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The study of fetal central nervous system anomalies by the means of antenatal 2D ultrasound examination in varying trimesters

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***Abstract*---**Aims and Objectives: 1) Objective is to study the range of Congenital neurological anomalies occurring in utero; 2) To evaluate the incidence of congenital anomalies in varying trimesters; 3) To confirm the diagnosis by post natal examination or autopsy as and when possible. Material and Methods: This is a prospective study which was conducted in the Radio-diagnosis department in Dhiraj General hospital, from January 2021 till February 2022. Ultrasound screening of pregnant women was performed. Pregnancy with ultrasound findings of central nervous system anomalies were followed up. Prenatal ultrasound findings were confirmed by post MTP examination in cases of therapeutic abortions and fetal losses. In case of live birth postnatal findings were noted. Results: After conducting a ultrasound screening in all three trimesters, a total of 30 congenital neurological anomalies were diagnosed. It was seen that majority of anomalies were detected in 2nd trimester. Among the neurological anomalies, most common anomalies were found to be hydrocephalus

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(4), Anencephaly (3) and lumbar myelo-meningocoele (3). Ultrasound findings were matching in 87 % of cases with post natal examination findings. In this study rare congenital neurological syndromes like Meckel Gruber syndrome and iniencephaly like disorders were also detected via ultrasound. Conclusion: Ultrasound is the most essential part of antenatal check-up. It is indispensable in screening of congenital anomalies in utero. Ultrasound can detect most of the congenital anomalies effectively and is monitor which practically gives us an anatomical record of the fetus's normal growth. Autopsy and post natal examination findings show high degree of correlation with ultrasound findings.

***Keywords*---**fetal central, system anomalies, antenatal 2D.

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. Central nervous system (CNS) abnormalities are among the most common congenital anomalies. A systematic evaluation of the head and spine should be performed to include, at a minimum, the following structures listed in the practice guideline for the standard obstetrical ultrasound examination: the lateral cerebral ventricles, the choroid plexus, the midline falx, the cavum septi pellucidi, the cerebellum, the cisterna magna, the upper lip, and the cervical, thoracic, lumbar, and sacral spine [4].

Materials and Method

The present study was conducted in Radio-diagnosis department of Dhiraj General hospital, SBKS Medical institute and research centre from January 2021 to February 2022. Total of 9850 patients were taken into study. Informed consent was taken. Ultrasound examination was done with 3.5 MHz curvilinear and 12 MHz linear probe on GE logic P5 and P9 ultrasound machines in the department of Radio-diagnosis. If required transvaginal probe of 5-9 MHz frequency was used in cephalic fetus especially in the first trimester. Post natal examination was

conducted in fetuses undergoing medical termination of pregnancy and also in fetuses which were stillborn and alive were compared with ultrasound findings.

Results

Routine ultrasound screening and dedicated anomaly scans detected various congenital neurological anomalies with or without associated anomalies. After conducting a ultrasound screening in all three trimesters, a total of 30 congenital neurological anomalies were diagnosed. The incidence of anomalies was seen in 0.3 %. Maternal age varied from 18 to 35 years with a mean of 24.3 years. It was seen that majority of anomalies were detected in 2nd trimester. Among the neurological anomalies, most common anomalies were found to be hydrocephalus (7), anencephaly (6) and lumbar myelo-meningocoele (6). Ultrasound findings were matching in 87 % of cases with autopsy findings and post natal examination findings. In this study rare congenital neurological syndromes like Meckel Gruber syndrome and iniencephaly like disorders were also detected via ultrasound. Ultrasound also detected a Arnold Chiari malformation type II in one of the fetuses of twin gestation. A case of Alobar holoprosencephaly was detected with associated cyclopia and bilateral cleft lip and cleft palate.

Anomalies detected by prenatal Ultrasound	Number
Hydrocephalus	7
Anencephaly	6
Lumbo-sacral meningio-myelocoele	6
Dandy walker malformations	2
Agenesis of corpus callosum	2
Hydrancephaly	2
Occipital encephalocoele	2
Holoprosencehaly	1
Iniencephaly	1
Rhombencephalosynapsis	1
Total	30

Majority of CNS anomalies were detected in the 2nd trimester:

Trimester	Number of anomalies
1st	3
2nd	16
3rd	11
Total	30

Pictorial depiction of few cases:

1. A case of **hydrocephalus** with dilatation of bilateral lateral ventricles and 3rd ventricle.
Normal measurement of lateral ventricle is < 10 mm and 3rd ventricle 3 mm.

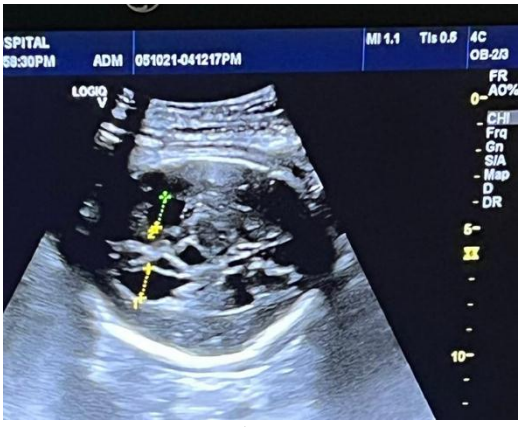


Fig 1.

Arnold chiari type II malformation-

Ultrasound signs- Banana cerebellum and lemon skull sign.

Fig 2.1

Lumbar meningio-myelocoele-

Below image shows splaying of posterior elements of lumbar vertebra (yellow arrow) with herniation of cystic structure with internal echoegenic strands s/o meningo-myelocoele.

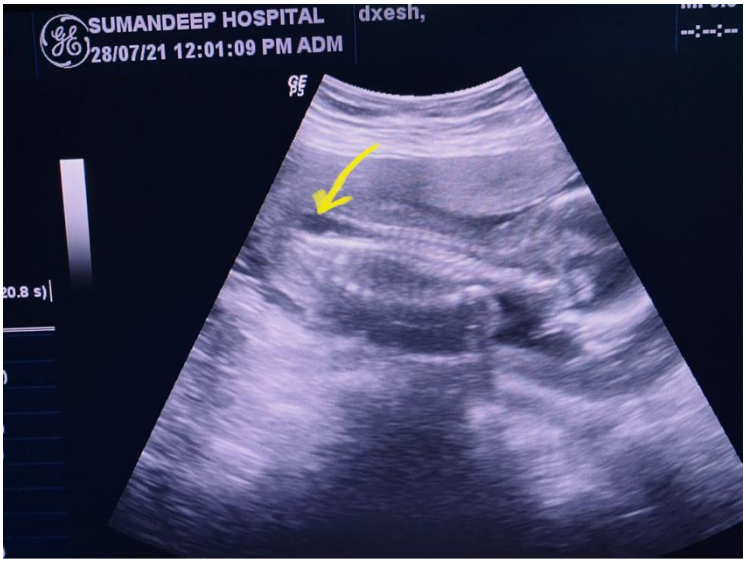


Fig 2.2

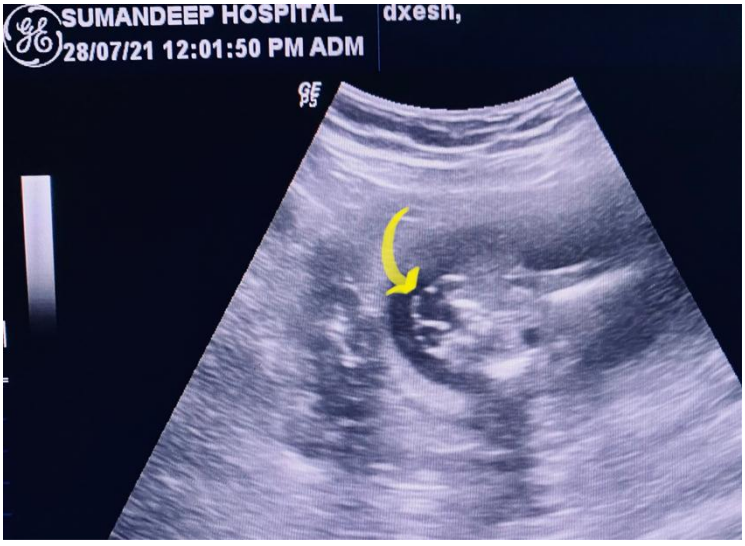
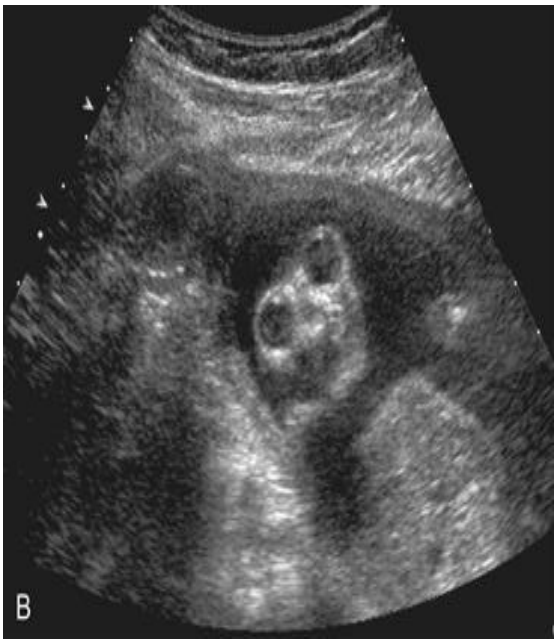


Fig 2.3 and 2.4.

Anencephaly

Ultrasound and post natal images shows a single uterine pregnancy of 16 weeks of gestation with an absence of brain tissue and bony calvarium giving a frog eye appearance.



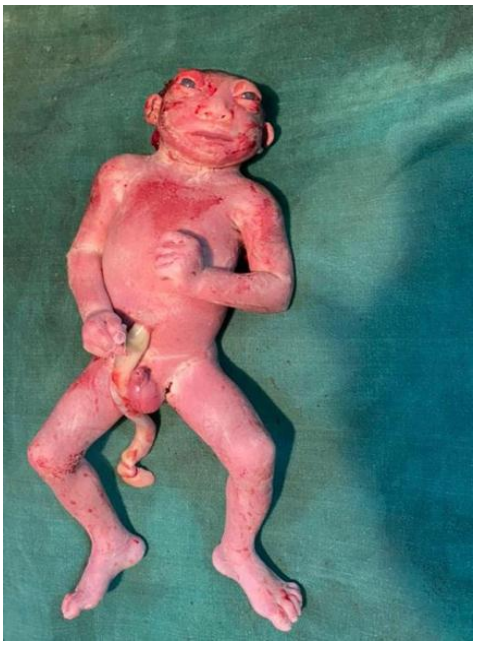
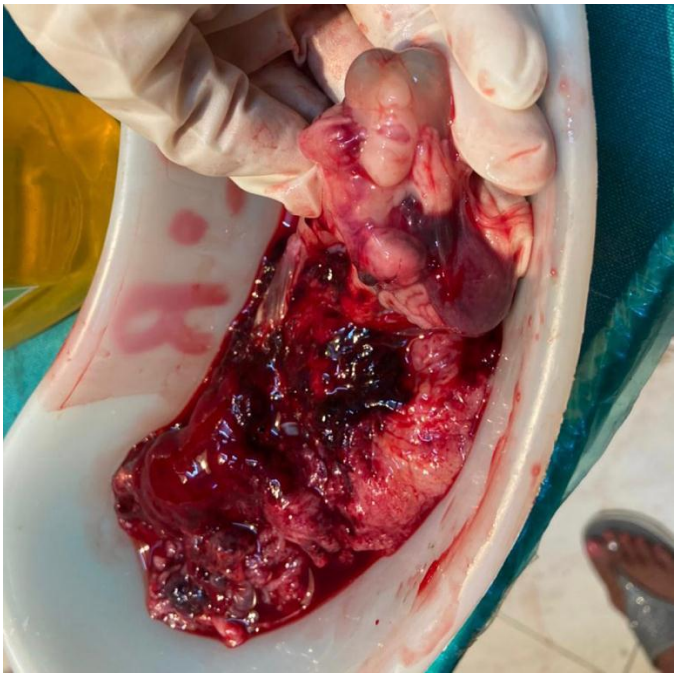


Fig 3.1 and 3.2

Iniencephaly apertus

Ultrasound and post natal images show absence of occipital part of calvarium and occipital encephalocoele (yellow arrow) with complete absence of cervical-thoracic vertebra (red arrow) (rachischisis) and fixed fetal head retroflexion. Images also show associated anomalies like gastroschisis with herniation of bowel and liver which can be seen on post MTP images.



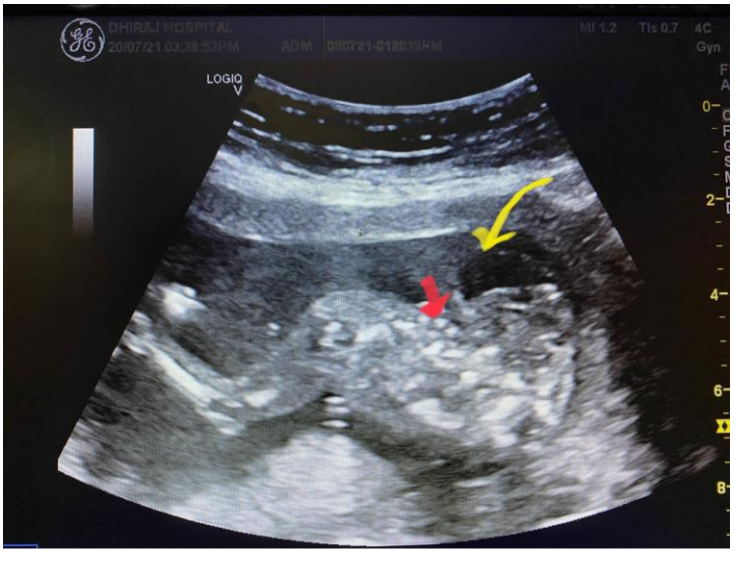


Fig 4.1 and 4.2

Occipital encephalocoele

Ultrasound images show herniation of brain tissue through a defect in occipital. There were associated anomalies like bilateral polycystic kidneys and polydactyly suggesting Meckel Gruber syndrome.

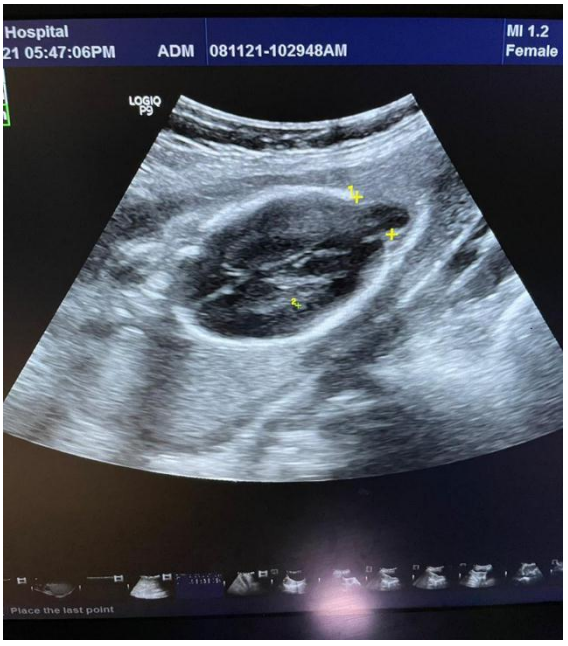


Fig 5

Hydrancephaly

Ultrasound images show gross dilation of bilateral lateral ventricle with near complete compression bilateral cerebral hemispheres.

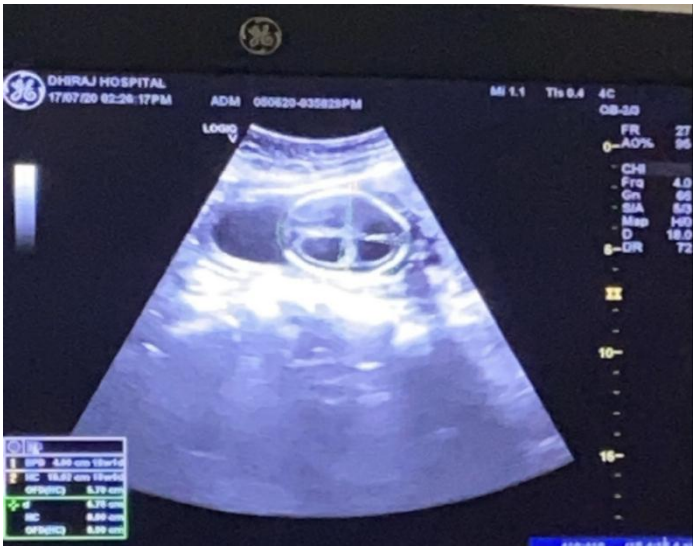


Fig 6.

Ultrasound findings matched in 87 % of cases in post MTP and post natal examination. A rare case of Meckel Gruber syndrome was diagnosed on prenatal ultrasound. A combination of occipital encephalocoele, polydactyly and polycystic kidneys were found. A rare case of twin pregnancy with one of fetus depicting sacral meningo-myelocoele and Arnold chiari malformation on examination of cranium and spine. Out of total 30 fetuses with CNS anomalies 7 (23.3%) patients showed associated extra-neurological anomalies. A total of 17 patients underwent medical termination of pregnancy while 4 were still birth. Remaining 6 fetuses were live births and 3 of the fetuses were lost due to follow up.

Discussion

Prenatal ultrasonographic examination is a part of routine antenatal screening or after alpha feto-protein screening. The basic US examination is performed via transabdomi- nal sonography by using 3 standard axial sections through the fetal brain. When an anomaly is suggested, additional sagittal and coronal planes should be attained [5,6]. If the fetus is cephalic, transvaginal imaging might improve imaging. When the fetus is breech, increased resolution with high-frequency probes in the maternal umbilicus might be helpful.

Routine scanning is currently recommended at 18 to 20 weeks of gestation. Although many cerebral anomalies are detectable in the first trimester and early second trimester, others develop or become apparent only later in pregnancy. Confirming ultrasound detected anomalies with post MTP examination and fetal MRI evaluation is definitely helpful in increasing the accuracy and for better educational purpose. It also helps in better counselling of patients. Fetal MRI is more accurate and better then ultrasound examination but ultrasound examination is cost effective and easily feasible. Further fetal MRI should be indicated in patients who are detected with abnormalities on preliminary antenatal ultrasound.

In the present study conducted at Dhiraj general hospital, the incidence of CNS anomalies were 0.3 percentage which is less in comparison to what is mentioned in previous studies and literature. The reason maybe due to increased awareness among obstetricians and physicians regarding neural tube defects and the role of frolic acid fortification in prevention of CNS anomalies.

Incidence of CNS anomalies:

Study	**Year**	**Incidence**
Hobbins et.al	1979	0.4 %
Rajan	1969	0.6%
Dhapate et.al	2007	0.4%
Deepali et.al	2011	0.3%
Present study	2021	**0.3%**

Ultrasound being the primary investigation to monitor the fetal growth and evaluate fetal anatomical structure is helpful in early detection of CNS anomalies which guides the obstetrician for further management and planning. Ultrasound also detects lethal CNS anomalies and malformation which would help obstetrician in better counselling of parents and suggesting them to undergo medical termination of pregnancy.

Ultrasound and post natal examination showed higher degree of correlation. Minor findings detected on ultrasound were correlated with fetal MRI as when it was possible. Ultrasound detected various minor CNS anomalies like mega cisterna magna and choroid plexus cysts which are important as they may be associated with syndromes like trisomies.

Ultrasound sometimes may be suboptimal in patients with difficult fetal head position, severe oligohydroamnios and obesity. Further newer 3D and 4D ultrasound techniques have increased the efficacy tremendously and further evaluation by MRI in suspected cases has revolutionized the present scenario of prenatal assessment. However Gray scale 2D ultrasound remains the primary investigation and with appropriate knowledge and technique, it is highly accurate in diagnosing CNS and spinal anomalies.

Conclusion

Ultrasound is the most essential part of antenatal check-up. It is indispensable in screening of congenital anomalies in utero. Ultrasound can detect most of the congenital anomalies effectively and is monitor which practically gives us an anatomical record of the fetus's normal growth. The incidence of CNS anomalies by ultrasound in present study was 0.30% and most of these anomalies were detected in the 2nd trimester at 24.3 weeks. Autopsy and post natal examination findings show high degree of correlation with ultrasound findings.

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