



EVALUATION OF FOETAL NEURAL AXIS IN ANTENATAL SCAN AND ITS CORRELATION WITH FOETAL OUTCOME

Radiology

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ABSTRACT

Central nervous system malformations are among the most frequent anomalies detected at antenatal sonography, with studies suggesting a prevalence as high as 1 in 1000. Ultrasound has become an increasingly valuable tool in identifying even the most subtle anomalies to a great extent. Most central nervous system malformations are discovered in the second trimester around 20 weeks of gestation, when the morphology is better recognized and targeted anomaly scans are performed as a routine practice. Even in early pregnancy, with effort and good equipment, it is now possible to move vital structures such as the spine, skull, interhemispheric fissure, choroid plexus, thalamus, and posterior cranial fossa. Sequential antenatal ultrasound starting early in first trimester has played a pivotal role in diagnosing these anomalies early.

KEYWORDS

foetal neural axis, neural tube defects, exencephaly, choroid plexus cyst, ventriculomegaly

INTRODUCTION:

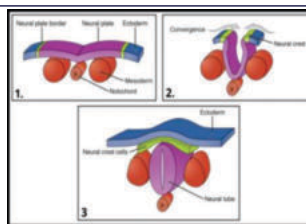
Most efforts to diagnose CNS malformations occur during the second trimester, during the detailed examination of the fetal morphology conducted at 20-24 weeks of gestation when more time is dedicated as well to the study. However, in the first trimester too with effort and good equipment, it is now becoming possible to identify some important structures like the spine, the cranium, the interhemispheric fissure, the choroid plexus, the thalamus, the posterior fossa etc. The common anomalies which are seen in the CNS are ventriculomegaly, Neural tube defects (NTDs), midline abnormalities like agenesis of the corpus callosum, Dandy Walker Malformation and other abnormalities like Intrauterine Cerebral injuries, intracranial cysts, tumors etc.



Ultrasound image of normal fetal central nervous system in midsagittal plane at 12 weeks of gestation. The thalamus (T), midbrain (M), brainstem (B), medulla oblongata (MO), and choroid plexus (CP) of the fourth ventricle are shown. Intracranial permeability (IT) of the fourth ventricle is shown.

Literature:

At the end of week two, a structure called the primitive streak appears as a groove in the epiblast layer of the bilaminar disc. Cells within the epiblast migrate downward through the primitive streak, giving rise to three layers from the initial two. These three germinal layers form the trilaminar embryonic disk: Endoderm – innermost layer, Mesoderm – middle layer and Ectoderm – outermost layer. The nervous system is derived from the ectoderm, which is the outermost layer of the embryonic disc.⁽¹⁾ In the third week of development, the notochord appears in the mesoderm. The notochord stimulates the differentiation of the overlying ectoderm into neuroectoderm, creating a thickened structure known as the neural plate. The lateral edge of the neural plate rises to the neural fold. Neural folds converge and meet in the midline, where they fuse to form the neural tube (the precursor to the brain and spinal cord). During fusion of neural folds, some cells migrate within the fold and form another cell population known as the neural crest cells.



Development of the neural tube



Normal choroid plexus (CP) & False choroid (FC) at 12 weeks.

At the fifth week of development, swelling occurs at the cranial end of the neural tube. The cranial end of the neural tube forms the brain and cerebellum, while the caudal end develops into the spinal cord. At 7 weeks, the cerebral hemispheres appear only as tiny buds which are more clearly visualised in all the foetuses by 9-10 weeks. As the hemispheres grow, they conceal the diencephalon and meet in the midline at 11 to 12 weeks. As the first trimester progresses from the 8th week onward, the mesencephalon gradually moves toward a more central location, and the third and fourth ventricles become more organized with a distinct isthmus visible between the cavities of the prosencephalon and mesencephalon by the 9th week. At 7 weeks, the falx has still not yet developed, due to which only a single lateral ventricle is visible. This continues upto 9-10 weeks with development of falx in a majority of cases. At 12 weeks, the corpus callosum is not visible, but the stalks that continue to develop can be seen. The Cavum septum pellucidum may be appreciated in upto 40% of foetuses by the end of second trimester. The Rhombencephalon is the most obvious structure in the early first trimester, where the growth of the Cerebellar hemispheres may be visualised by 10-11 weeks, which fuse in the midline by 11-12 weeks. The choroid plexus of the 4th ventricle is visible by the tenth week as a hyperechoic region across the roof of the 4th ventricle. The spine can be visualised as two parallel lines as early as 8 weeks, period of gestation, which are further appreciated better by 11-12 weeks.

NTD results from failure of cranial and/or caudal neural tube closure. Their etiology is unknown. However, associations with folic acid deficiency and differential incidence of structural abnormalities and karyotypic/genetic defects have been reported.⁽²⁻⁴⁾ The neural tube comprises a bundle of nerve sheath which closes to form brain caudally and spinal cord rostrally. The closure should occur at around the 28th day of conception failing which the brain or spinal cord does not form properly.

Numerous types of neural tube defect are recognised including

- Myelomeningocele (50%)

- Anencephaly (40%)
- Encephalocele (5%)
- Tethered cord
- Exencephaly
- Meningoencephalocele
- Meningocele

Few other abnormalities in the brain are enumerated below:

- Blake pouch cyst
- Arachnoid cyst
- Choroid plexus cyst
- Vermian agenesis/hypoplasia
- Dandy Walker syndrome

MATERIALS AND METHODS:

- Type of study- Prospective study of Foetal Neural axis evaluation and its correlation with foetal outcome.
- Period of study- April 2021 to December 2022
- Institute Ethics Committee approval was obtained before start of the study.

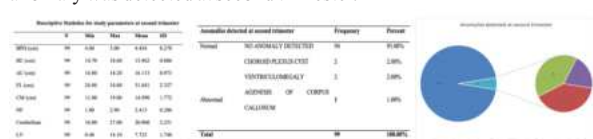
The data was obtained from the antenatal cases coming to the obstetric unit of MGM Kalamoli, Navi Mumbai. The ladies coming for their routine ultrasound examinations for ANC checkup were evaluated in all trimesters for the ultrasound study; 100 cases were studied and followed up. The patients underwent detailed NT-NB and anomaly scan.

RESULTS:

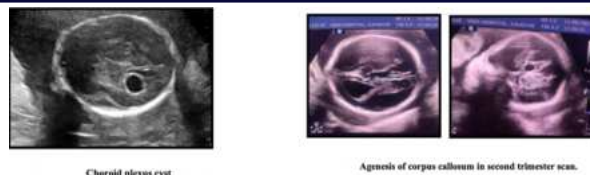
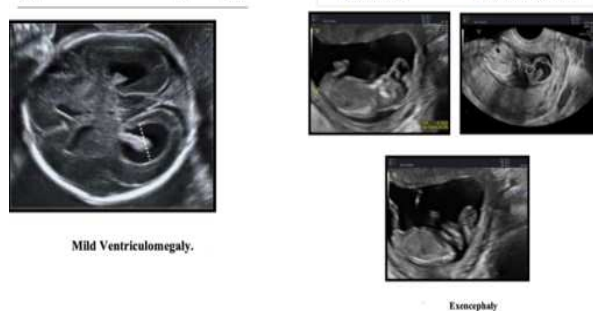
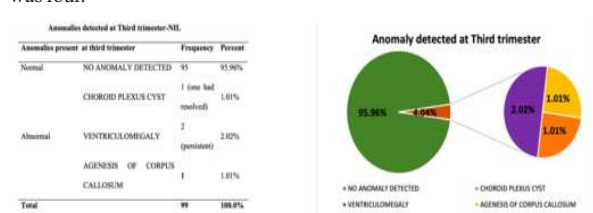
Out of 100 study subjects, in case of 97 (97%) no anomaly was detected at first trimester, and in case of 3 (3%) subjects anomaly was detected. Out of these three anomalies, 1 (1%) was Ventriculomegaly, 1 (1%) was Agenesis of corpus callosum and 1 (1%) was Exencephaly.



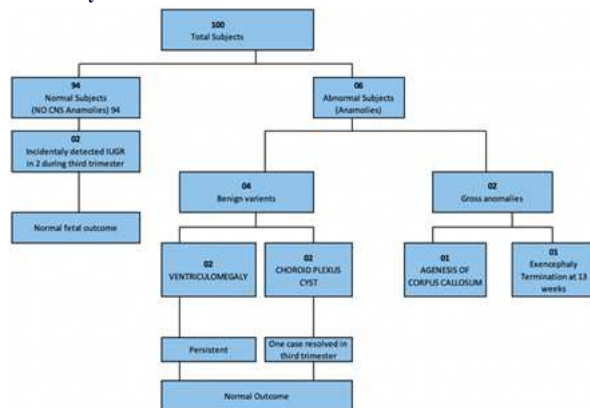
Among three anomalies detected at first trimester, in one subject with Exencephaly anomaly, the pregnancy was terminated. At second trimester, in case of 94 (94%) subjects no anomaly was detected and 3 (3%) anomalies were detected during second trimester, in case of 2 (2%) subject, the Choroid Plexus Cyst anomaly was detected and in case of 1 (1%) subject, new ventriculomegaly was detected. Hence till second trimester total six anomaly were detected out of which 3 anomaly was detected at second trimester.



During the third trimester no new anomaly was detected, but in case of one subject, the previous anomaly of Choroid Plexus Cyst was resolved. Hence, total number of anomalies seen in the third trimester was four.



Summary:



DISCUSSION:

In the 12-13 week scan, which is also known as Nuchal translucency scan, is considered to be the first level anomaly scan in routine practice. In the selected cases, which were followed up, there was one case of Exencephaly detected. The findings were immediately conveyed to the referring clinician, and the patient was counselled regarding it, and was advised to undergo termination, which she did. This management was similar to a study by Zapp⁽⁵⁾, where all cases of exencephaly were diagnosed by 13 weeks of gestation and all underwent termination. In the second trimester scan, done at 19-20 weeks of gestation all ladies were thoroughly evaluated for all possible anomalies. In our study, which now included only 99 subjects, there were two cases of mild Ventriculomegaly, one of which was newly added as compared to the first trimester scan. These patients were carefully assessed for any other abnormalities, family history and were found to be within normal limits. They were now considered to be benign etiology, likely to be normal variants. This was in concordance with the study by Melchiorre K et al, according to which most cases of mild Ventriculomegaly have a normal fetal outcome, whereas 10% have some neurodevelopmental abnormalities⁽⁶⁾. Also, there were two cases of Choroid Plexus cysts newly detected at the anomaly scan. The incidence of CPC in the present study was 2%, which is similar to a study by Michael Entezami⁽⁷⁾. The case of agenesis of corpus callosum was thoroughly evaluated for other abnormalities, as a detailed anatomical scan was possible at this week of gestation. The various features were noted and assessed and the patient was counselled regarding the outcome & prognosis. The patient chose to continue with the pregnancy. At the second trimester, there were three new anomalies detected, all of which were normal variants, and also the diagnosis of ACC was confirmed. It is a comparatively rare anomaly with incidence of 1.4 per 10,000 live births, and 2 % in developmentally disabled, as per Jeret JS et al⁽⁸⁾. In the third trimester scan, all patients were assessed for interval growth. All the subjects were followed in the third trimester, which included 99 patients, of which five had anomalies detected in the previous trimesters, while 94 of them were normal. One case of Choroid plexus cyst which was detected in the anomaly scan was now resolved, while one was persistent. The case of agenesis of corpus callosum was persistent. The two cases of ventriculomegaly were persistent. All cases had full term uneventful deliveries, except for the case of Exencephaly which was terminated. This is in keeping with a systematic review by Sotiradis A⁽⁹⁾, where 74.7 % of cases have a normal developmental outcome. This study shows that with careful sonographic examination, with addition of TVS, 50% of the anomalies were detected in the first trimester itself, while the rest were detected in the second trimester anomaly scan. This is especially important since the legal limit for undergoing MTP in India is 20 weeks, & only under special circumstances, it can be upto 24 weeks. Hence, early detection of these anomalies is crucial. Also 100% of the gross anomalies were detected in the first trimester itself at 12-13 weeks scan. With early detection the process of termination becomes less complicated, with adoption of minimally invasive techniques, & lower risk to the mother.

No new anomalies were detected in the third trimester, which was expected since all ladies were regularly called for their antenatal scans with appointments given before.

CONCLUSION:

The anomalies of the fetal neural axis can be diagnosed antenatally by careful clinical assessment and serial Ultrasonography. The addition of TVS in routine first trimester scans provides greater anatomical details, and increases chances of picking up anomalies at an earlier stage. Extensive supervised training is required in order to achieve competence in assessment of fetal neural axis. Knowledge of normal anatomy & embryology is crucial in evaluation of the fetal neural axis. It also emphasized counselling the patients, and good communication with the referring obstetrician, regarding the findings. Any abnormal finding must be correlated with other systems & also supplemented with clinical, as well as family history.

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