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Original Research Paper

Radio-Diagnosis

A WALK THROUGH THE FETUS: A PICTORIAL REVIEW OF CONGENITAL ANOMALIES BY ULTRASONOGRAPHY

Dr. Vinay. M	Assistant Professor, Department of Radiodiagnosis, SIMS&RC/ RGUHS, India
Dr. Sumanth Gowda R*	Junior Resident, Department of Radiodiagnosis, SIMS&RC/RGUHS, India *Corresponding Author
Dr Mallikarjunappa B	Professor and Head of the Department, Department of Radiodiagnosis, SIMS&RC/ RGUHS, India
Dr. Akshay Gupta	Junior Residents, Department of Radiodiagnosis, SIMS&RC/RGUHS, India
Dr. Rakshitha C	Junior Residents, Department of Radiodiagnosis, SIMS&RC/RGUHS, India
Dr. Darshan H S	Junior Residents, Department of Radiodiagnosis, SIMS&RC/RGUHS, India

ABSTRACT In today's obstetrics, the ultrasound is one of the most often utilized diagnostic tools for identifying fetal developmental abnormalities. However, it is crucial to understand which defects may be diagnosed with great certainty during pregnancy and which ones can only be discovered partially or not at all before giving birth. Our study aims to evaluate the role of ultrasound in detecting congenital fetal anomalies and to find out the most common system involved / most common fetal anomaly encountered in our institution. Prenatal diagnostics also requires a great deal of medical professional responsibility because the effects of an unintentionally incorrect diagnosis can have a negative impact on both the fetus and the family.

KEYWORDS : Fetal anomalies, Early pregnancy, NT scan, Nasal bone, soft markers, Obstetric scan

INTRODUCTION

Ultrasonography (US) is performed during early pregnancy between 6 – 10 weeks for dating, Fetal heart rate assessment (FHR), determination of the number of fetuses, and for assessment of early complications.

The next diagnostic ultrasound screening is often performed between 11 and 13 weeks of pregnancy when the thickness of the nuchal translucency is assessed and the presence of the nasal bone is confirmed. In the first trimester, nuchal translucency (NT) is an excess of fluid under the nuchal skin of the fetus.

The next ultrasound screening is normally performed between 18 and 20 weeks of pregnancy. The purpose of this screening is to discover congenital deformities as well as other symptoms of chromosomal abnormalities and other syndromes. Therefore, this ultrasound examination is called a "TIFFA scan" (Targeted Imaging for Fetal Anomalies).

Heart abnormalities, for example, are frequently simpler to detect later in pregnancy as the organ develops and grows. It is also easier to discover anomalies when the problem develops with gestational age (for example, pyelectasis).

Aims and Objectives

- To evaluate the role of ultrasound in detecting congenital Fetal anomalies.
- To find out the most common system involved / most common fetal anomaly encountered in our institution.

METHODS AND MATERIALS

The study was carried out in Radiodiagnosis Department of Sapthagiri institute of medical science and research centre, Bangalore. Study was given ethical clearance from the ethical committee of the above-mentioned institution.

Conventional, B mode ultrasonography images stored in the Medsynapse PACS (Picture archiving and communication system) system were reviewed, data from 25 cases with fetal abnormalities were retrospectively reviewed, and data were retrieved. All images presented in this article were obtained using GE Voluson S8.

RESULTS

In our review of 25 cases, who were referred for Obstetric scans to the department of radiology, for the evaluation of the fetal congenital anomalies.

Out of 25 cases, 28 % of the cases had anomalies of the central nervous system, about 12 % involved the urogenital tract, 16 % had anomalies involving the extremities, and about 16 % constituted cases having an isolated soft marker.

System	No of cases
Central nervous system	7
Face	3
Cardiovascular system	2
Respiratory system	1
Urogenital tract	3
Extremities	4
Isolated soft markers	4
Lymphatic malformation	1

SYSTEM:

1. Central Nervous System:

a) Arnold Chiari Malformation: They are a group of disorders associated with congenital inferior displacement of the cerebellum and brainstem in the posterior fossa. Classic Chiari malformations come in three different forms. Without affecting the fourth ventricle or medulla, the cerebellar tonsils are simply displaced downhill in a Chiari I malformation. Caudal displacement of the medulla, fourth ventricle, and cerebellar vermis through the foramen magnum with lumbosacral myelomeningocele is Chiari II malformations, Chiari III is similar to Chiari II but with a high cervical/occipital encephalomeningocele [1].

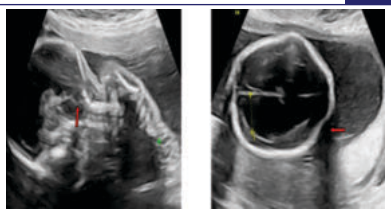

Figure 1a: Herniation Of Cerebellar Tonsil(arrow). Figure 1b: Dilated Lateral Ventricle In The Brain – Hydrocephalus. Frontal Bone Indentation (arrow) Bilaterally Gives The Appearance Of A Lemon Sign.

b) Lumbosacral Myelomeningocele: Spina bifida cystica, or myelomeningocele, is a complicated congenital spinal defect that causes spinal cord deformity. Results from the dorsal neural tube failing to fuse during development. There is a persistent neural placode and a localized failure in the closure of the caudal neuropore. With regard to 80%–98% of instances, the lower lumbar and higher sacral regions are the most often affected portions. In the upper thoracic and cervical spine, myelomeningocele is uncommon [2].

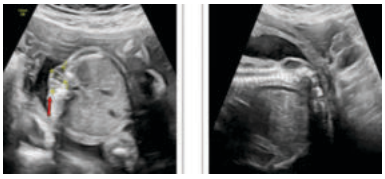

Figure 2 a & b: Defect In The Posterior Aspect Of The Spine At The Lumbosacral Level With Herniation Of Echogenic Cystic Structures (arrow) - S/o Spina Bifida With Lumbosacral Myelomeningocele.

c) Sub Occipital Encephalocele: Encephaloceles are congenital anomalies characterized by a defect in the skull and dura with the extracranial dissemination of intracranial structures. Occipital encephalocele is the most common form and is manifested as a swelling of different sizes over the occipital bone. Occipital encephaloceles are more common in females than in males. The incidence is between 1 in 3000 to 1 in 10,000 live births [3].

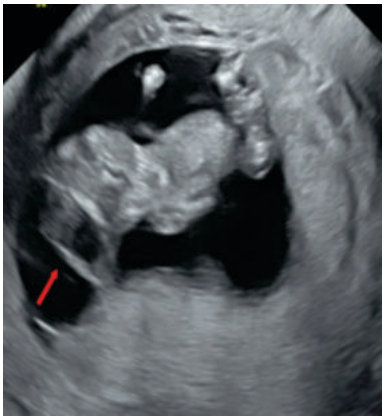

Figure 3: Defect In The Suboccipital Region (arrow) With A Cystic Protrusion Of Meninges And Cerebral Parenchyma Through The Defect - S/o Suboccipital Encephalocele.

d) Corpus Callosum Agenesis: The corpus callosum, which connects the two hemispheres of the brain, is the largest white matter structure and has 200 million axons. Agenesis of the corpus callosum is one of the most common congenital cerebral malformations [4]. Development of the corpus callosum occurs between 8 and 20 weeks of gestation. Sonographic Findings include Widely spaced lateral ventricles, extremely narrow frontal horns (slit-like), Colpocephaly, elevated third ventricle extending between lateral ventricles, and absent cavum septum pellucidum[5].

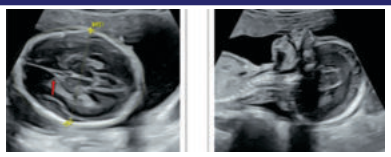

Figure 4 A & B: Non-visualisation Of Cavum Septum Pellucidum (arrow) – Corpus Callosum Agenesis.

e) Acrania Exencephaly Complex: Exencephaly is a congenital fetal brain developmental anomaly considered to be a precursor to anencephaly in the acrania-exencephaly-anencephaly sequence. Acrania is characterized by a partial or complete absence of the cranium[6].

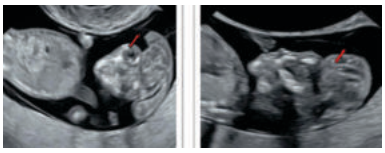

Figure 5 A & B: Cranial Vault Is Not Visualized (arrows) With Freely Floating Brain Parenchyma In The Amniotic Fluid

f) Rachischisis with Hydranencephaly and Iniencephaly: Iniencephaly is a fatal neural tube defect characterized by occipital bone defects at the foramen magnum, spinal dysmorphism, fixed retroflection of head and lordosis of cervicothoracic vertebrae [7]. Hydranencephaly is a rare fetal condition in which the cerebral hemispheres are fully or nearly completely absent, resulting in a membrane sac filled with cerebrospinal fluid, glial tissue, and ependyma.

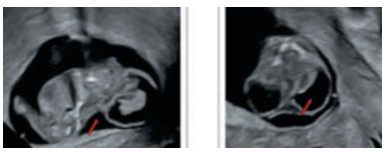

Figure 6 A: Exaggerated Lordosis Of The Cervical Spine (arrow) With Resultant Short Neck And Fixed Hyper-extended Head – Iniencephaly. Figure 6 B: Fluid-filled Cranial Cavity With Non-visualization Of The Cerebral Parenchyma (arrow), However, Bilateral Thalami And Posterior Fossa Contents Are Visualized - Hydra-nencephaly.

g) Dandy Walker Malformation: Classically Dandy-Walker malformation consists of the triad of hypoplasia of the vermis and cephalad rotation of the vermian remnant; cystic dilatation of the fourth ventricle extending posteriorly; enlarged posterior fossa with circular-lambdoid inversion[8].

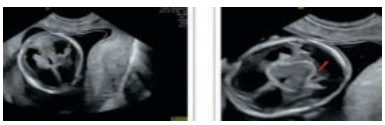

Figure 7 A & B: Dilated Bilateral Lateral Ventricles. Hypoplastic Cerebellum With Prominent Cisterna Magna (arrow) - S/o Dandy Walker Malformation.

2) Cardiovascular System:

a) Hypoplastic Left Heart Syndrome: The term "HLHS" refers to a group of cardiac abnormalities that include hypoplasia of the left ventricle, ascending aorta, and hypoplasia or atresia of the aortic and mitral valves. The following are the most significant sonographic findings: a tiny, thick-walled, hyperechoic left ventricle with poor contractility; absent or minute left ventricle and an anterior mitral leaflet, of 5 mm or less; enlarged right ventricle with an increased excursion of the tricuspid valve, absence of antegrade flow through the aortic valve, and poor motion of aortic valve; ascending aorta hypoplasia of variable degree, which is a small or not visible, and small amount of flow through it[9].

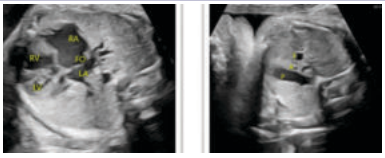

Figure 8 A: Small La And Lv In 4 Chamber View; Figure 8 B: Dilated Pulmonary Artery And Small Proximal Aorta In 3 Vessel View; S- Svc; A – Aorta; P – Pulmonary Artery (from Fetal Right To Left).

3) Respiratory System:

a) Pulmonary sequestration: Pulmonary sequestration is characterized by a non-functioning mass of lung tissue that either shares the normal lung's pleural envelope (intra lobar) or has its own pleura (extra lobar), lacks normal communication with the tracheobronchial tree, and receives arterial supply from one or more systemic vessels [10]. Typically, the sequestered section of the lung is more echogenic than the rest of the lung. A feeder vessel from the aorta can be identified using a colour Doppler.

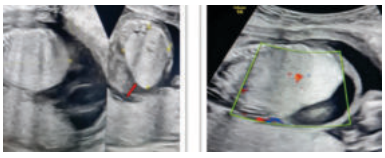

Figure 9 A & B: Well Defined Hyperechoic Lesion Is Seen Occupying The Right Hemithorax With An Arterial Supply From Systemic Circulation – S/o Sequestration.

4) Face:

a) Cleft lip and palate: One of the most frequent neonatal facial defects is cleft lip and palate. In 80% of situations, the two coexist. The lip develops between the fourth and seventh weeks of pregnancy. Between the sixth and ninth weeks of pregnancy, the palate (the roof of the mouth) develops. The condition develops between the fourth to sixth weeks of pregnancy when one or both medial nasal prominences fail to fuse [11].

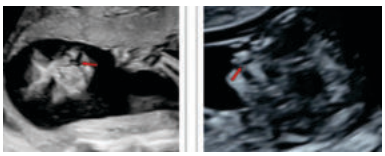

Figure 10 A: Median Cleft Lip Seen Involving The Upper Lip Extending To Right Para Median Location With Involvement Of The Palate (arrow) – Cleft Upper Lip. Figure 10 B: Hypoechoic Region Through The Fetal Palate (arrow) – Cleft Palate.

5) Lymphatic System

a) Cystic Hygroma: Cystic hygroma is a type of lymphangioma, which is a vascular anomaly caused by fluid accumulation in the cervicofacial and axillary regions. Cystic hygroma is most often located in the neck (75%), followed by the axilla (20%), retroperitoneum and intra-abdominal organs (2%), limbs and bones (2%), and mediastinum (1%) [12]. They may appear as a nuchal cyst on prenatal ultrasonography and may include septations +/- signs of fetal anasarca/ hydrops fetalis.

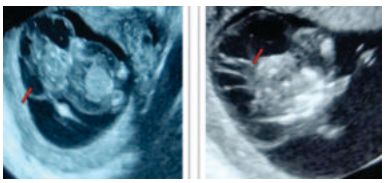

Figure 11 A & B: Well-defined Cystic Lesion With Internal Septation (arrows) Is Seen In The Posterior Cervical Space – Suggestive Of Cystic Hygroma/lymphangioma.

6) Urogenital Tract:

a) Multicystic dysplastic kidney: Multicystic dysplastic kidney (MCDK) is a kind of renal dysplasia characterized by multiple noncommunicating cysts separated by dysplastic parenchyma. The criteria for MCDK were proposed to include echogenic renal parenchyma, several noncommunicating cysts of varying sizes at the kidney's periphery, and no ultrasonic evidence of obstructive nephropathy [13].

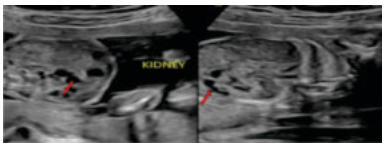

Figure 12 A & B: Transverse And Sagittal Section Showing Multiple Small Cysts (arrows) In A Slightly Enlarged Right Kidney

7) Extremities:

a) Club Foot: It is considered the most common anomaly affecting the feet diagnosed on antenatal ultrasound. The sonographic features may differ depending on the severity. The tibia and fibula may be in the same view as the medially deviated foot, and the foot may appear plantar flexed as well [14].

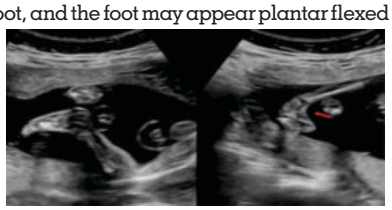

Figure 13 A & B: Right Foot Is Normal. The Left Foot Is Medially Deviated (arrow) And Is Seen In The Same Plane As The Tibia And Fibula. The Sole Of The Foot Is Not Perpendicular To The Lower Leg.

8) Isolated Soft Markers

a) Choroid Plexus Cyst: Antenatal choroid plexus cysts are normally benign and transitory, originating in utero from neuroepithelial infolding. They are frequently discovered in the second trimester as sonolucent cysts, particularly around the lateral ventricles. They usually disappear by 26-28 weeks in utero and are of limited concern in the majority of cases [15].

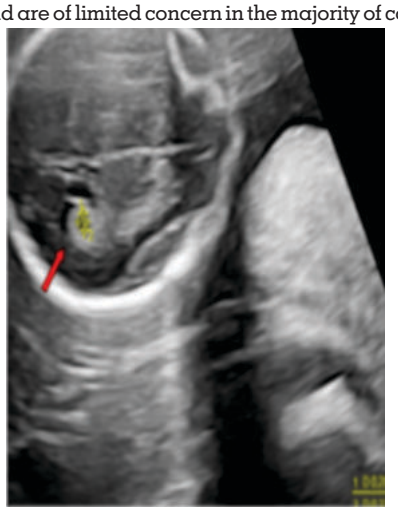

Figure 14: A Cyst (arrow) Measuring 2.8 X 2.1 Mm Is Noted In The Left Choroid Plexus.

b) Intracardiac Echogenic Focus: It is a relatively common sonographic finding. On a four-chamber image, they are often observed as small bright echoic foci within the fetal heart. The vast majority of echogenic intracardiac foci are unilateral in nature. In terms of location, the left ventricle is the most common of the heart chambers [16]. There is no recognized direct link between an EIF and congenital cardiac disease [17].

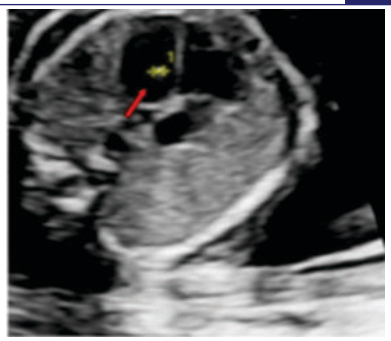

Figure 15: Intracardiac Echogenic Focus (arrow) In Left Ventricle.

c) Increased NT translucency: Nuchal translucency (NT) is the typical fluid-filled subcutaneous area between the back of the fetal skin and the overlying skin. Between 11- 14 weeks gestation, NT is measured using ultrasonographic imaging. Increased NT has been associated to various fetal chromosomal and non-chromosomal disorders [18]. Increased NT is considered when it measures >95th percentile for a given crown rump length [19].

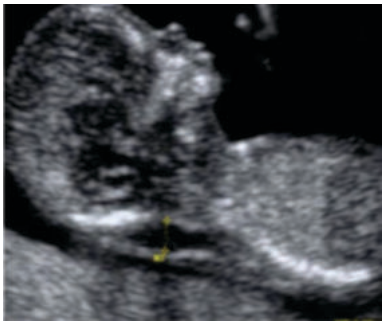

Figure 16: Increased NT Translucency Measuring 3.9 Mm.

d) Fetal Renal Pyelectasis: Fetal pyelectasis is assessed as an AP measurement of the renal pelvis on an axial plane ultrasound image. According to the Society of Fetal Urology (SFU) consensus, fetal pyelectasis is considered present if the AP diameter of the renal pelvis measures:>4 mm up to 28 weeks or >7 mm at or after 28 weeks gestation [20].

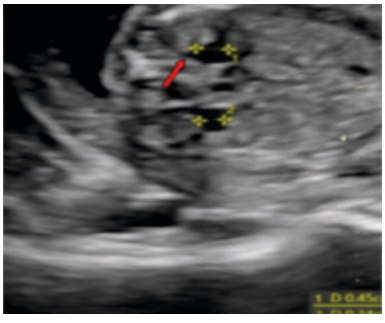

Figure 17: Right Sided Fetal Renal Pyelectasis (arrow) In A Fetus Of 24 Weeks Of Gestation

CONCLUSION

Ultrasound is a useful tool for screening for fetal congenital abnormalities.

Understanding the natural history of fetal malformations is crucial for accurate diagnosis. For example, the diagnosis of hydrocephalus in the first trimester is very unlikely as a result of the physiologically large proportions of the lateral ventricles to the calvarium. Similarly, indirect signs of spina bifida such as the effaced cisterna magna, lemon sign and small cerebellum have been described as useful markers in the evaluation of spinal defects.

However, because these cranial signs are not visible in the first trimester, the sensitivity of detection of spina bifida with early ultrasonography is unlikely to be as high as in the second trimester.

Furthermore, a normal appearance of cardiac anatomy at any point of pregnancy does not preclude heart abnormalities that may develop with advancing gestational age and can be discovered later in pregnancy or even after birth.

Detailed examination of fetal anatomy during the routine 11–14 weeks of gestation scan can provide a comprehensive assessment of fetal anatomy and can detect approximately half of major structural defects in both low- and high-risk pregnancies.

First-, second-, and third-trimester ultrasounds all provide information on potential fetal abnormalities; because of the late development of some organ systems and the delayed onset of a significant number of major anomalies in the second and third trimester, standard second-trimester anatomy ultrasound is the most accurate at finding structural abnormalities, while third-trimester ultrasound provides information on fetal growth. A normal ultrasound does not rule out fetal genetic abnormality. For the definitive diagnosis of genetic disorders, ultrasound does not substitute invasive testing.

Declaration of Patient Consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of Interest: There are no conflicts of interest.

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