Original Research Paper

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Radio-Diagnosis

IMAGING WITH USG, DIAGNOSTIC ACCURACY IN DETECTING FETAL CNS ANOMALIES

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ABSTRACT Anomalies of the CNS are the most common cause of referral for prenatal diagnosis. CNS anomalies occur with a frequency of about 1.4 to 1.6 per 1000 live births but are seen in about 3% to 6% of stillbirths.1 The increased use of maternal serum alpha protein screening and first trimester nuchal translucency screening has resulted in increased number of pregnancies being referred for evaluation of CNS and suspected anomalies. Routine scanning is 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 later pregnancy. 2-6

Prenatal diagnosis of central nervous system (CNS) anomalies is very important in making decision about therapeutic termination. In the past several decades, prenatal abnormalities were mainly detected by maternal serum analysis, amniocentesis, cordocentesis and chorionic villous sampling.

Ultrasound (US) detection of prenatal abnormalities is a non-invasive technique, which is more acceptable by patients. Several studies have shown an accuracy of 92% to 99.7% for US detection of CNS anatomic anomalies.

This study was designed to assess the role of US in detection of gross prenatal CNS anatomic anomalies and its impact on outcome in southern belt of rajasthan.

KEYWORDS :

INTRODUCTION:.

Embryological anatomy: Morphological changes start from about 5th menstrual week when the cells destined to form the notochord infiltrate into the embryonic disc and induce the overlying tissue to thicken, fold over and fuse as the neural tube. Zipper like fusion starts in the mid trunk of the embryo and then extends to the cranial and caudal ends. [7,8]

Common CNS anomalies detected in a routine ultrasound scan are as under:

(1)Neural tube defects.

Definition: These include anencephaly, spina bifida and cephalocele.

Anencephaly:

Diagnosis:

- Anencephaly- acrania exencephalyanencephaly
- Demonstration of absent cranial vault and cerebral hemispheres.
- However, the facial bones, brain stem and portions of the occipital bones and mid-brain are usually present.
- Associated spinal lesion- 50% of cases.
- The diagnosis can be made after 11 weeks, when ossification of the skull normally occurs.

Spina bifida-

- The neural arch is "U" shaped.
- There is an associated bulging meningocele (thin-walled cyst) or myelomeningocoele.
- Associated kyphoscoliosis are best assessed in the longitudinal scan.

Encephaloceles- cranial defects with herniated fluid-filled or brain-filled cysts. They are most commonly found in an occipital location (75% of the cases) but alternative sites include the frontoethmoidal and parietal regions.

(2)Hydrocephalus/ ventriculomegaly

In hydrocephalus there is pathological increase in the size of the cerebral ventricles.

Diagnosis:

A transventricular plan: demonstrate the dilated lateral ventricles, defined by a diameter of 10 mm or more. [9,10]

a. Mild- 10mm; b.Borderline ventriculomegaly-10-15 mm; c.Overt ventriculomegaly or hydrocephalus- >15 mm.

(3)Holoprsencephaly

- There are three types according to the degree of forebrain cleavage. [11]

1. Alobar ; 2. Semilobar ; 3. lobar holoprosencephaly.

Diagnosis:

- There is a single dilated midline ventricle replacing the two lateral ventricles or partial segmentation of the ventricles. The alobar and semilobar types are often associated with facial defects, such as hypotelorism or cyclopia, facial cleft and nasal hypoplasia or proboscis. [12-16]

(4) Agenesis of corpus callosum:

The corpus callosum is a bundle of fibers that connects the two cerebral hemispheres. It develops at 12-18 weeks of gestation.

Diagnosis:

- The absence of the cavum septum pellucidum
- The 'teardrop' configuration of the lateral ventricles (enlargement of the posterior horns).
- Agenesis of the corpus callosum is demonstrated in the mid-coronal and mid-sagittal views, which may require vaginal sonography.

(5) Dandy walker malformation:

- The Dandy-Walker complex refers to a spectrum of abnormalities of the cerebellar vermis, cystic dilation of the fourth ventricle and enlargement of the cisterna magna.

Diagnosis:

- Cystic dilatation of the fourth ventricle.
- Partial or complete agenesis of the vermis.
- Associated hydrocephalus and other extracranial defects.
- Enlarged cisterna magna is diagnosed if the vertical distance from the vermis to the inner border of the skull is more than 10 mm. [17]

- Prenatal diagnosis of isolated partial agenesis of the vermis is difficult and a false diagnosis can be made prior to 18 weeks gestation, when the formation of the vermis is incomplete and anytime in gestation, if the angle of insonation is too steep.

(6) Vein of galen aneurysm:

This is a midline aneurismal dilation of the vein of Galen due to an

arteriovenous malformation with major hemodynamic disturbances. Vein of Galen aneurysm is a very rare abnormality.

Diagnosis:

The diagnosis is made by the demonstration of a supratentorial midline translucent elongated cyst. Color Doppler demonstrates active arteriovenous flow within the cyst.

MATERIALS AND METHODS:

- This is a prospective study of diagnostic accuracy of US in detecting prenatal gross CNS anomalies in pregnant women with gestational age of 15 weeks or more.
- This study was conducted in Geetanjali medical college and Hospital between May 2018 to Jan 2019. 8000 pregnant women were scanned during this period of time.
- Cases were all referred by obstetricians/gynaecologists for routine work-up of pregnancy
- All patients were scanned by professors.
- Brains were scanned in axial, coronal and sagittal sections through most important anatomic areas; i.e., transventricular, transthalamic and transcerebellar planes.
- Whenever possible delivered fetuses were followed up to confirm the anomalies.
- Because of lack of speciality of fetal medicine, still born and dead fetuses were not autopsied to conform the diagnosis. So in such cases, ultrasonographic diagnosis was considered as final diagnosis.

Technique- Brain scans:

- 3 transverse planes- transthalamic, *transventricular* and the *transcerebellar* plane.

1) The transthalamic plane- Used for measurement of the biparietal diameter (BPD), head circumference (HC),
2) The *transventricular* plane- obtained at the level of the cavum septum pellucidum.

- Demonstrate - frontal horns, atria of the lateral ventricles, the choroid plexuses and the Sylvian fissures.

3)The *transcerebellar* (or suboccipitobregmatic) view obtained through posterior fossa.

- Demonstrates: mid-brain and posterior fossa;
- Used for measurement of the transverse cerebellar diameter (TCD) and cisterna magna (CM).

Additional scanning planes along different orientations may be required from time to time to better define subtle details of intracranial anatomy in selected cases.

Technique spine scan:

- Axial scan of entire spine starting from cervical to sacral region must be done as spina bifida is better demonstrated in axial view (U shape instead of V shape).
- Sagittal scan-
  The normal spine- 'double railway' appearance
  - the intact soft tissues above it.
- Coronal plane, the three ossification centers of the vertebra form three regular lines that tether down into the sacrum.

RESULTS:

CNS ANOMALY	FREQUENCY	PERCENT
ANENCEPHALY	4	14.8%
CHOROID PLEXUS MASS	1	3.7%
CORPUS CALLOSAL AGENESIS	3	11.1%
DANDY WALKER MALFORMATION	1	3.7%
ENCEPHALOCELE	2	7.4%
HOLOPROSENCEPHALY	5	18.5%
HYDROCEPHALUS	3	11.1%
SPINA BIFIDA	7	25.9%
VEIN OF GALEN MALFORMATION	1	3.7%
Total	**27**	**100.0%**

CNS ANOMALY	NORMAL	POLYHYDRAMNIOS	TOTAL
ANENCEPHALY	0	4	4
CHOROID PLEXUS MASS	1	0	1
CORPUS CALLOSAL AGENESIS	3	0	3
DANDY WALKER MALFORMATION	1	0	1
ENCEPHALOCELE	1	1	2
HOLOPROSENCEPHALY	5	0	5
HYDDROCEPHALUS	3	0	3
SPINA BIFIDA	0	7	7
VEIN OF GALEN MALFORMATION	1	0	1
TOTAL	15	12	27

CNS ANOMALY	NO FA supplementation	FA supplementation	TOTAL
ANENCEPHALY	3	1	**4**
ENCEPHALOCELE	1	1	**2**
SPINA BIFIDA	7	0	**7**
TOTAL	**11**	**2**	**27**

gestational week	Frequency	Percent
<20	5	18.5%
>20	22	81.5%

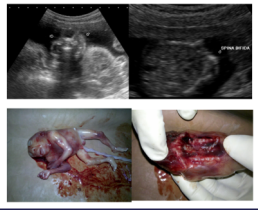
SUMMARY:

- Overall incidence of CNS anomalies as detected in our study was 0.35%
- Ultarsound was found quite accurate for detection of congenital CNS anomalies with accuracy approaching 100%.
- Majority of CNS anomalies are neural tube defects like spinabifida, anenecephaly and encephalocele followed by holoproscencephaly.
- Majority of patients with anomalies came for antenatal USG for the first time after 20 weeks.
- Non of the patients have taken folic acid supplementation before conception.
- Only few patients have taken folic acid supplementation after conception.

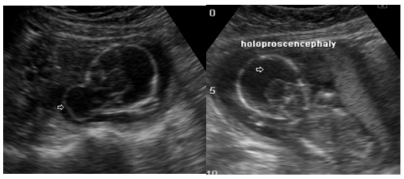
CONCLUSION:

- Majority of the CNS anomalies are either lethal (i.e. anencephaly, alobar and semilobar holoproscencepahly , encephaloceles) or associated with major morbidity (i.e. spina bifida, hydrocephalus) to the child in the later life, so their early and timely detection in antenatal period is must so that they can be timely intervened.
- Majority of patients with CNS anomalies in our study came for antenatal USG for the first time in late second trimester. This is major problem in developing countries like us mainly due to lack of awareness on part of patient.
- Folic acid supplementation is recommended before conception and after pregnancy to reduce risk of neural tube defects. In our study majority of patients with neural tube defects have not taken folic acid supplementation.
- Advent of 3D and 4D ultrasound has been a new revolution in fetal medicine being most accurate especially in detection of spinal dysraphism.

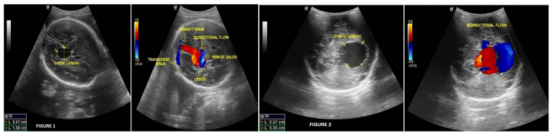
ANENCEPHALY 15 WEEKS:



OCCIPITO-CERVICAL MENINGOCELE 16 WEEKS:



Vein of Galen malformation 25 weeks



REFERENCES:

1. Chitty LS, Pilu G. The challenge of imaging the fetal central nervous system: an aid to prenatal diagnosis, management and prognosis. Prenat Diagn 2009:29(4):301-302.
2. Malinger G, Lerman-Sagie T, Watemberg N, et al. A normal second trimester ultrasounf does not exclude intracranial structural pathology. Ultrasound Obstet Gynecol. 2002;20(1):51-56
3. Bilaas HG, Eik-Nes SH. Sonoembryology and early prenatal diagnosis of neural anomalies. Prenat Diang.2009;29(4):312-325.
4. Pooh RK, Kurjak A. 3D/4D sonography moved prenatal diagnosis of fetal anomalies from the second to the first trimester of pregnancy. J Matern Fetal Neonatal Med.2012;25(5):433-455.
5. Saltvedt S, Almstrom H, Kublicaks M et al. Detection of malformations in chromosomally normal foetuses by routine ultrasound at 12 or 18 weeks of gestation – A randomised control trial in 39,527 pregnancies. BJOG.2006;113(6):664-674.
6. Carletti A, Colleoni GG, Perolo A, et al. Prenatal diagnosis of cerebral lesions acquired in utero and with late appearance. Prenat Diagn.2009;29(4):389-395.
7. Greene ND, Copp Aj. Development of the vertebrate central nervous system: formation of the neural tube. Prenat Diagn.2009;29(4):303-311.
8. O'Rahilly R, Muller R, editors. Human embryology and teratology. 2nd ed. New York: Weily- Liss; 1996.
9. Gagloiti P, Oberto M, Todors T. The significance of fetal ventriculomegaly: etiology, short and long term outcomes. Prenat Diagn 2009;29(4):381-388.
10. Melchiorre K, Bhide A, Gika AD, et al. Counselling in isolated mild fetal ventriculomegaly. Ultrasound Obstet Gynecol.2009;34(2):315-317.
11. Demyer W, Zeman W, Palmer CD. Familial alobar holoprosencephaly with median cleft lip and palate. Report of patient with 46 chromosomes. Neurology. 1963;13:913-918.
12. Winter TC, Kennedy AM, Woodward PJ. Holoprosencephaly: a survey of the entity, with embryology and fetal imaging. Radiographics.2015;35(1):275-290.
13. Dubourg C, Bendavid C, Pasquier L, et al. Holoprosencephaly. Orphanet J Rare Dis. 2007;2:8.
14. Blaas HG, Eriksson Ag, Salvesen KA, et al. Brains and faces in holoprosencephaly: pre and postnatal description of 30 cases. Utrasound Obstet Gynecol. 2002;19(1):24-38.
15. Hahn JS, Barnes PD. Neuroimaging in holoprosencephaly: refining the spectrum of the midline malformation. Am J Med Genet C Semin Med Genet. 2010;154C(1):120-132.
16. Volpe P, Campobasso G, De Robertis V, Rembouskos G. Disorders of promessencephalic development. Prenat Diagn.2009;29(4):340-354.
17. Engels AC, Joyeux L, Brantner C et al. sonographic detection of central nervous system defects in the first trimester of pregnancy. Prenat Diagn.2016;36(3):266-273.