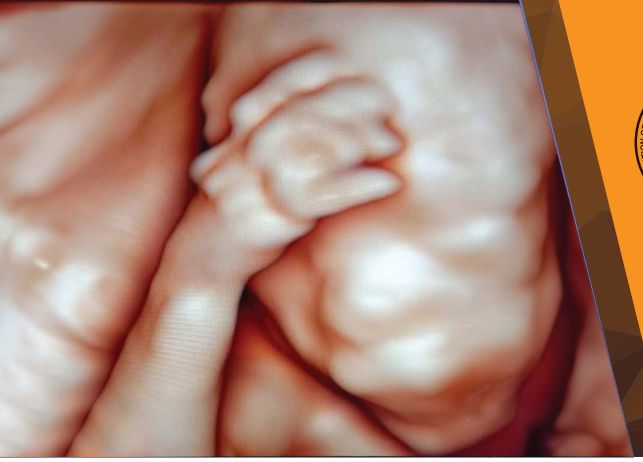


Vol.1 • Issue.2 • June 2023

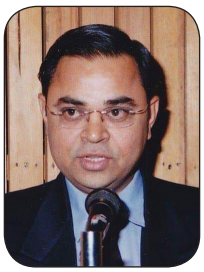
e-Bulletin

FOGSI Genetics & Fetal Medicine Committee 2023-25

First Trimester Anomalies

Dr Hrishikesh Pai
President, FOGSI

Dr Madhuri Patel
Secretary General, FOGSI



Dr Geetendra Sharma
Vice President FOGSI

Dr Seetha Ramamurthy Pal, Chairperson GFMC

Contents

Foreword

The Genetics and Fetal Medicine Committee is priveleged to share with you the second edition of our newsletter.

Traditional anomaly scan also known as level II or Targeted Imaging For Fetal Anomalies (TIFFA) scan has been performed in second trimester of pregnancy at 18–22 weeks of gestation. However, due to improvement in technology, many structural anomalies can now be detected in the first trimester. Early detection of fetal anomalies in antenatal period allows for better perinatal management and provides couples with options for decision making.

Many recent studies support the utility of sonographic detection of major fetal structural abnormalities in the first trimester of pregnancy. However, it should be kept in mind that some anomalies will not be evident until later in the pregnancy due to ongoing fetal anatomical development. Detection rates of anomalies vary according to the organ system being examined, equipment settings, and radiologist experience.

The first trimester provides a great window of opportunity to assess the fetus and mother for a myriad of conditions. With the inverted pyramid of antenatal care, the first trimester forms a strong base for this. With improved machines and operator skill, the focus of detecting anomalies has gradually shifted to the first trimester. More and more anomalies are being detected in the first trimester and those suspected can be reconfirmed in the early second trimester. Following guidelines laid by International bodies like ISUOG can help improve the same.

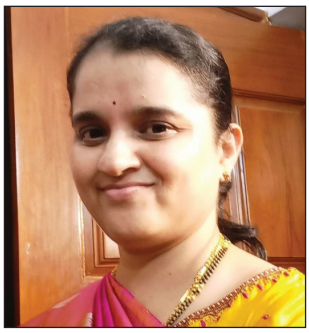
This edition deals with some common and some uncommon anomalies detected in the first trimester and their approach to the same. I am extremely thankful to all the contributors for sharing their cases and a special thanks to Dr Sreelakshmi for coordinating and putting this all together.

There is a crossword again at the end to tickle your brains.

Happy reading folks!

Dr. Seetha Ramamurthy Pal, *Chairperson,*
Genetic & Fetal Medicine Committee, FOGSI

FROM THE EDITOR'S DESK



Sreelakshmi KN
MS, DNB, FICOG, MRCOG (UK)
Fellow in Medical Genetics Consultant Fetal Medicine
Malnad Hitech Diagnostic Centre

Myself Dr Sreelakshmi K N, take immense pleasure in writing this message as the editor for this newsletter. My heartfelt thanks to Dr Seetha Ramamurthy Pal for this opportunity and guidance. I truly appreciate her commitment and valuable words of wisdom.

It is well known fact that with more knowledge, experience, data and high resolution ultrasound, it is possible to detect many structural abnormalities at 11-14 weeks scan. We, in this newsletter, have made a sincere attempt to present some of the fe common and uncommon fetal abnormalities and also the approach to such abnormalities. It is important to understand that ultrasound findings in the first trimester are different from the second trimester and many a times patients have to be recalled at 16 week to confirm the diagnosis.

Table showing detectable anomalies of different systems is helpful, and useful to counsel patients.

Here I would like to highlight the importance of fetal evaluation after termination of pregnancy. It is essential to offer all patients, detailed counseling, complete work up including fetal autopsy, storage of DNA and appropriate genetic testing. We have tried in all cases to report appropriate fetal evaluation after termination. In many cases this will enable us to offer appropriate prenatal diagnosis and also establish definite diagnosis.

With this, I thank again

Long live FOGSI

Sreelakshmi KN

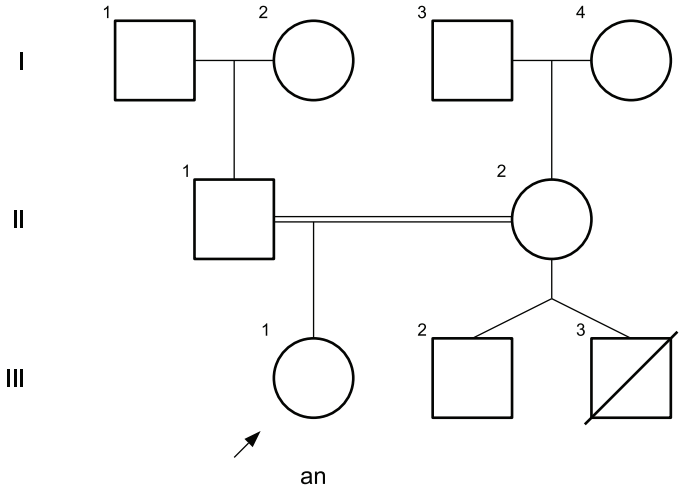
FIRST TRIMESTER DIAGNOSIS OF SKELETAL DYSPLASIA

Sreelakshmi KN
MS, DNB, FICOG, MRCOG (UK)
Fellow in Medical Genetics, Malnad Hitech Diagnostic Centre, Shimoga, Karnataka

Introduction:

Skeletal dysplasias are a heterogeneous group of diseases of genetic etiology affecting bone and cartilage. They show diversity in their form of inheritance, natural history, treatment and prognosis. More than 800 types are reported so far. They are different both in phenotype and genotype. Many are diagnosed after birth, many by 20 weeks of gestation, but with more experience, knowledge, data and good resolution ultrasound machines it is possible to detect lethal skeletal dysplasia in 11-14 weeks scan. We present a rare case of lethal skeletal dysplasia in the first trimester and also discuss approach to diagnosis. It is important to know presentation is different in second trimester from first trimester.

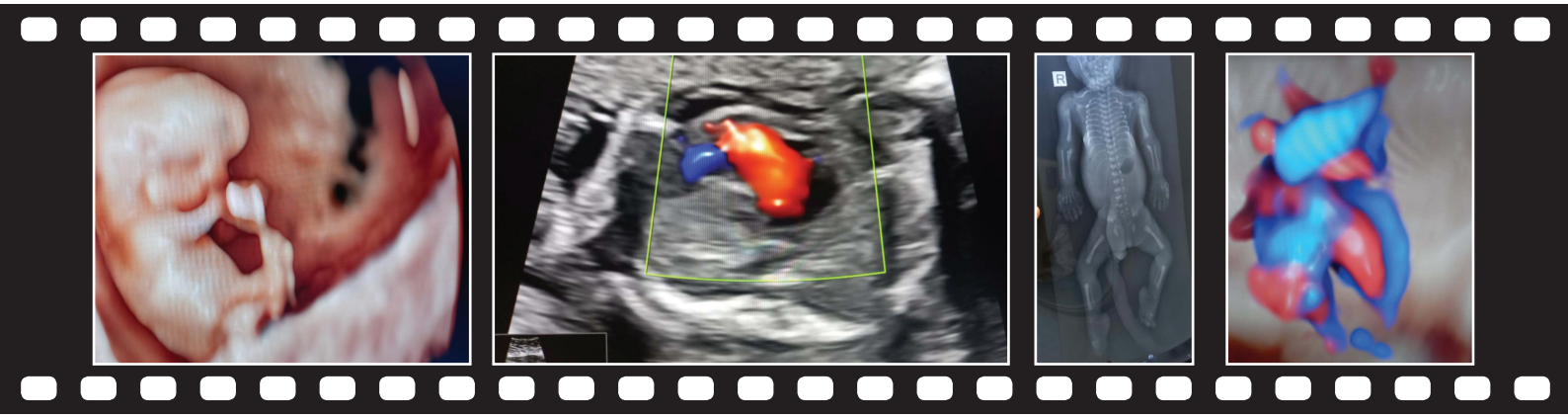
Case Report:



- Consanguineous marriage
- Past history of twin pregnancy with neonatal death of one of the twins with similar affected and was suspected to have lethal skeletal dysplasia
- At ultrasound: short long bones, abnormal nuchal translucency, reversal of flow in ductus venosus, and absent mineralization of spine
- Fetal autopsy showed abnormal nuchal translucency, absent nasal bone, short thorax
- Radiography of the expired fetus showed short long bones
- Absent mineralization of spine, and short thorax and was suspected to have achondroplasia or lethal skeletal dysplasia
- **Whole exome sequencing revelaed-likely pathogenic: c.836-1G>A (3' splice site) Desbuquois dysplasia-1 (OMIM#251450); Multiple epiphyseal dysplasia-7 (OMIM# 617719)**

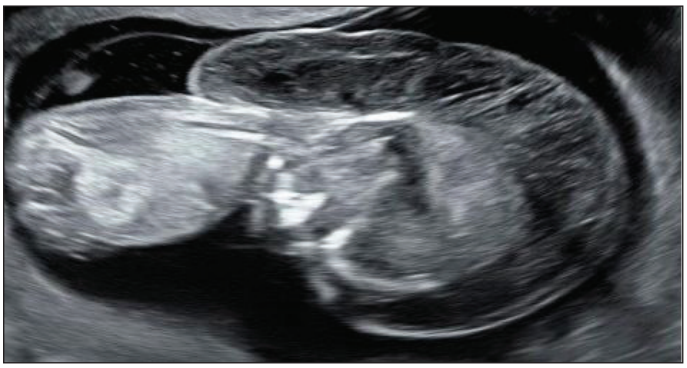
Desbuquois dysplasia-1 (OMIM#251450) and multiple epiphyseal dysplasia-7 (OMIM#617719)

- Homozygous or compound heterozygous mutations in the CANT1 gene (OMIM*613165)
- Desbuquois dysplasia-1 is characterized by severe prenatal and postnatal growth retardation (stature less than -5 SD), joint laxity, short extremities, and progressive scoliosis
- Main radiologic features are short long bones with metaphyseal splay, a 'Swedish key' appearance of



the proximal femur (exaggerated trochanter), and advanced carpal and tarsal bone age with a delta phalanx

- Multiple epiphyseal dysplasia-7 is characterized by mild short stature, anterior wedging of vertebral bodies, minimal platyspondyly, mild scoliosis, flat acetabulae, hypoplastic capital femoral epiphyses, irregularly shaped capital femoral epiphyses, short femoral neck, mild 'Swedish key' appearance of proximal femur, small epiphyses at knees, metaphyseal flare at knees, medial condylar epiphyseal dysplasia, genu varum, and advanced carpal bone age



A similar case of lethal skeletal dysplasia was picked up by 18 weeks anomaly scan. At retrospection, femur length was <1st centile in the first trimester and was a clue which was possibly missed. ***A case of absent nasal bone was referred for amniocentesis: at detail scan, large head and short long bones were documented suspecting thanatophoric dysplasia. Fetal DNA is stored as well.***

Discussion:

Huge thickened NT; with absent Nasal bone was present in all cases of achondrogenesis. It is to be noted that ultrasound feature of skeletal dysplasia in first trimester is different from second trimester.

Konstantinidou1 et al. found that the diagnosis of SD is possible in the first trimester with lethal abnormalities having the highest detection rate, although the majority of diagnoses is usually made in the second or third trimester. Ultrasound suspicion and diagnosis of SD is possible in the first trimester based on biometric criteria (short long bones) and morphological abnormalities of long bones, such as multiple fractures, bowed femur, abnormal hands and feet. Measurements of the femur must be included in the first-trimester ultrasound in addition to nuchal translucency, and these results should be reported in percentiles.

Increased NT in the first trimester scan also has a high association with dysplasia but when within normal limits does not prove effective in excluding it.

Whenever a skeletal dysplasia is suspected in the first trimester, a follow up Ultrasound should be done at around 16-17 weeks as the abnormalities become more obvious then.

Summary of ultrasound features of skeletal dysplasia in first trimester

Thickened Nuchal Transluscency
Short Femur
Abnormal Ductus Venosus flow
Short Arms, Legs, hands and feet
Reduced ossification of skull and spine.

Reference:

1. Konstantinidou AE, Agrogiannis G, Sifakis S, Karantanas A, Harakoglou V, et al. (2009) Genetic skeletal disorders of the fetus and infant: pathologic and molecular findings in a series of 41 cases. Birth Defects Res A Clin Mol Teratol 85: 811-821.
2. Khalil A, Pajkrt E, Chitty LS. Early prenatal diagnosis of skeletal anomalies. Prenat Diagn 2011; 31: 115–124
3. Ngo C, Viot G, Aubry MC, Tsatsaris V, Grange G, Cabrol D, Pannier E. First-trimester ultrasound diagnosis of skeletal dysplasia associated with increased nuchal translucency thickness. Ultrasound Obstet Gynecol. 2007;30(2):221–6
4. Kashyap N, Pradhan M, Singh N, et al. Early detection of fetal malformation, a long distance yet to cover! present status and potential of first trimester ultrasonography in detection of fetal congenital malformation in a developing country: experience at a Tertiary Care Centre in India. J Pregnancy 2015;2015:1–9. DOI: 10.1155/2015/623059

FIRST TRIMESTER DIAGNOSIS OF LIMB ANOMALIES: TRANSVERSE LIMB DEFECT

Charitha U
MS (OBG), Fellow in Maternal & Fetal Medicine

Introduction:

Overall incidence of congenital limb reduction deformities ranges from 0.49 to 3.5 per 10,000 births. ***Reduction deformity is the congenital absence or shortening of a limb or a part of limb.*** Lower limbs are affected less often than upper limbs. The commonest limb reduction defect is terminal transverse defect and this is isolated and sporadic in occurrence and unilateral. We describe isolated transverse limb defect, identified at NT scan done.

Case Report:

A 26-year-old primigravida, who had conceived after ovulation induction with clomiphene citrate for 5 days, was referred for NT scan. The fetal activity and liquor were normal and the CRL was corresponded to 12 weeks and 6 days. Left upper limb appeared abnormal and on close up observation, distal to the elbow, forearm and arm were absent. There was no amniotic band. Right upper limb was imaged and confirmed to be normal in proximal, mid and distal segments. Lower limbs were also normal in structure and movements. There were no other associated anomalies for the present gestational age. They deferred further testing.

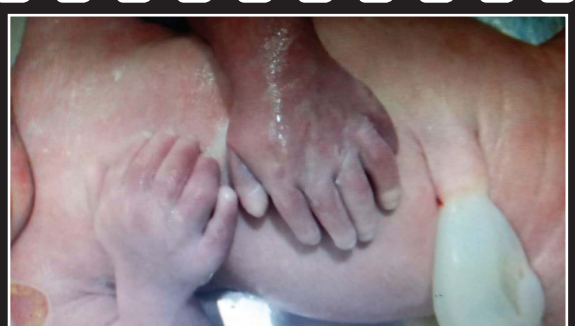
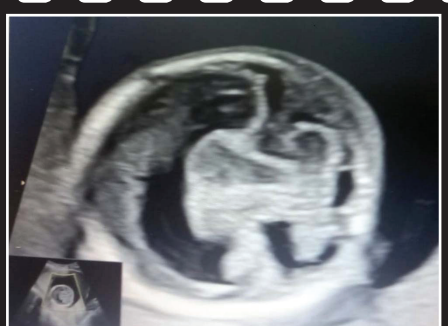
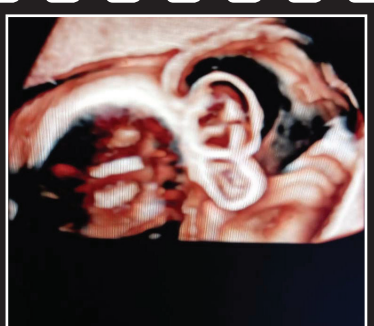
Couple was counseled. Problems with isolated limb defect and probable solutions were explained. Option of upper limb prosthesis was explained, and was reassured about other limbs being normal. However, couple opted for termination of pregnancy. Fetus showed transverse limb defect with forearm and absent hand. There were no other associated anomalies. Couple were counseled that transverse limb defect is sporadic and recurrence rates are low. They were advised NT scan in all subsequent pregnancies.

Approach to Limb Anomalies

- Both upper and lower limbs to be imaged in all cases
- All three segments in all four limbs to be imaged
- Imaging fingers and digits is not mandatory but assessment of thumbs may be useful, if possible, to rule out Fanconi's anaemia and Roberts syndrome
- Many a times, TVS is essential to complete survey
- One good flexion and extension movement is a good practice
- Detailed family and drug history
- If isolated, can consider Genetic testing
- If associated anomalies, genetic counselling to rule out genetic syndromes

Discussion:

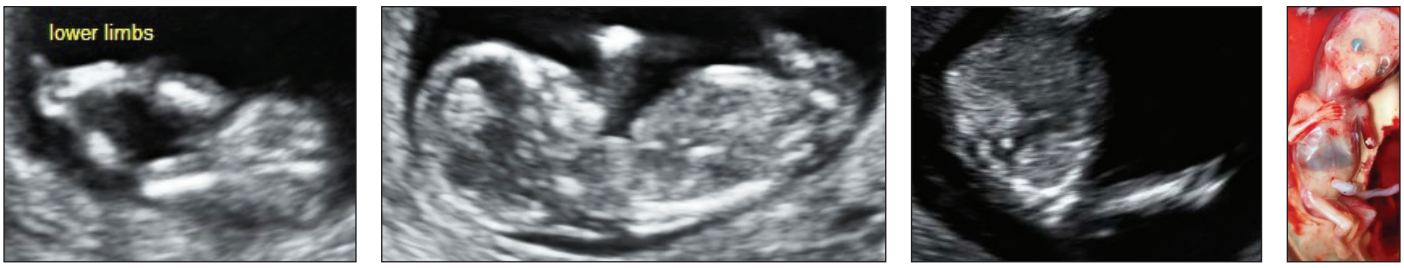
Etiology of limb abnormalities is complex. Single gene disorders, chromosomal abnormalities, intrauterine factors, vascular events, maternal diseases and exposure to certain teratogens cause these. In many, cause is idiopathic. Fetal limb survey is essential at 11-14 weeks of gestation. Systematic approach is helpful. Knowledge of etiology and thus prognosis,

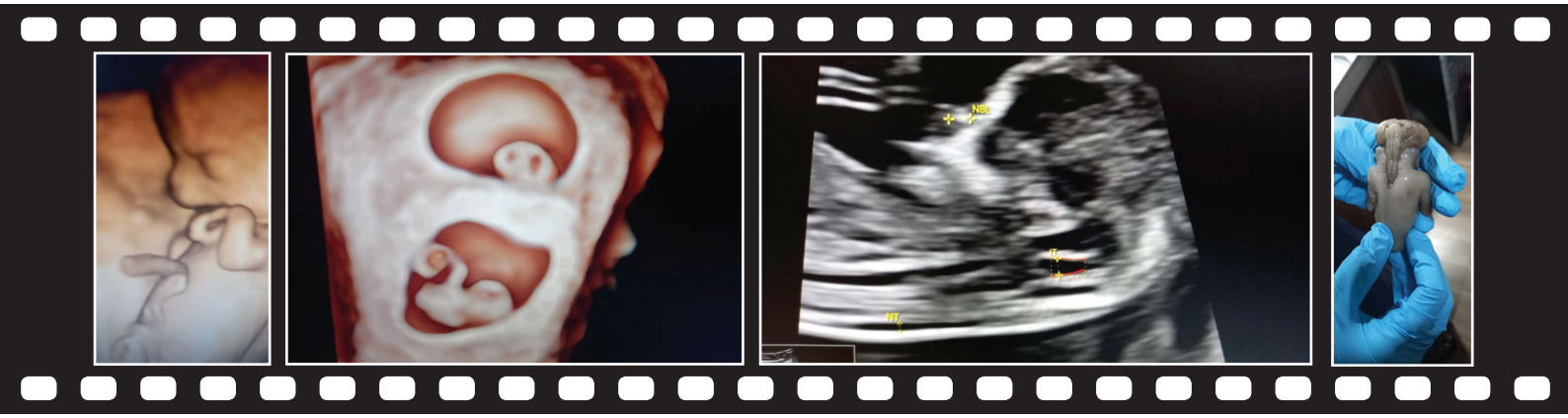
will provide parents prenatal diagnosis for their future pregnancies. Most limb abnormalities diagnosed in the first trimester and reported in the literature were usually associated with other abnormalities like hydrops, cardiac abnormalities and megacystis. Enlarged NT is a common finding again. Fetuses with bilateral forearm defects or those with unilateral lesions and other abnormalities detected prenatally have a high incidence of aneuploidy and genetic syndromes. Isolated, unilateral lesions usually have a good prognosis. Patient's body habitus, quality of USG machine and the operator skill have a main role in the detection and diagnosis of limb anomalies using prenatal ultrasonography. A multidisciplinary approach involving obstetrician, sonologist, clinical geneticist, neonatologist / pediatrician and pediatric orthopedic surgeon is beneficial.

Reference:

1. Rice KJ, Ballas J, Lai E, et al. Diagnosis of fetal limb abnormalities before 15 weeks: Cause for concern. Journal of Ultrasound in Medicine. 2011;30(7):1009-1019
2. Stoll C, Wiesel A, Queisser-Luft A, et al. Evaluation of the prenatal diagnosis of limb reduction deficiencies. EUROSCAN Study Group. Prenatal Diagnosis. 2000;20(10):811-818
3. Pajkrt E, Cicero S, Griffin DR, van Maarle MC, Chitty LS. Fetal forearm anomalies: prenatal diagnosis, associations and management strategy. Prenat Diagn. 2012 Nov;32(11):1084-93.
4. Vij N, Goncalves LF, Llanes A, Youn S, Belthur MV. Prenatal radiographic evaluation of congenital transverse limb deficiencies: A scoping review. World J Orthop. 2023 Mar 18;14(3):155-165. doi: 10.5312/wjo.v14.i3.155. eCollection 2023 Mar 18.



Ultrasound Images and Fetus with Limb Defect



11-14 WEEKS SCAN: CNS ANOMALIES: HOLOPROSENCEPHALY WITH CYCLOPIA WITH PROBOSCIS

Roopadevi Hosamani
MBBS DMRD DNB, Consultant Radiologist Shree Scan Centre Haveri

Introduction:

Alobar holoprosencephaly is a rare lethal congenital anomaly occurs as a result of incomplete separation of prosencephalon into two halves of hemispheres during organogenesis leading to failure of cleavage of orbit into two cavities of eyes. Typically, nose is either missing or replaced with a nonfunctioning nose in the form of a proboscis. Such a proboscis that generally appears above the central eye is a characteristic of a form of cyclopia called rhinencephaly or rhinocephaly. While holoprosencephaly affects one in 16,000 live newborns, cyclopia is seen as rarely as one in 100,000 newborns.

Case Report:

Young primigravida, spontaneous conception referred for NT scan.

Ultrasound:

- Incomplete cleavage of the forebrain with fused thalami and single ventricle.
- Gross dilatation of the lateral ventricle.
- Cyclopia- the orbits of the eye are not properly divided into two cavities, they can be seen either as two bilateral fields that are very close to each other
- Proboscis -anterior appendage-like structure is seen projecting from the midline fetal face. Depending on the exact location, it was. Interorbital proboscis.

Couple was counseled regarding lethal nature of the anomaly and possible associated chromosomal abnormalities, deferred further testing and opted for termination of pregnancy.

Discussion:

A holoprosencephaly, cyclopia with proboscis is a rare form of HPE.

Prevalence:

- Holoprosencephaly affects one in 16,000 live newborns
- Cyclopia is seen as rarely as one in 100,000 newborns.

Alobar holoprosencephaly, represents the most severe form and constitutes undifferentiated cerebral hemispheres with monoventricles, a fusion of thalamus, and other cranial midline abnormalities. Most of the cases are sporadic and incompatible with life, and the etiology of this condition remains largely unknown. However, various heterogeneous risk factors have been implicated. The possible culprits include defective inheritance and environmental factors. Maternal exposure to teratogenic drugs (such as salicylates, amidopyrine, corticosteroids, aspirin, lithium, anticonvulsants, retinoic acid, anticancer agents), alcohol, toxoplasmosis, rubella, cytomegalovirus, and herpes simplex (TORCH) infections, ionic radiation, and maternal diabetes are the notable risk factors in previously reported cases.

Associated Abnormalities:

- Chromosomal defects, mainly trisomies 13 or 18, are found in > 50% of cases at 12 weeks' gestation.
- Genetic syndromes are found in 20% of cases.
- Alobar and lobar holoprosencephaly are associated with microcephaly and midfacial defects in 80% of cases. Extracerebral defects are particularly common in fetuses with trisomies 13 and 18 and those with genetic syndromes.
- Alobar and semilobar: usually lethal within the first year of life.

Recurrence:

- Isolated: 6%.
- Part of trisomies: 1%.
- Part of genetic syndromes: 25-50%.

First Trimester evaluation of the Central Nervous system is challenging as the cerebral structures evolve considerably and continuously change in shape and form during pregnancy. However, most of the lethal

anomalies that can be detected in the first trimester are in the Nervous system. According to the ISUOG recommendations for the first-trimester scan, the examination of the fetal head and central nervous system is best achieved using a combination of axial and midsagittal planes. The standard axial and sagittal planes can help in detection of the major cranial and spinal defects. Inclusion of the non axial planes, multiplanar neurosonography can be performed. Additional use of volume imaging can help detect early signs of abnormalities that are normally detected in the second trimester. Recent studies have shown a 72% detection rate of fetal brain anomalies using the various CNS parameters. Findings that raise the suspicion of a cranio-spinal defect in the first trimester are:

Axial Plane:

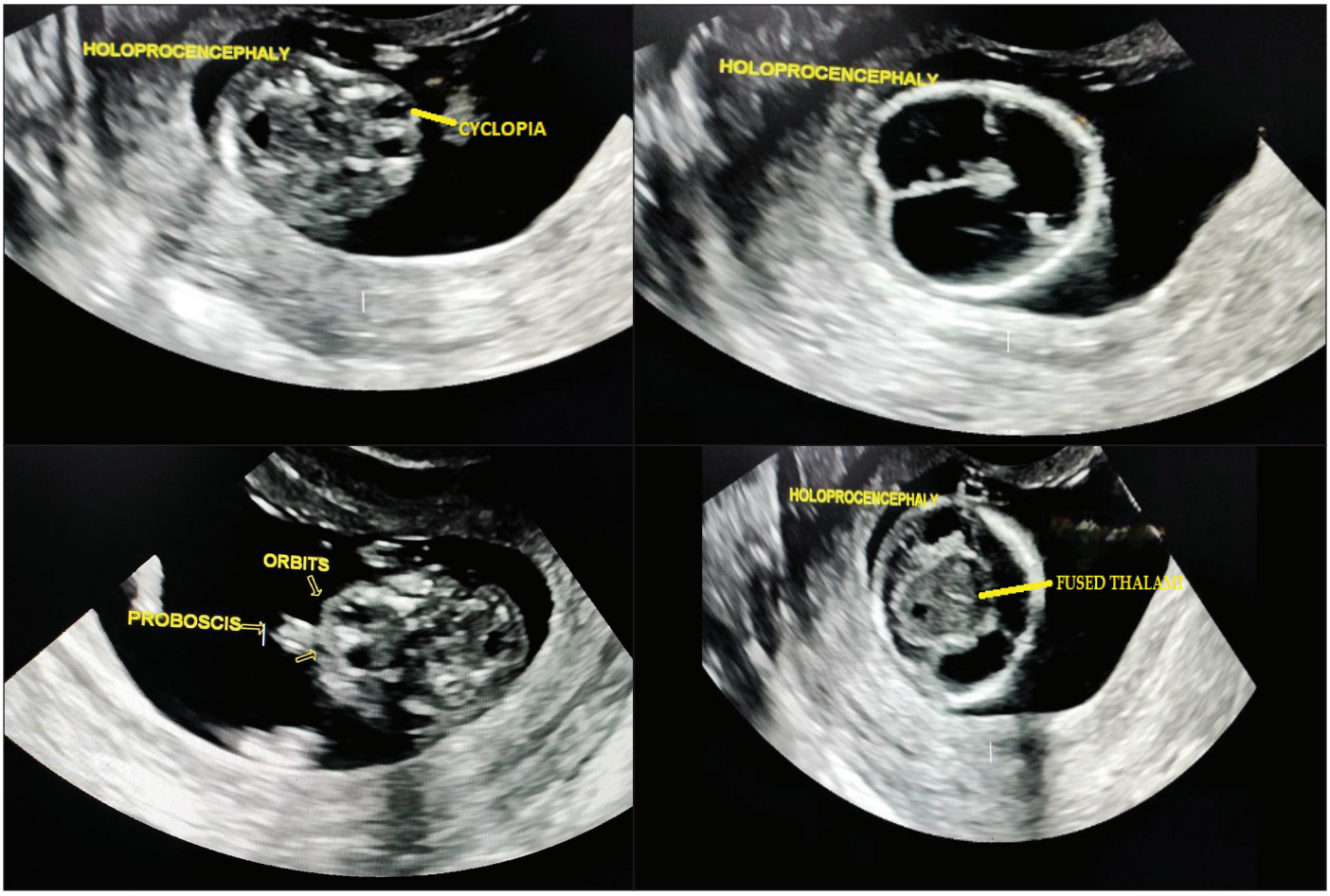
- Small BPD, BPD/AC
- Parallel cerebral peduncles
- Dry Brain
- Aqueduct of Sylvius- Occipital bone distance
- Crash sign

Sagittal Plane:

- Obliterated or Increased IT
- Brain stem/Brain stem- occipital bone ratio (BS/BSOB > 95th centile)

References:

1. Dubourg C, Bendavid C, Pasquier L, Henry C, Odent S, David V. Holoprosencephaly. Orphanet J Rare Dis. 2007;2:8.
2. N. Volpe, A Dall'asta et al. First-trimester fetal neurosonography: technique and diagnostic potential: Ultrasound Obstet Gynecol 2021;57: 204 – 214.
3. Ungureanu DR, Drăgușin RC, Căpitănescu RG, Zorilă L, Ofițeru AMI, Marinaș C, Pătru CL, Comănescu AC, Comănescu MC, Sîrbu OC, Vrabie MS, Dijmărescu LA, Streață I, Burada F, Ioana M, Drăgoescu AN, Iliescu DG. First Trimester Ultrasound Detection of Fetal Central Nervous System Anomalies. Brain Sci. 2023 Jan 9;13(1)



11-14 WEEKS SCAN: GASTROINTESTINAL ANOMALIES

Sharath Kumar
HS DNB, Consultant Radiologist, Malnad Hitech Diagnostic Centre, Shimoga, Karnataka

Introduction:

Timing of prenatal diagnosis for Gastrointestinal anomalies, namely: omphalocele & gastroschisis is possible to almost 100% accuracy by 11-14 weeks of gestation. Advanced and high resolution ultrasound machines have with more knowledge and experience have increased the detection rates. However, physiological omphalocele has to be kept in mind when period of gestation is <10 weeks.

Case Report:

A 35 year old second gravida with long standing infertility had a spontaneous conception. At 11-14 weeks scan, omphalocele and hypoplastic left heart syndrome were diagnosed and amniocentesis was done by 16 weeks. FISH was positive for trisomy 18. Patient opted for termination.

Discussion:

Two most common congenital abdominal wall defects are gastroschisis and omphalocele. Both have reported incidences around 1 in 4,000 live births. Though both are the result of errors during embryologic development of the foetal abdominal wall, main differential on sonography being midline and para-midline defect. Appropriate imaging at umbilical cord insertion, both can be differentiated. As it is well known that, Omphalocele being midline defect with sac, gastroschisis is a para-midline defect with no covering layers.

It is important to differentiate between omphalocele and gastroschisis as prognosis and further management are different in both conditions. Omphalocele is associated with other anomalies and a chromosomal defect in many as compared to gastroschisis which is usually is not.

Embryologically, omphalocele is thought to be the result of a folding defect that occurs as the bowel is returning to the abdominal cavity during normal development. In general, omphaloceles are classified as either small, giant, or ruptured. Postnatal outcomes in infants with omphalocele are predominantly dependent on other concurrent anomalies or comorbidities associated with larger-sized defects.

Concurrent congenital anomalies can be present in up to 40–80% of omphalocele cases and most commonly include chromosomal (15–57%), cardiac (11–23%), genitourinary (6–21%), musculoskeletal (21%), gastrointestinal (7–19%), and neurologic (4–8%) abnormalities. Omphalocele can also be associated with certain genetic syndromes, such as Beckwith-Wiedemann, pentalogy of Cantrell, and cloacal extrophy.

Conclusion:

Comprehensive prenatal workup and early detection as well as any associated anomalies or comorbidities is critically important for continued surveillance, prognostication, counselling, and further management. Genetic counselling and fetal autopsy are essential to know the cause and risk of recurrence in subsequent pregnancies.

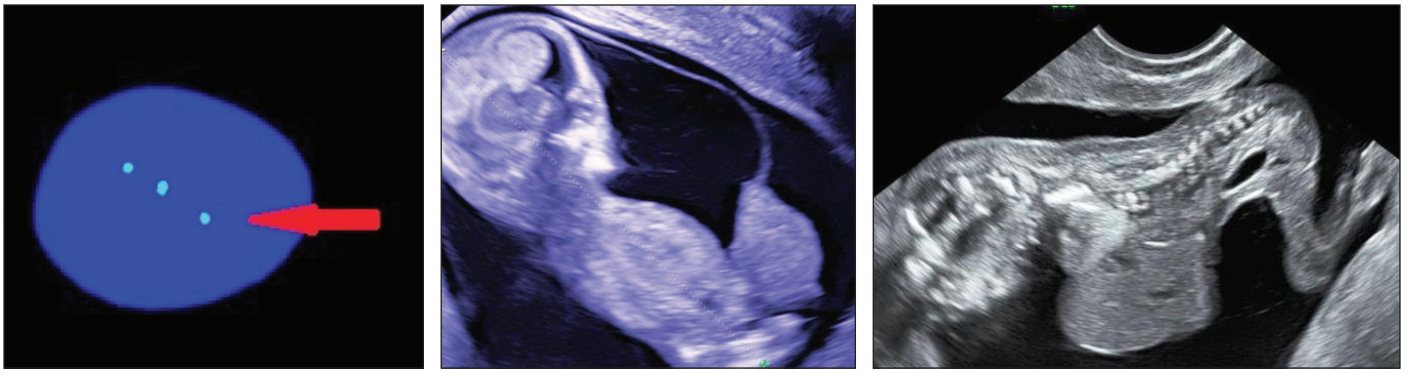
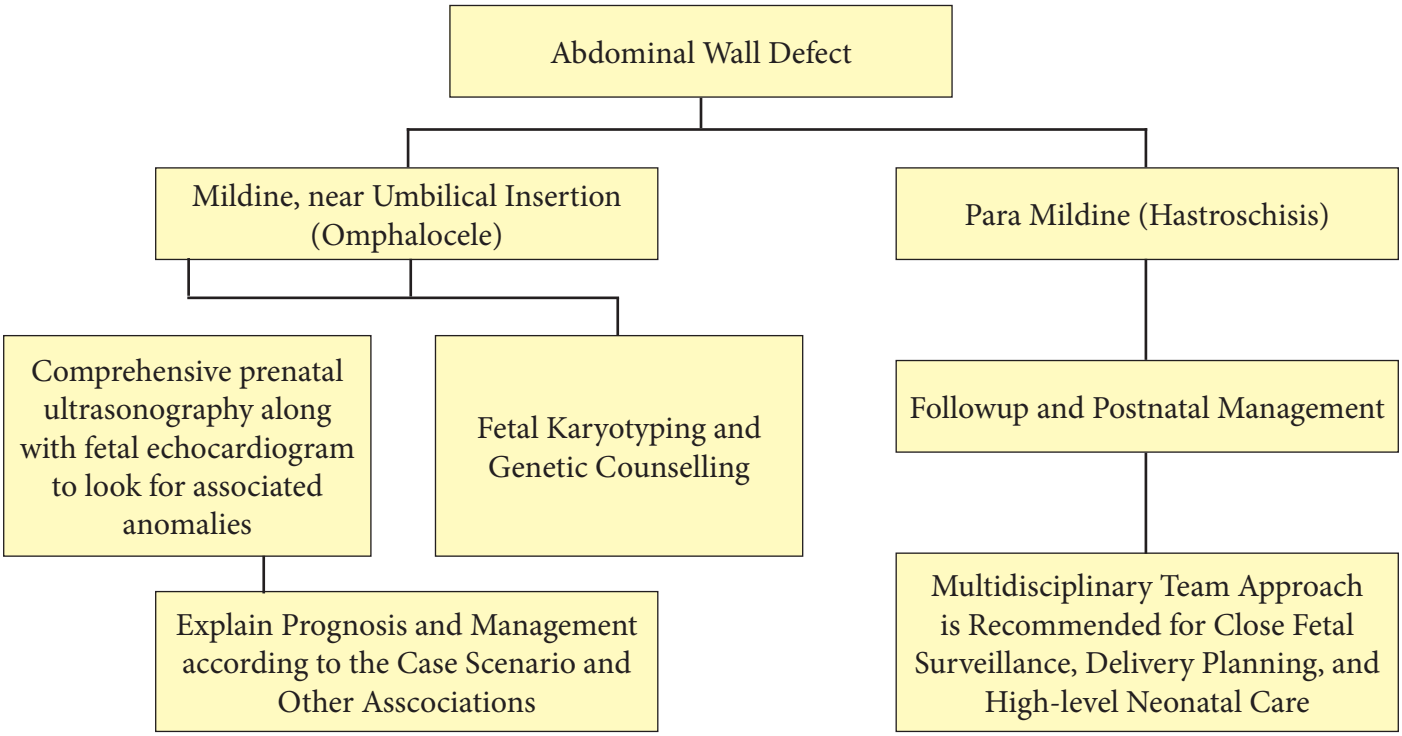


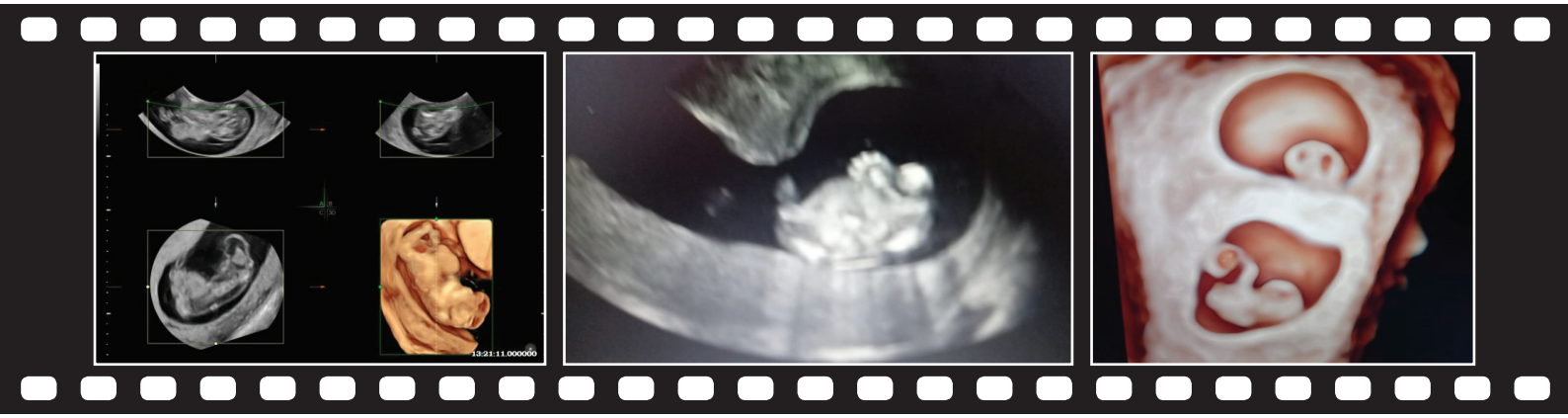
Fig. 1: A FISH Positive for Trisomy 18, b: omphalocele: c: omphalocele with kyphoscoliosis (courtesy: Dr Sreelakshmi KN)

Diagnostic approach on suspected omphalocele in first trimester:



References:

- American Pediatric Surgical Association. Omphalocele. (https://apsapedsurg.org/parents/learn-about-a-condition/f-o/) Accessed 5/24/2021.
- Centers for Disease Control and Prevention. Birth Defects: Omphalocele. (https://www.cdc.gov/ncbddd/birthdefects/omphalocele.html) Accessed 5/24/2021.
- Merck Manual. Abdominal Wall Defects (Omphalocele and Gastroschisis). (https://www.msdmanuals.com/home/children-s-health-issues/birth-defects-of-the-digestive-tract/abdominal-wall-defects-omphalocele-and-gastroschisis) Accessed 5/24/2021.



FIRST TRIMESTER DIAGNOSIS OF LIMB BODY WALL COMPLEX

Shantha Shekar
MD, Consultant Gynecologist, Shantha Nursing Home, Shimoga, Karnataka

Introduction:

Limb body wall complex (LBWC) is rare lethal anomaly. It is incompatible with life. Features are:

- Exencephaly and or/ Encephalocele
- Thoraco and /or Abdominoschisis
- Limb defects

Case Report:

Young primigravida at 13 wks of pregnancy had the following features in ultrasound:

- Huge septated Cystic hygroma with Inencephaly, star gazing appearance of the face, developing exencephaly
- Face showed protruding Eye, no facial clefts
- Absent vertebral column
- Protruding chest and cardia being placed anteriorly near the sternum
- Both upper limbs were seen and only 1 lower limb was seen

The couple opted for termination of pregnancy.

Limb body wall complex:

Sonographic features of LBWC are:

- Neural tube abnormality
- Severe scoliosis
- Positional deformities

Diagnosis is based on any 2 of the 3 previously mentioned features.

LBWC is suspected if a short or absent umbilical cord is seen at the time of your ultrasound, if the baby is in a fixed and awkward position throughout the scan or if there appear to be any abnormalities of the face or abdomen.

Risk factors:

Young maternal age, cocaine abuse, smoking and Alcohol

Sporadic inheritance with normal karyotype

May be caused by any of the following

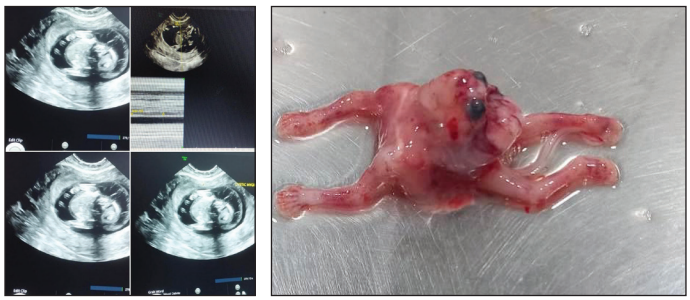


Fig: Ultrasound and fetus images of limb body wall complex

- Germ disc defect with early embryonic maldevelopment
- Primary rupture of amnion leading to formation of amniotic bands
- Vascular disruption
- Disturbances of embryonic folding process

The terminology of BSA has been variably interchanged with LBWC in the literature. When limb abnormalities predominate, a diagnosis of LBWC is made, whereas fetal BSA refers to fetuses in which the abdominal wall defect predominates.[1] They appear to be pathogenetically similar and are likely manifestations of the same spectrum. Both share a normal genotype and are uniformly lethal. LBWC is a lethal fetal anomaly that is not compatible with life; therefore, an early antenatal sonographic diagnosis is extremely important in all pregnant women to counsel them on the options of continuation or termination of pregnancy. With early diagnosis, the parents can be counseled on early termination of pregnancy and be reassured of nonrecurrence of the lesion in the future pregnancies.[2]

References:

1. Murphy A, Platt LD. First-trimester diagnosis of body stalk anomaly using 2- and 3-dimensional sonography. J Ultrasound Med 2011; 30:1739–1743
2. Chen CP, Lin CJ, Chang TY, Hsu CY, Tzen CY, Wang W, et al. Second-trimester diagnosis of limb-body wall complex with literature review of pathogenesis. Genet Couns 2007;18:105-12.

EARLY DIAGNOSIS OF TETRALOGY OF FALLOT AT NT SCAN

Megha Kamalpurkar
Devata Hospital, Kalaburagi, Karnataka

Introduction:

Congenital Heart disease is the most common birth defect and a leading cause of infant mortality, with Tetralogy of Fallot being the commonest. It was described by the French physician Étienne-Louis Arthur Fallot, after whom it is named.

Four parts of Tetralogy of Fallot (TOF) are:

- Pulmonary Stenosis
- Ventricular Septal Defect (VSD)
- Overriding Aorta
- Right Ventricle Hypertrophy

However RVH is not seen prenatally and develops postnatally.

TOF is found in 1in 3600 live births and accounts for 3% to 7 % of infants with CHD, here I present a case of TOF in the First Trimester.

Case Report:

- Second gravida with previous one abortion came for routine NT scan
- Uneventful pregnancy so far
- It was a non-consanguineous marriage
- Visceral and cardiac Situs was normal
- 4 Chamber view: left axis deviation and no other abnormality
- LVOT view showed overriding of aorta
- 3 Vessel View: narrow pulmonary artery, anteriorly placed larger aorta
- 3 VT: narrow ductal arch and Aberrant right Subclavian Artery
- No other extra cardiac abnormality was seen
- NT was Normal

In View of Cardiac abnormality, couple was advised further genetic testing, deferred but opted for termination of pregnancy.

Fetal Autopsy was done and prenatal findings were confirmed.

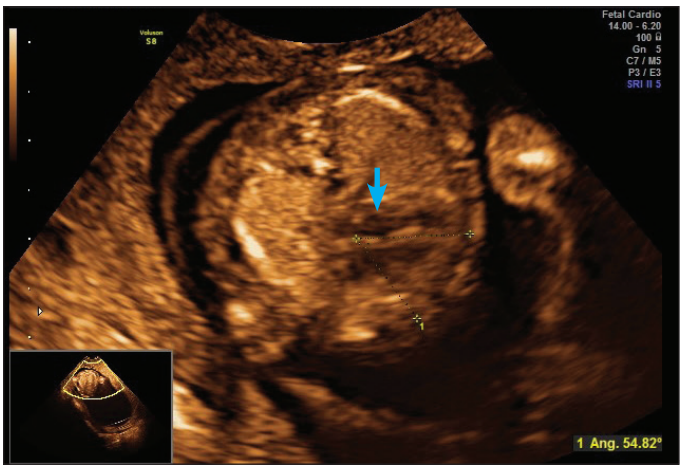


Fig. 1: Shows Lt axis deviation otherwise normal 4 CV

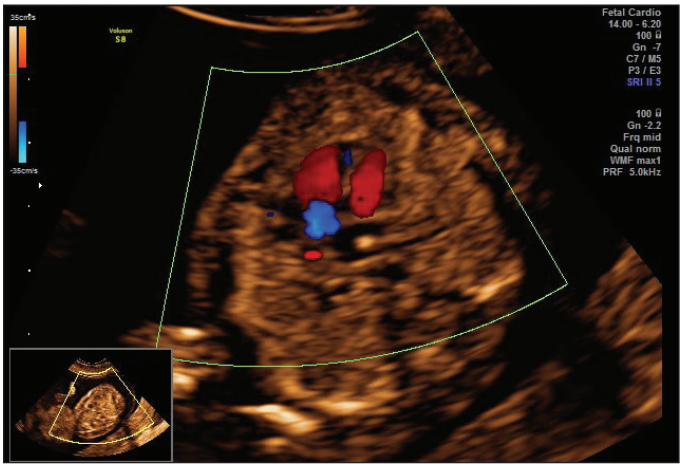


Fig. 2: Normal 4 CV with color

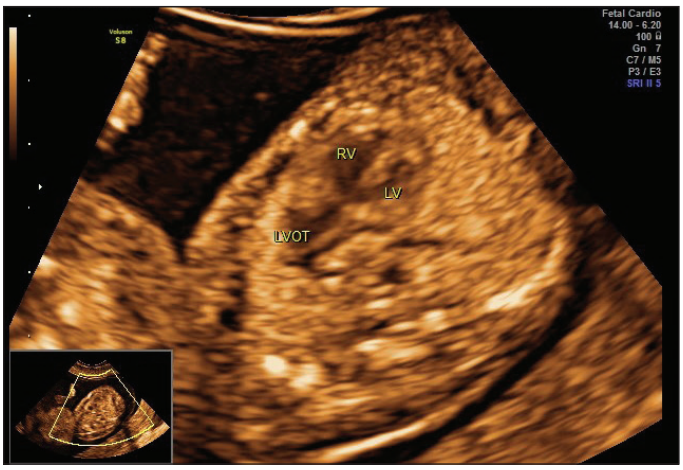


Fig. 3: Aortic override seen with perimembranous VSD.

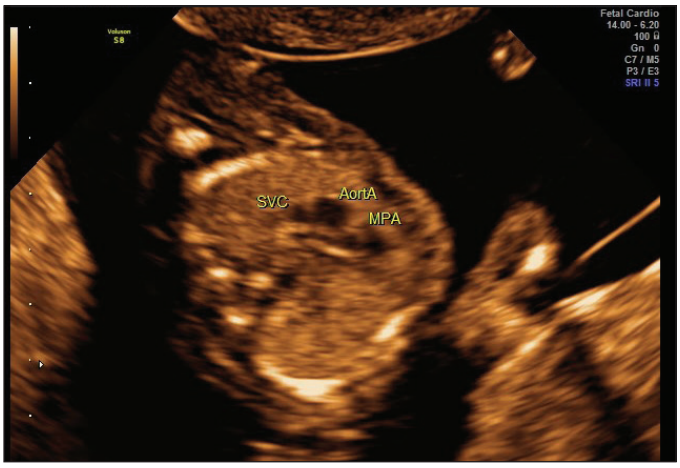


Fig. 4: Aorta placed anteriorly, bigger than MPA

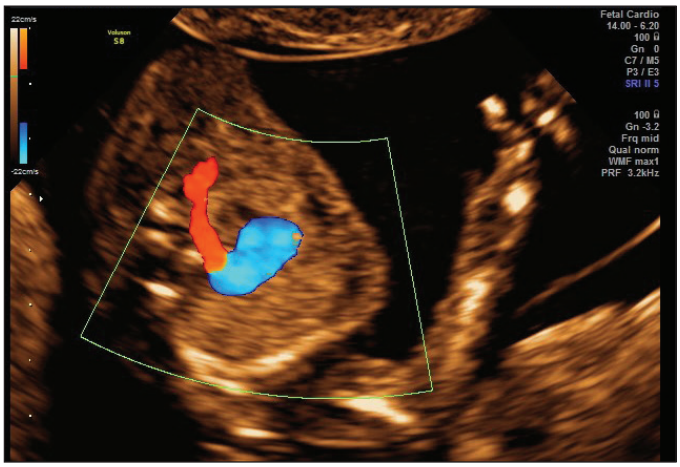


Fig. 5: Shows 3 VT view with dilated aortic arch, narrow Ductal arch and ARSA

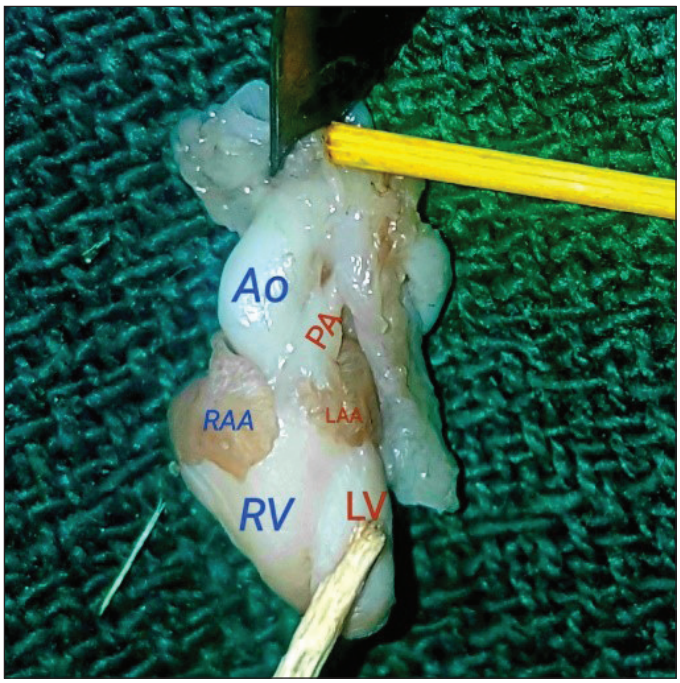


Fig. 6: Autopsy picture of fetal heart showing Dilated aorta, anteriorly and narrow pulmonary artery posteriorly.

Discussion:

Sequential segmental anatomy of fetal heart is possible in skilled hands, Transvaginal route and favorable fetal position and helps in early diagnosis of Tetralogy of Fallot.

Key Points:

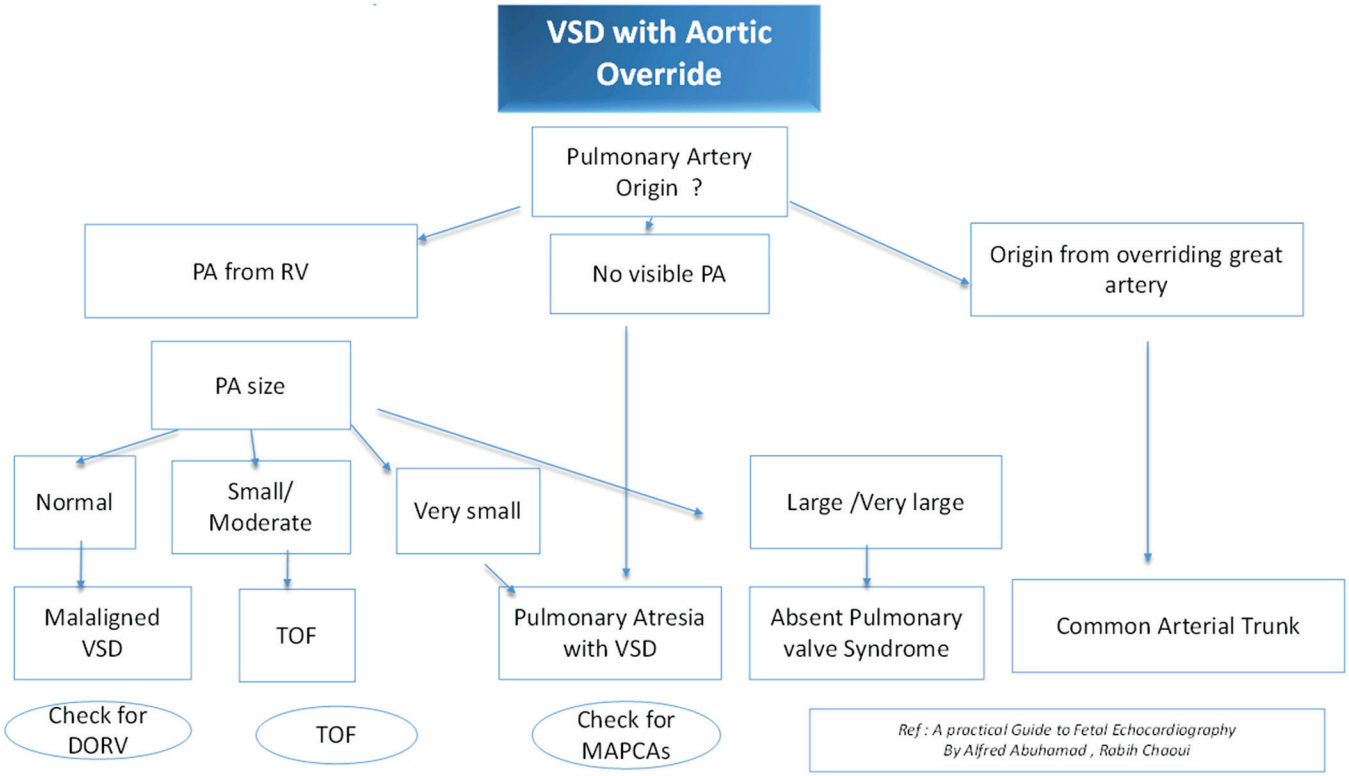
1. Four chamber view appears normal in TOF unless the VSD is large and visible in this plane.
2. TOF is typically detected in the five chamber view, demonstrating a perimembranous VSD and aortic override.
3. There is strong association in raised nuchal translucency and TOF.
4. Other cardiac and extra cardiac abnormalities are common
5. Chromosomal abnormalities are found in 30% cases of TOF.
6. Microdeletion of 22q 11is found in 10% 15% of cases.

Approach to Cardiac defects in first trimester:

Many studies have shown that detection of major CHDs is feasible in the first trimester. However, this still remains a challenge and the detection rate varies according to the type of te cardiac abnormality. Recently the International Society of Ultrasound in Obstetrics and Gynaecology has recommended the use of multiplanar Imaging with the use of Colour Doppler. Systematic step by step approach, as in the second trimester should be done with adequate optimization of grey scale and Colour Doppler.

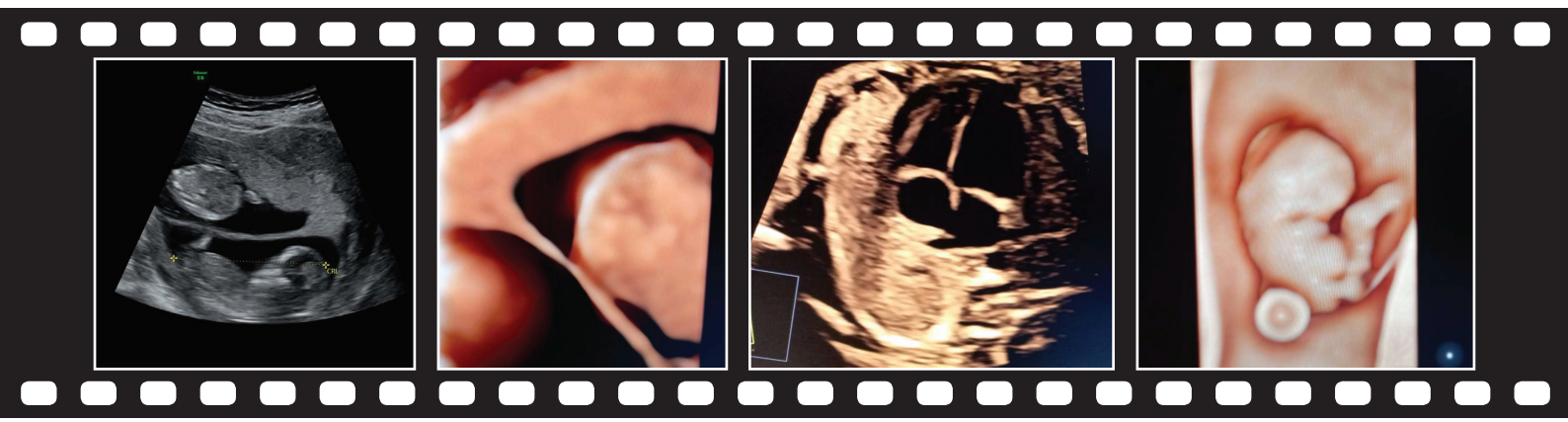
- Cardiac situs and Axis
- 4 chamber view in grey scale and colour doppler.
- 3VT is imaged by V sign
- Abnormal axis, Increased NT (above the 95th centile), DV reversal are clues to do a detailed cardiac survey.

However, the limitations of fetal echocardiography in the first trimester must be borne in mind, and follow-up at mid-gestational echocardiography is prudent in some cases. Also some cardiac abnormalities are not evident until later in pregnancy.



References:

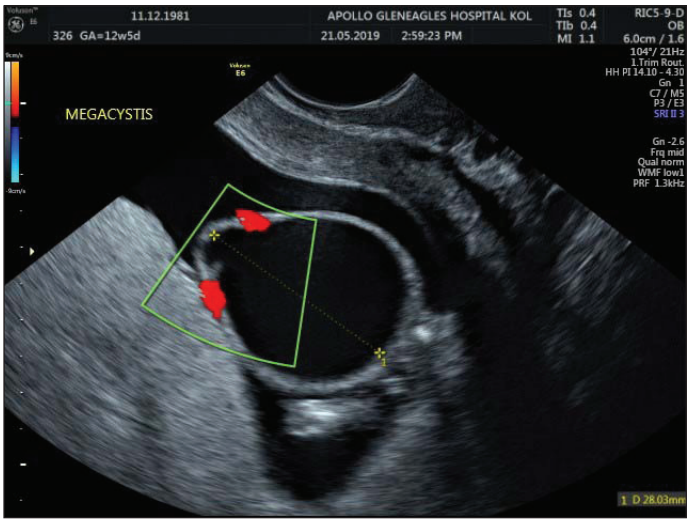
1. Becker R, Wegner R. Detailed screening for fetal anomalies and cardiac defects at the 11–13-week scan. Ultrasound Obstet Gynecol 2006;27(6):613–618. DOI: 10.1002/uog.2709
2. Asma Khalil, Kypros H. Nicolaides. Fetal heart defects: Potential and pitfalls of first-trimester detection. Seminars in Fetal & Neonatal Medicine 2013;(18): 251-260.
3. International Society of Ultrasound in Obstetrics and Gynecology, Bilardo CM, Chaoui R, Hyett JA, Kagan KO, Karim JN, Papageorghiou AT, Poon LC, Salomon LJ, Syngelaki A, Nicolaides KH. ISUOG Practice Guidelines (updated): performance of 11–14-week ultrasound scan. Ultrasound Obstet Gynecol 2023; 61: 127–143.'
4. Alfred Abuhamad, Rabih Chaoui. First Trimester Ultrasound Diagnosis of Fetal Abnormalities.

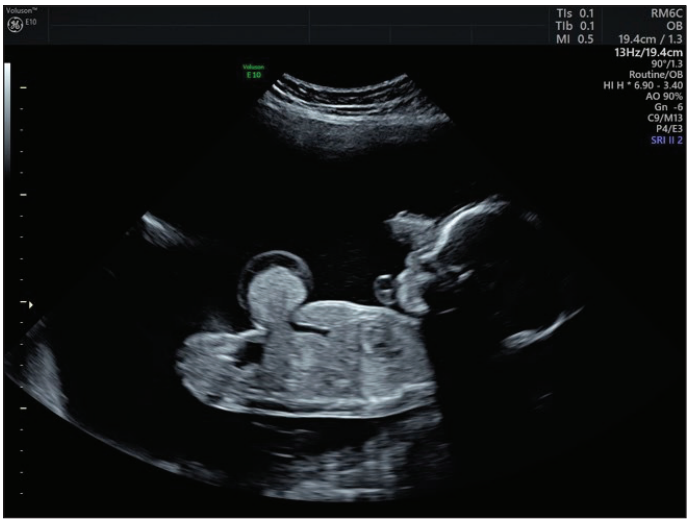


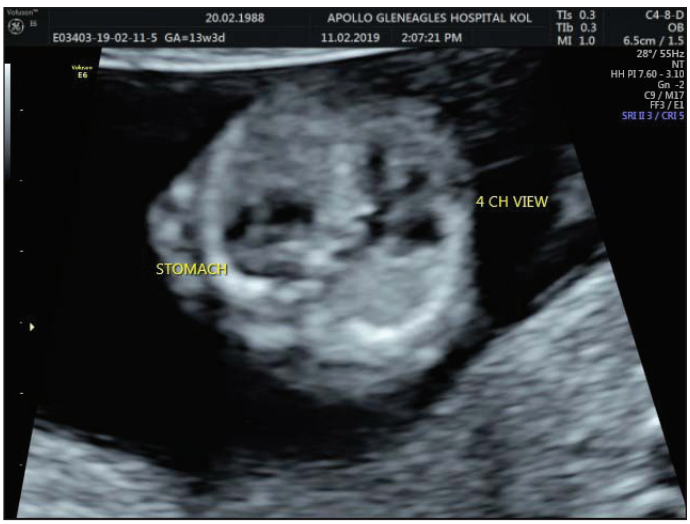
DETECTABLE ANOMALIES IN FIRST TRIMESTER

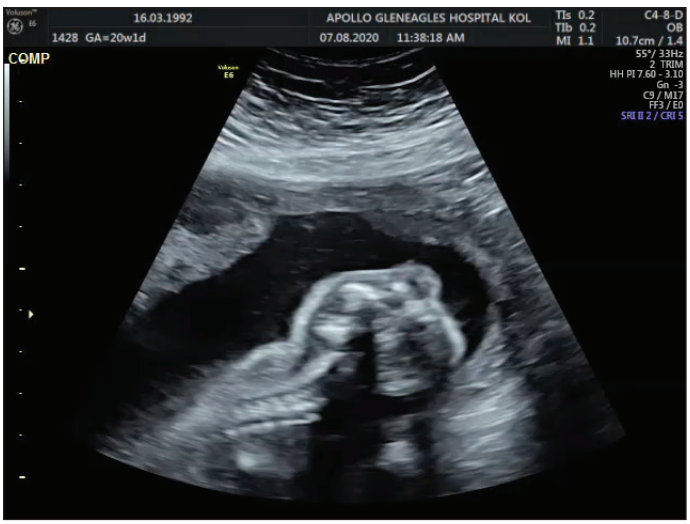
Always Detectable	Potentially Detectable	Undetectable
• Anencephaly • Body stalk Anomaly • Alobar Holoprosencephaly • Exomphalos • Gastroschisis • Megacystis	• Posterior fossa defects • Spina bifida • Facial clefts • Cardiac defects • Renal defects • Limb defects	• Microcephaly • ACC • Fetal Tumours • Ventriculomegaly due to infection/Haemorrhage • Fetal Ovarian cysts • CCAM, Sequestration • Duodenal obstruction, small bowel obstruction

Reference:
Syngelaki A, Chelemen T, Dagklis T, Allan L, Nicolaides KH. Challenges in the diagnosis of fetal non-chromosomal abnormalities at 11-13 weeks. Prenatal Diagnosis. 2011;31(1):90-102

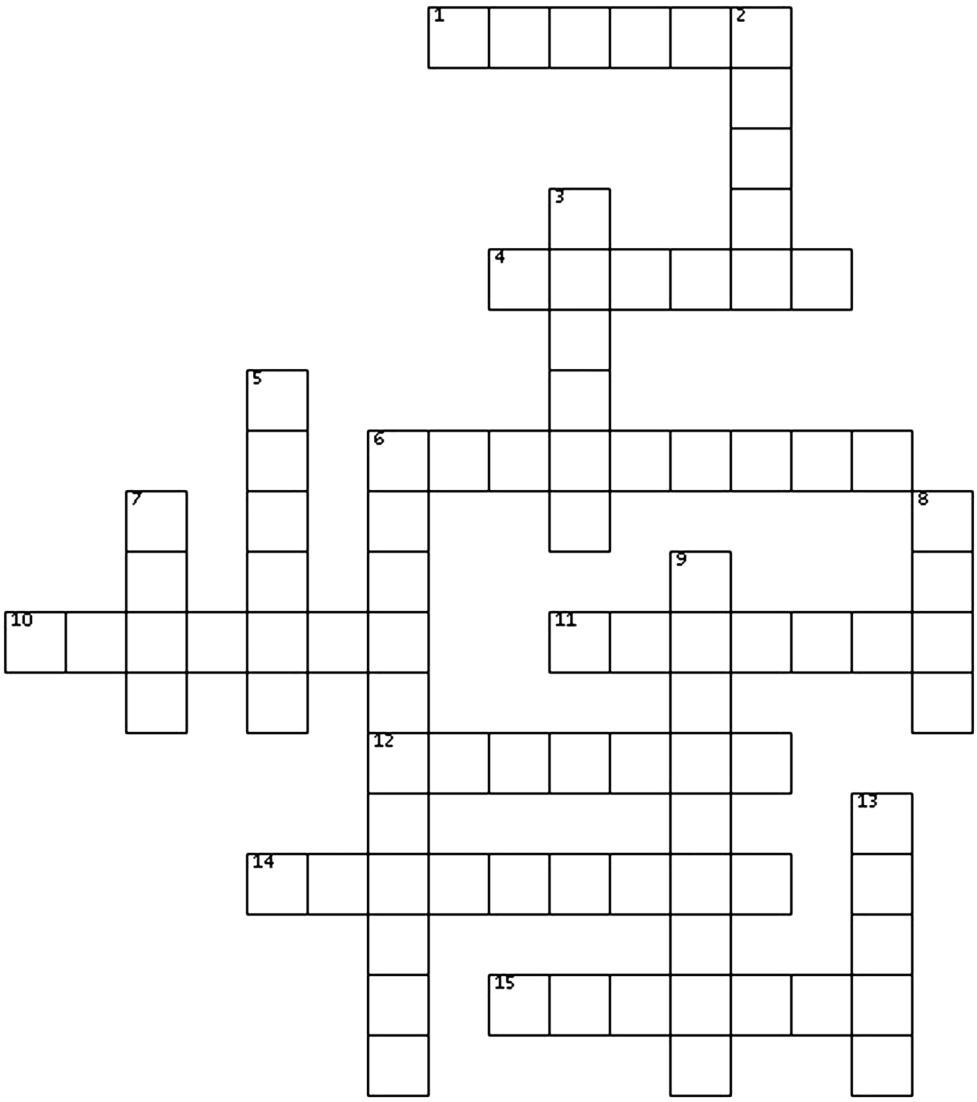








CROSSWORD



ACROSS

1. Sign of placentation in DCDA Twins
4. This syndrome can cause High NTs
6. Five defects described by Cantrell
10. The base of the PMT formed by the _____ palate
11. This needs to be cleaved by the first trimester
12. First detectable anomaly in the first trimester
14. Sign that describes the choroid plexus in the first trimester
15. Feature of Turners Syndrome in the first trimester

DOWN

2. Safety principle in Ultrasound
3. IT in the first trimester is the future __ ventricle
5. Part of this should be seen in the Nuchal Image
6. Feature of Meckel Gruber syndrome
7. Nasal bone forms an 'equal sign with this echogenic part on the nasal bridge'
8. Complex of severe midline defects
9. Gap in this can raise suspicion of palatal defects
13. Early sign of Open NTD in the first Trimester

CROSSWORD ANSWERS: ***(Across):*** 1. LAMBDA; 4. NOONAN; 6. PENTOLOGY; 10. PRIMARY; 11. THALAMI; 12. ACRANIA; 14. BUTTERFLY; 15. HYDROPS. ***(Down):*** 2. ALARA; 3. FOURTH; 5. THORAX; 6. POLYDACTYLY; 7. SKIN; 8. OEIS; 9. MAXILLARY; 13. CRASH