally by ultrasound [2]. Sankar et al., [3] confirmed most of the CNS malformations postnatally by autopsy. Confirmation of the anomalies aids in counseling for the next pregnancy. This study was undertaken to identify and evaluate the incidence of CNS anomalies in utero by ultrasound and to confirm them postnatally.

MATERIALS AND METHODS

The present study was carried out at NKP Salve Institute of Medical Sciences and Research Centre Nagpur, India from May 2009 to April 2011. The protocol was approved by the institutional ethics committee. Total 7,485 pregnant women with gestational age more than 10 weeks were evaluated by ultrasound. Informed consent was taken. Ultrasound examination was done with 3.5 MHz convex transducer on Toshiba Core Vision and Esaote My Lab 50 ultrasound machines in the Department of Radiodiagnosis. If required transvaginal ultrasound was done with 5-9 MHz transducer especially in first trimester pregnancy. Fetuses with CNS anomalies with or without associated other anomalies were included. Their family and maternal history was noted. Autopsy was performed in case of termination of pregnancy in first and early second trimester,

spontaneous abortions, intrauterine deaths and still births. It included photograph, whole body radiograph, external and internal examination. Microscopy was done wherever necessary. Autopsy findings were compared with ultrasound findings. If pregnancy was continued, the postnatal findings were noted by external examination, radiography and ultrasound. These findings were compared with prenatal ultrasound findings.

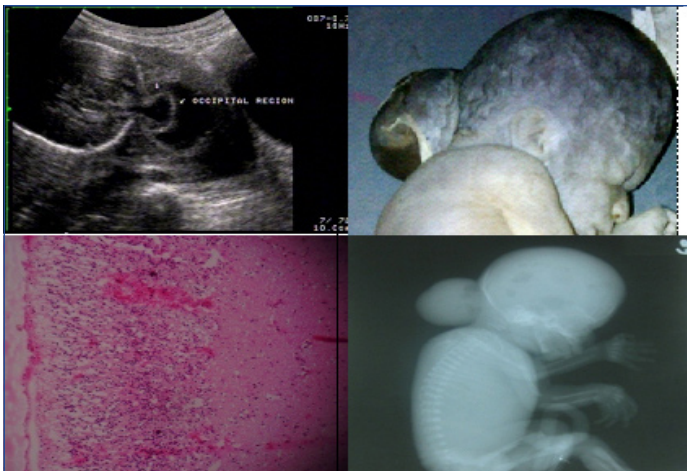
RESULTS

Ultrasound detected CNS anomalies with or without associated anomalies in 24 fetuses [Table/Fig-1-3]. The incidence of CNS anomalies was 0.31%. The mother's age varied from 19 to 32 years, with a mean of 21.3 years. The average gestational age for diagnosis of CNS anomalies was 23.54 weeks. The trimester wise distribution was as per [Table/Fig-4]. Four patients had previous history of miscarriages. None had previous birth with CNS malformation or any family history of the same. Three were lost to follow up and 3 continued the pregnancy. Eighteen autopsies were performed.

Ultrasound findings matched in 18 (85.7%) cases with autopsy

Anomalies detected by Ultrasound	Number
Anencephaly	9
Occipital encephalocele	2
Hydrancephaly	3
Hydrocephalous	2
Dandy-Walker variant	3
Agenesis of corpus callosum with interhemispheric cyst	1
Holoprosencephaly	1
Caudal regression syndrome	1
Lumbar meningo-myelocele	2
Total	24

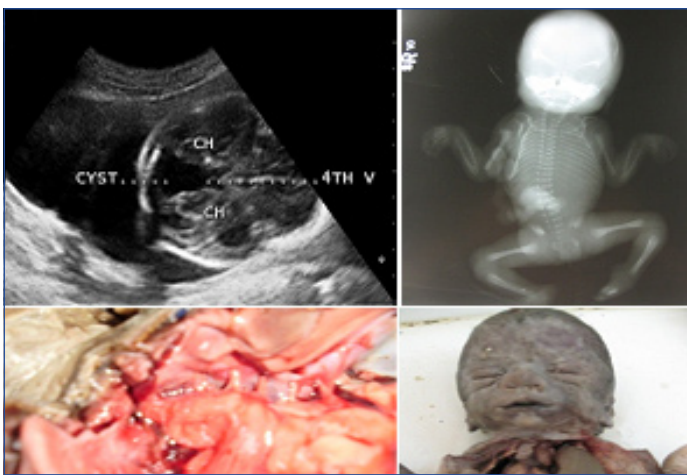
[Table/Fig-1]: Distribution of cases as per ultrasound diagnosis



[Table/Fig-2]: Occipital encephalocele on ultrasound, autopsy, microscopy and radiography



[Table/Fig-6]: Ultrasound (transabdominal and transvaginal) shows anencephaly with deformed spine. Autopsy showing anencephaly with rachischisis



[Table/Fig-3]: Dandy-Walker variant on ultrasound. Radiogram showing opaque hemithorax. Autopsy showing Dandy-Walker variant, pulmonary hypoplasia and dextrocardia.

Trimester	Number
First	1
Second	12
Third	11
Total	24

[Table/Fig-4]: Trimester wise distribution

S. No	Ultrasound finding	Autopsy
1	Anencephaly	Occipital encephalocele
2	Anencephaly with spinal defect	Craniorachischisis
3	Interhemispheric cyst with agenesis of corpus callosum	Interhemispheric cyst with partial agenesis of corpus callosum

[Table/Fig-5]: Variation in ultrasound and autopsy findings

along with postnatal findings. In 15 (83%) cases findings matched between ultrasound and autopsy. Slight variation was found in three cases. [Table/Fig-5,6].

One case of Dandy-Walker variant diagnosed on ultrasound had additional finding of micrognathia and absent right ear on autopsy. In other case diagnosed as caudal regression syndrome on ultrasound also had hemivertebra on autopsy.

In three cases of live birth, two cases had lumbar meningomyelocele and one had hydrocephalous on ultrasound. The postnatal findings exactly correlated in all three cases.

DISCUSSION

CNS anomalies occur in significant frequency and some of them are incompatible with life whereas others are life limiting. 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 [4].

In the present study the incidence of CNS anomalies on ultrasound was 0.31% which is lesser than other studies in the literature [Table/Fig-7]. The decreasing incidence in recent studies may be explained by increased awareness amongst treating physicians about folic acid fortification [10].

Study	Year	Incidence
Hobbins et al., [4]	1979	0.4%
Rajan [5]	1989	0.6%
Dhapate et al., [6]	2007	0.4%
Present study	2011	0.3%

[Table/Fig-7]: Incidence of CNS 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 23.54 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.

Autopsy and postnatal findings were matching with ultrasound in 85.7% cases. The ultrasound findings were exactly matching with autopsy in 83%. This is higher than the other reported studies [Table/Fig-8]. The possible reason for the increased rate of detection could be technological development in the ultrasound and increased experience of ultrasonologists over the years. The studies in literature have variable rate of detection. This can be due

Study	Year	Percentage
Carroll et al., [7]	2000	77
Yeo et al., [8]	2002	80
Szigeti et al., [9]	2007	65
Present study	2011	83

[Table/Fig-8]: Agreement between ultrasound and autopsy

to difference in their study population. Some of them are population based, i.e. they cover all expectant mothers in a given period; while others are carried out at a hospital level and include pregnancies with high risk of congenital anomalies [2].

On autopsy findings varied slightly in three cases [Table/Fig-5,6]. It did not alter the prognosis. Attention to minor defect of CNS anomalies can be difficult on ultrasound due to factors like fetal positioning [4]. Poor visibility due to oligohydramnios or obesity is an important cause of error in ultrasound diagnosis [3].

There were additional minor associated anomalies in two cases. Discovering minor abnormalities is important as they can be associated with a syndrome. With the present level of ultrasound the false positive diagnosis is extremely rare. Their detection on autopsy leads to refinement in aetiological diagnosis [3].

Newer ultrasound techniques like 3D and 4D ultrasound can also be used in diagnosis of birth defects. Magnetic Resonance Imaging (MRI) is being increasingly used to supplement ultrasound in diagnosis of congenital anomalies [11]. In future ultrasound is likely to be used as a primary investigation and MRI will be used to clarify the findings.

CONCLUSION

CNS anomalies are of great concern due to their severity. The incidence of CNS anomalies by ultrasound in present study was 0.31%. It is showing decreasing trend over the years. Neural tube defects were the commonest. Overall 85.7% agreement between ultrasound and autopsy along with postnatal findings was

observed. The agreement between ultrasound and autopsy alone was 83%. Confirmation of findings by autopsy can be helpful in better preconceptual counseling for next pregnancy. Autopsy at times provides additional information which may lead to enrichment of diagnosis or indicate a syndrome.

REFERENCES

- [1] Rumack C. The Fetal Brain. In Diagnostic Ultrasound, 4th edition. Elsevier Mosby Philadelphia, 2011; 1197-1244.
- [2] Neto CN, De Souza A, Filtro O, Noronha A. Validation of ultrasound diagnosis of fetal anomalies at a specialist center. *Rev Assoc Med Bras.* 2009; 55(5):541-6.
- [3] Sankar VH, Phadke SR. Clinical utility of fetal autopsy and comparison with prenatal ultrasound findings. *J Perinatol.* 2006; 26(4):224-9.
- [4] Hobbins JC, Grannum P, Berkowitz RL, Silverman R, Mahoney MJ. Ultrasonography in the diagnosis of congenital anomalies. *Am J Obstet Gynecol.* 1979; 134:331-45.
- [5] Rajan R. Ultrasound diagnosis of fetal anomalies. *J Obst Gynec India.* 1989; 39(4):461-6.
- [6] Dhapate SS, Shingare AK, Sanjay D. Early diagnosis of anencephaly - Value of ultrasound in rural area. *J Anat Soc India.* 2007; 56(2):4-7.
- [7] Carroll SG, Porter H, Abdel-Fattah S, Kyle PM, Soothill PW. Correlation of prenatal ultrasound diagnosis and pathologic findings in fetal brain abnormalities. *Ultrasound Obstet Gynecol.* 2000; 16:149-53.
- [8] Yeo L, Guzman ER, Shen-Schwarz S, Walters C, Vintzileos AM. Value of a complete sonographic survey in detecting fetal abnormalities- Correlation with perinatal autopsy. *J Ultrasound Med.* 2002; 21:501-10.
- [9] Szigeti Z, Csaba A, Pete B, Hajdú J, Papp Z, Papp C. Correlation of prenatal sonographic diagnosis and morphologic findings of fetal autopsy in fetuses with trisomy 21. *J Ultrasound Med.* 2007; 26:61-8.
- [10] De Wals P, Tairou F, Margot L, et al. Reduction in neural-tube defects after folic acid fortification in Canada. *N Engl J Med.* 2007; 357:135-42.
- [11] Salomon LJ, Garel C. Magnetic Resonance Imaging of the foetal brain. *Ultrasound Obstet Gynecol.* 2007; 30:1019-32.

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