

Prenatal Detection OF Central Nervous System Malformations on Ultrasound

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Abstract: Central nervous system(CNS) malformations are one of the commonest malformations that frequently encountered in pregnancy. It can vary from subtle to very severe defects. Most of them can be diagnosed with prenatal ultrasound.

Objective: The purpose of our study was to describe the incidence of various CNS malformations and their imaging findings on antenatal ultrasound.

Materials and Methods: This prospective study was carried out in a tertiary care hospital in Kanchipuram, India from January 2012 to December 2014. Total of 3469 cases underwent antenatal ultrasound in the department of radiology. Fetuses with central nervous system malformations were included in the study and detailed clinical history recorded from the mother. Cases with congenital malformations without involvement of central nervous system were not included in the study. Antenatal ultrasound findings were confirmed after the delivery of fetus with clinical examination. Autopsy of delivered fetuses were not done in our study.

Results: Out of 3469 cases, 16 were diagnosed with central nervous system anomalies. Postnatal examination showed higher degree of correlation with antenatal Ultrasound findings.

Conclusion: Incidence of central nervous system anomalies was 0.46%. Ultrasound is the diagnostic modality of choice in the detection of congenital central nervous system anomalies. Proper screening with ultrasound during antenatal period prevents the delivery of child with central nervous system anomalies.

Keywords: Spina bifida; Anencephaly; Spinal Dysraphism; Sacral agenesis; TORCH; Cephalocele;

I. Introduction

Central nervous system(CNS) malformations are one of the commonest congenital anomalies encountered in pregnancy. CNS malformations are due to insult in the development during embryogenesis^[1,2]. 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]. Severe and Lethal CNS malformations requires termination of pregnancy, however minor defects can be managed in the earliest possible postnatal period. Recent advances in the ultrasound made earlier detection of CNS malformations in pregnancy and increased the sensitivity of detection of CNS malformations.

II. Objective

The purpose of our study was to describe the incidence of various central nervous system anomalies and their imaging findings on antenatal ultrasound.

III. Materials And Methods

This prospective study was carried out in a tertiary care hospital in Kanchipuram, India from January 2012 to December 2014. Total of 3469 cases underwent antenatal ultrasound in the department of radiology. Ultrasound screening was done with GE Logic P6 and Aloka – Prosound Alpha 6. Fetuses with central nervous system anomalies were included in the study and detailed clinical history recorded from the mother. Cases with congenital anomalies without involvement of central nervous system were not included in the study. Antenatal ultrasound findings were confirmed after the delivery of fetus with clinical examination. Autopsy of delivered fetuses was not done because none of the patient or patient's relatives gave consent.

IV. Results

Out of 3469 cases, 16 were diagnosed with central nervous system anomalies. Age groups of mothers having fetus with CNS malformations varied from 18 – 36 years in our study(**Table – 1**). Gestational age during which CNS malformations detected was in between 12 – 33 weeks in our study. 2 patients had history of consanguineous marriage. Two patients had history of one previous abortion. 4 patients were primi gravida, 8 patients were second gravida, 2 patients were 3rd gravida and 2 patients were 4th gravida(**Table – 2**). 8 patients(50%) were not registered in antenatal clinic for regular checkup. No patient had history of congenital CNS malformation in previous child or family history. Various CNS malformations detected in our study were listed below(**Table – 3**).

3 cases were diagnosed as **Anencephaly**(**Fig - 1**), on ultrasound complete absence of calvarium with dysplastic brain parenchyma floating in the amniotic fluid and positive “Mickey Mouse sign”. Dysplastic cerebral hemispheres as two semicircular structures above the orbits floating in amniotic fluid described as Mickey Mouse Sign by Chatzipapas et al^[3].

1 case was diagnosed as **isolated hydrocephalus** due to moderate dilatation of lateral ventricles(16 mm at the level of atria) without any associated malformations.

In 4 cases, splaying of posterior spinal elements(3 cases at lumbo-sacral level and 1 case at cervical level) involving 3-4 vertebrae with dura and neural elements floating freely in the amniotic fluid(**Fig - 2**). All of them were diagnosed as **open spina bifida with meningocele**. All the 4 cases were associated with moderate hydrocephalus. One case also showed corpus callosal agenesis. One patient came for first time ultrasound examination at 33 weeks of gestation.

In 2 cases, focal splaying of posterior elements involving one vertebra at lumbar spine level with intact overlying skin and subcutaneous tissue(**Fig - 3**). No hydrocephalus seen in both the cases. Hence both of them diagnosed as **Closed Spina bifida**.

In 1 case, the fetus had no cranium, the brain parenchyma appears dysplastic but it is covered with a thick layer of membranes(**Fig - 4**) and we diagnosed this case as **Acrania** since the dysplastic brain parenchyma was covered with a layer of scalp^[2]. Unfortunately we lost follow up of this patient.

In 1 case, focal calvarial defect seen in the occipital region through which herniation of brain covered with dura and diagnosed as **Occipital cephalocele**.

In 1 case, complete agenesis of sacrum(**Fig - 5**) seen and diagnosed as **caudal regression syndrome**.

In 1 case, the curvature of the fetal spine is altered with disrupted parallel line pattern and a smaller size of vertebral body at upper dorsal spine level(**Fig - 6**) seen and diagnosed as **Hemivertebra**^[4].

In 1 case, both lateral ventricles fused to form single ventricle with partially fused thalamus and absent cavum septum pellucidum(**Fig - 7**), hence it is diagnosed as **Holoprosencephaly**.

In 1 case faint periventricular calcifications are seen in the brain(**Fig - 8**) with no other associated abnormalities, we suspected some **TORCH** infection^[5] but we lost follow up of this patient.

Incidence of CNS malformations in routine pregnancy was 0.46% in our study. It is well correlated with other recent studies.(**Table – 4**)^[1,6,7,8]

V. Discussion

Congenital CNS malformations are very frequently encountered among the congenital malformations, in which neural tube defects are the most common. Open spina bifida and anencephaly 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 12 weeks and another at 18 weeks of gestational age. Hydrocephalus is the commonest associated finding in cases with open spina bifida. However hydrocephalus not always seen in cases with closed spina bifida, hence normal ventricles will not rule out spinal anomalies. In our study ventricles appear normal in both the cases of closed spina bifida.

Fetal head should be evaluated in three planes namely transventricular, transthalamic and transcerebellar planes to identify the important structures such as Head shape, Bone density, ventricles, Cavum septi pellucidi, Thalamus, Cerebellum & vermis and Cisterna magna especially at 18 – 22 weeks of gestational age^[2,9].

Fetal spine should be evaluated in at least two planes axial and sagittal, if possible coronal plane can be included. In axial planes spine should be examined from the base of skull to sacrum and each vertebra appear as three echogenic dots(three ossification centers). In sagittal plane whole spine appears as continuous two or three parallel lines with smooth tapering at sacral spine level. Always the intactness of the skin overlying the spine should be examined in axial and sagittal planes^[9].

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^[10]. Unfortunately it was not possible in our study due to religious customs and the patients not willing for the procedure. However the incidence of CNS malformations especially the neural

tube defects was decreased nowadays due to proper folic acid supplementation in the females before conception^[11]. Proper registration in antenatal clinics with regular antenatal checkups and standard ultrasound screening at 11 – 14 weeks and 18 – 22 weeks will help in the detection of most of the congenital malformations. Ultrasound is the most preferable modality of choice to assess the fetal well being.

VI. Conclusion

Incidence of CNS malformations was 0.46%. Neural tube defects were the commonest. Ultrasound is the diagnostic modality of choice in the detection of congenital central nervous system anomalies. It is easily available, cheaper, cost effective, safer during pregnancy and repeatable. But the main drawback with ultrasound is completely observer dependent and high interobserver variation. Also the visibility of fetus depends on the position of fetus, amount of amniotic fluid and maternal obesity^[1,10,12]. Recent advances in Ultrasound such as 3D/4D techniques will improve its diagnostic efficacy even in minor congenital anomalies. In future MRI can be used as a modality for confirmation of findings and ultrasound used as a first line of investigation in pregnancy. Proper screening with ultrasound in a specified period of gestation prevents the delivery of child with central nervous system anomalies.

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Tables

Table – 1:

Age groups	No. of Cases
18-20 years	5
21-30 years	7
31–36 years	3

Table – 2:

Obstetric History	No. of Cases
Primi	4
2 nd Gravida	8
3 rd Gravida	2
4 th Gravida	2

Table – 3:

CNS Malformations	No. of Cases	Percentage %
Anencephaly	3	18.75
Acrania	1	6.25
Open Spina bifida	4	25
Closed Spina bifida	2	12.5
Holoprosencephaly	1	6.25
Sacral Agenesis	1	6.25
Hemivertebra	1	6.25
Suspected TORCH Infection	1	6.25
Occipital Encephalocele	1	6.25
Isolated Hydrocephalus	1	6.25

Table – 4:

Various Studies	Incidence
Ghavami et al 2007	0.5 %
Dhapate et al 2007	0.19 %
Deepali Onkar et al 2014	0.31 %
Nuzhat Amer et al 2014	0.57 %
Our Study	0.46 %

Figures

Fig - 1



Fig – 1: Anencephaly - Coronal section fetal head showing absent calvarium above the level of orbits.

Fig - 2



Fig – 2: Open spina bifida with Hydrocephalus – Brain axial section(2a) showing dilated lateral ventricles, Sagittal(2b) and axial(2c) images of spine showing splaying of posterior spinal elements covered with dura.

Fig - 3

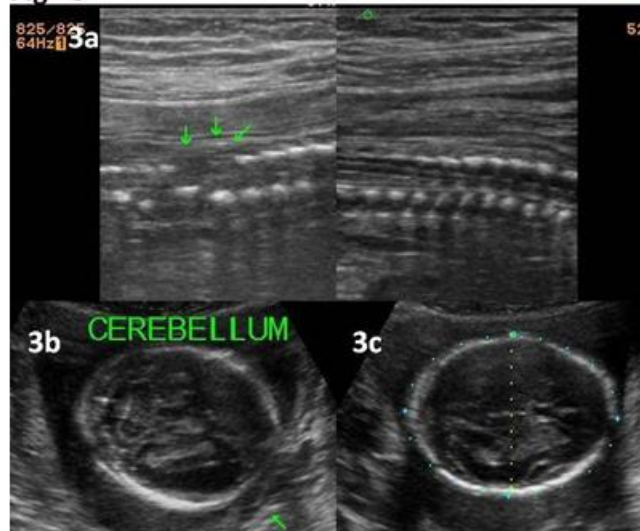


Fig – 3: Closed spina bifida without Hydrocephalus – Sagittal(2a) images of spine showing absence of posterior spinal elements with intact overlying skin and Brain axial sections(2b & 2c) showing normal lateral ventricles.



Fig – 4: Acalvaria – Sagittal(4a) and Axial brain(4b & 4c) images showing absent calvarium with dysplastic brain covered with a thick membrane probably scalp.



Fig – 5: Sacral Agenesis – Sagittal image of spine showing absence of sacral spine.

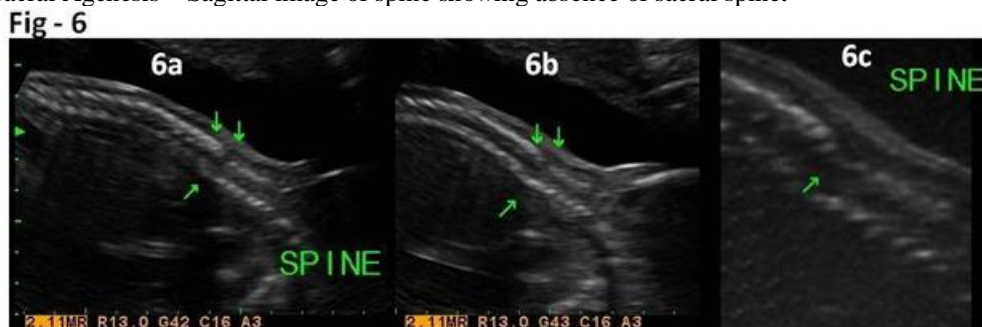


Fig – 6(a,b &c): Hemivertebra – Sagittal images of spine showing altered curvature with disrupted parallel line pattern and a smaller size of vertebral body at upper dorsal spine level.

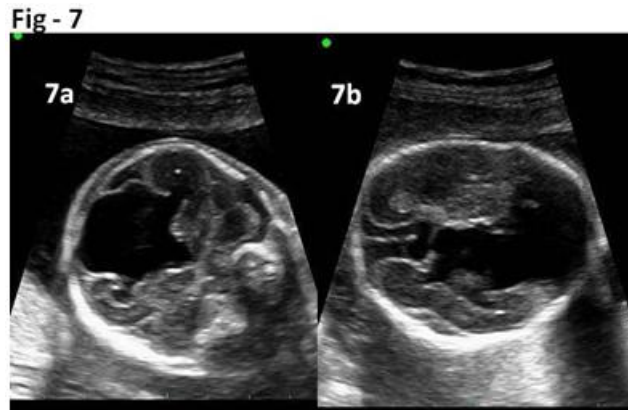


Fig - 7(a&b): Holoprosencephaly - Coronal(a) & axial(b) images of brain showing fused single lateral ventricle with partially fused thalami.

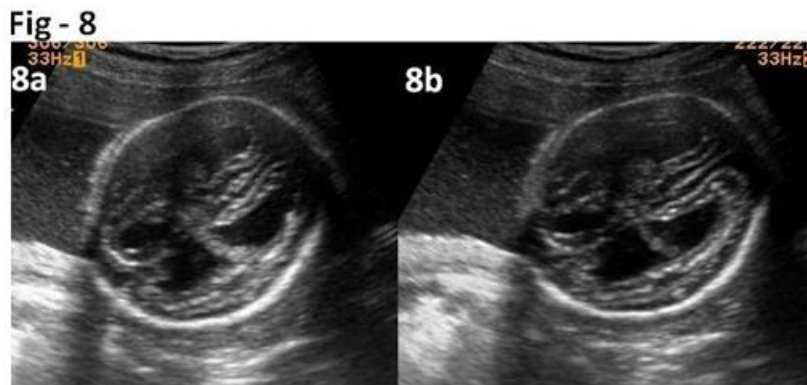


Fig - 8(a&b): Axial images of brain showing faint echogenic soft calcification in bilateral periventricular region.