Guidelines & Protocols in OBGY

A Ready Reckoner

Fetal Medicine Update - Part 1

Dr. Vaidehi Marathe
President

Dr. Rajasi Sengupta
Hon. Secretary

Volume 6

Team NOGS 20-21

From The President's Desk ...

Dear Members,

It gives me immense pleasure to release the sixth volume our **"READY RECKONER - The Guidelines and Protocols in Ob-Gy"**.

In this era of evidence based medicine, it is expected that all treatment modalities be guidelines based. To have a quick access to the standard guidelines and have them well sorted out, we will be releasing this ready reckoner on various essential topics every month. This **first of its kind and unique attempt** is our small effort to simplify protocols.

With great pleasure we announce the release of its sixth volume : **"Fetal Medicine Update - Part 1 & Part 2".** I am sure, this release will be valuable in your clinical practice and help quick amending.

I will fail in my duty if, I don't acknowledge the tremendous efforts and contributors Dr. Unnati Shende, Dr. Neelam Chhajed, Dr. Neha Puniyani, Dr. Kunda Shahne, Dr. Amee Rahatekar. They have toiled very hard to compile these guidelines for your benefit.

Happy reading...Wishing you all Safe and Ethical Clinical Practice...

Academically yours,
Dr. Vaidehi Marathe
President NOGS-2020-21

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FIRST TRIMESTER SCREENING

- DR.KUNDA SHAHNE

FIRST TRIMESTER SCREENING

First trimester screening is first important step of evaluation in all pregnancies.1ST Trimester screening is offered to all pregnant ladies from 11 to 13 +6 weeks

With the help of 1st trimester screening, we will be able to achieve methodical, uniform, cost effective and better fetal evaluation. We are also aiming at lowering the prenatal invasive diagnostic procedures because of the introduction of effective screening methods.

Training and registration:

As per the law in India today, those who fulfill the criteria according to PCPNDT act, can perform the scan. He/ she should be sufficiently trained to do Sonography and must be registered with appropriate authority.

This is advised for

- **Screening for chromosomal defects**
- **Diagnosis of major fetal abnormalities**
- **Screening and diagnosis in twin pregnancies**
- **Screening for preeclampsia**
- **Baseline before CVS**
- **Baseline before Fetal reduction in multifetal pregnancy**

GOALS :

- Provide the informed choices to the couple at risk of having abnormal child
- Provides reassurance and remove anxiety, especially among high risk groups

- Allow couples at high risk to know that the presence or absence of the disorder can be confirmed by testing
- Allow the couple the option of appropriate management(psychology of pregnancy, delivery, postnatal)
- To enable the prenatal treatment of the fetus

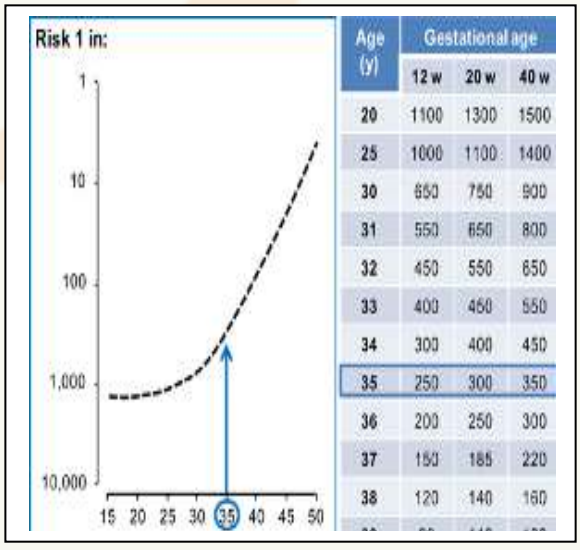
Screening modalities

- Maternal age
- Ultrasound
- Combined
- Biochemical markers
- NIPT

The risk for trisomy 21 increases with maternal age.

The risk for trisomy 18 and 13 increases with maternal age and decreases with gestation. Turner syndrome and Triploidy is unrelated to maternal age is unrelated to maternal age

Age	Frequency (live births)
< 35	< 0.3 %
37	0.5 %
40	1 %
50	10 %



Age (y)	Gestational age		
	12 w	20 w	40 w
20	1100	1300	1500
25	1000	1100	1400
30	650	750	900
31	550	650	800
32	450	550	650
33	400	450	550
34	300	400	450
35	250	300	350
36	200	250	300
37	150	185	220
38	120	140	160

Combined screening

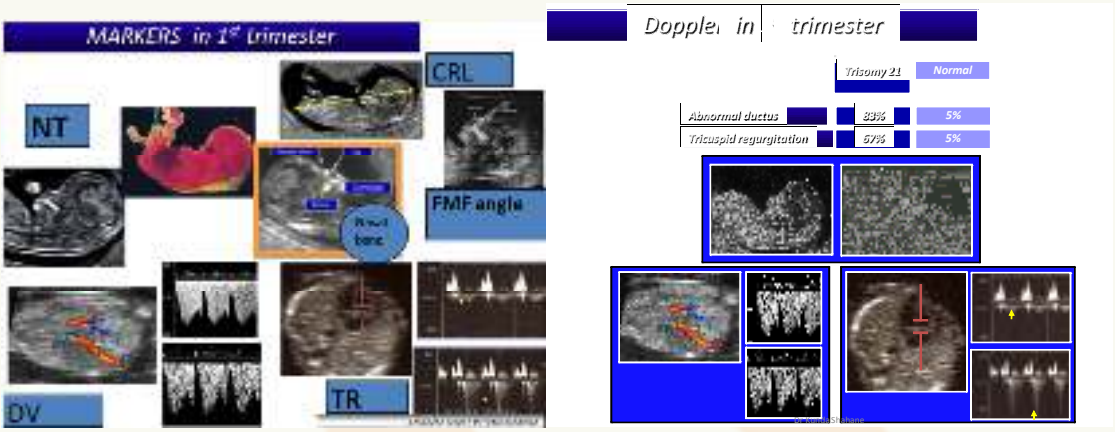
- ❑ SOFT MARKER ASSESMENT ON ULTRASOUND
- ❑ BIOCHEMICAL MARKER(double marker)
- ❑ MATERNAL AGE
- ❑ DR AROUND 95%

Noncombined screening

- ❑ ULTRSOUND
- ❑ MATERNAL AGE
- ❑ DR AROUND 90%

Before 13 weeks, gestational age can be accurately assessed from the measurement of crown rump length. However from 14th weeks crown rump length (CRL) should not be used because the fetus becomes increasingly flexed making the measurement unreliable. As an alternative to CRL, bi-parietal diameter, and/or head circumference should be used .The early scan can usually be performed by transabdominal and /or transvaginal route.

Correct measurement of crown rump length- CRL, nuchal translucency- NT, heart rate must be done NT is measured when CRL is between 45 to 84 only Fetal anomaly scan to detect the gross fetal anomaly should be performed Additional study of nasal bone, Ductus venosus and tricuspid regurgitation can improve the detection rate of fetal aneuploidy .Study of uterine artery Doppler is to be done for prediction of preeclampsia and cervical length for prediction of preterm delivery. Reporting of 11-, 13 +6 week scan must include the risk assessment of Trisomy 21. Consultant who is performing NT scan must be properly trained, preferably certified by Fetal Medicine Foundation United Kingdom (FMF UK). The software for calculating the risk of Trisomy 21 is available free for those accredited by FMF in 11-13+6 week scan regular audit report ensures continuity of license for the use of software.



95th percentile for nuchal traslucency is 2.5 mm

99th percentile is 3.5 mm

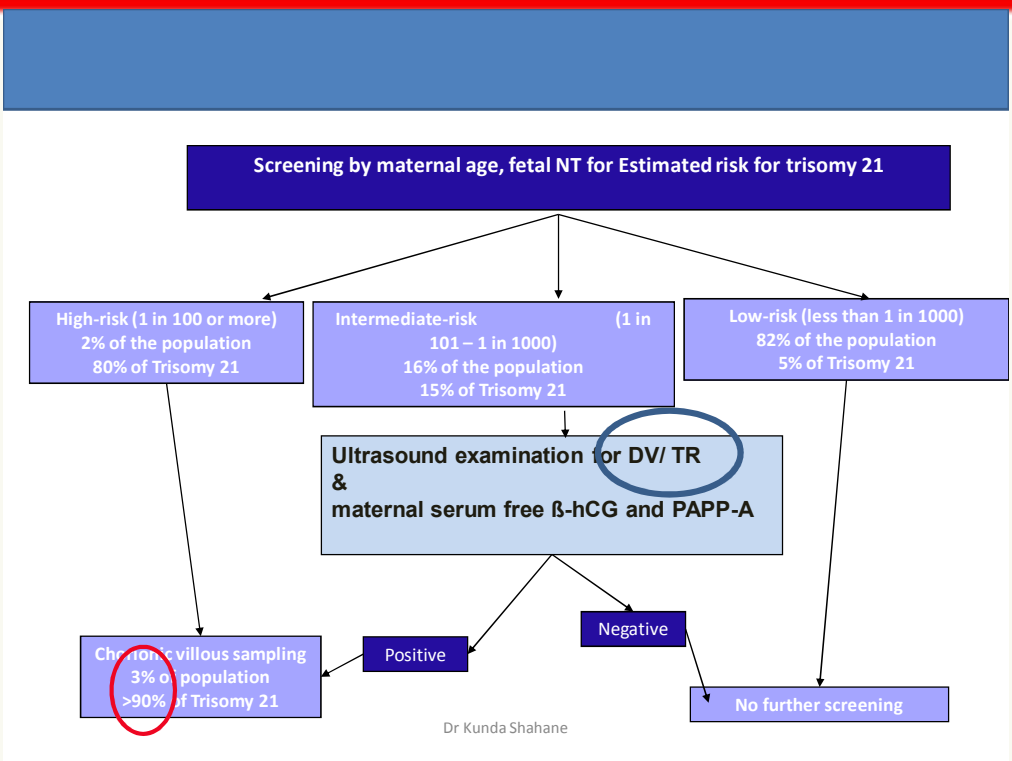
Increase NT and chromosomal defects:

Increased fetal NT

- Trisomy 21 and other major chromosomal abnormalities.
- Cystic hygroma
- Congenital heart disease
- Fetal infection
- Fetal hydrops

Apriory risk is the background risk with the consideration of maternal age

Final modified risk is o be calculated with combined risk assessment and further management is decided.



BIOCHEMICAL SCREENING AND NIPT:

Biochemical marker is a double marker which includes free B hcg MoM and papp-a MoM

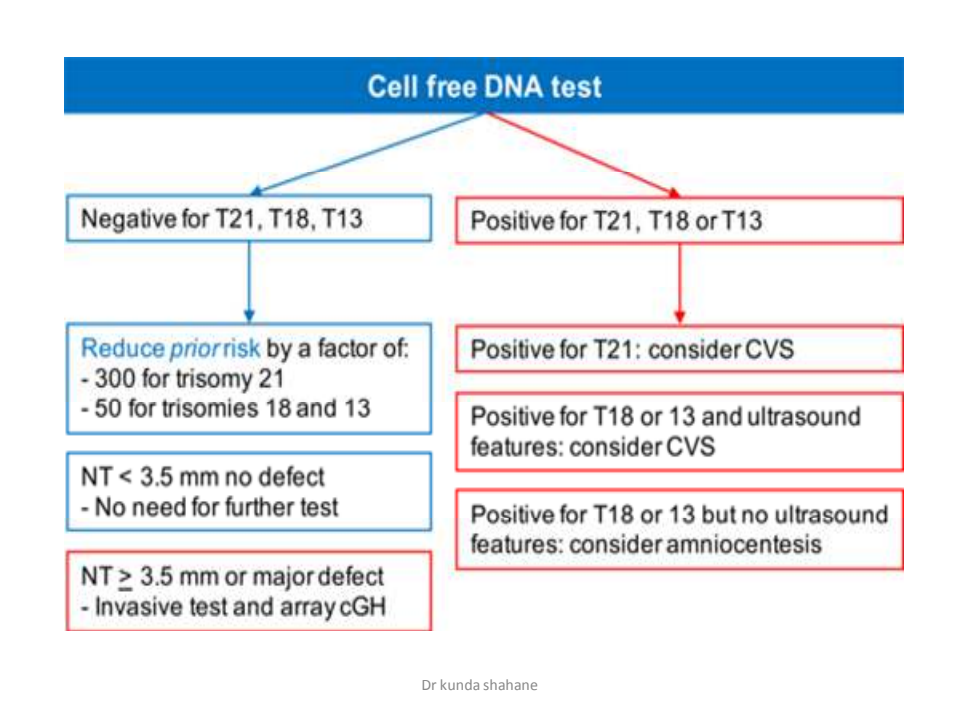
NIPT (NON INVASIVE PRENATAL TESTING)

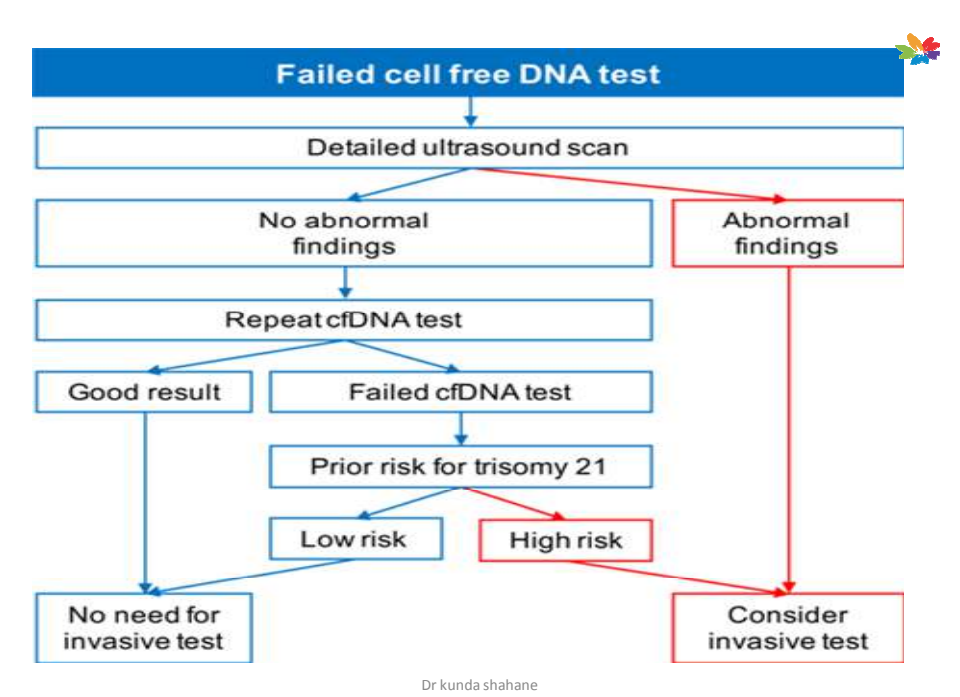
- This is a screening test of higher sensitivity. **Cell free fetal DNA in maternal blood** can detect about 99% of fetuses with trisomy 21 and 98% of fetuses with trisomy 18 or 13 at a false positive rate (FPR) of 0.1-0.2%
- screen for sex-chromosome aneuploidies and certain microdeletions, such as 22q11 (Di George syndrome).

INDICATIONS:

- Advanced maternal age
- Have a marker screen risk i.e. FTS/IPS/MSS (1:250 TO 1:1000)
- Have had a previous pregnancy or child with aneuploidy

RESULTS:





BENEFITS :

- Fewer women having diagnostic tests with associated risk of pregnancy loss
- Increased access
- Early test result (drawn at ≥ 9-10 weeks at earliest)
- No risk of miscarriage
- Detects the most common chromosomal aneuploidies
- Higher detection rates than IPS or MSS

LIMITATIONS :

NIPT **cannot**:

- Detect chromosome differences other than aneuploidy of chromosomes 13, 18, 21, X and Y
- Completely rule out aneuploidy
- Detect single gene conditions

- Detect congenital anomalies
- Possibility of no result (~6%)
  1/2- 2/3 can be successfully resolved with redraw at later gestation
- False positives and false negatives
- Twins pregnancies

Pre- and post-test counseling is important as Invasive testing following positive results needs confirmation by diagnostic invasive testing

EARLY ANOMALY SCAN

OUR AIM IS TO DIAGNOSE MAXIMUM POSSIBLE ANOMALIES THAT CAN BE DETECABLE AT EARLY 12 WEEKS SCAN WITH ALL THE STANDARD VIEWS STUDIED

- ❑ Major cardiac defects
- ❑ Diaphragmatic hernia
- ❑ Exomphalos
- ❑ Megacystis
- ❑ Body stalk anomaly
- ❑ Skeletal abnormalities

In other abnormalities fetal NT is usually normal:

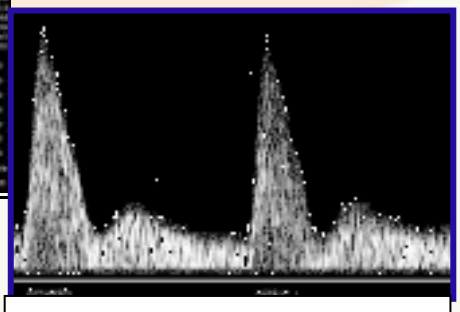
- Acrania / anencephaly
- Ventriculomegaly
- Holoprosencephaly
- Spina bifida
- Gastroschisis

Screening for preeclampsia:

UTERINE ARTERY DOPPLER IS USED AS A PREDICTOR OF PREECLAMSIA, OLIGOAMNIOS, FGR

Uterine artery Doppler study reflects implantation and maternal arterial remodeling

Normal waveform



High resistance flow

10

Women with uteroplacental insufficiency are at risk for having complications like Preeclampsia, IUD, Prematurity, IUGR, Placental abruption. (50-67 % is PPV **more than 93% is NPV)**

Mean uterine artery PI is considered and cutoff is 2.5 in first trimester. Interpretation is important to start treatment in the form of aspirin for prevention of abovementioned complications and close follow up is suggested.

⇨ **TO SUMMARISE**

1ST TRIMESTER SCREENING IS AN IMPORTANT MILESTONE OF PREGNANCY TO SCREEN CHROMOSOMAL ABNORMALITY, CONGENITAL ANOMALIES AND PREDICTON OF PRECLAMSIA

KEY POINTS:

INCREASED NT >99TH PERCENTILE (3.5MM)REQUIRES CONFIRMATION BY CHORION BIOPSY AT 12/13 OR AMNOCENTESIS AT 16 WEEKS

IF CHROMOSOMAL EVALUATION IS NORMAL THEN NEXT STEP IS TO DO TARGEDTED SCAN AND FETAL ECHO TO RULE OUT CONGENITAL ANOMALIES AND CARDIAC ABNORMALITES

CLOSE FOLLOW UP IS TO BE DONE

SECOND TRIMESTER SCREENING

- DR. KUNDA SHAHNE

2ND TRIMESTER SCREENING

An attempt must be made to evaluate the fetus completely with standard views to exclude major structural anomalies in transverse, sagittal, and coronal plains The "18-20 weeks Anomaly Scan is to reassure the woman that the fetus appears to have no obvious structural abnormalities. The primary aim should be to prove the "normality"

SECOND TRIMESTER SCREENING INCLUDES:

⇨ ANOMALY SCAN WITH SOFT MARKER ASSESMENT

⇨ BIOCHEMICAL MARKER - QUADRUPLE MARKER

⇨ UTERINE ARTERY DOPPLER

⇨ CERVICAL SCREENING

(COMBINED SCREENING ANOMALY SCAN WITH QUADRUPLE MARKER IF DUAL MARKER IS NOT DONE IN 1ST TRIMESTER OR AS A INTEGRATED SCREENING IF DUAL MARKER IS IN INTERMEDIATE RISK)

The minimum standard for an 18-20 week Anomaly Scan, Gestational age can be established by measurement of bi-parietal diameter, head Circumference, abdominal circumference and femur length. Uterine artery Doppler and the measurement of cervical length should be included in the scan

Anomaly Scan should be carried out at the clinic with minimum standard described below If a clinic considers that it cannot deliver scans to this minimum standard as described below then the 18-20 week scan should be referred to an appropriate unit for The Procedure.

OPTIMAL TIME FOR ROUTINE SECOND TRIMESTER SCREENING:

- **Earliest gestation** at which the necessary **measurements** and a full **fetal anatomy** survey can be performed.
- **Latest gestation** at which an acceptable range of **options can be offered** to the pt if an abnormality is detected.

- **Measurements after 15 weeks.**
- **Fetal anatomy at 18-20 weeks.**
- **Fetal heart 18-24 weeks.**
- **Preferable time is 18/19 weeks**

AIMS OF PERFORMING ROUTINE SECOND TRIMESTER SCREENING :

- Basically to **define and document** the **normality/ abnormality of**

1. Fetal structure
2. Fetal growth
3. Fetal environment

- For prediction of risk of perinatal morbidity and mortality

RATE OF PICKING AN ABNORMALITY IS DIRECTLY PROPORTIONAL TO :

1. **Level of knowledge** and experience of sonographer.
2. **Quality of the machine** used.
3. **The time spend** during sonography.
4. **Adherence to protocol** (Which is made on the bases of international **"Guide lines")**

ROUTINE SECOND TRIMESTER SCREENING PROTOCOL :

1. Fetal life, Number and presentation
2. Amniotic fluid volume
3. Placental location and relation to cervix
4. Estimation of GA
5. Estimation of Fetal Growth and Wt.
6. Uterus and adnexae

7. Specified fetal anatomic survey
8. Soft marker assessment and screening for chromosomal abnormality

DOCUMENTATION :

- Permanent records of images
- Date, Name or identifier
- Written report in medical records
- According to ACR communication standard

RULE OF 2 FOR ANATOMICAL SURVEY:

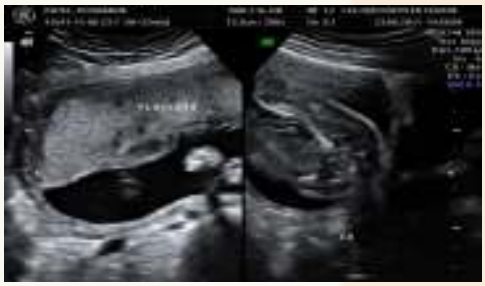
Fetal environment 1. Placenta 2. liquor

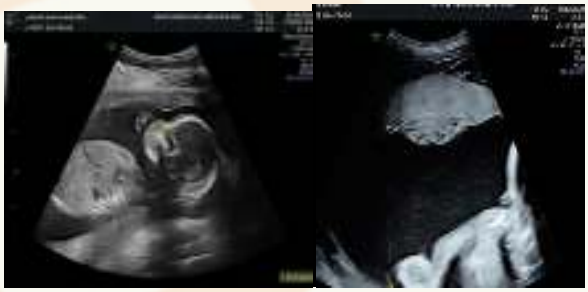
Placenta 2 points measurment

Localisation and thickness

Two methods of liquor

Eyeballing and single large pocket

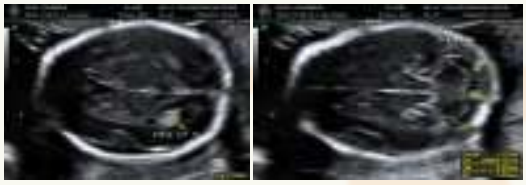




FETAL STRUCTURE:

Head :

1.transventricular 2.transcerebellar

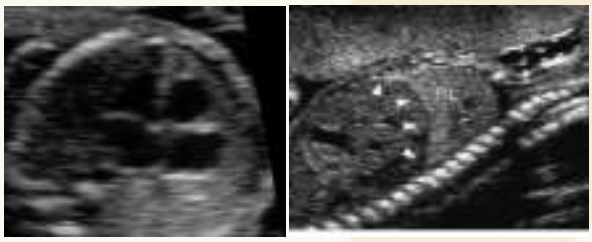


Face 2 views:

1. Profile 2 .PMT

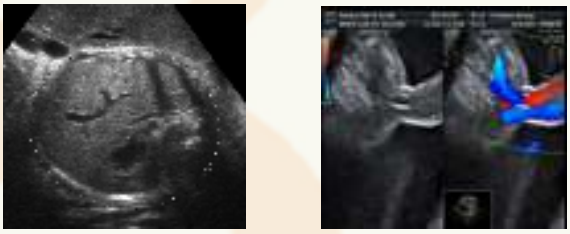
Thorax :

1 .transverse 2. sagittal



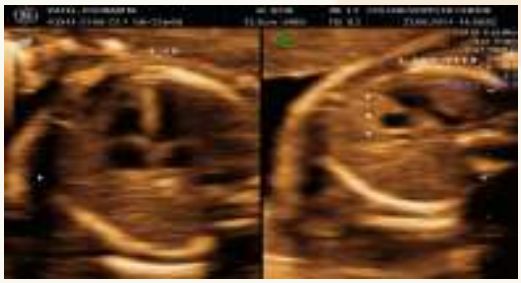
Abdomen transverse views :

1. Stomach bubble 2.cord insertion



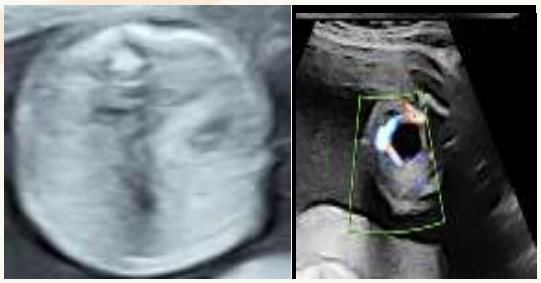
Cardiac two views :

1. Four chamber 2.three vessel



Renal system :

1.kidney transaxial view 2.bladder transaxial



SKELETAL 2 PARAMETERS

SPINE :

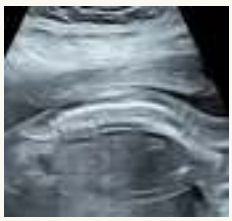
Sagittal
sets of long bones

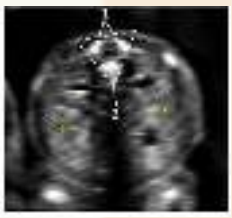
Transverse
sets of long bones

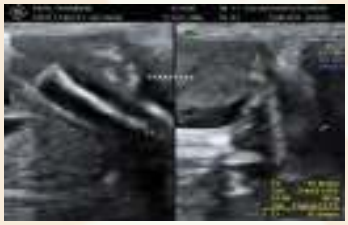
LIMBS :

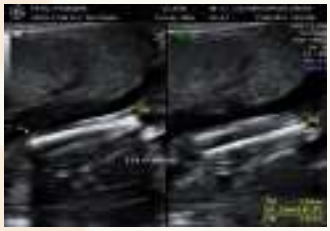
2 upper limbs with 2 segments with 2

2 lower limbs with 2 segments with 2

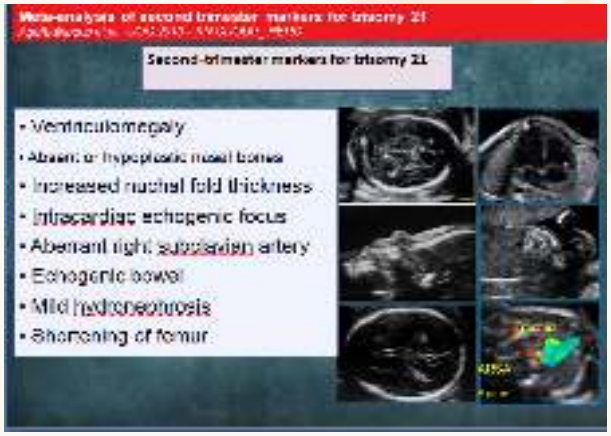








SECOND TRIMESTER SOFT MARKERS



The risk of aneuploidy should be mentioned when scan is performed between 16-20 weeks with the help of software or can be calculated with the help of likelihood ratio for all the soft markers

BIOCHEMICAL SCREENING :

QUADRUPLE MARKER	TRIPLE MARKER
AFP	AFP
ESTRIOL	ESTRIOL
B HCG	B HCG
INHIBIN A	
DR 81% FPR 5%	DR 67-70 % FPR 5%

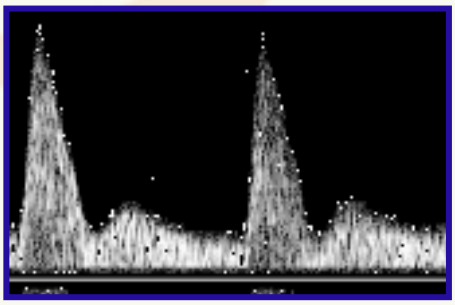
QUADRUPLE MARKER IS BETTER SCREENING TEST IN SECOND TRIMESTER WITH HIGHER SENSITIVITY THAN TRIPLE MARKER

SCREENING FOR PRE-ECLAMPSIA:

UTERINE ARTERY DOPPLER IS USED AS A PREDICTOR OF PREECLAMSIA ,OLIGOAMNIOS,FGR

Uterine artey Doppler study reflects implantation and maternal arterial remodeling

Normal waveform



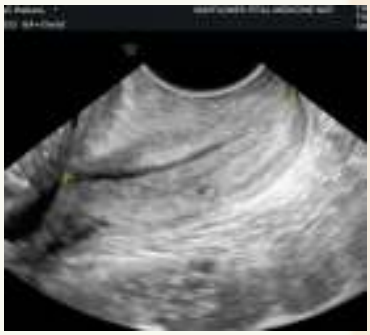
High resistance flow

Womens with uteroplacental insufficiency are at risk for having complications like Preeclampsia,IUD,Prematurity,IUGR,Placental abruption.(50-67 % is PPV

more than 93% is NPV)

Mean uterine artery PI is considered and cutoff is 1.75 in first trimester.Interprtation is important to start treatment in the form of aspirin for prevention of abovementioned complications and close follow up is suggested.

CERVICAL SCREENING :



- Cervical length and competency should be evaluated for prediction of preterm birth or need of cervical encirclage.
- Cervical length less than 25 mm is considered to be high risk for preterm labour

Women should receive written details about their scan result. All scans should be carefully documented and archived Accurate record keeping is needed to with the pregnancy outcome is recorded with sufficient detail. Use of the computer based record keeping with the use of software should be encouraged and preferred which also helps in checking the quality and the audit of the unit consultant Regular audit of pregnancy outcome should be checked

The "Genetic Sonogram" should be a part of "The 18-20 weeks anomaly scan". The genetic Sonogram will help in identifying the fetus at the risk of fetal aneuploidy The genetic Sonogram involves the evaluation of presence of absence of soft markers between 16 to 20 weeks. Soft markers are the obstetric ultrasound findings, which are considered variants of normal but are noteworthy because they also increase the risk or underlying fetal aneuploidy. Presence of soft markers may be associated with nonchromosomal malformations also.

The presence of soft markers increases the risk for fetal aneuploidy but is not diagnostic. Individual soft markers will vary in the degree of association with fatal aneuploidy It has become practice to estimate the degree of association as a likelihood ratio (L.R) by which the a priori background risk is altered. Detection of multiple solt markers will increase the significance of the finding, compared with seeing the same marker in isolation. In addition, “maternal serum testing screening tool can complement and enhance the overall screening l process. Providing an accurate assessment of fetal genetic risk require the ability to integrate known factors before patients can make an informed choice about proceeding with invasive diagnostic testing.

GUIDANCE ON SCREENING FOR ANEUPLOIDY :

A. **THICKENED NUCHAL FOLD-LIKELIHOOD RATIO : 10**

1. A thickened nuchal fold significantly increases the risk of fetal aneuploidy. Expert review is recommended, and Karyotyping should be offered.

2. A thickened nuchal fold is associated with congenital heart disease and rarely with otheri genetic syndromes.

B. **MID VENTRICULOMEGALY-LIKELIHOOD RATIO : 9**

1. Cerebral ventricles greater than or equal to 10 mm are associated with chromosomal and central nervous system pathology. Expert review should be initiated to obtain a detailed anatomic evaluation looking for additional malformations or soft markers, laboratory investigation for the presence of congenital infection or fetal aneuploidy. Fetal MRI as and additional imaging technique may be of help.

2. Neonatal assessment and follow-up are important to rule out associated abnormalities and

are important because of the potential for subsequent abnormal neuro development.

C. **ECHOGENIC INTRACARDIAC FOCUS (ECF) LIKELIHOOD RATIO : 1**

1. ECF should be evaluated and reported as part of the 4-chamber cardiac review.

2. Women with right-sided, biventricular, multiple, particularly conspicuous, or nonisolated

ECF should be offer referral for expert review and possible Karyotyping.

D. **MILD PYELECTASIS LIKELIHOOD RATIO :1**

1. If pylectesis is visualized, the renal pelvis should be measured in the anterior/posteriori diameter.

2. All fetuses with renal pelvic measurements > 5 mm should have a neonatal ultrasound, and

those having measurements > 10 mm should be considered for a regular follow up scan.

3. Isolated mild pylectesis does not require fetal Karyotyping.

4. Referral for pylectesis should be considered with additional ultrasound findings and (or) in women at increased risk for fetal aneuploidy owing to maternal age or maternal serum screen results.

E. **SINGLE UMBILICAL ARTERY (SUA)**

I. Assessment of cord vessels is considered a part of the routine obstetric ultrasound at 18 to 20 weeks.

2 The finding of a SUA requires a more detailed review of fetal anatomy including kidneys and fetal heart (fetal echo)

3 An isolated SUA does not warrant invasive testing for fetal aneuploidy.

F. **ECHOGENIC BOWEL. LIKELIHOOD RATIO : 3**

1. Echogenic bowel should be identified by comparison with the echogenicity of surrounding bone using an appropriate transducer and gain setting. Bowel echogenicity equal to or greater than bone is signifcant.

2. Echogenic bowel is associated with both chromosomal and nonchromosomal abnormalities. Expert review is recommended to initiate the detailed ultrasound evaluation looking for additional structural anomalies or other sol markers of aneuploidy, detailed evaluation of the fetal abdomen looking for signs of bowel obstruction or perforation, detailed evaluation of placental characteristics. Detailed maternal work up for serum screening tests, evaluation for cystic fibrosis and infection should be done.

The genetic counseling and fetal karyotype should be considered.

G. **CHOROID PLEXUS CYSTS (CPC)**

1. Isolated CPCs require no further investigation when maternal age or the serum screen equivalent is less than the risk of a 35-year-old.

2. Fetal Karyotyping should only be offered if isolated CPCs are found in women 35 years or older or if the maternal serum screen is positive for either trisomy 18 or 21

3. All women with fetal CPCs and additional malformation should be offered referral and Karyotyping

H**. MEGA CISTERNA MAGNA**

1. An isolated mega cisterna magna is not an indication for fetal Karyotyping.

2. With a mega cisterna magna, expert review is recommended for follow-up ultrasound, fetal I MRl and investigations.

G. **SHORT FEMUR LENGTH and SHORT HUMERUS LENGTH**

LIKELIHOOD RATIO : 1 and LIKELIHOOD RATIO : 1.5

If a femur and / or humerus appear abnormal or measures short on screening ultrasound, otherl long bones should be assessed and referral with follow-up ultrasound considered.

SOFT MARKERS &THEIR INTERPRETATION

- DR.NEHA MUNIYAR PUNIYANI

INTRODUCTION :

- Ultrasonographic evaluation of fetal anatomy between 18 & 22weeks of gestation is part of routine prenatal care.
- The scan provides an opportunity for careful evaluation of fetal anatomy and major congenital malformations.
- It also provides and opportunity to evaluate for so-called “soft markers”.

WHAT ARE THESE “SOFT MARKERS”

- Anatomical variants & not malformations
- Mostly present transiently
- Associated with ↑ risk for aneuploidies & genetic abnormalities
- Mainly - trisomy 21 (Downs’ syndrome) , trisomy 18 & 13.
- Also seen in 15%- normal euploid fetuses
- Harder to ignore a soft marker than to see it
- Reporting and counselling is challenging & needs to be individualized
- Disclosure of soft markers of no clinical significance to low risk patient leads to

- Reporting & counselling requires knowledge of :
- patient’s prior risk status for aneuploidy
- clinical significance of each specific marker

WHAT IS LIKELIHOOD RATIO ?

- Likelihood ratio is a number given to each marker
- It tells the probability/chance of a women having a Downs baby because she has tested positive for a marker
- Ex. – if the LR for marker ‘X’ is 5 any woman who tested positive for this marker has a 5 times higher chance of having a Down’s baby

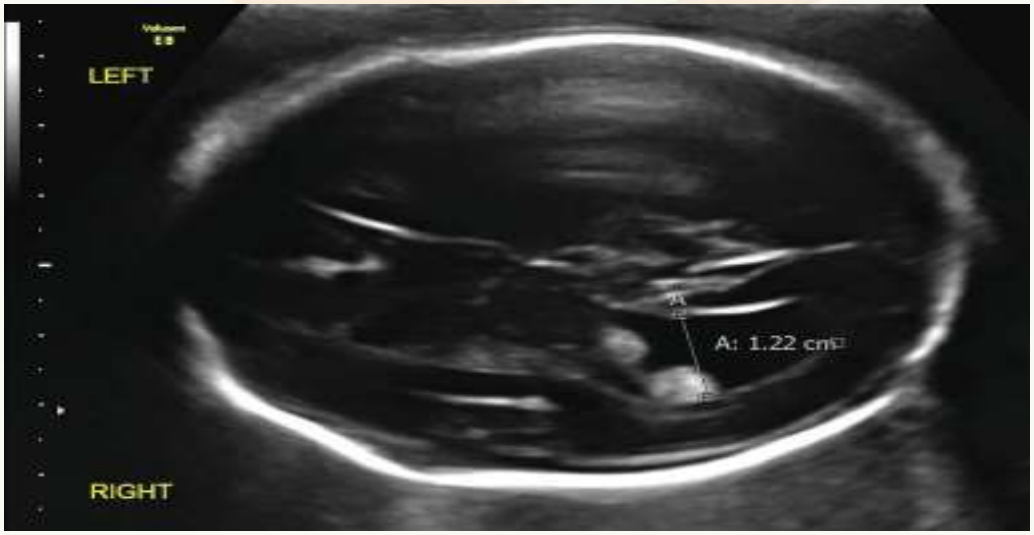
INTERPRETING LIKELIHOOD RATIO

RATIOS	INTERPRETATION
0-1	Decreased evidence for disease; very likely not to have the disease
1	No diagnostic value
>1	Increased evidence for disease; more likely to have the disease

SOFT MARKERS :

- Ventriculomegaly
- Thickened nuchal fold
- Echogenic intra-cardiac focus
- ARSA (aberrant right subclavian artery)
- Echogenic bowel
- Mild pylectasis
- Hypoplastic / absent nasal bone
- Short femur/humerus
- Choroid plexus cyst

MILD VENTRICULOMEGALY :



- **DEFINITION**- width of cerebral lateral ventricle at the level of atrium/parieto-occipital sulcus between 10-15 mm .
- **Correct measurement requires** -

-inner to inner border,

-perpendicular to ventricular cavity

-cross section at the level of thalami/lateral ventricle

-CSP should bisect midline falx

-regular skull contour

ASSOCIATIONS :

- Aneuploidies-mainly Down's syndrome (LR- 3.81, one of the strong marker)
- Structural malformations
- Congenital infections (TORCH)
- Cerebral hemorrhage
- Developmental brain abnormalities
- Neuronal migration abnormalities

APPROACH TO MILD VENTRICULOMEGALY :

- Detailed anatomical evaluation + fetal neurosonography & genetic counselling
- Karyotype due to increased risk for aneuploidy
- Evaluation for congenital infection(TORCH)- maternal TORCH test
- Follow up USG after 3 weeks – to assess any increase in the size of lateral ventricles
- Consider fetal magnetic resonance (MRI) in late 2^{nd} or 3^{rd} trimester to rule out migration & gyration abnormalities

THICK NUCHAL FOLD (NF) :

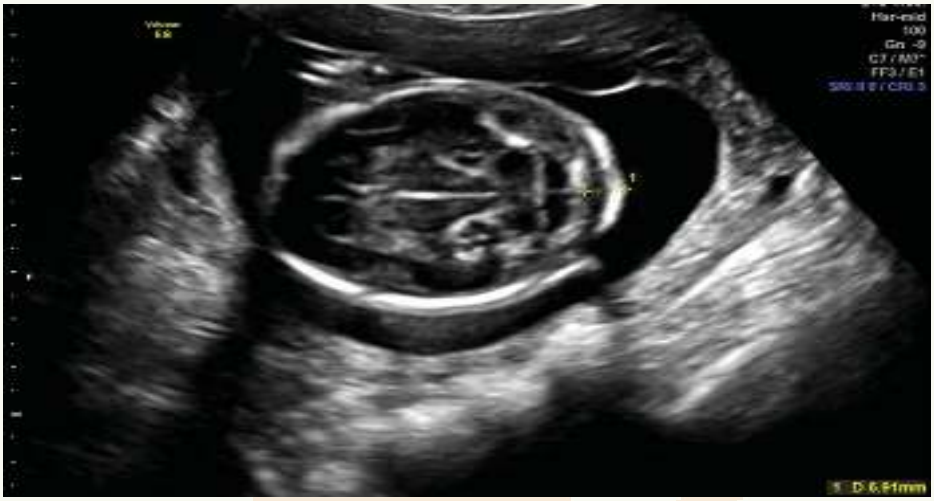
- NF is measured **on Trans-cerebellar plane**
- With transducer tilted 30 degree towards fetal occiput
- **Correct measurement requires**-

- regular skull contour
- CSP, midline falx and cerebellum should be seen
- from outer edge of occipital bone to outer skin edge

- **Thick NF** – when NF ≥ 6 mm at 15-20 weeks
- NF –normally thicker in fetuses with breech presentation & nuchal cord

- **NF ≥ 5mm** - associated with ↑risk of congenital heart disease
- Increased NT in 1st trimester with thick NF in the 2nd trimester – consider **Noonan syndrome**
- **Thick NF** - **One of the strongest second trimester marker for Down's** syndrome with LR - 3.79

WHAT NEXT ?



- Detailed anatomical scan to look for other markers of aneuploidy
- Fetal echocardiography
- Genetic counselling
- Definitive testing- Amniocentesis

A. If any other marker present

↓

Readjust risk based on published LRs

Consider genetic counselling & definitive testing

B. Increased NT in 1st trimester with normal karyotype but thick NF in 2nd trimester

↓

Look for subtle abnormalities of Noonan on ultrasound

Consider Noonan syndrome mutation detection

If isolated thick NF

No prior screening , regardless of age:

Given the high +ve LRs genetic counselling & Amnio for fetal KT should be done

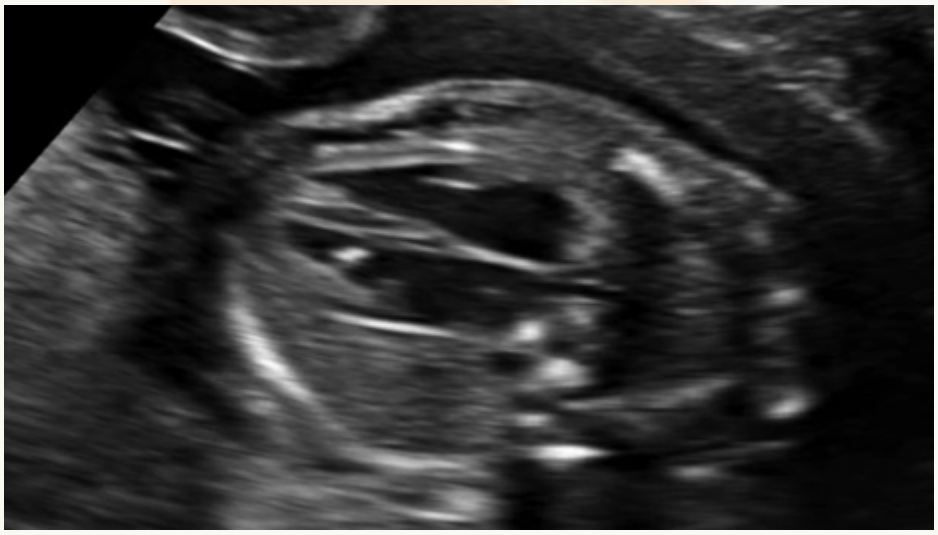
Women with prior 1st tri or 2nd tri screening

Readjust the risk based on previous testing, Genetic counselling & Amnio to be done

Women with negative CVS Amnio or NIPT for Down's

No further assessment

ECHOGENIC INTRA-CARDIAC FOCUS (EIF) :



- **DEFINED** – as focal hyperechogenic spot within the heart with brightness comparable to that of bone
- Occurs due to microcalcification of papillary muscles and/or thickening of the chordae tendinae.
- Frequently seen in the left ventricle (88%) ; right ventricle (5%) or both ventricles (7%)

- **Small association with Down's** – LR-0.95, generally no other concerns therefore further USG evaluation not needed if isolated
- May be seen in 3% to 5% of normal fetuses
- More common in twins
- **Imaging requirement**-

  -EIF should be visible from multiple angles at the 4 chamber view

APPROACH FOR EIF :

Perform a detailed USG looking for other markers

Other markers present

Readjust the risk ,
Genetic counselling
& definitive testing- Amnio

If isolated, then review maternal age & result of prior screening (1st or 2nd tri screening for Down's)

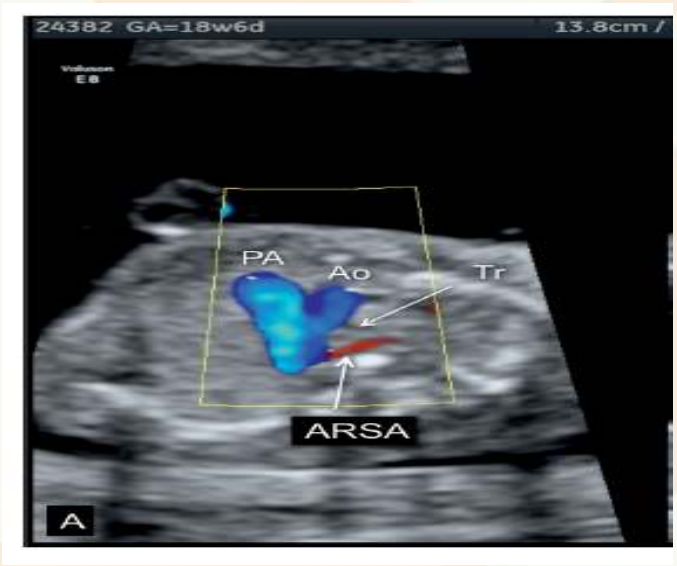
As risk of Down's with isolated EIF is only 1.5 times higher

a. Most cases will be low risk, no further work-up will be required.

b. If readjusted risk is ≥1:250 , consider genetic counselling & invasive testing or NIPT

ABERRANT RIGHT SUBCLAVIAN ARTERY (ARSA) :

- Occurs in the setting of a *left*-sided aortic arch
- When the last branch of the aortic arch, instead of being the left subclavian artery, is the right subclavian artery.
- ARSA crosses behind the trachea & esophagus & does not form a vascular ring
- Diagnosed on three-vessel & trachea view (3VTV) with color Doppler & low PRF
  - seen as a fourth vessel originating from a left aortic arch.
- In the 3VTV, no vessels should be seen behind the trachea.
- Prevalence of ARSA among fetuses with trisomy 21 ranges from 7.8% to 37.5%, with LR of 3.94
- Seen in 0.5% to 1.3% of the normal population



- ARSA – seen from junction of the aortic arch & ductus arteriosus with a course behind the trachea (Tr) toward the right clavicle and shoulder

WHAT NEXT ??

1. Do a detailed anatomical scan to look for other markers of aneuploidy

↓

Other markers present

↓

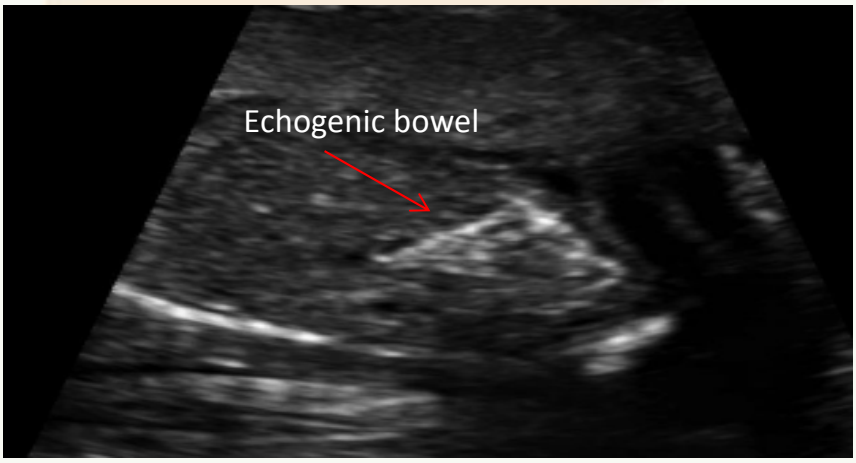
Readjust the risk ,Genetic counselling & Definitive testing- Amnio

If isolated ARSA

No prior screening , regardless of age:	Women with prior 1st tri or 2nd tri screening	Women with negative CVS Amnio or NIPT for Down's
Given the high +ve LRs genetic counselling & Amnio for fetal KT should be done	Readjust the risk based on previous testing, Genetic counselling & Amnio to be done	No further assessment

ECHOGENIC BOWEL :

- **Definition** - Bowel as bright or brighter than the surrounding bone
- Seen in 0.4 – 1.8 % of normal fetuses
- **Imaging essentials for diagnosis :**
  - Transducer with lowest frequency possible (5MHz or less)
  - Decrease gain , turn off harmonic imaging
  - Turn of any other digital imaging enhancers
- **Incidence** of aneuploidy in fetuses with isolated echogenic bowel - 3.3% to 16%



- **Chromosomal abnormalities** –mainly Down's (LR for Down's) – 1.65,but also Tri 18, 13 & Turner syndrome
- Cystic fibrosis ,Congenital intrauterine infections (TORCH, mainly CMV)
- Intrauterine growth restriction (IUGR) , Intrauterine fetal demise (IUD)
- GI abnormalities like bowel obstruction, meconium peritonitis
- Swallowing of amniotic fluid contaminated by blood- (e.g. following invasive procedure) can also lead to echogenic bowel in both euploid & aneuploid fetus .

APPROACH TO ECHOGENIC BOWEL :

Perform a detailed ultrasound

If other anomalies +nt

Readjust the risk
Genetic counselling,KT
Evaluation for congenital
Intrauterine infections

If isolated finding

a. Determine parental cystic fibrosis carrier status

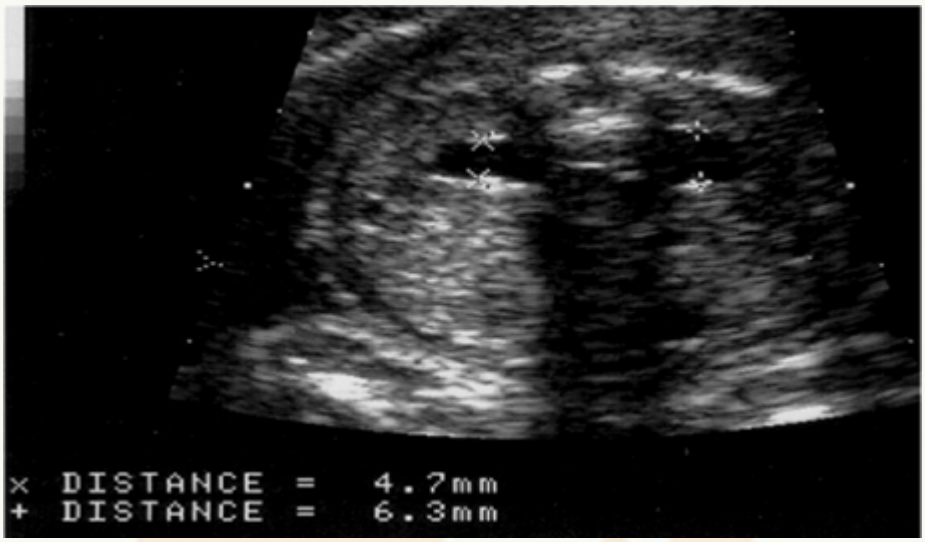
b. Assess prev. aneuploidy risk & offer Amnio if the readjusted risk is high

c. Evaluate for congenital infections particularly CMV

d. F/U assessment for
-Fetal growth,
-GI abnormalities ,
- ↑ risk of IUFD

e. Inform pediatrician at birth

RENAL PYLECTASIS :



- Also known as ***renal pelviectasis, full renal pelvis or renal pelvis dilatation***
- **Definition**- anteroposterior (AP) diameter of the renal pelvis on the transverse section/axial plan with spine either anterior or posterior
  - **≥ 4 mm to 6.9mm before 28 weeks (2nd tri)**
  - **≥7 mm to 10mm after 32 weeks (3rd tri**)
  - inner to inner measurement
- *Hydronephrosis* - when the AP diameter of the renal pelvis is ≥ 10 mm and/or calyceal dilatation is present.
- 0.6% to 4.5% - among normal fetuses, more common among male fetuses
- Has mild association with Down's , LR – 1.08
- Other associations are of urinary tract abnormalities & part of syndromes like VATER syndrome, prune belly disorder
- Postnatal pathologies associated with mild isolated pyelectasis :
  - ureteropelvic junction (UPJ) obstruction
  - vesicoureteral reflux
  - ureteral pathology
  -posterior urethral valves (0.2%).

Patients should be counselled that :

- Isolated mild pylectasis in 2nd tri-
  - will resolve by 32 weeks in 50 % of fetuses
  - may persist in 30% as mild pylectasis
  - 15% fetuses likely to progress to severe hydronephrosis

- Once pyelectasis is encountered, detailed USG should be done to look for -
  - presence of dilated ureters
  - appearance of the renal parenchyma/ cortico-medullary junction and calyces
  - bladder size and wall thickness
  - presence of an ureterocele
  - assessment of amniotic fluid volume

APPROACH TO MILD PYLECTASIS –

Perform a detailed ultrasound

If other markers/anomalies +nt

Readjust the risk
Genetic counselling
Definitive testing
(Amnio/NIPT)

If isolated pylectasis

a. Assess previous aneuploidy risk & offer genetic counselling & Amnio/NIPT if the readjusted risk is high

b. If low risk for chromosomal anomalies or prior normal CVS, Amnio or NIPT

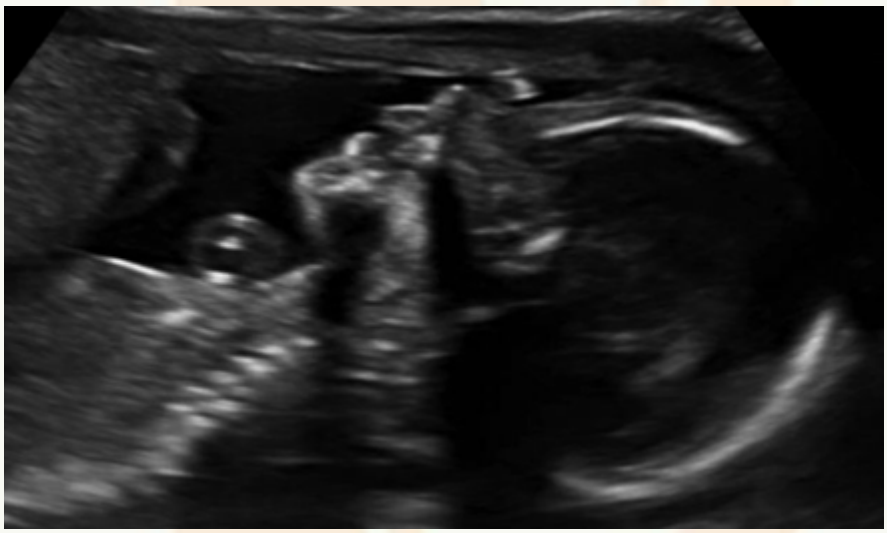
Re-evaluate at 32 weeks, if pylectasis persists or evolves hydronephrosis ,recommend postnatal evaluation atleast 7 days after delivery

ABSENT/HYPOPLASTIC NASAL BONE (NB) :

- **Definition - NB< 2.5th percentile or 5th percentile**
  Or NB< 2.5 mm or < 0.75 MoM
- **Strongest marker for Tri 21** (Down's), LR – 6.58

- Sensitive & specific marker for trisomy 21
- Size of NB varies with race & ethinicity
- Incidence of absent or hypoplastic NB in normal fetuses at 15-22 weeks among Asians – 2.8% .

IMAGING ESSENTIALS –



- **Correct plane**
- Should be seen in profile view
- Strict mid- sagittal plane
- Tip of nose should be visible
- Nasal bone should be more echogenic than skin
- Angle of insonation – close to 45 degree

- Essentially all pts with this finding, categorize as " screen positive" regardless of MA

WHAT NEXT ??

- Do a detailed anatomical scan to look for other markers of aneuploidy

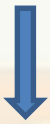
- Other markers present

- Readjust the risk ,Genetic counselling & Definitive testing- Amnio

If isolated absent/hypoplastic nasal bone

No prior screening ,

regardless of age:



Women with prior 1st tri or 2nd tri screening

Women with negative CVS Amnio or NIPT for Down's

Given the high +ve LRs genetic counselling & Amnio for fetal KT should be done

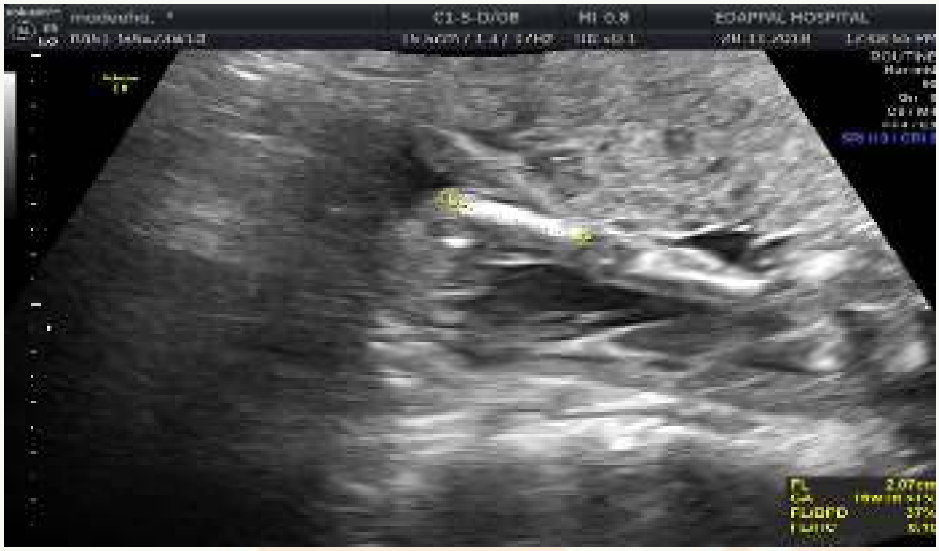
Readjust the risk based on previous testing, Genetic counselling & Amnio to be done

No further assessment

SHORT HUMERUS LENGTH (HL) / FEMUR LENGTH (FL) :

- 40- 50% of fetuses with Down's have short long bones
- **Definition** - FL or HL < 5th percentile for gestational age
- Shortening for HL is more sensitive & specific than FL
- Since both measurements are correlated, the Humerus length LR for Down syndrome is usually used
- Long bone length varies with ethnicity & race
- Therefore, short long bone - difficult marker to use
- in isolation, particularly in low-risk populations
- Also,short HL/FL has very minor association with Tri 21(Down's)
- **LR for Down's** - 0.61 for FL & 0.78 for HL
- - Also associated with
  - ✔ Fetal growth abnormalities
  - ✔ Skeletal dysplasias

SHORT LONG BONE – FEMUR



- **Imaging requirement**
  - Longest axis
  - 0-15 degree horizontal
  - Both ends of ossified metaphysis clearly visible
  - Distal femoral epiphysis to be excluded

WHAT NEXT ??

- Do a detailed anatomical scan to look for other markers of aneuploidy

↓

- Other markers present

↓

- Readjust the risk ,Genetic counselling & Definitive testing- Amnio

IF ISOLATED SHORT HL/FL :

- Measure all long bones to r/o skeletal dysplasia
- Assess the overall growth to r/o FGR/IUGR
- Isolated short HL/FL does not significantly changes the aneuploidy risk

↓

Invasive testing not required

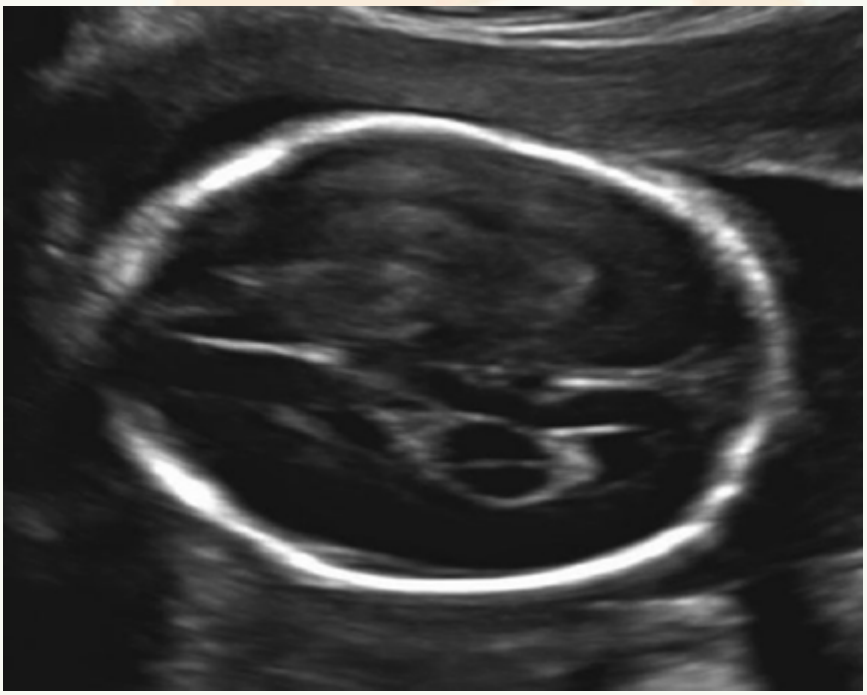
- Only f/u ultrasound to assess fetal growth in 3rd trimester

CHOROID PLEXUS (CP) CYST :

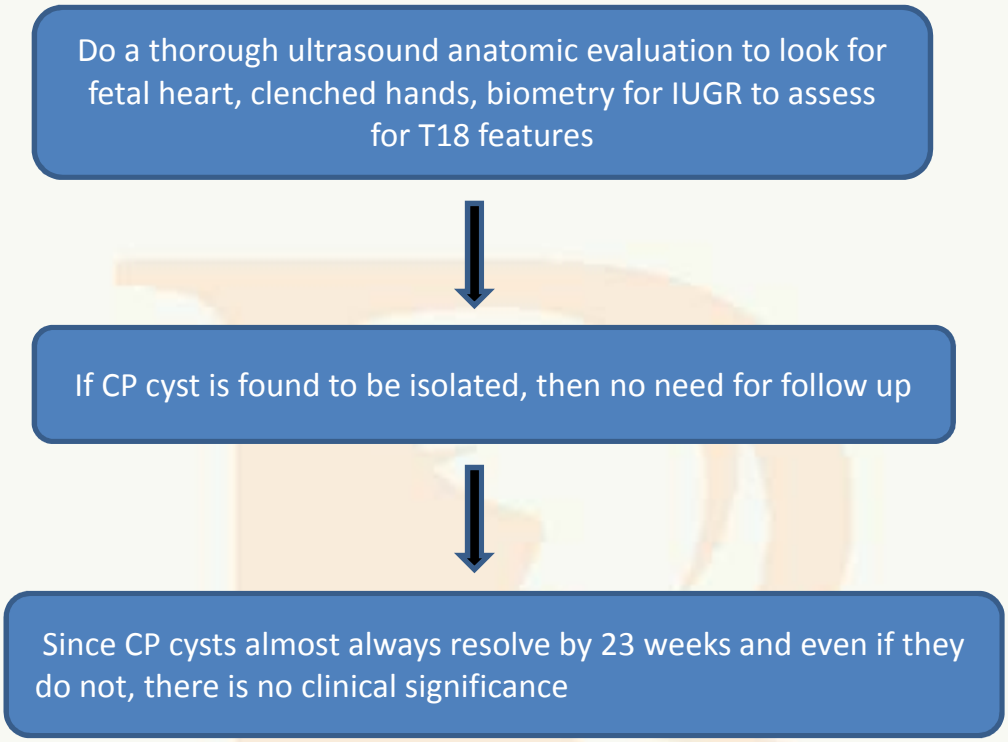
- **CP cyst** - small fluid-filled cystic structures seen within choroid plexus of the lateral ventricles of the fetal brain
- **US appearance** - echolucent cysts within the hyperechogenic choroid
  - single or multiple
  - unilateral or bilateral
  - less than 1 cm in diameter
- Seen in 0.3-3.6% fetuses during 2nd trimester ultrasound

ASSOCIATIONS :

- ***Not associated with DS***
- CP cyst is present in **30-50% of fetuses with Trisomy 18**
- T18- however is characterised by major structural heart defects, clenched hands, talipes deformity of feet, growth restriction & polyhydramnios
- When CP cyst is seen with structural anomaly probability of Trisomy 18 is 37%
- In the presence of isolated CP cyst – likelihood of T18 is extremely low



WHAT NEXT AFTER FINDING CP CYST ??



APPROACH FOR PATIENTS WITH CP CYST :

1. Perform a detailed ultrasound examination and fetal echocardiogram.

i. **If other anomalies present**- do genetic counselling and definitive testing.

ii. **If isolated**:

a. Maternal age < 35 years-old and trisomy 18 screen negative: no further genetic testing.

b. Maternal age < 35 years-old with no prior screening for trisomy 18 : additional genetic testing (e.g. NIPT or amniocentesis) may be considered for patients who are strongly motivated to pursue definitive diagnosis.

c. Maternal age 35 years-old and/or trisomy 18 screen positive: genetic counselling with consideration for amniocentesis or NIPT

LIMITATIONS OF SOFT MARKERS :

- Data is acquired from retrospective studies in tertiary centres (high prevalence)
- Lack of standardization in definitions
- Wide variation in population demographics
- Inconsistencies in study results

CONCLUSION :

- During 2nd trimester anomaly scan , thorough search for markers should be made
- Once a marker is found, detailed ultrasound to look for other markers should be carried out.
- Readjust the risk using LR
- Do a genetic counselling & give patient a cafeteria approach (choice of testing, it's pros & cons) for further management
- Patients should be informed that invasive testing is very safe in experienced hand with miscarriage rate is 0.11- 0.22%
- In the case of most isolated markers, including intracardiac echogenic focus, echogenic bowel, mild hydronephrosis & short femur, there is only a small effect on modifying the pre-test odds.

PRENATAL DIAGNOSTIC TESTS

-DR. KUNDA SHAHNE

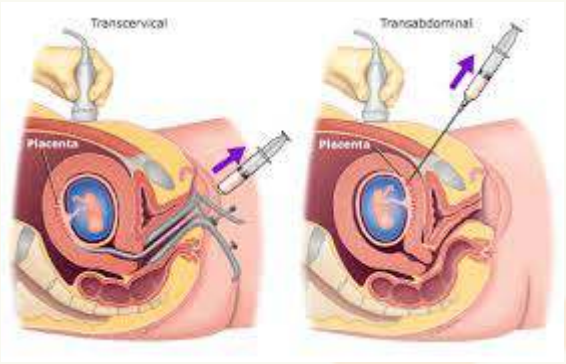
Prenatal Diagnostic Tests

Commonly practiced invasive prenatal diagnosis techniques are chorionic villus sampling (CVS), amniocentesis, less commonly cordocentesis or percutaneous umbilical blood sampling (PUBS), fetal tissue sampling. Invasive Testing can be performed in the first trimester by chorionic villus sampling or in the second trimester by amniocentesis; and have been the two most common prenatal diagnostic procedures for decades. Both procedures are safe, with and equivalent 0.5% risk of procedure-induced pregnancy loss. When performed prior to the routine sampling window of 15 weeks, amniocentesis may increase the risk of talipes equinovarus, the highest risk being encountered prior to 13 weeks' gestation. When chorionic villus sampling is performed prior to 9 weeks' gestation, there may be an increased risk of limb reduction defects. There are wide variations in utilization, operator skills, quoted procedure risks, actual observe risks and patient choices that come from highly variable counseling as to those risks. The laboratory analysis of both procedures is reliable.

Chorionic villus sampling has a 1-2% incidence of confined placental mosaicism, requiring additional evaluation in some cases. Most studies comparing CVS to amniocentesis in skilled hands have found equivalency of risks. Cordocentesis has fewer indications, is performed in the late second trimester of pregnancy but allows direct laboratory testing from fetal blood. Experienced operators should perform all invasive procedures under continuous ultrasound guidance. Patient counseling should include an evaluation of the procedural risk associated with each individual case with its background risk. In general, patients are allowed to resume most daily activities after the procedure.

- Invasive prenatal diagnosis procedures are to be done at the recognized place and person under PCPNDT only. Formal informed consent for invasive procedure should be obtained before the procedure.
- Forms F and G of the PNDT Act should be correctly filled, signed, and sent to the appropriate health authority.

CHORIONIC VILLUS SAMPLING




Chorionic villus sampling (CVS) is a test where a small piece of chorion frondosum (placentall tissue) is removed and used for genetic testing. Chorionic villus sampling (CVS) is the most common first trimester invasive prenatal diagnosis technique for evaluation of fetal Karyotype, molecular, and biochemical abnormalities. CVS should not be performed before 10 weeks gestation because of the risk of transverse limb reduction defects. The ideal timing is around 11 to 12 weeks after baseline NT scan.CVS should be performed or closely supervised by an operator using concurrent ultrasound. The operator should have adequate training and should-continue performing sufficient numbers annually to maintain expertise.

INDICATIONS FOR CVS:

• An abnormal first trimester screening by USG with/without serum biochemistry indicating increased risk for chromosome problems (Screen positive)

•Finding of fetal abnormality on ultrasound a previous child with a chromosome abnormality

• Parents carry a chromosome translocation (rearrangement) or evaluation for the single gene disorder like Thallasemia major, Tay-Sachs, sickle cell anemia, and DMD/CAH/CFetc.

•Skin disorders: epidermolysis bullosa dystrophica, albinism, lcthyosis

ADVANTAGES OF CVS :

An early result is advantageous for the patient, in that, in case of an unaffected pregnancy the anxiety is relieved and in cases of affected preqnancy early termination

of pregnancy can be undertaken with lower complication rate and less emotional stress than when termination follows amniocentesis at a later gestational age.

DISADVANTAGES & RISKS OF CVS :

A) Confined Placental Mosaicism - A discrepancy between the chromosomes in the chorionic and fetal tissues, is a biologic placental factor, which is present in 1% to 2% of pregnancies

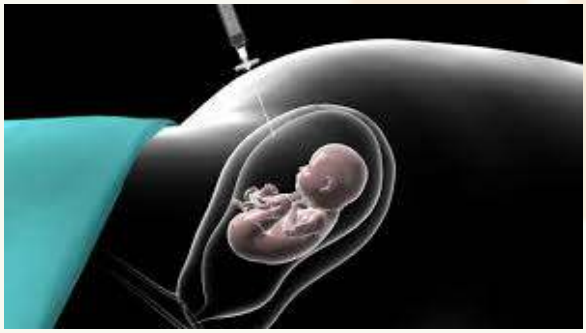
B) Maternal Contamination with decidual tissue

C) Pregnancy Loss In addition to the background risk of spontaneous pregnancy loss in the advanced maternal age group. The procedure related loss is about 1% to 2% in comparison to the0.5 to 1% risk for amniocentesis.

D) Limb or Facial Anomalies

The risk of limb or facial anomalies is higher if CVS is done at a gestational age earlier than nine weeks, hence universal CVS' is generally restricted to greater than or equal to 10 weeks. These anomalies may be due to a vascular disruption sequence event, which may be associated with the CVS procedure.

AMNIOCENTESIS :



Amniocentesis is usually performed for determination of fetal Karyotype, molecular, and biochemical abnormalities. The two most common tests performed on the amniotic fluid are the l fetal Karyotype from fetal and membrane cells in the amniotic fluid after tissue culturing or fetal chromosomal evaluation by direct fluorescent in situ hybridization (FISH) techniques.

Amniocentesis should be performed with concurrent ultrasound should be used Amniocentesis is usually performed from 16 weeks gestation and should not routinely be performed before 14 weeks gestation because of the increased risk of adverse outcome.

Some of the most common Indications for amniocentesis are:

For Chromosomal analysis in the fetus that is screen positive after USG and /or serum biochemistry

•A previous child with a chromosome abnormality or metabolic disorder ,One or both parents carry a chromosome translocation (rearrangement)

• Both parents carriers of a genetic disease such as Thalasemia minor, Tay-Sachs sckecll anemia, etc

•Finding of a fetal abnormality on ultrasound suggestive of Chromosomal anomaly

•Risk of fetal infection

•Sex determinations (only for X-linked disease, CAH, DMD)

•Biochemical disorders and inborn errors of metabolism screening in fetus.

•Study of micro deletions in fetus

RISKS OF AMNIOCENTESIS :

A) Fetal Loss

Fetal loss after amniocentesis is 0.5% to 1% above the background loss.

B) Infection

The risk of infection introduced at the time of the amniocentesis is estimated to be 1 to n 3000 procedures.

C) Fetal Injury

Serious fetal injuries at the time of amniocentesis are rare with continuous ultrasound guidance.

D) Other Complications include leakage of amniotic fluid, bleeding, and uterine irritability. These complications are estimated to occur in 1% of procedures and are generally self limited.

Comparing various approaches of prenatal diagnostic techniques: A. Transabdominal.CVS versus second trimester amniocentesis:

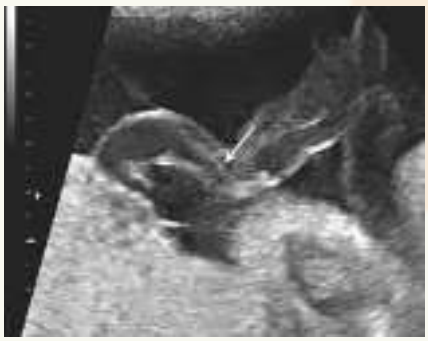
A subgroup of Denmark compared Transabdominal CVS with second trimester amniocentesis and found no significant difference in the total pregnancy loss between the two procedures (6.3% versus 7%; RR 0.90: 95% CI 0.66 to 1.23).

B. **Transabdominal versus Trans cervical CVS**:

Compared with Transabdominal CVS, total pregnancy loss and spontaneous miscarriages were higher after transcervicalCvs. Vaginal bleeding following the procedure was much more common after transcervical CVS, though there was no difference in the incidence of vaginal bleeding later in pregnancy There was no significant difference in the amniotic fluid leakage i following the procedure and prelabour spontaneous rupture of membranes before 28 weeks. i C. Early amniocentesis (EA) versus Transabdominal CVS:

Spontaneous miscarriages after early amniocentesis are more common

CORDOCENTESIS :



Fetal blood sampling provides information that is not obtainable by other techniques for fetal assessment. It is performed after 20 weeks of gestation when umbilical cord is accessible .It has tremendous fetal diagnostic and therapeutic applications, and exciting research potential. It allows the direct estimation of fetal hemoglobin, hematocrit, blood group, platelet count, reticulocyte, and white blood cell count for prenatal

diagnosis of fetal anemia, thrombocytopepia, etc/Cord blood gives a better and quicker chromosomal preparation than with chorionic villi or amnnotic fluid. Congenital Infections can be diagnosed by serology, direct identification of the viral particles by electron microscopy of fetal blood, cultures of fetal blood and indirect parameters like platelet count, total leukocyte count, differential count, and liver enzymes are carried out to arrive at a diagnosis. Fetal blood sampling (direct ultrasound guided fetal blood sampling) should also be performed or closely supervised by operators trained in this procedure who perform a sufficient number of such samplings to ensure technical success (i.e. Sampling fetal blood), and to minimize the complication rate.

INDICATIONS:

• Rh immunization- Hb, Blood Group, Intra Uterine Transfusion

•Rapid fetal Karyotype (late pregnancy)

•Hematology-Hb, Factor VIII, IX deficiency. Platelets

•Congenital infections- PCR, IgM (TORCH), parvovirus

•All indications similar to amniocentesis

Indications are decreasing as prenatal diagnosis of these conditions can now be done by CVSor by amniocentesis.

The procedure related fetal loss rate for cord blood sampling is 1 to 2.6 %. The overall mortality (including background morbid condition of a diseased fetus) appears to be around 5.0% (between 3.84 and 5.87%), but fetal loss rate directly related to the procedure seems to be around 1% (between 0.88 and 0.98%) only, Fetal loss rate is closely related to the state of the l fetus and indication of the procedure.

Transient bradycardia varying from 15 to 134 seconds may be seen in 3 to 9 percent. Complications and success in obtaining the blood sample depends on the experience of the operator.

Rhesus status

Rhesus status should be available or obtained in every case before the prenatal invasive diagnosis. Anti-D Ig should be given to all non-sensitized RhD negative women with Rh positive husband after the invasive prenatal diagnosis like amniocentesis, chorion vllus

sampling. Fetal blood sampling and other intrauterine procedures e.g. insertion of shunts, embryo reduction. A dose of 50 mcg is recommended for prophylaxis following sensitizing events up to 20 weeks of pregnancy and for all events after 20 weeks, at least 100mcg anti-D lg should be given followed by a test to identify Feto Maternal Hemorrhage. Final dose has to be calculated after the quantification of Feto Maternal Hemorrhage. Feto Maternal Hemorrhage l greater than 4ml red cells of fetal blood, additional anti-D lg should be given as per requirement.

CONCLUSION:

Amniocentesis and CVS are very useful techniques for Fetal care, quite safe Obstetric Procedures in expert hands, with backup requirement of a good Genetic Laboratory. They require skill, and should preferably be done in referral centers to maximize safety, and optimize patient management. In addition, there is need for improved and more specific noninvasive screening methods to identify women whose fetuses are at risk of congenital or genetic disease, to minimize number of women requiring PND procedures. Obstetricians play a key role in Prenatal Diagnostic and Genetic services, by screening, counseling, and timely referrals. General principles for prenatal diagnosis programs

1. All patients considering prenatal diagnosis should have access to professionals who are knowledgeable in the field appropriately minimally trained for doing the procedures. The prenatal diagnostic service units should use state of the art ultrasound equipment. Each specialized prenatal diagnostic service requires the services of a multi-disciplinary team of a specialist in obstetric ultrasound, clinical geneticist, genetic counselor, obstetrician (with specializing in prenatal diagnosis and management of fetal abnormality prefered,pediatrician, pediatric surgeon and laboratory. There should be at least one specialized prenatal diagnostic service center for all states of India.

2. A suggested minimum caseload of 50 invasive procedures per year is recommended per practitioner and 100 prenatal specimens for the genetic lab in order to maintain an appropriate level of competence. Exceptions to this minimum caseload may be justified because of unique geographic circumstances.

3. Each patient should have an appropriate assessment of family history and genetic counseling i prior to undergoing invasive prenatal diagnosis.

4. Counseling should be given in a non-directive manner in order to allow an informed choice by the couple.

5. The distinction between screening and diagnostic investigations should be clarified, including the frequency of abnormal results, false positive and false-negative tests. Accuracy of results, frequency of need for repeats testing, and risks of pregnancy loss are of particular relevance with invasive prenatal diagnosis procedures. The couple should be reminded that normal test results do not rule out every genetic or structural abnormality in their fetus.

6. Prior to embarking on prenatal diagnosis testing, couples should be made aware of the full range of options when confronted with an abnormal test result. Prior commitment to termination of pregnancy following the diagnosis of fetal abnormality is not a prerequisite for prenatal diagnosis. Each centre must be aware of the local, regional, national, and international policies and protocols related to termination of pregnancy and advise the couple of such before undertaking prenatal diagnosis. This is particularly important at gestations beyond 20 weeks.

7. Determination of fetal sex for the purpose of sex selection procedures on a non-medical basis is inappropriate and against the law. Genetic lab should not carry out the testing for sex determination unless indicated for the sex link genetic disorders.

8. When a fetal anomaly is found, a multi-disciplinary group should be involved for the management of the patient and the fetus

FETAL CENTRAL NERVOUS SYSTEM

- DR AMEE RAHATEKAR

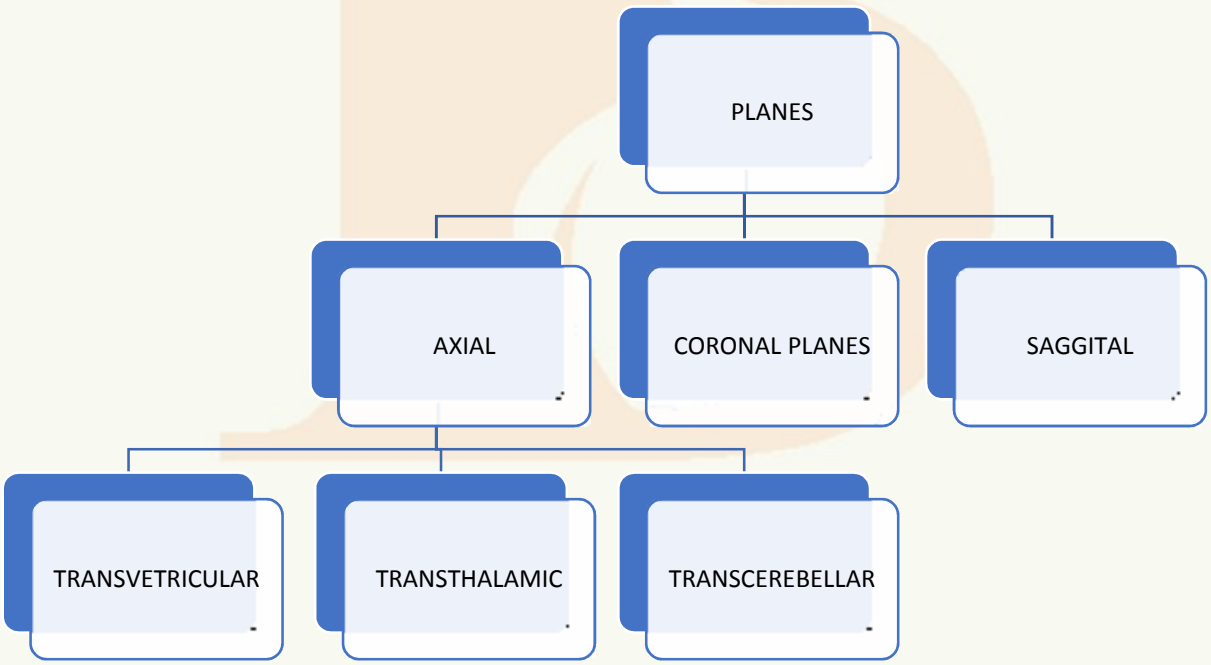
CNS EXAMINATION

The CNS examination should include evaluation of the fetal head and spine.

Ultrasound screening for fetal brain malformations is commonly performed at 19–21 weeks' gestation.

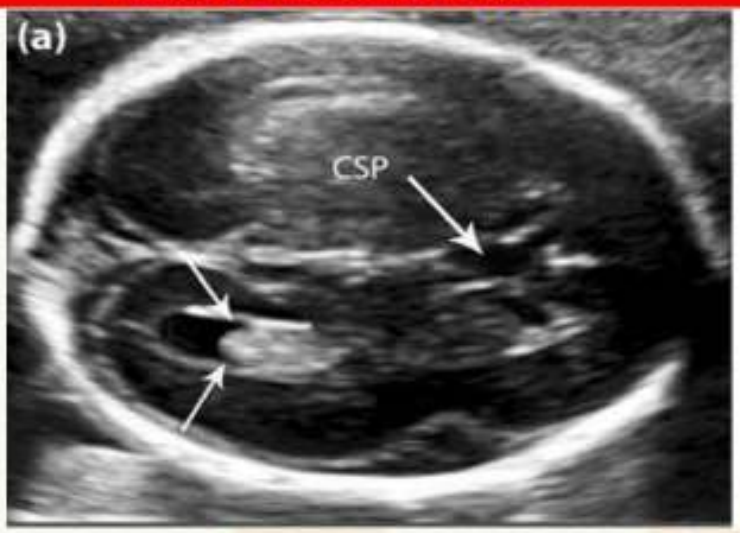
The second-trimester anomaly scan includes the following axial planes: transthalamic, transventricular, and transcerebellar.

Coronal and sagittal views may become necessary in a targeted examination, known as *fetal neurosonogram*, in patients with an increased risk of CNS anomalies

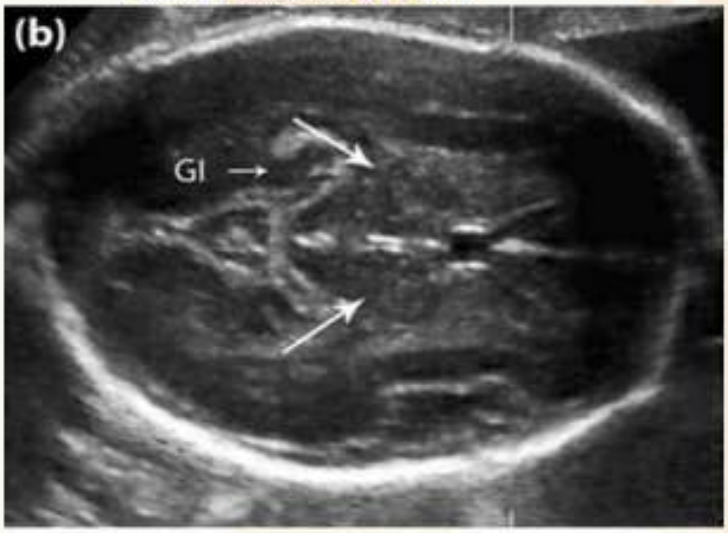


AXIAL PLANES

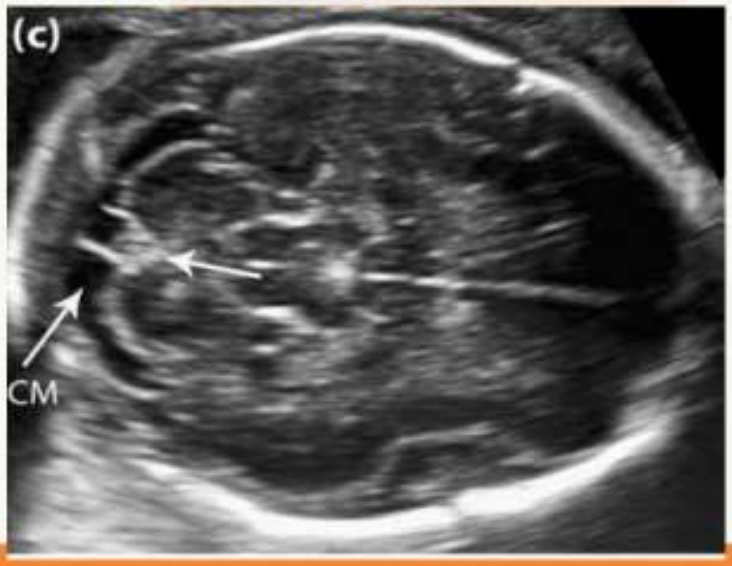
TRANSVENTRICULAR PLANE



TRANSTHALAMIC PLANE

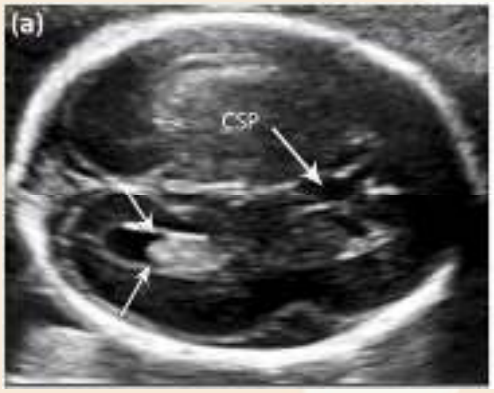


TRANSCEREBELLAR PLANE

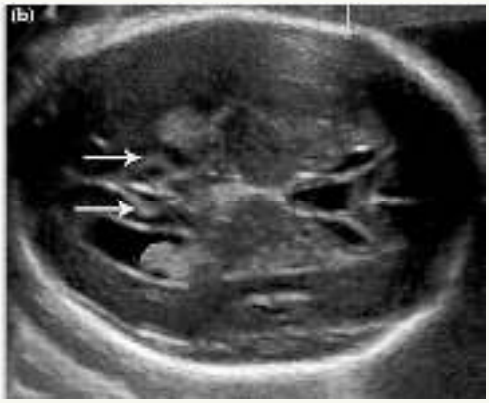


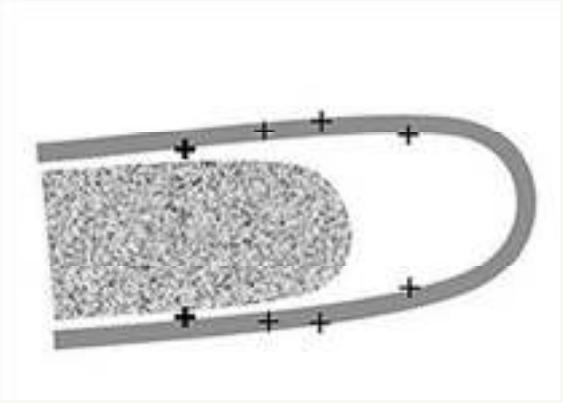
AXIAL TRANSVENTRICULAR VIEW

- This is the most cephalad axial scan plane of the fetal head.
- It allows visualization of the sonolucent lateral ventricles with the echoic choroid plexuses, filling the ventricular bodies and atria
- They are separated medially by the cavum septi pellucidi (CSP).
- The CSP is a fluid-filled cavity between two thin membranes.

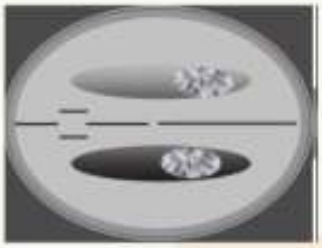


- To exclude the presence of ventriculomegaly (VM), the width of the atria is measured at the level of the glomus. The measurement is made perpendicular to the ventricular axis by positioning the calipers on the inner sides of the echogenic ventricular walls (inner to inner borders and should be less than 10 mm.
- The site for the measurement, at the level of the glomus, is chosen because, regardless of the etiology, dilatation of the lateral ventricle generally involves the caudal portion (atrium and posterior horn) first [2].
- The 10 mm cutoff applies throughout gestation; any measurement of 10 mm or more indicates the presence of VM

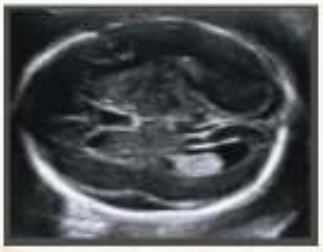


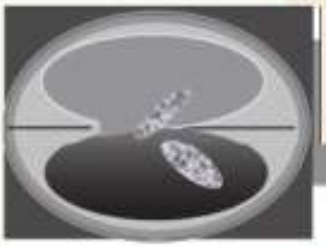


Ventricular anomalies
(Trans-ventricular scanning plane)

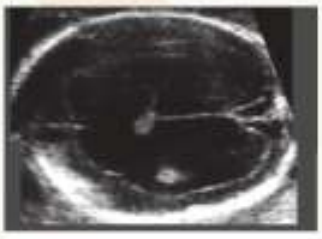


Normal

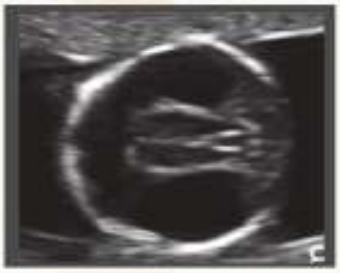




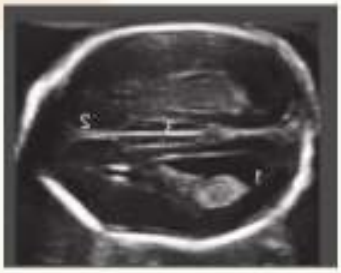
Biventricular ventriculomegaly

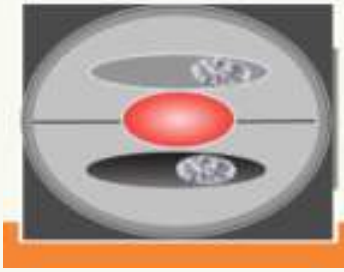


Holoprosencephaly

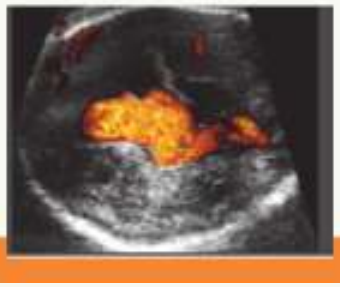


Corpus callosum agenesis



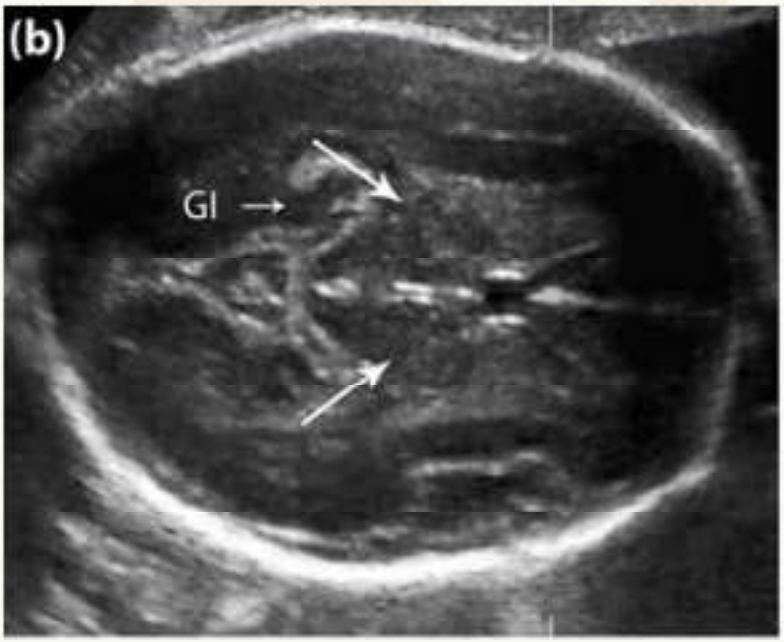


Vein of Galen aneurysm

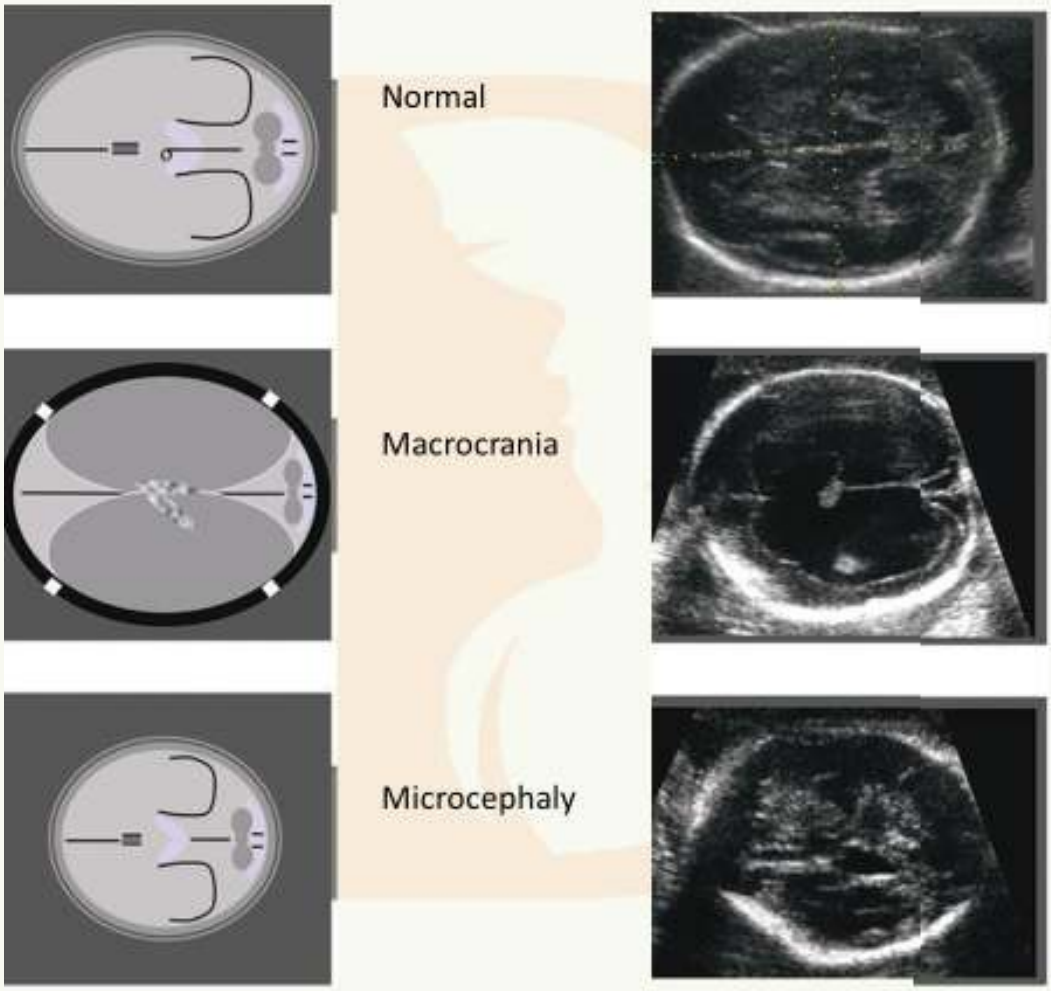


AXIAL TRANSTHALAMIC VIEW

- The axial transthalamic view is the classic plane in which the biparietal diameter (BPD) and the fetal head circumference (HC) are measured. On this view, the CSP, the thalami, and the symmetry of the cerebral hemispheres can also be assessed.
- This plane helps in detection of-
- **microcephaly**, which is characterized by a marked reduction of the fetal HC, and
- **hemimegalencephaly** in which the two cerebral hemispheres are of different size,
- Some skull deformities, due to **craniosynostoses** or to other anomalies of the CNS, can also be more easily recognized.

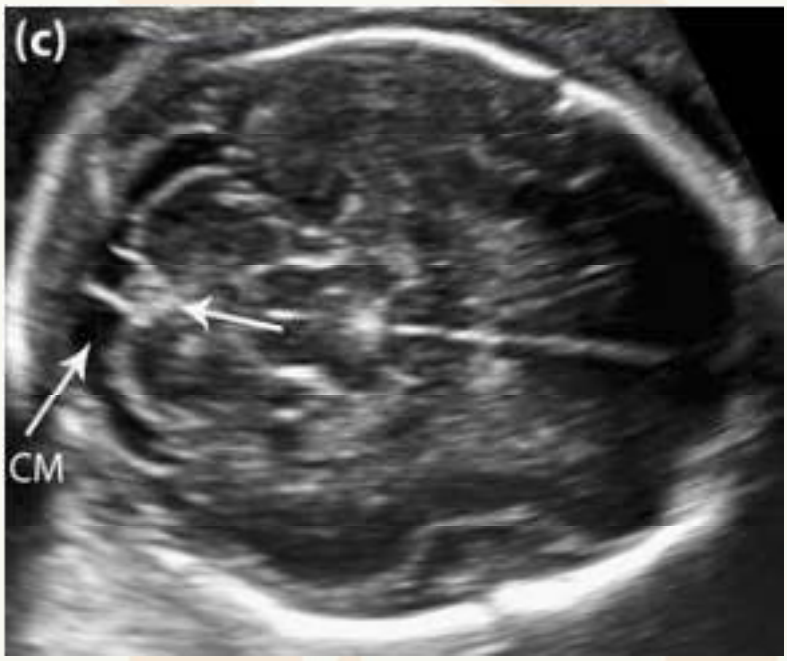


Head and skull anomalies
(Trans-thalamic scanning plane)

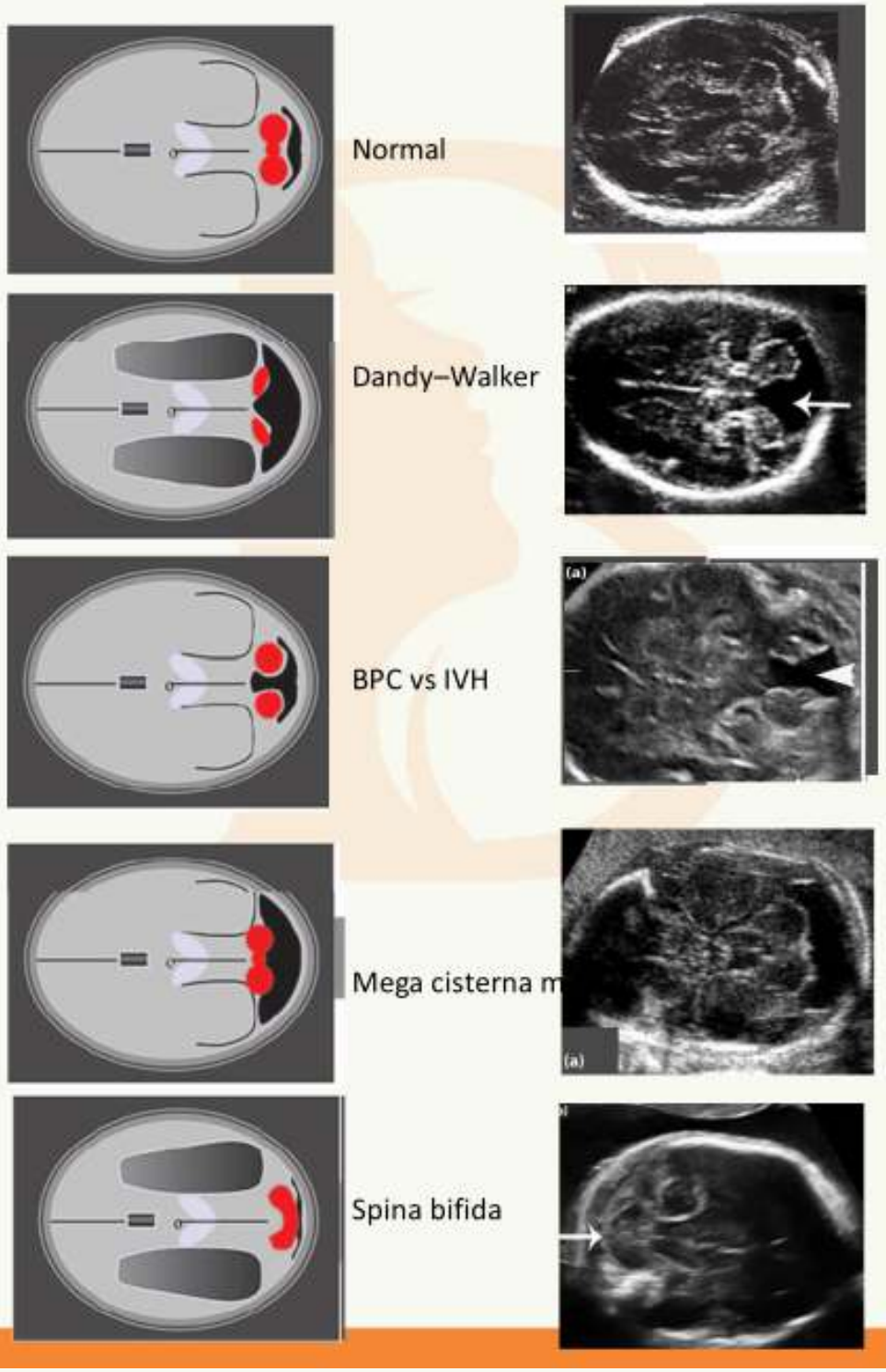


AXIAL TRANSCEREBELLAR VIEW

- This plane is obtained at a slightly lower level than that of the transventricular plane and with a slight posterior tilting.
- This view is used to assess the posterior cranial fossa and the related structures, namely the cerebellum (including the vermis) and the cisterna magna



Cerebellar anomalies
(Trans-cerebellar scanning plane)



COMMONLY DETECTED LESIONS OF SKULL AND BRAIN AND THEIR FURTHER MANAGEMENT

- ACRANIA
- VENTRICULOMEGALY
- DANDYWALKER MALFORMATION
- ARNOLD CHIARI MALFORMATION
- CHOROID PLEXUS CYST
- HOLOPROSENCEPHALY
- MICROCEPHALY
- BLAKE POUCH CYST
- MEGACISTERNA MAGNA

ACRANIA

- **Prevalence:**

1 in 1,000 at 12 weeks' gestation.

- **Ultrasound diagnosis:**

Absence of cranial vault and cerebral hemispheres.

At 12 weeks acrania is suspected by absence of the normally ossified skull and distortion of the brain (exencephaly). At >16 weeks the brain is destroyed (anencephaly).

- **Associated abnormalities:**

Chromosomal defects in isolated acrania are rare.

CNS or other defects are found in about 50% of cases, including spina bifida in 25%.

- **Investigations:**

Detailed ultrasound examination.

- **Follow up:**

If the pregnancy continues, follow-up should be standard.

- **Delivery:**

Standard obstetric care and delivery.

- **Prognosis:**

Lethal condition with death within the first week of life.

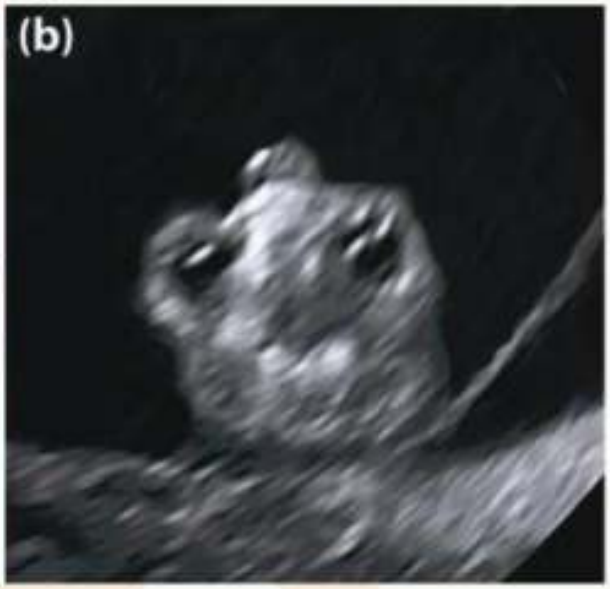
- **Recurrence:**

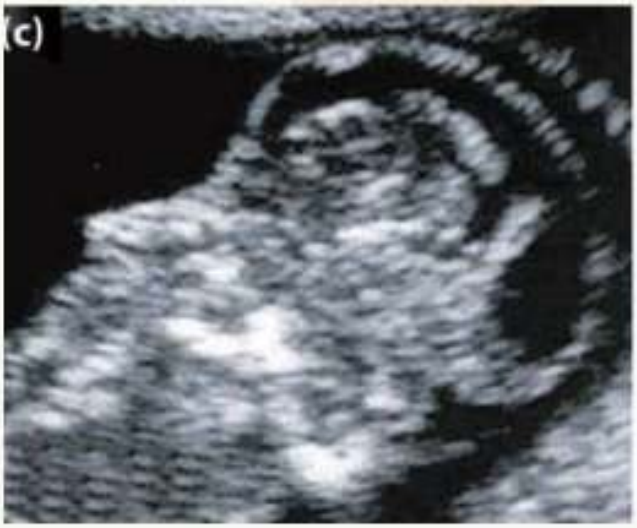
One previous affected sibling: 5%.

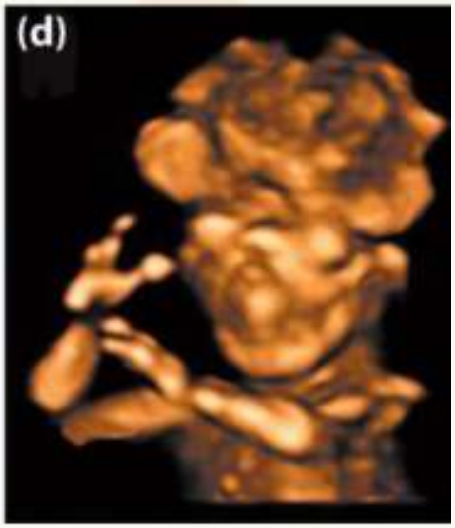
Two previous affected siblings: 10%.

Supplementation of the maternal diet with folate (5 mg/day) for 3 months before and 2 months after conception reduces the risk of recurrence by about 75%.

ACRANIA







VENTRICULOMEGALY

- **Prevalence:**

1 in 100 fetuses at 20 weeks' gestation.

1 in 1,000 births.

- **Ultrasound diagnosis:**

Bilateral or unilateral dilation of the lateral cerebral ventricles observed in the standard transverse section of the brain.

Subdivided according to the diameter of the lateral ventricle into mild (10-12 mm), moderate (13-15 mm) and severe (>15 mm).

- **Associated abnormalities:**

Chromosomal defects, mainly trisomies 21, 18 or 13, are found in 10% of cases. In isolated ventriculomegaly there is a 4-fold increase in risk for trisomy 21. The risk is inversely related to the severity of ventriculomegaly.

Cerebral and non-cerebral defects and genetic syndromes are found in 50% of cases.

- **Investigations:**

Detailed ultrasound examination, including neurosonography.

Invasive testing for karyotyping and array.

TORCH test for fetal infections.

Maternal blood testing for antiplatelet antibodies in cases with evidence of brain hemorrhage.

Fetal brain MRI at ≥32 weeks for diagnosis of abnormalities of neuronal migration, such as lissencephaly.

- **Follow up:**

Ultrasound scans every 4 weeks to monitor the evolution of ventriculomegaly.

- **Delivery:**

Standard obstetric care and delivery.

Cesarean section if the fetal head circumference is >40 cm.

- **Prognosis:**

Isolated mild / moderate: neurodevelopmental delay in 10% of cases, this may not be higher than in the general population.

Isolated severe: 10 year survival 60%, severe mental handicap 50%.

- **Recurrence:**

Isolated <1%. Increases to 5% if there is a history of affected fetus or sibling.

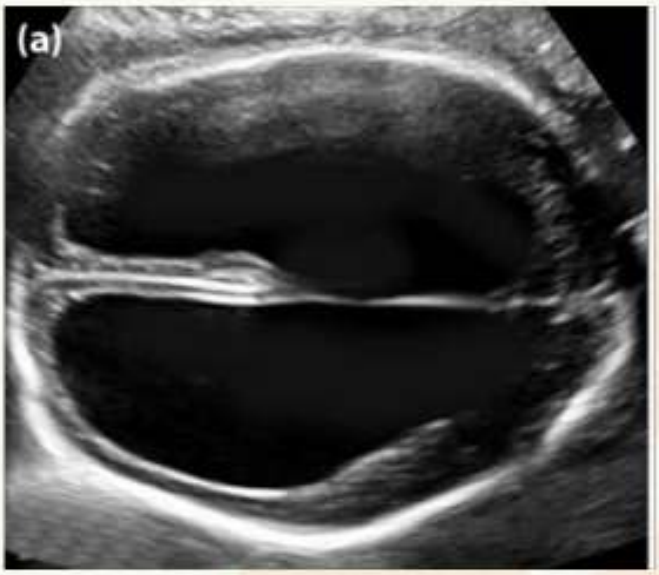
Part of infection: no increased risk.

Part of trisomies: 1%.

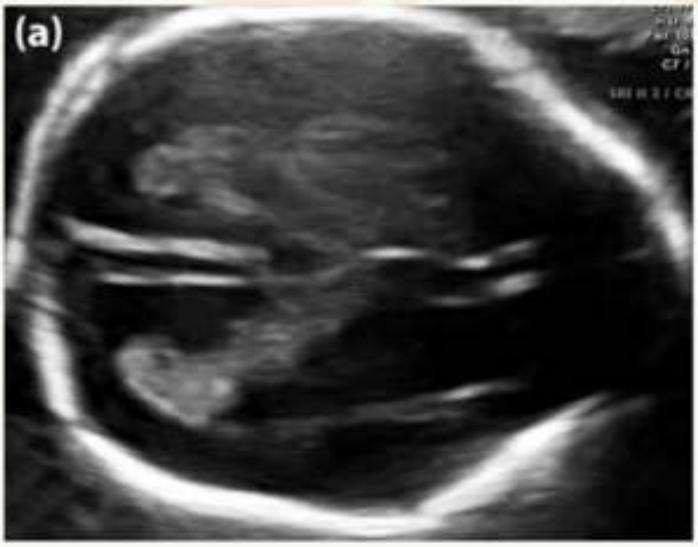
X-linked hydrocephaly: 50% of males.

Associated with alloimmune thrombocytopenia and no treatment: 100%.

SEVERE VENTRICULOMEGALY



MILD VENTRICULOMEGALY



DANDYWALKER MALFORMATION

- **Prevalence:**

1 in 30,000 births.

- **Ultrasound diagnosis:**

Cystic dilation of the fourth ventricle that fills the posterior fossa and extends into the cisterna magna.

Hypoplasia or complete agenesis of the cerebellar vermis.

- **Associated abnormalities:**

Chromosomal defects, mainly trisomies 13 or 18, are found in about 30% of cases.

Genetic syndromes (most common: Walker–Warburg syndrome, Meckel–Gruber syndrome, Aicardi syndrome, Neu–Laxova syndrome) and defects (brain, heart, gastrointestinal and genitourinary) are found in >50% of cases.

Severe ventriculomegaly is common and postnatally develops in >80% of cases.

- **Investigations:**

Detailed ultrasound examination, including neurosonography.

Invasive testing for karyotyping and array.

Fetal brain MRI at ≥32 weeks' gestation for the diagnosis of neuronal migration disorders.

- **Follow up:**

Ultrasound scans every 4 weeks to monitor possible development of severe ventriculomegaly.

- **Delivery:**

Standard obstetric care, but delivery in a hospital with neonatal intensive care.

Cesarean section if the fetal head circumference is >40 cm.

- **Prognosis:**

Isolated: neurodevelopmental delay in >50% of cases.

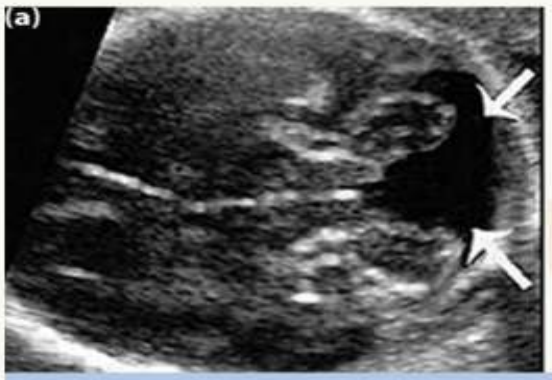
Severe ventriculomegaly: mortality rate >50% and neurodevelopmental delay in most survivors.

- **Recurrence:**

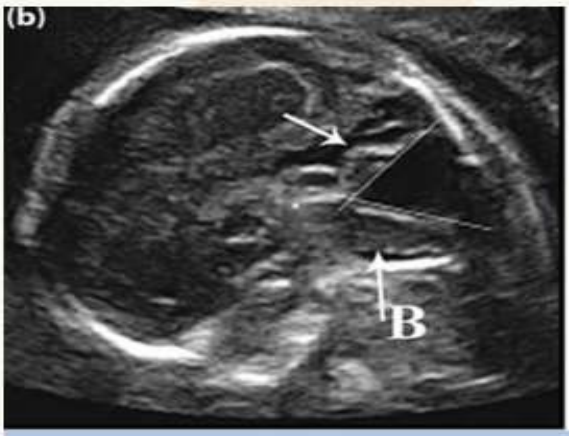
Isolated: 3-5%.

Part of trisomies: 1%.

DANDYWALKER MALFORMATION



Axial view of the posterior fossa showing a CSF collection (arrows) and a V-shaped cerebellum.



Midsagittal view of the posterior fossa showing an upward displacement of the small vermis (arrow) and an open fourth ventricle, communicating with the cisterna magna.

ARNOLD CHIARI MALFORMATION

- **Incidence.**

  1 in 1000 at birth.

- **Ultrasound diagnosis.**

  Indirect cerebral signs: Lemon sign (frontal bossing), banana sign (cerebellar dysmorphism), and effacement of the cisterna magna.

  Direct signs: "C" or "U" shape of the affected vertebra, which is due to absence of the dorsal arches; interruption of the cutaneous contour with/without a meningocele; splaying of the lateral processes.

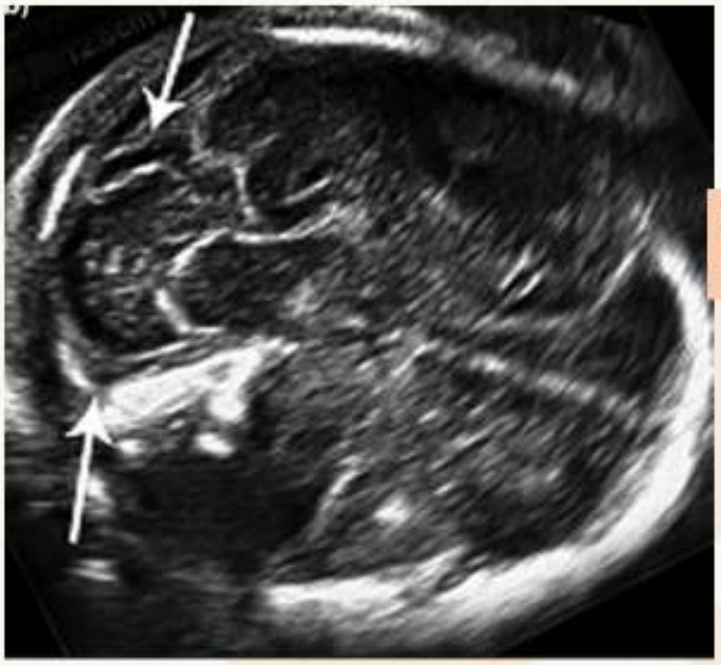
- **Risk of chromosomal anomalies.** Relatively high at 8%–16%.

- **Risk of nonchromosomal syndromes.** Low.

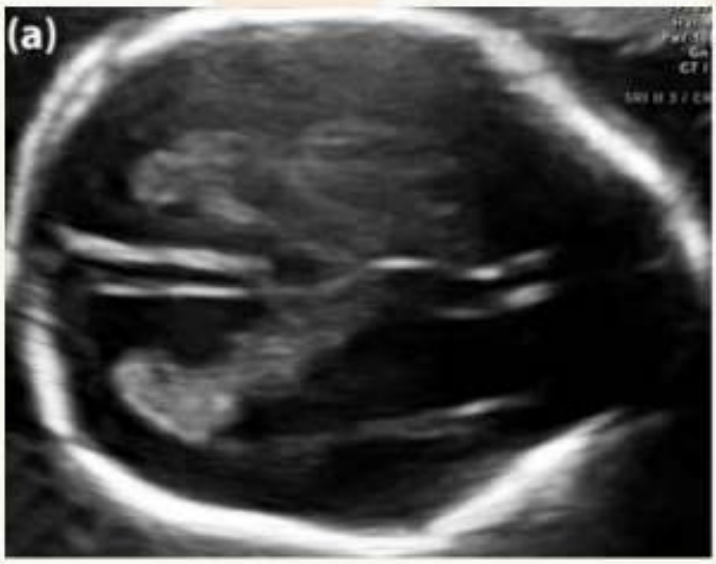
- **Outcome**. Poor

  14% to 20% of children with open spina bifida die within the 5 years of life due to shunt complications, in a significant number of cases. If brainstem disfunction related to Chiari II malformation is present the mortality rate rises to 35–40%. Motor disability depends on the level and complexity of the spinal defect

ARNOLD CHIARI MALFORMATION

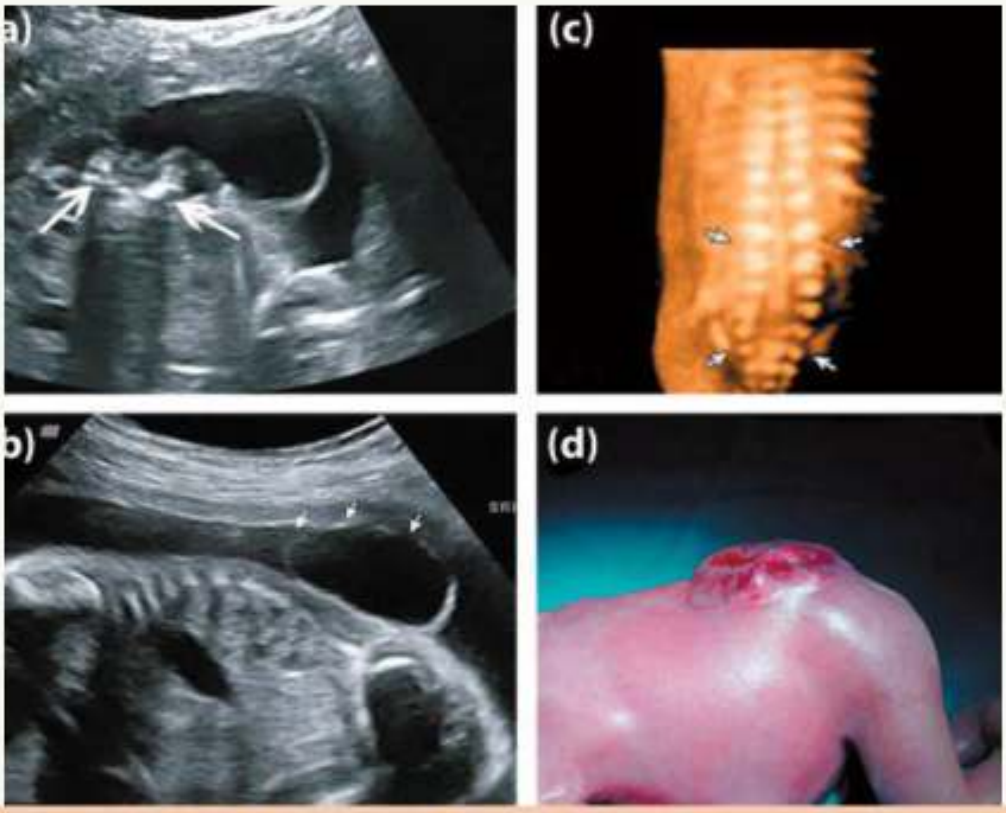


Banana cerebellum



Lemon shaped skull- scalloping of frontal bones due to drainage of csf from open spinal defect

ARNOLD CHIARI MALFORMATION



Axial scan of the lumbosacral spine demonstrating the open vertebra (arrows) and membranous coverage of the meningocele. Sagittal view of the lumbar spine showing the bulging membranes of the myelomeningocele (arrows)

CHOROID PLEXUS CYST

- **Prevalence:**

1 in 50 fetuses at 20 weeks' gestation.

More than 90% resolve by 26 weeks.

- **Ultrasound diagnosis:**

Single or multiple cystic areas (>2 mm in diameter) in one or both choroid plexuses of the lateral cerebral ventricles.

- **Associated abnormalities:**

Associated with increased risk for trisomy 18 and possibly trisomy 21.

- **Investigations:**

Detailed ultrasound examination for presence of other markers of trisomies 18 and 21. In the absence of other markers there is no need for invasive testing.

- **Follow up:**

  Follow-up should be standard.

- **Delivery:**

Standard obstetric care and delivery.

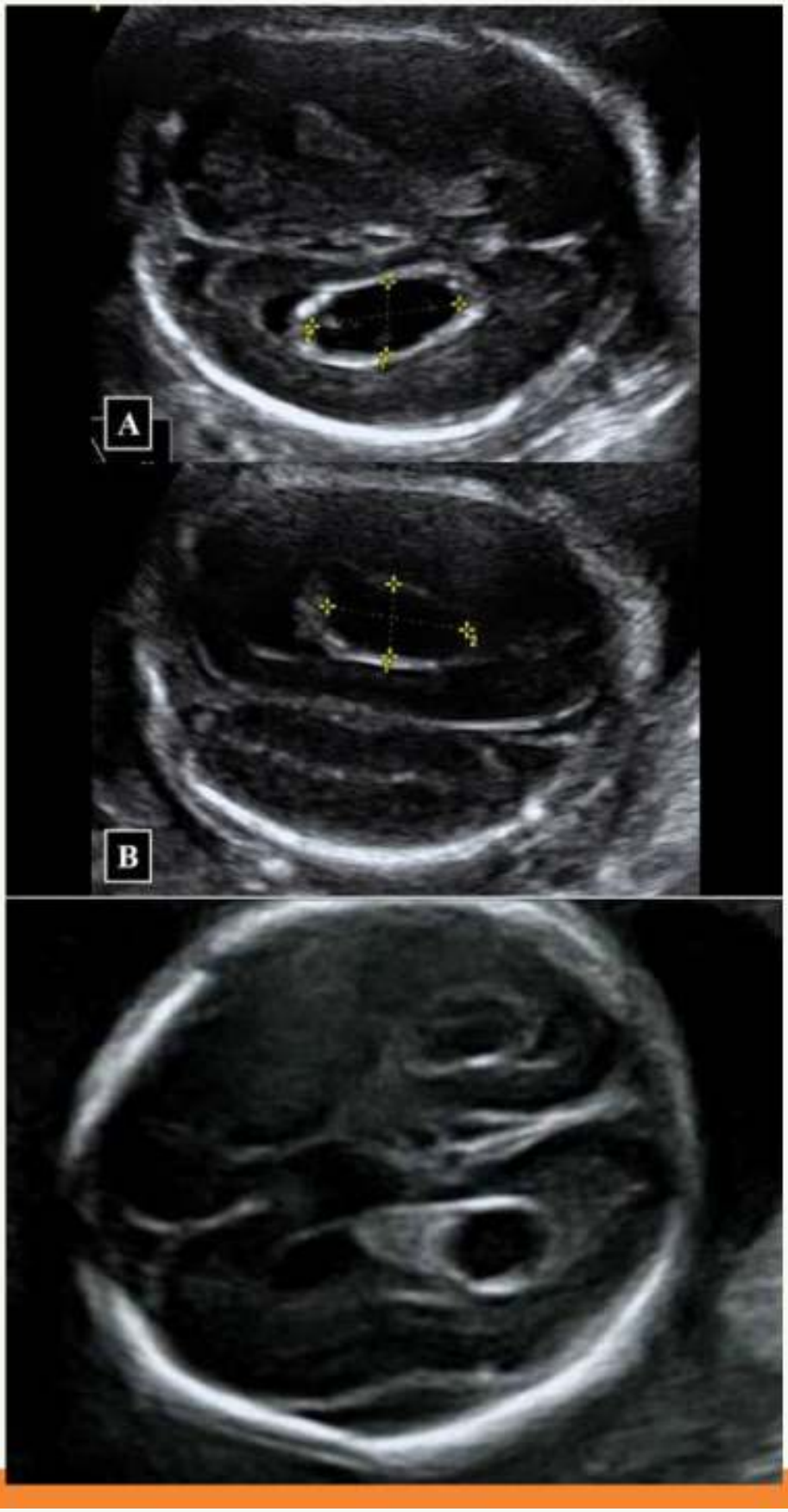
- **Prognosis:** Good.

- **Recurrence risk:**

Isolated: no increased risk of recurrence.

Part of trisomies: 1%.

CHOROID PLEXUS CYST



HOLOPROSENCEPHALY

- **Prevalence:**

1 in 1,300 fetuses at 12 weeks' gestation.

1 in 10,000 births.

- **Ultrasound diagnosis:**

Abnormalities from incomplete cleavage of the forebrain observed in the standard transverse sections of the brain.

There are 4 types:

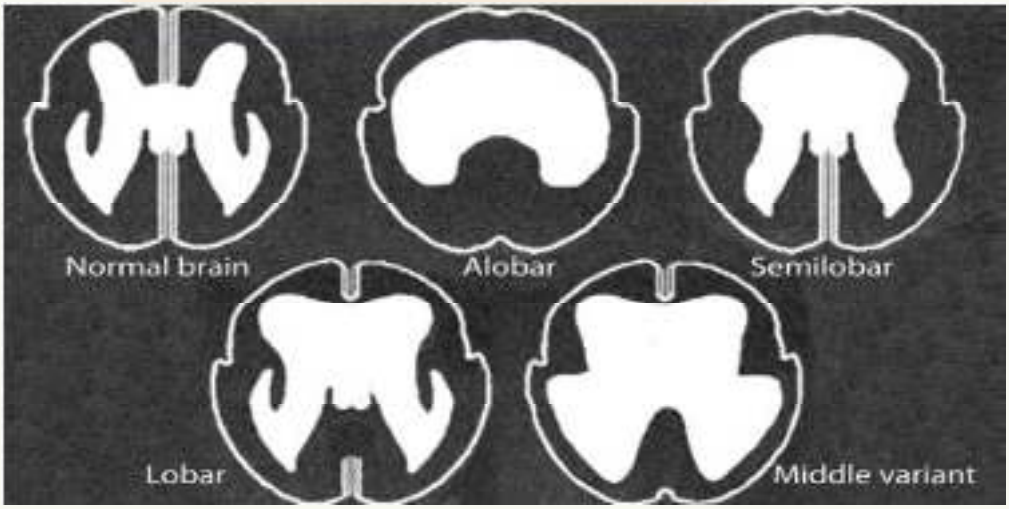
Alobar: fusion of the cerebral hemispheres with a single ventricle.

Semilobar: cerebral hemispheres and lateral ventricles are fused anteriorly but separated posteriorly.

Lobar: cerebral hemispheres are separated both anteriorly and posteriorly, but there is partial fusion of the frontal horns of the lateral ventricles, absence of septum pellucidum and abnormalities of the corpus callosum, cavum septum pellucidum and olfactory tract. The main differential diagnosis is septo-optic dysplasia and therefore an attempt should be made to examine optic chiasm and optic nerves by MRI.

Syntelencephaly, the anterior and occipital areas of the brain are fully cleaved as in the lobar type, but unlike this, there is no parietal cleavage and therefore the Silvian fissures are vertically oriented and abnormally connected across the midline over the vertex of the brain.

Lobar holoprosencephaly is detectable at ≥18 weeks' gestation, but the other three types can be detected at the 11-13 weeks scan.



TYPES OF HOLOPROSENCEPHALY

- **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.

- **Investigations:**

Detailed ultrasound examination, including neurosonography.

Invasive testing for karyotyping and array.

Fetal MRI may be useful for confirmation of diagnosis in cases of suspected lobar holoprosencephaly.

- **Follow up:**

If pregnancy continues, follow-up should be standard.

- **Delivery:** Standard obstetric care and delivery.

- **Prognosis:**

Alobar and semilobar: usually lethal within the first year of life.

Lobar: life expectancy may be normal but usually with severe developmental delay and visual impairment.

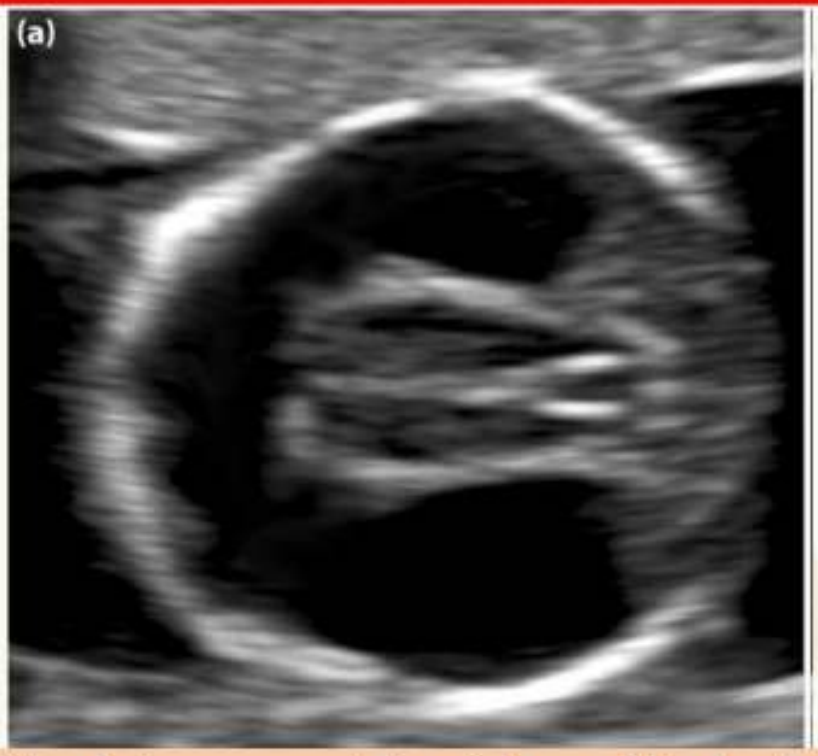
- **Recurrence:**

Isolated: 6%.

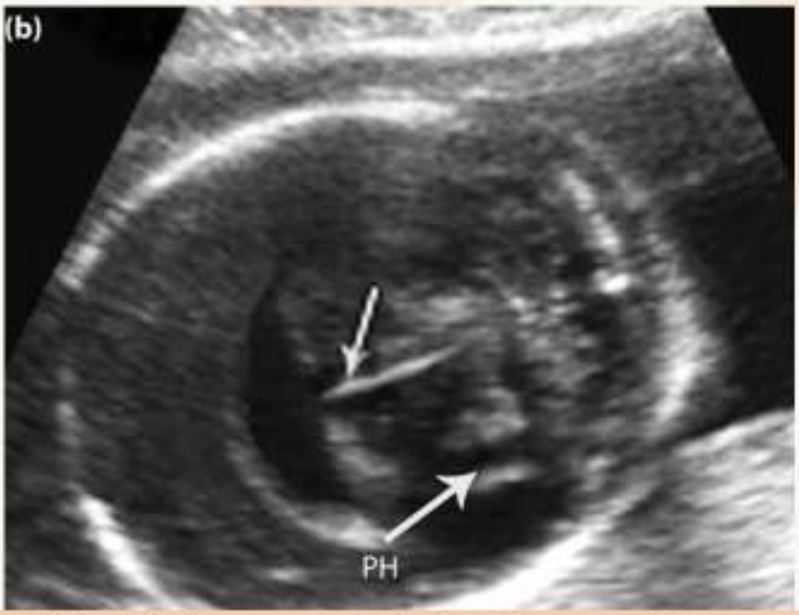
Part of trisomies: 1%.

Part of genetic syndromes: 25-50%.

HOLOPROSENCEPHALY



Alobar holoprosencephaly: axial scan at the level of the thalamus showing the single ventricle, absence of midline structures, and fused thalamus;



Semilobar holoprosencephaly: ultrasound image showing the tw cerebral hemispheres partially separated posteriorly; the rudime lateral ventricles with sketchy posterior horns (PH) and posterior cerebri can be seen

MICROCEPHALY

- **Prevalence:**

1 in 1,000 births.

80% of affected infants have a normal head circumference at birth and 90% of affected individuals had a normal head circumference in the second trimester.

- **Ultrasound diagnosis:**

Ultrasound diagnosis is made usually in the late second and third trimesters.

The fetal head circumference to abdominal circumference ratio is below the 3rd percentile (2 standards deviations below the normal mean for gestational age).

There is slopping forehead due to the disproportion of the frontal lobes and the face.

In most cases presenting in the second trimester there is associated holoprosencephaly or encephalocele and in those presenting in the third trimester there are abnormalities of sulcation or neuronal migration.

- **Associated abnormalities:**

Chromosomal abnormalities are rare and the most common are trisomies 13, 18 and 21.

Genetic syndromes are very common, most of them being caused by single gene defects with either autosomal recessive or X linked patterns of inheritance. The most common are: Meckel-Gruber, Walker-Walburg, Miller-Diecker, Smith-Lemli-Opitz, Seckel syndrome.

- **Investigations:**

Detailed ultrasound examination, including neurosonography.

Invasive testing for karyotyping and array.

TORCH test for fetal infections.

Fetal brain MRI at ≥32 weeks' gestation for diagnosis of abnormalities of neuronal migration, such as lissencephaly and polymicrogyria.

- **Follow up:**

Ultrasound scans every 4 weeks to monitor the evolution of head circumference.

- **Delivery:**

Standard obstetric care and delivery.

- **Prognosis:**

Isolated: the risk of neurodevelopmental delay inreases with decreasing head circumference from 10% if the circumference is 2 to 3 standard deviations (SD) below the normal mean for gestational age, to 100% if >4 SD's.

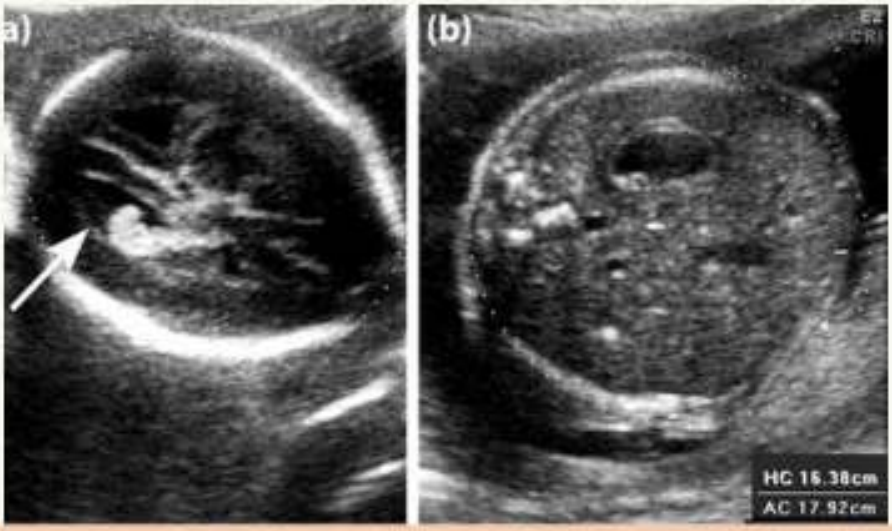
Syndromic: the prognosis depends on the underlying condition.

- **Recurrence:**

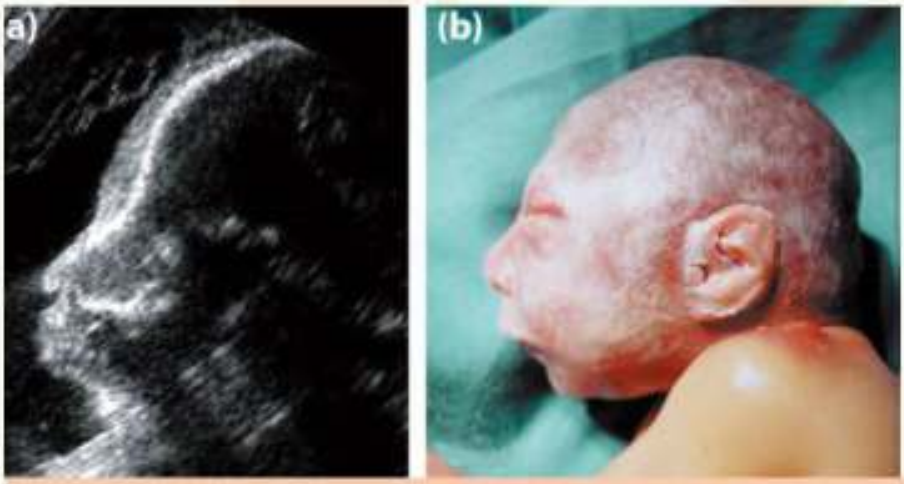
No associated structural defects: 5-10%.

Familial form of isolated microcephaly: 25%.

MICROCEPHALY



The sonographic diagnosis of microcephaly is based mainly on biometric parameters such as an HC/AC ratio below the first percentile



Side view of a 24-week fetus showing a typical sloping (or slanting) forehead

BLAKE POUCH CYST

- **Prevalence:**

1 in 1,000 births.

- **Ultrasound diagnosis:**

Expansion of the 4th ventricle into the cisterna magna resulting in a unilocular, avascular cyst in the posterior fossa – 'key-hole' sign in the transverse cerebellar view.

Vermis: normal size with mild to moderate upward rotation.

Cisterna magna: normal.

- Differential diagnosis: mega cisterna magna (>10 mm; normal vermis), arachnoid cyst (cyst in the cisterna magna with mass effect on surrounding structures; normal vermis).

- **Associated abnormalities:**

It is usually an isolated finding.

Risk of chromosomal abnormalies, mainly trisomy 21, in up to 5% of cases but usually in the presence of other suggestive markers.

- **Investigations:**

Detailed ultrasound examination, including neurosonography.

Fetal brain MRI may be useful if other brain abnormalities are suspected.

Invasive testing and array is recommended in non-isolated cases.

- **Follow up:**

Ultrasound scans every 4 weeks to monitor the size of the cyst and possible compression resulting in ventriculomegaly.

Spontaneous resolution by 24-26 weeks in 50% of cases.

- **Delivery:**

Standard obstetric care and delivery.

- **Prognosis:**

Neurodevelopment: good in 90% of cases, mild impairment in 10%.

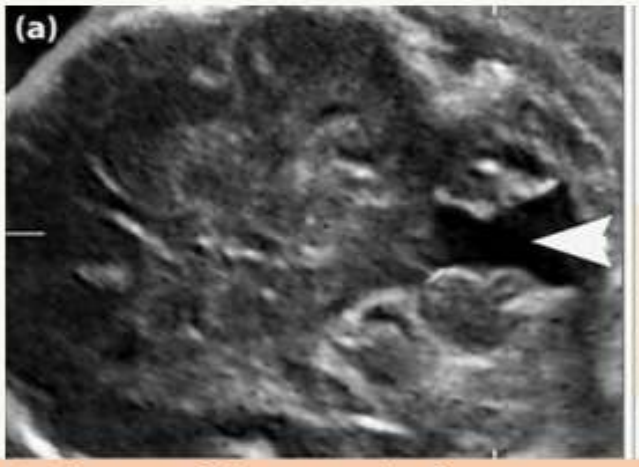
Small risk of postnatal hydrocephalus with the need to shunt.

- **Recurrence:**

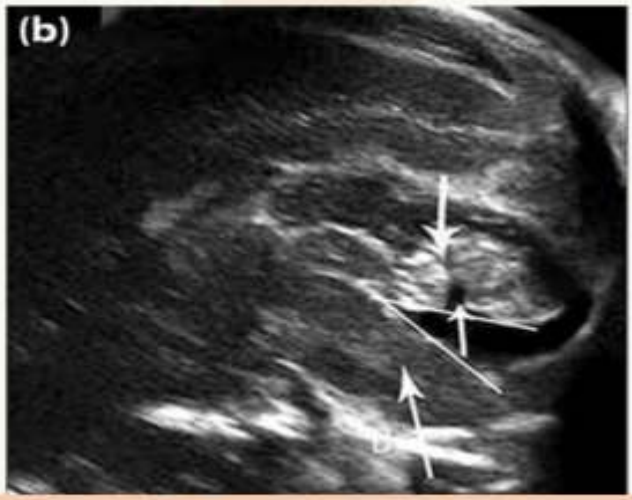
Isolated: no increased risk of recurrence.

Part of trisomies: 1%.

BLAKE POUCH CYST



Axial scan of the posterior fossa showing an hourglass opening fourth ventricle (arrowhead).



Sagittal scan of the posterior fossa showing an upward displacement of a normal vermis (arrow) and an open fourthventricle, communicating with the cisterna magna. The upward rotation of the vermis is mild;

MEGACISTERNA MAGNA

- **Prevalence:**

1 in 5,000 births.

- **Ultrasound diagnosis:**

The cisterna magna is >10 mm in the transverse cerebellar view.

Vermis: normal.

- Differential diagnosis: Blake's pouch cyst (expansion of the 4th ventricle into the cisterna magna resulting in a unilocular, avascular cyst in the posterior fossa; vermis normal in size with upward rotation), arachnoid cyst (cyst in the cisterna magna with mass effect on surrounding structures; normal vermis).

- **Associated abnormalities:**

It is usually an isolated finding, but in up to 10% of cases there is ventriculomegaly.

- **Investigations:**

Detailed ultrasound examination, including neurosonography.

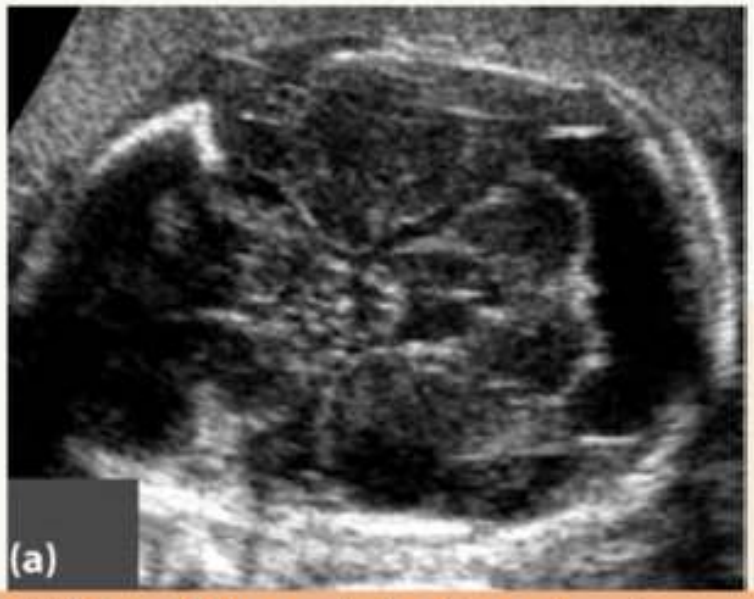
Fetal brain MRI may be useful if other brain abnormalities are suspected.

- **Follow up:**

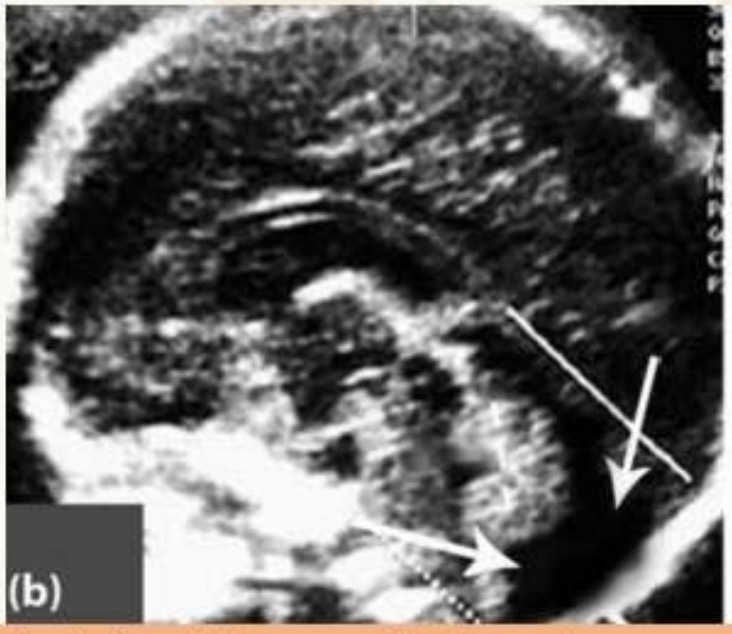
Ultrasound scans every 4 weeks to monitor the size of the cisterna magna and possible development of ventriculomegaly.

- **Delivery:** Standard obstetric care and delivery.
- **Prognosis:** Normal neurodevelopment.
- **Recurrence:** No increased risk of recurrence.

MEGACISTERNA MAGNA



Axial view of the posterior fossa showing enlargement of the cisterna magna; the cerebellarvermis appears intact.



Sagittal view of the posterior fossa demonstrating increased size of the cisterna magna (arrows), a closed fourth ventricle, and an intact vermis.

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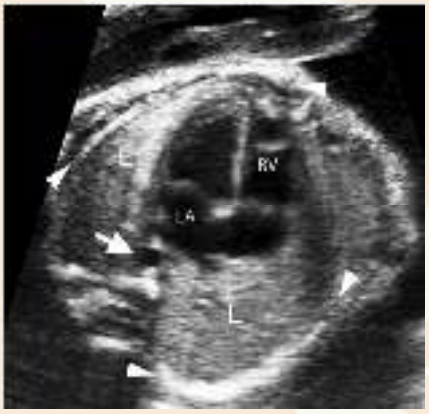
FETAL THORAX

-DR AMEE RAHATEKAR

VIEW- AXIAL FOUR CHAMBER VIEW OF HEART

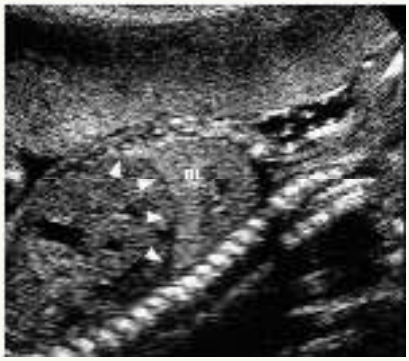
Axial four-chamber - fundamental plane where heart and lung anatomy is assessed. On this view, the following structures can and should be checked in addition to the **cardiac anatomy**

- **the thoracic outline**, consisting of the two displayed ribs and the overlying soft tissues and skin;
- **the two lungs**, shown in cross-section;
- **the thoracic aorta**, lying in the prevertebral area just left of the midline and behind the left atrium.



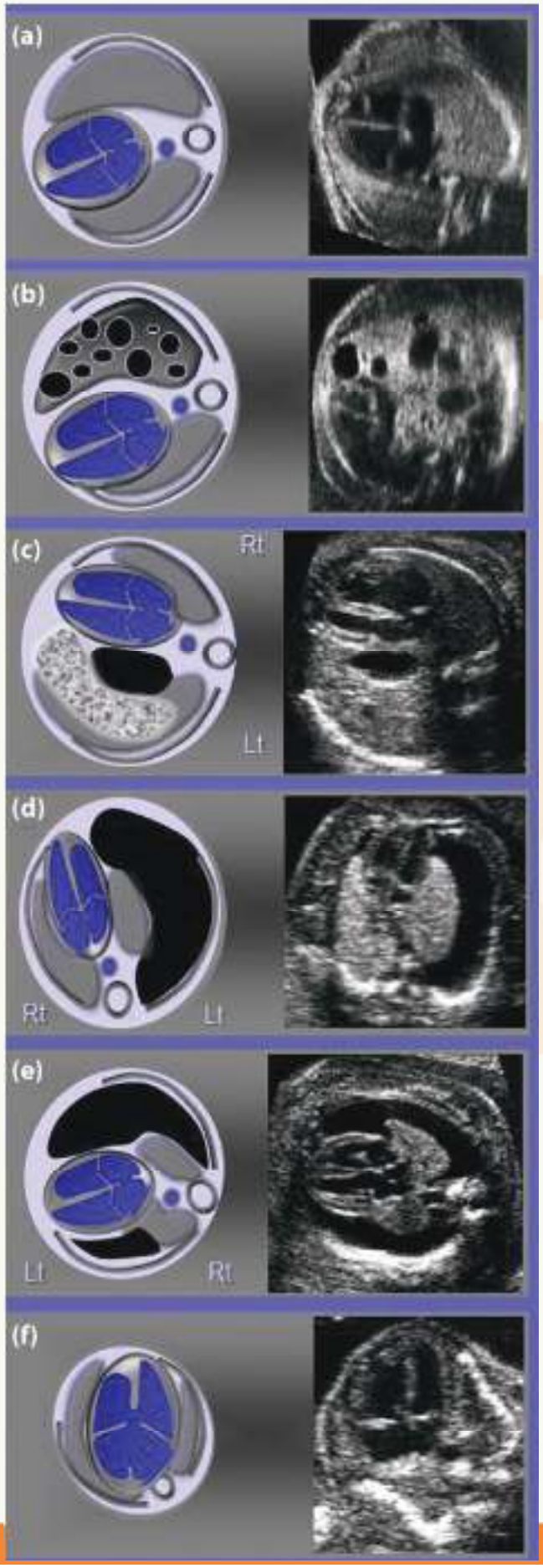
RIGHT PARASAGITAL VIEW

- This view allows detection of the hypoechoic muscular layer represented by thediaphragm (arrowheads), just below the right lung
- Care should be taken to consider the identification of the diaphragmatic plane in this view as a demonstration of an intact diaphragm: if the hernia is located on the other side, as in the Bochdalek type ,the contour of the right hemidiaphragm is normal.
- This view can be employed advantageously to disclose the severe thoracic hypoplasia typical of lethal skeletal dysplasia.



ANOMALIES OF THORAX

- Congenital diaphragmatic hernia
- CPAM
- Pleural effusion
- Pulmonary sequestration
- CHAOS
- Lung hypoplasia



Normal view of Thorax

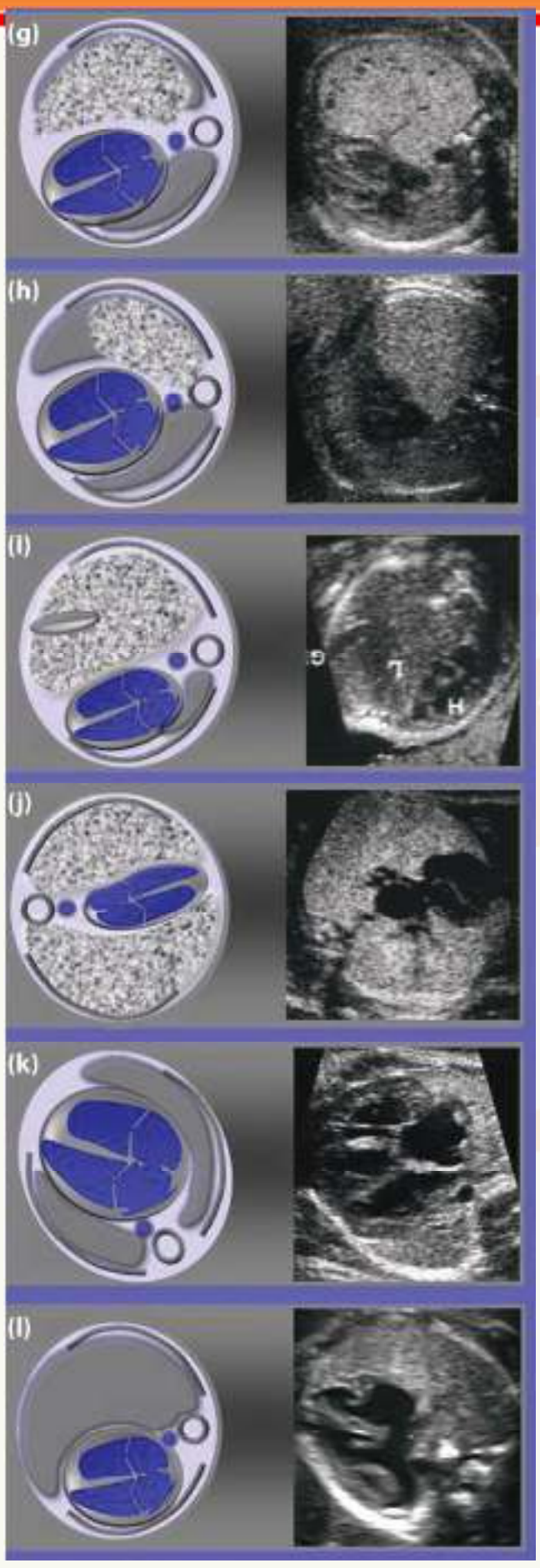
Cystic adenomatoid malformation of the lung (CAML), macrocystic type

Congenital diaphragmatic hernia (CDH), left-sided

hydrothorax, unilateral

hydrothorax, bilateral

thoracic hypoplasia;



pulmonary sequestration

CAML microcystic type

CDH, right-sided

Laryngeal atresia- CHAOS

Cardiomegaly

Right lung agenesis

CONGENITAL DIAPHRAGMATIC HERNIA

Incidence- Common.

Ultrasound diagnosis :

Stomach in the thorax (left-sided hernias);

right liver lobe in the thorax (right-sided hernias)

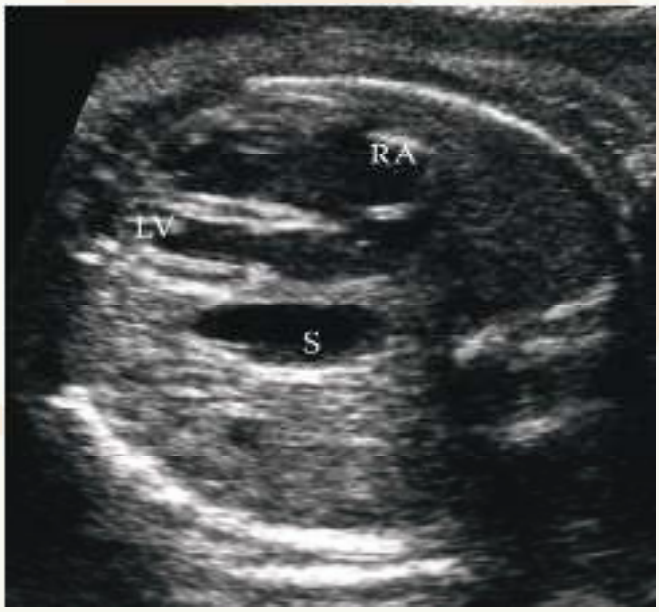
heart displaced in right hemithorax or hyper-rotated in the left hemithorax. Sometimes, ileal loops and/or the spleen in the thorax as well.

Risk of chromosomal anomalies - Relatively high (5%–15%): trisomies 18 and 21.

Risk of nonchromosomal syndromes - High (25%–30%): Fryns, Pallister–Killian, Beckwith–Wiedemann.

Outcome - Extremely poor in syndromic cases. Overall survival rate of 40%–60% in nonsyndromic cases;

prenatal endoscopic approach seems to improve survival in cases with the poorest prognosis.



OBSTETRIC MANAGEMENT

- Karyotyping is mandatory because of the relatively high risk of aneuploidy. (5%-15%)
- A thorough anatomic scan should be performed by an expert, in order to detect major and/or minor signs possibly leading to the diagnosis of one of the associated syndromes. (25%-30%)
- Mode of delivery, in the past, it had been apparently shown that delivery by Cesarean section was associated with a better outcome.- currently there is no recommendation to deliver fetuses with CDH by Cesarean section.
- Timing of delivery: In a recent study, survival was significantly higher for deliveries occurring later than 40 weeks of gestation than for those occurring at 38–40 week.
- Due to compression effect patient may develop polyhydramnios and sometimes hydrops
- Amnioreduction may be done in patients with respiratory discomfort due to severe polyhydramnios

POSTNATAL MANAGEMENT

- Once the neonate has been stabilized, surgery is performed.
- The timing of the surgical intervention depends on the severity of the desaturation (O2) and on the presence of pulmonary hypertension.
- The surgical approach usually involves the use of a synthetic patch when the diaphragmatic defect is too large to be closed by primary tissue approximation.
- Occasionally, rotational muscle flaps can be used to repair the defect.
- Unfortunately, because prosthetic patches do not grow with the neonate, re-herniation will occur in a significant number of survivors.

PROGNOSTIC FACTORS FOR CDH- BASIS FOR COUNSELLING

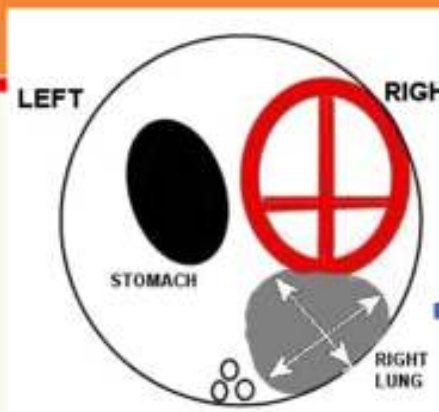
- Association with chromosomal or non chromosomal anomalies- poor prognosis
- Early gestational age at diagnosis
- Presence of a mediastinal shift
- Higher pulmonary artery resistance at pulsed wave doppler
- Intrathoracic position of the liver (liver up)
- Lung-to-head ratio (LHR)
- Observed/expected LHR (o/e LHR)
- Right sided CDH –worse prognosis

PROGNOSIS DEPENDS ON PREDICTION OF LETHAL PULMONARY HYPOPLASIA AND OF PULMONARY HYPERTENSION

SURVIVAL AND LONGTERM SEQUELAE

- Long-term sequelae include growth retardation (18%), gastroesophageal reflux (27%), chronic lung disease (22%), and, rarely, oxygen requirement (2%).

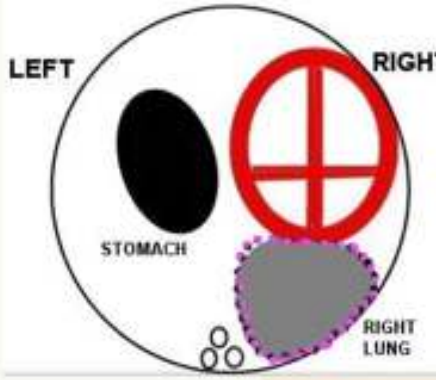
Methods to get lung area



LONGEST DIAMETER METHOD

Lung area = length 1 x length 2

LHR = LUNG AREA / HEAD CIRCUMFERENCE



TRACING METHOD

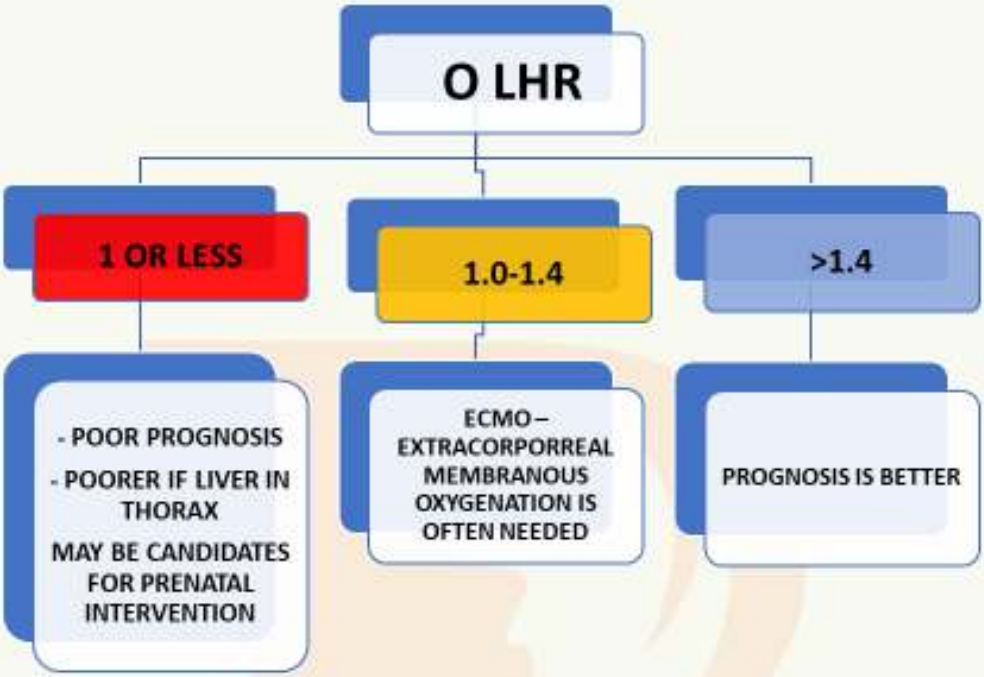
METHOD OF CALCULATING THE O/E LHR

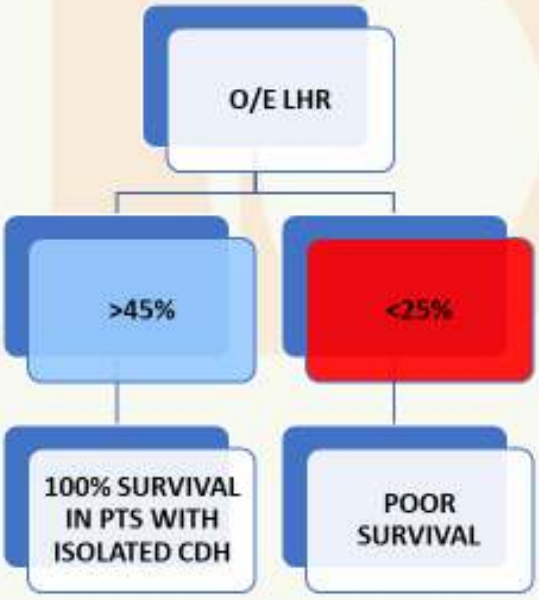
1. Select Congenital diaphragmatic hernia (CDH) is on the ● **Left side** ○ **Right side**

Enter Parameters for Lung Area	
2. Longest Diameter Method Enter: Length 1 ____ mm Enter: Length 2 ____ mm	**3.** Tracing Method Enter: Area traced= ____ mm2
4. Enter Fetal Head Circumference ____ mm	
5. Enter Gestational Age ____ weeks ____ days	

Calculate Clear (1 cm ^2 = 100 mm^2)

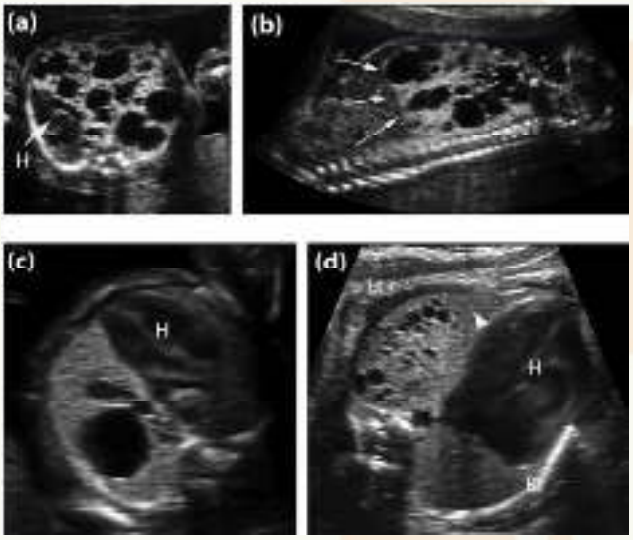
INTERPRETATION



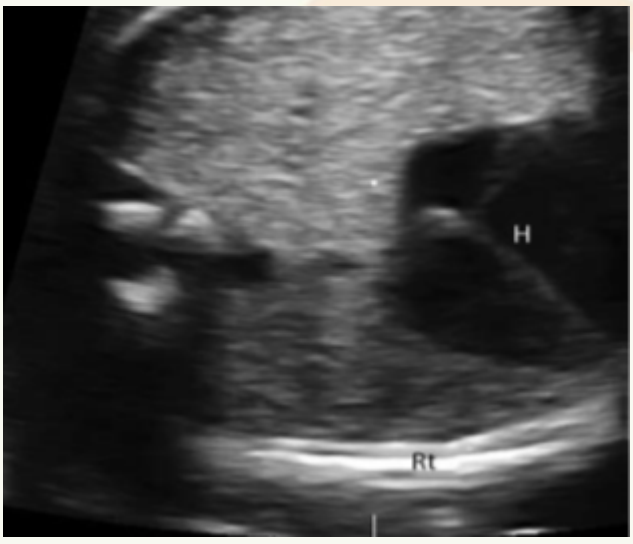


CPAM- CYSTIC PULMNARY ADEMTOID MALFORMATION

- **Incidence** - Relatively rare
- **Ultrasound diagnosis** - On the *4-chamber view*, unilateral uni- or multilocular cystic lung mass; unilateral,homogeneously hyperechoic lung mass.
- **Risk of chromosomal anomalies** - Extremely low.
- **Risk of nonchromosomal syndromes**-Extremely low.
- **Outcome** - Good/very good, with spontaneous complete regression or surgical removal after birth.



MACROCYSTIC CPAM



MICROCYSTIC CPAM

OBSTETRIC MANAGEMENT

- Should a CAML be detected in a fetus, it is of the utmost importance to search further for very early signs of hydrops.
- Karyotping is not mandatory.
- Prenatal counseling sessions regarding the relatively high rate of spontaneous regression and the consequently very good outcome
- This is done in cases where hydrops develops as a result of increased intrathoracic pressure; it may consist of simple cyst aspiration or, more often, placement of a thoracoamniotic shunt

POSTNATAL MANAGEMENT

- The first step is to confirm the volume of the lesion by MRI
- CAML should be removed, even if asymptomatic, by 12 months of age as advised by Pediatric surgeons.
- The need to resect the nonfunctioning lobe is based on the high incidence of infection and on the very low risk of neoplastic transformation

PROGNOSTIC FACTORS FOR CPAM- BASIS FOR COUNSELING

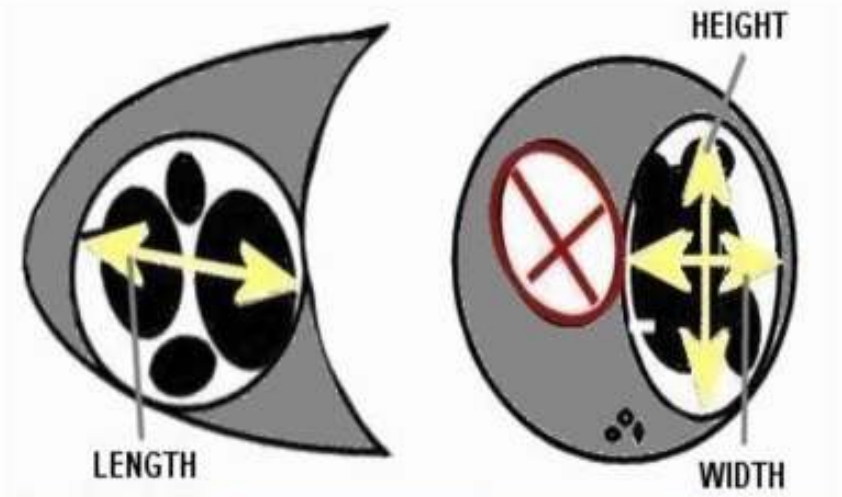
- If hydrops is absent, survival is generally unaffected, with no functional limitations
- If hydrops is present neonatal death is almost certain and no intrauterine procedure has succeeded in reversing it.
- The Congenital Pulmonary Airway Malformation Volume is a sonographic indicator that has been proposed for the evaluation of fetuses at risk for hydrops and possible intervention.

SURVIVAL AND LONGTERM SEQUELAE

- Overall survival rates for prenatally diagnosed CAML are greater than 80% in most series.

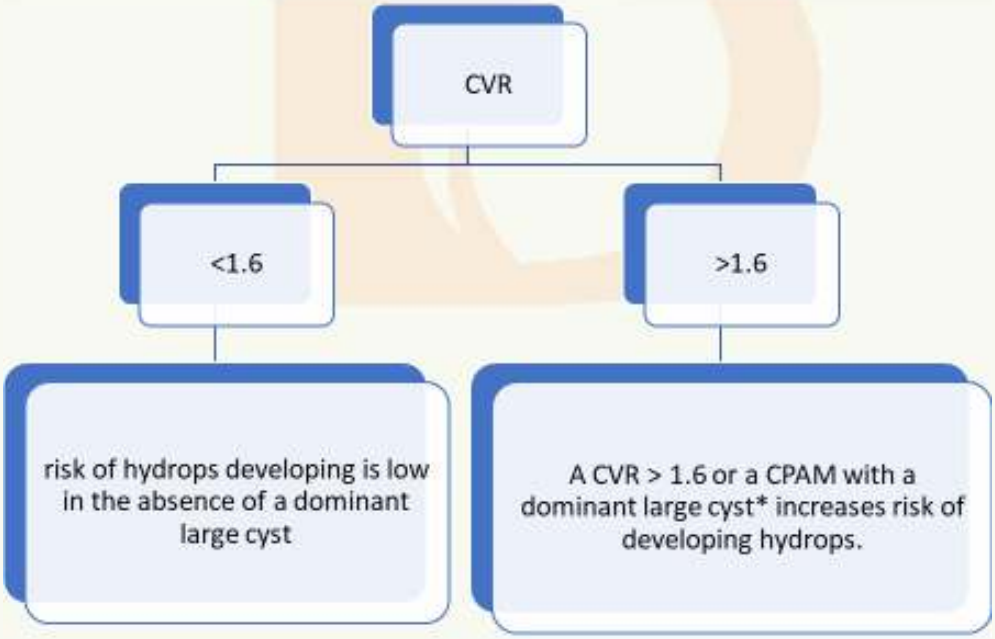
CVR- CPAM Volume Ratio

- CVR = (Length X Height X Width X 0.52)/Head Circumference



The CVR is obtained by dividing the CPAM volume by the head circumference (measured in cm)

- CVR = (Length X Height X Width X 0.52)/Head Circumference



The cyst is dominant if it constitutes greater than a third the size of the CPAM.

PLEURAL EFFUSION

- **Prevalence:**

Isolated congenital chylothorax is found in about 1 in 10,000 births.

- **Ultrasound diagnosis**:

Usually presents with polyhydramnios at around 26 weeks' gestation.

Unilateral (25% of cases) or bilateral anechoic area surrounding the lung. Subjectively classified as mild, moderate or severe and in the latter case if unilateral there is mediastinal shift.

In half of the cases the effusion is isolated and in the other half there is associated hydrops with skin edema and / or ascites.

- **Associated abnormalities:**

Chromosomal defects, mainly trisomy 21 and monosomy X, are found in 10% of cases.

Noonan syndrome (autosomal dominant but >90% are due to *de novo* mutations; cystic hygromas, hypertelorism, pulmonary stenosis, fetal growth restriction), is found in <5% of cases of isolated hydrothorax.

In the case of associated hydrops there is a wide range of genetic conditions, especially if there are other defects.

- **Investigations:**

Detailed ultrasound examination, including echocardiography.

Invasive testing for karyotyping and array.

Specialist investigations, including infection screen and assessment for anemia and metabolic disorders, may be necessary in the case of hydrops.

- **Fetal therapy:**

In severe unilateral or bilateral pleural effusions placement of thoraco-amniotic shunts restores the normal intrathoracic anatomy and results in resolution of associated hydrops and polyhydramnios. An alternative to shunting is pleurodesis in which a sclerosant substance is injected in the pleural cavity.

- **Follow up:**

Ultrasound scans every 2 weeks to monitor the evolution of the pleural effusions and associated hydrops. Insertion of new shunts may become necessary if these are blocked or displaced.

- **Delivery:**

Place: hospital with neonatal intensive care.

Time: 38 weeks.

Method: cesarean section, if there is severe hydrops. In fetuses with shunts, these need to be clamped at delivery to prevent the development of pneumothorax.

- **Prognosis:**

Isolated effusion: survival is >90%.

Effusions with hydrops: in 50% of cases the hydrops resolves after thoraco-amniotic shunting and in these cases the prognosis is good. If there is no resolution of hydrops, there may be an underlying genetic syndrome or infection and in these cases the prognosis is poor.

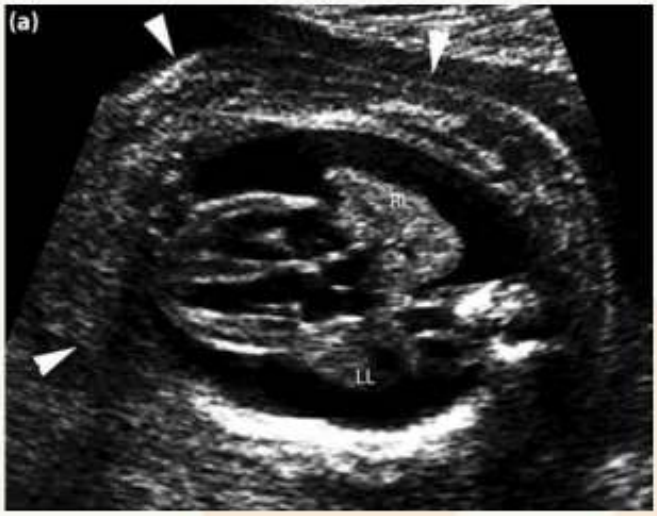
- **Recurrence:**

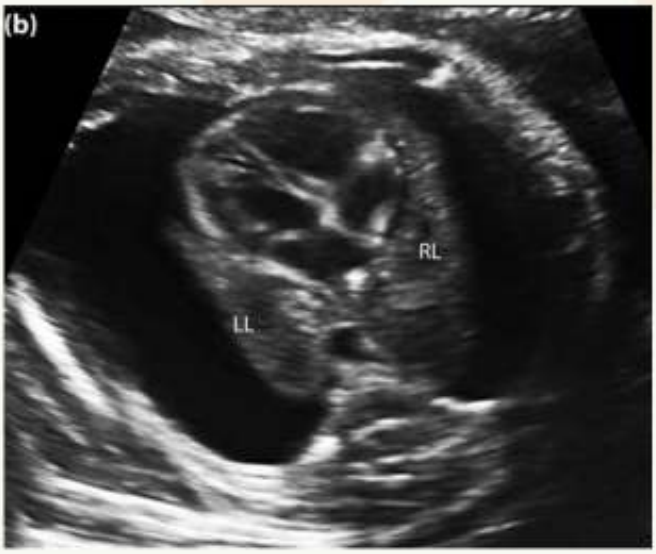
Isolated or associated with infection: no increased risk.

Part of trisomy: 1%.

Part of genetic syndrome: up to 25%.

PLEURAL EFFUSION





PULMONARY SEQUESTRATION

- **Prevalence:**

1 in 15,000 births

- **Ultrasound diagnosis**:

Hyperechogenic mass in the lung, mostly in the left lower lobe.

Color Doppler demonstrates a feeding vessel that arises from the descending aorta.

In 75% of cases it is intralobar, making it indistinguishable in appearance from microcystic CPAM.

In 25% of cases it is extralobar, located outside the normal lung with its own visceral pleura; in most of these cases there is an associated pleural effusion.

- **Associated abnormalities**:

The incidence of chromosomal abnormalities and genetic syndromes is not increased.

Defects in other systems, mainly diaphragmatic hernia and cardiac or vertebral anomalies are found in up to 50% of cases with extralobar sequestration.

- **Fetal therapy:**

Ultrasound guided laser coagulation of the feeding vessel in cases of severe hydrothorax or hydrops.

- **Follow up:**

Ultrasound scans every 4 weeks to monitor growth of the tumor and hydrothorax or hydrops.

In >30% of cases there is regression or disappearance of the tumor during the 3rd trimester.

- **Timing and route of delivery:**

Place: hospital with neonatal intensive care and pediatric surgery.

Time: 38 weeks.

Method: induction of labor aiming for vaginal delivery.

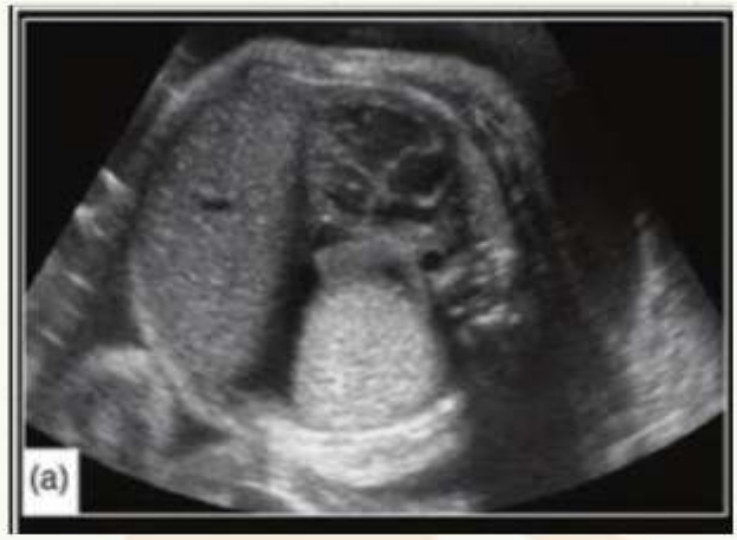
- **Prognosis:**

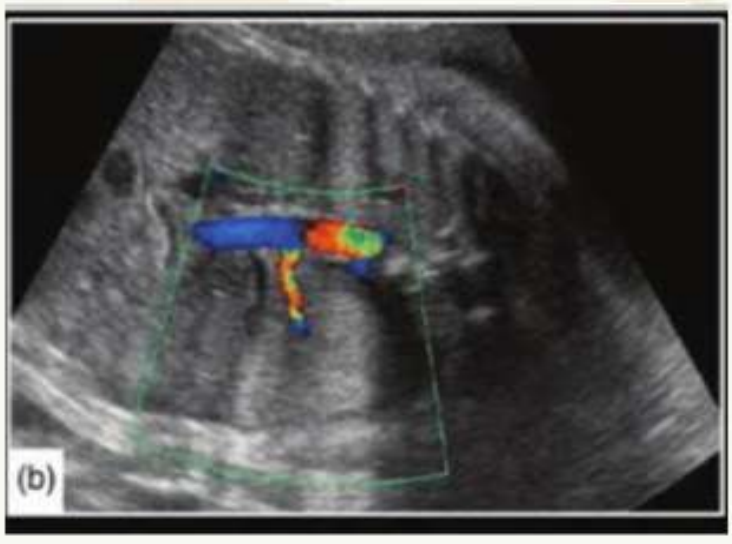
Survival: >95%.

Postnatal therapy: endoscopic removal of mass or selective embolization of the feeding artery.

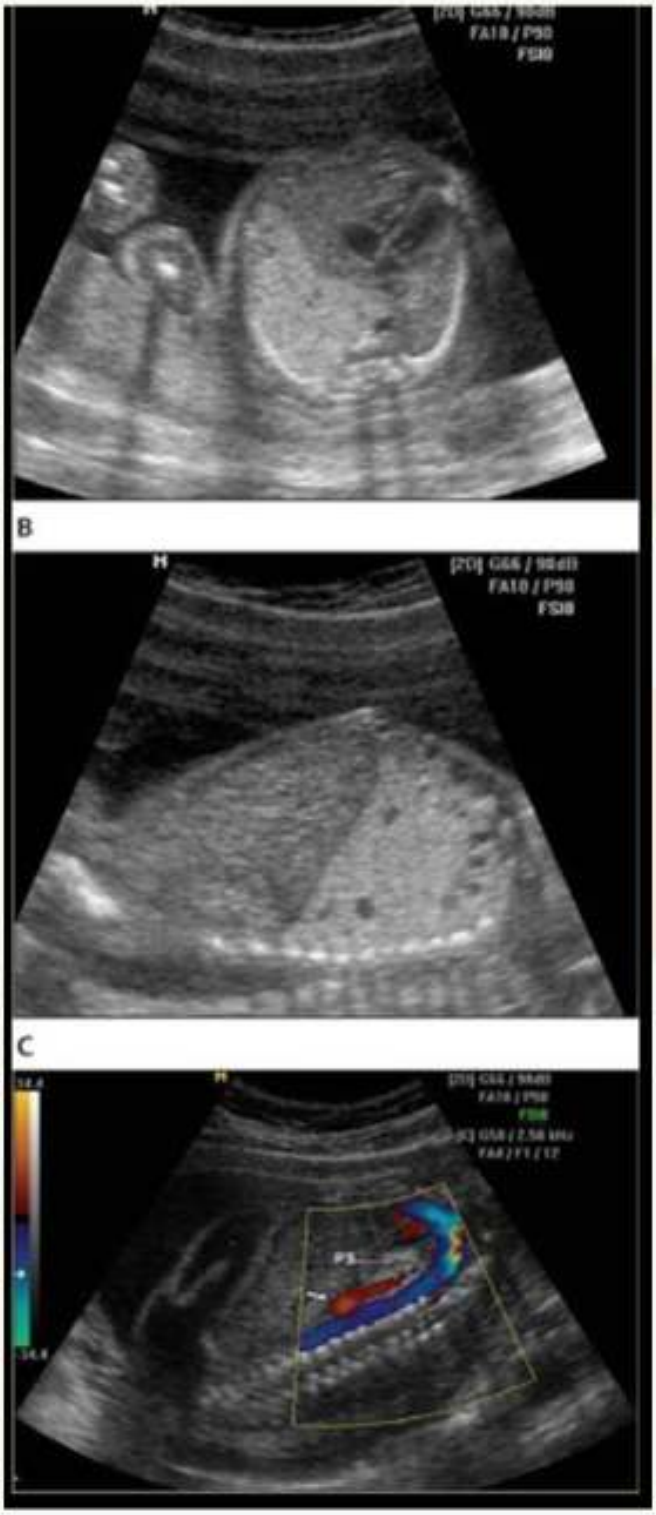
- **Recurrence:**

No increased risk of recurrence.





PULMONARY SEQUESTRATION



B

C

CHAOS- Congenital high airway obstruction syndrome

- **Prevalence:**

1 in 50,000 births.

- **Ultrasound diagnosis:**

Agenesis or stenosis of a segment of the upper airways at the level of the trachea or larynx. In a few cases the condition results from the presence of laryngeal cysts or laryngeal web.

Ultrasound features become evident ≥16 weeks' gestation. The lungs are massively enlarged and hyperechogenic resulting in compression of the heart and development of ascites. The bronchial tree is dilated and the diaphragm inverted.

- **Associated abnormalities:**

The incidence of chromosomal abnormalities is not increased.

Genetic syndromes are found in >50% of cases. The most common is Fraser syndrome (autosomal recessive; microphthalmia, facial cleft, tracheal atresia, bilateral renal agenesis, heart defects, syndactyly or polydactyly).

- **Investigations:**

Detailed ultrasound examination.

Fetal MRI could help identify the location and type of obstruction.

- **Follow up:**

If the pregnancy continues, serial scans should be carried out to define the best time for delivery based on evolution of hydrops.

- **Delivery:**

Place: hospital with neonatal intensive care and pediatric surgery.

Time: 38 weeks.

Method: cesarean section with EXIT procedure and tracheostomy.

- **Prognosis:**

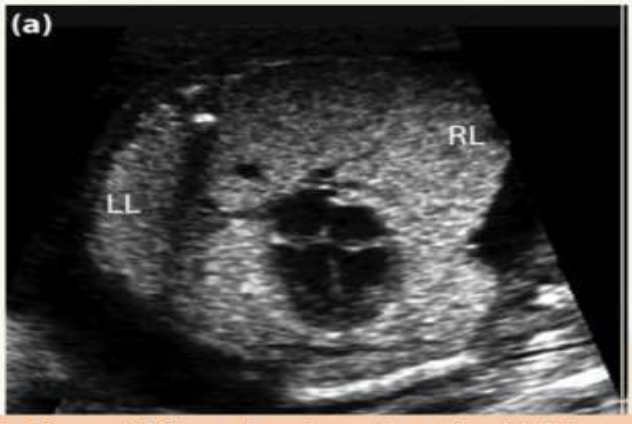
Highly lethal condition with almost all cases dying in the neonatal period.

There are a few case reports of spontaneous resolution in utero, either because of dilatation of a stenotic segment between the larynx and the trachea or even development of a tracheoesophageal fistula with drainage of the bronchial fluid into the esophagus.

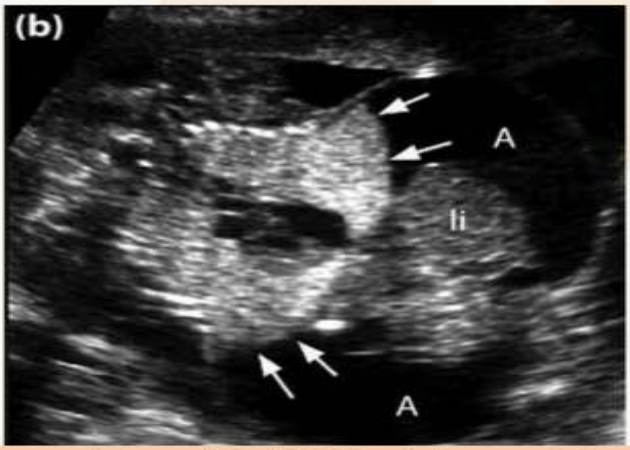
- **Recurrence:**

Isolated: no increased risk.

Fraser syndrome: 25%.



On the axial four-chamber view, the highly hyperechoic lungs appear severely enlarged. The (normal) heart is squeezed into the mediastinum.



The coronal view of the fetal trunk demonstrates eversion of the diaphragm (arrows), due to the massive enlargement of the lungs, and ascites, which is almost ubiquitous in laryngeal atresia

LUNG HYPOPLASIA

- **Prevalence:**

1 in 50,000 births.

- **Ultrasound diagnosis**:

Complete absence or hypoplasia of one lung, usually the right one, with major mediastinal shift.

- **Associated abnormalities:**

The incidence of chromosomal abnormalities is not increased.

The association of hypoplastic right lung with hypoplastic right pulmonary artery and partial pulmonary venous drainage of the right lung into the inferior vena cava is referred to as Scimitar syndrome.

- **Investigations:**

Detailed ultrasound examination, including echocardiography.

- **Follow up:**

Ultrasound scans every 4 weeks to monitor the evolution of the condition.

- **Delivery:**

Place: hospital with neonatal intensive care and pediatric surgery.

Time: 38 weeks.

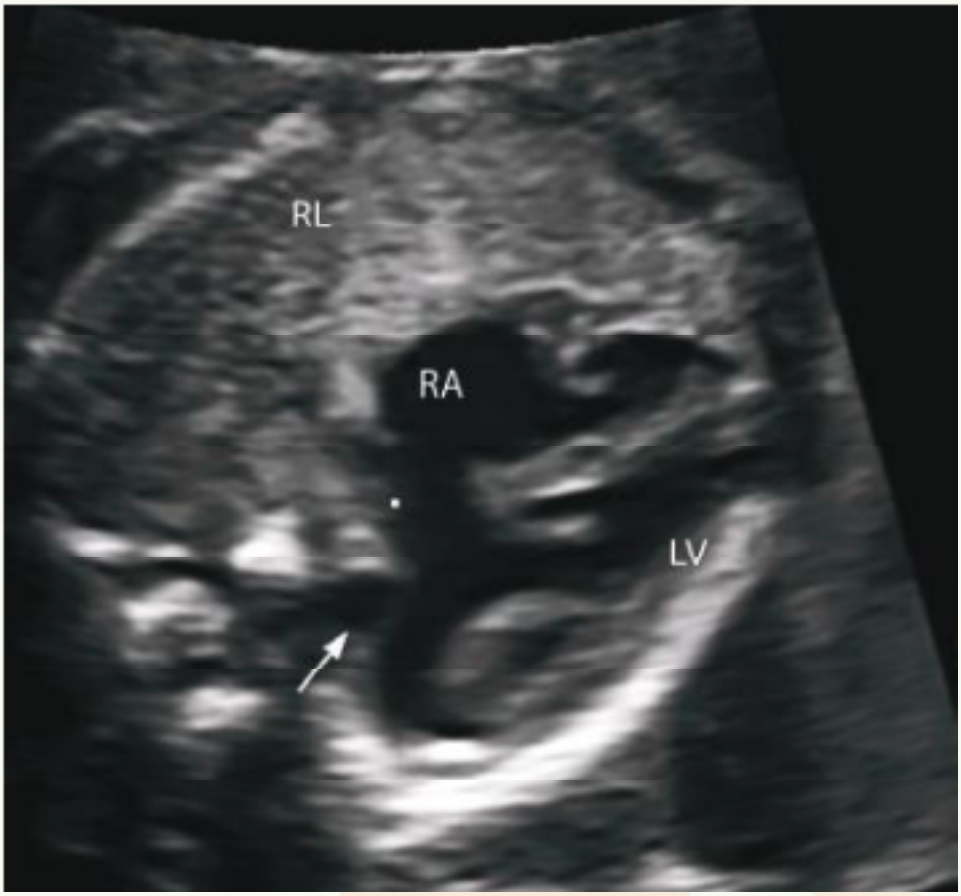
Method: induction of labor aiming for vaginal delivery.

- **Prognosis:**

Prognosis is worse for right than left lung agenesis probably because of greater degree of heart and mediastinal displacement. In Scimitar syndrome, mortality is about 10% due to severe pulmonary hypertension.

- **Recurrence:**

No increased risk of recurrence.



The left lung is absent, and the right lung (RL) is larger than normal. Note the evident mediastinal shift toward the left,with the heart completely in the left hemithorax

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FETAL HEART

- DR.NEHA MUNIYAR PUNIYANI

INTRODUCTION

- CHD is a leading cause of infant mortality, with an estimated incidence of about 4–13 per 1000 live births.
- Prenatal detection of CHD has been shown to improve the outcome of fetuses with specific types of cardiac malformations.
- The cardiac screening examination of the fetus is designed to maximize the detection of heart anomalies during a second-trimester scan.
- Screening programs are intended to be applied to the low-risk population, and thus should be part of the routine care.
- Cardiac screening examination is performed optimally between 18 and 22weeks.

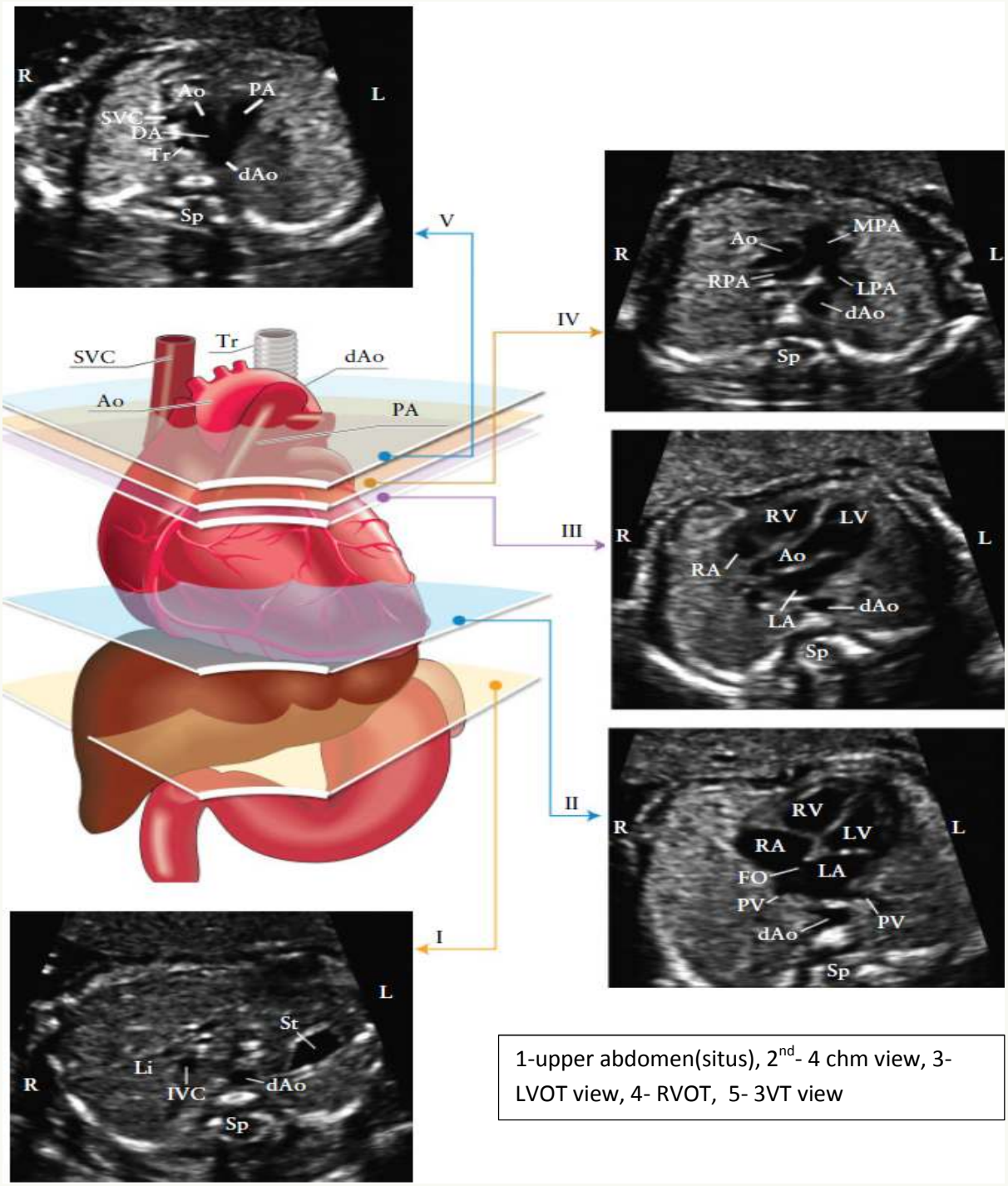
TECHNICAL FACTORS TO BE CONSIDERED FOR CARDIAC EXAMINATION

- Use higher frequency probes
- Turn on harmonics imaging – especially in obese women
- Cross- sectional grey scale imaging remains the basis
- High frame rate, with increased contrast & high resolution.
- Low persistence, a single acoustic focal zone & a relatively narrow image field should be used
- Images should be magnified until the heart fills at least one third to one half of the screen

CARDIAC SCREENING – BASICS

- Cardiac screening involves a series of transverse sections/axial sections in caudal to cranial direction to include

- Cardiac situs
- Four chamber view (4 Chm)
- Left ventricular outflow tract (LVOT)
- Right ventricular outflow tract (RVOT)
- 3 vessel view (3VV)
- 3 vessel & trachea view (3VT)



1-upper abdomen(situs), 2nd- 4 chm view, 3- LVOT view, 4- RVOT, 5- 3VT view

BASIC POINTS TO REMEMBER WHILE DOING A CARDIAC EXAMINATION

- Fetal heart is best examined with
- spine posterior i.e apex of heart pointing up (apical view)

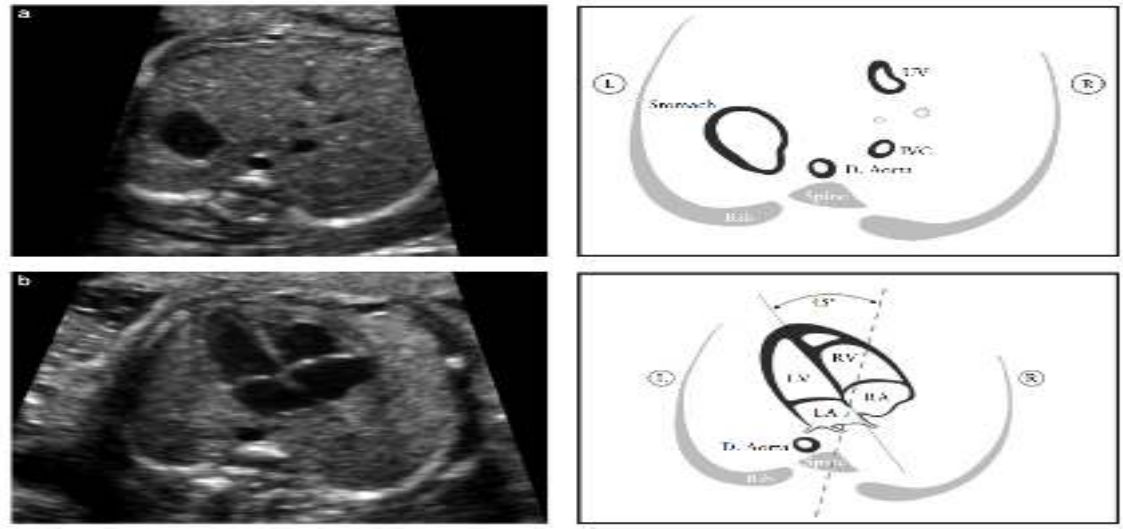
  OR
- spine in rt. lateral or lt .lateral , i.e – lateral view with rt. side of the heart being anterior

- To avoid false-positive diagnosis
- avoid examining heart in basal view(spine ant) &
- lateral view when left side of heart is anterior
- Select proper probe/mode
- like fetal echo/ fetal heart
- optimise the settings

CARDIAC SCREENING- STEPWISE APPROACH

CARDIAC SITUS – STEPWISE APPROACH

- Determine fetal presentation
- Confirm the right & left side of fetus
- Obtain a transverse/axial section of fetal abdomen
- Stomach should be seen on fetal lt. side
- Move the transducer cranially to thorax to visualise fetal heart
- Apex of the heart should point towards left side (as stomach)
- DA should be to the lt. & IVC to the rt. of spine



Cardiac situs check list	Situs abnormalities
• Fetal stomach & cardiac apex on the left	• Situs abnormality is suspected when fetal heart and/or stomach are not found on the left side
• Descending aorta to the left of spine	• Situs abnormalities include - Situs inversus - situs ambiguous - heterotaxy syndromes
• Inferior vena cava to the right of spine	

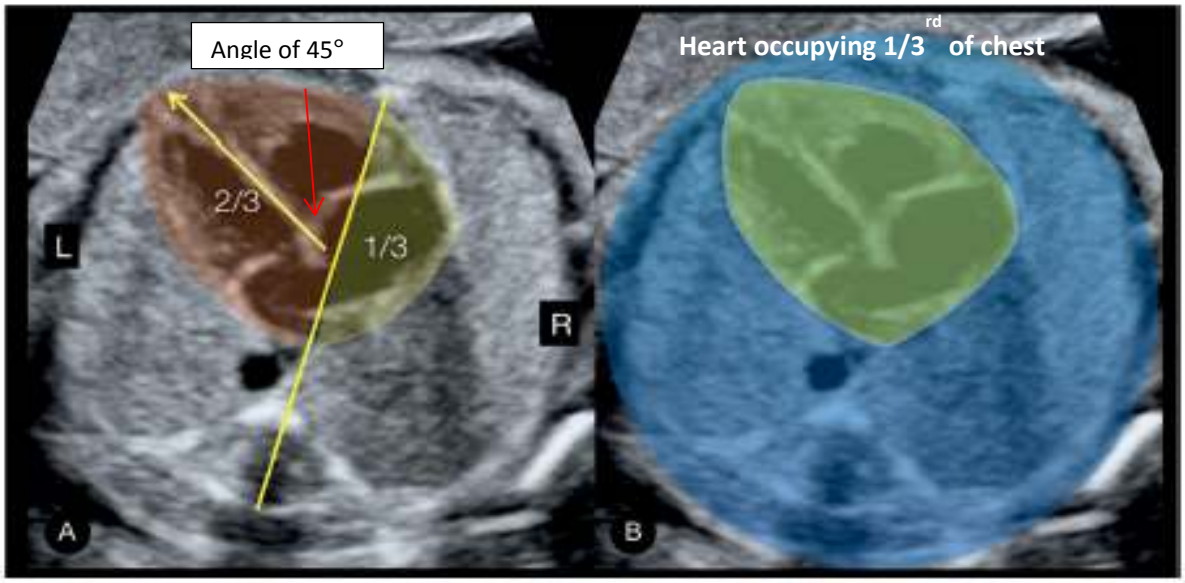
CARDIAC AXIS, POSITION & SIZE

- Normal heart occupies one third of the area of the chest
- 2/3rd of heart -situated in left side of the chest
- 1/3rd of heart in rt.side of the chest
- Normal cardiac axis points to left with an angle of about 45±20 degrees
- Small hypoechoic/anechoic rim of upto 2mm around heart is normal

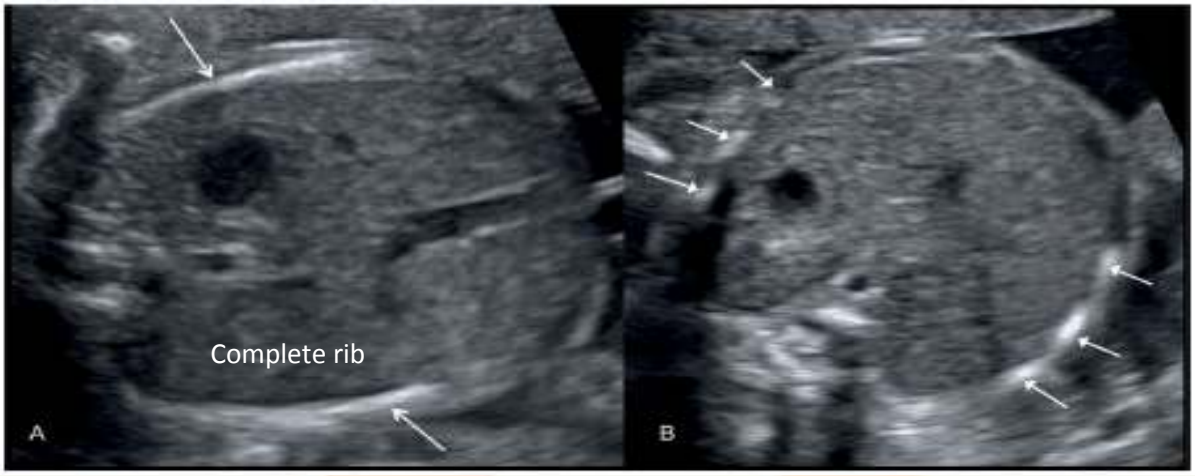
ABNORMALITIES OF CARDIAC AXIS, POSITION &SIZE

- **Abnormal axis** - ↑risk of cardiac malformation (outflow tracts & chromosomal anomaly)
- **Abnormal displacement** of heart from its normal anterior left position can occur due to
  - diaphragmatic hernia or
  - space-occupying lesion, such as cystic adenomatoid malformation of lung (CCAM
- **Position abnormalities** can be secondary to
  - fetal lung hypoplasia or agenesis.
- **Shift of axis** to the left can be due to
  - fetal gastroschisis & omphalocele

CARDIAC AXIS & POSITION

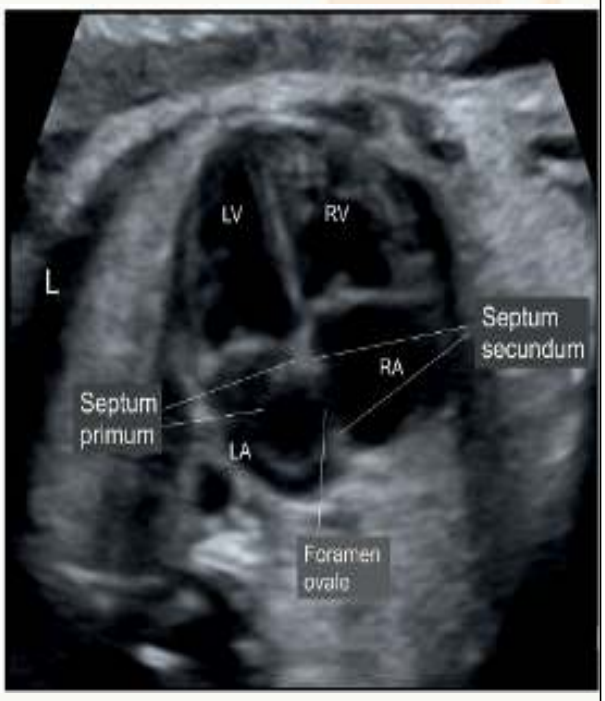


FOUR CHAMBER VIEW



- Four chamber view is one of the most important plane to be visualised during cardiac examination
- Scanning technique:
  -Slide the transducer cranially from transverse plane of fetal abdomen until 4 chamber view is seen
- Optimum plane for visualisation of 4 chm view requires:
  - one complete rib on each side of fetal chest wall
  - 2 inferior pulmonary veins entering the left atrium
  - Apex of the heart

FOUR CHAMBER VIEW CHECKLIST :



Four cardiac chambers- 2 atria & 2 ventricles

- ✓ **Atrial chambers**

-Two atria, almost equal in size
-Foramen ovale flap in left atrium
-Atrial septum primum present (near to crux)
-Pulmonary veins entering left atrium

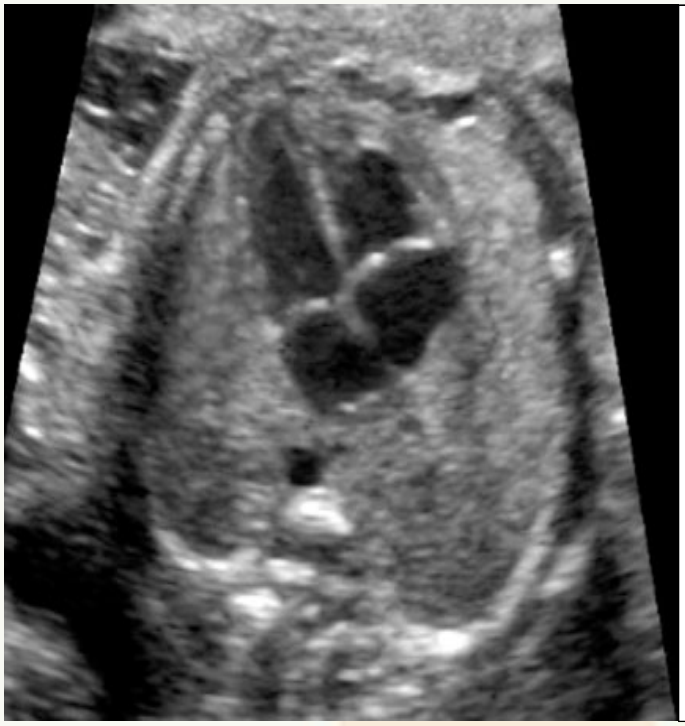
- ✓ **Ventricular chambers**

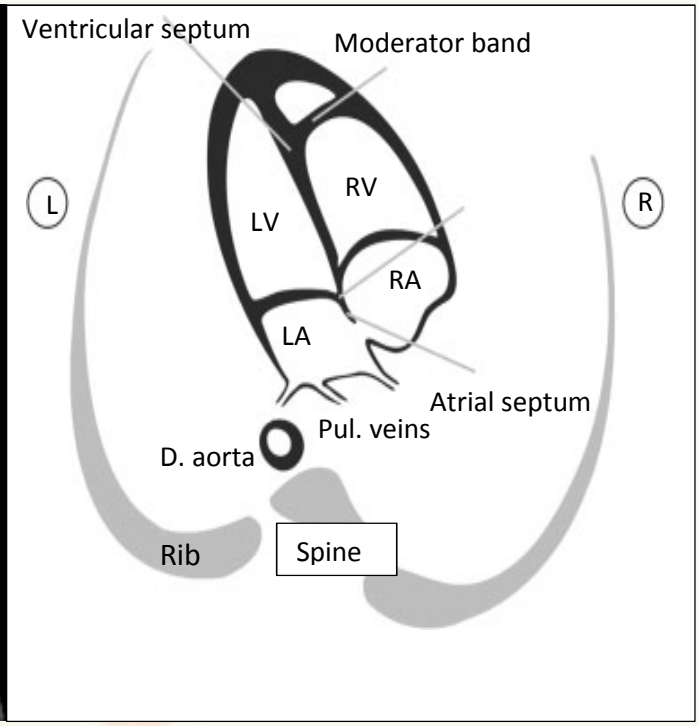
-Two ventricles, almost equal in size
-No ventricular wall hypertrophy
-Moderator band in rt. Ventricle
-Smooth margins of lt. ventricle
-Apex formed by lt. ventricle
-Ventricular septum intact (apex to crux)

- ✓ **Atrioventricular (AV) valves & junction**

-Intact cardiac crux
-Two separate AV valves orifices
-AV valves opening & moving freely
-Normal AV valve offset: tricuspid valve leaflet inserts on ventricular septum closer to cardiac apex than does mitral valve

NORMAL FOUR CHAMBER VIEW





ABNORMALITIES COMMONLY ASSOCIATED WITH NORMAL 4 CHAMBER VIEW :

- Tetralogy of fallot (TOF)
- Transposition of great arteries (TGA)
- Double outlet right ventricle (DORV)
- Small VSD's
- Common arterial trunk
- Aortic arch anomalies

ABNORMALITIES COMMONLY ASSOCIATED WITH ABNORMAL 4 CHAMBER VIEW

- Mitral/aortic atresia
- Tricuspid/pulmonary atresia
- Ebstien's anomaly/tricuspid valve dysplasia
- Atrioventricular septal defects
- Large ventricular septal defect (VSD)
- Severe Aortic/Pulmonary stenosis
- Single ventricle
- Severe coarctation of aorta

- Hypoplastic left heart disease
- Total anomalous pul. Venous connection
- Cardiac tumours

OUTFLOW TRACT VIEWS

- Views of the left and right ventricular outflow tracts are considered an integral part of the fetal cardiac screening examination.
- Evaluation of outflow tracts increases the detection rates for major cardiac malformations above those achievable by the four-chamber view alone
- At the very least, examination of the outflow tracts requires that the great vessels are approximately equal in size and cross each other at right angles from their origins as they exit from the respective ventricles.

LEFT VENTRICULAR OUTFLOW TRACT (LVOT)

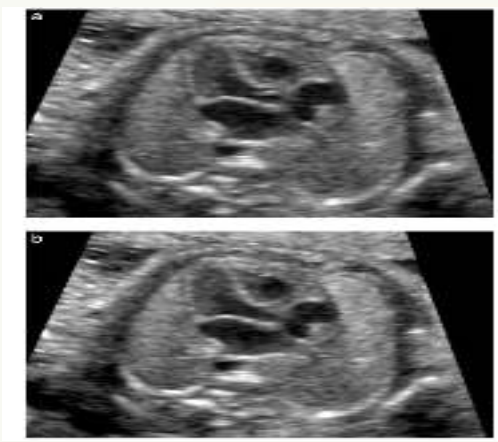
How to acquire LVOT view:

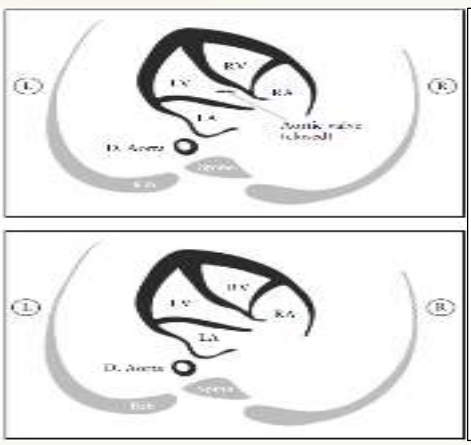
- Perform a transverse sweep (sweep technique) with cranial movement of the transducer from the fetal abdomen through the 4 cham view & towards the upper mediastinum to get LVOT view where aortic valve is seen moving freely

LVOT – check list :

- LVOT view is also known as **5 chamber** view
- LVOT arises from the centre of heart & courses towards the right shoulder
- LVOT view confirms the presence of a great vessel originating from the morphological left ventricle.
- Continuity should be documented between the ventricular septum & the anterior wall of aorta.
- The aortic valve moves freely and should not be thickened
- Normally the two great vessels(Aorta & Pul.artery) cross each other at rt angle

LVOT VIEW

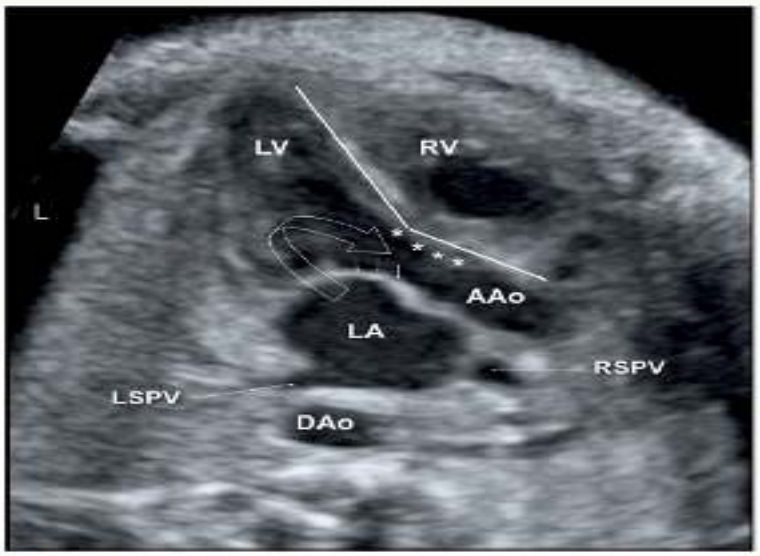




A. Aortic valve is closed in picture(a)

B. Aortic valve is open in picture(b)

NORMAL LVOT VIEW



Normally wide angle is seen between direction of ventricular septum & ant. wall of aorta (white line in the above diagram)

ABNORMALITIES OF LVOT :

- Loss of this angle + loss of continuity between ant. Wall of ventricular septum & ant.wall of aorta: Conotruncal anomalies like **Aortic override as in TOF,**
- Origin of aorta from morphological left ventricle rules out **Double outlet right vetricle (DORV) & Transposition of great arteries (TGA)**

RIGHT VENTRICULAR OUTFLOW TRACT (RVOT)

How to acquire RVOT view :

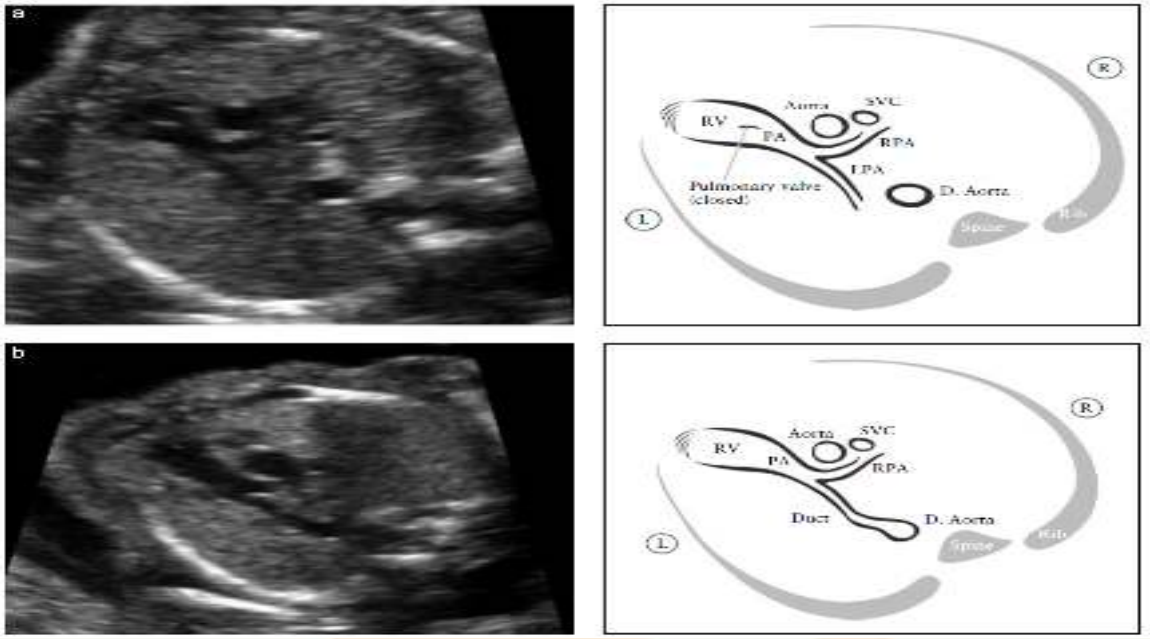
- RVOT view is obtained by sliding the transducer cranially from LVOT
- RVOT arises close to ant. wall of fetal thorax as compared to LVOT (which arises posteriorly to PA)

RVOT – check list :

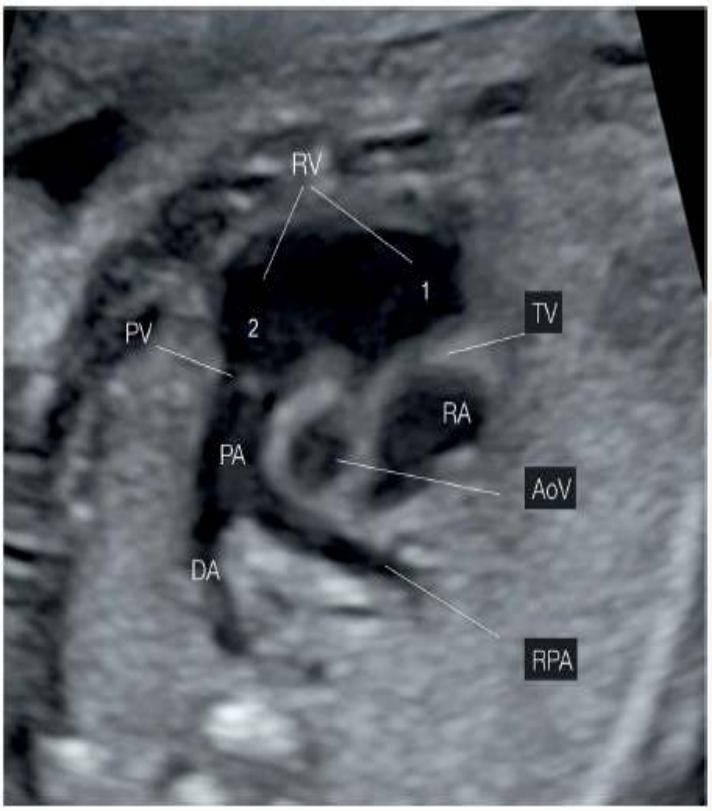
- RVOT view confirms the presence of a great vessel (Pulmonary artery) originating from the morphological right ventricle
- Pulmonary artery normally arises anteriorly & courses directly posteriorly to left of ascending aorta.
- The vessel originating from the RVOT can be confirmed as the pulmonary artery only if it branches into rt. & lt. branch after a short course
- PA continues posteriorly as the duct arteriosus & connects to descending aorta
- It is usually slightly larger than the aortic root during fetal life & crosses the ascending aorta at almost right angle just above its origin

- Ascending aorta is seen to the rt. of PA
- Pulmonary valve moves freely and should not be thickened.

RVOT VIEW



NORMAL RVOT WITH BOTH INFLOW (1) & OUTFLOW (2)



ABNORMALITIES OF RVOT VIEW

- Origin of great vessel (PA) from rt. Ventricle & it's division into rt. PA & lt. PA helps to rule out **Transposition of great arteries (TGA**)
- Cross section of asc. Aorta seen near the origin of PA – excludes abnormalities associated with parallel orientation of vessels like **TGA, DORV**
- **Stenosis/ hypolasia of PA** can be seen if present
- Small PA than aorta - **TOF**
- **Dilated PA** – as in **TOF with absent pulmonary valve syndrome** can also be picked up

THREE VESSEL VIEW (3VV) :

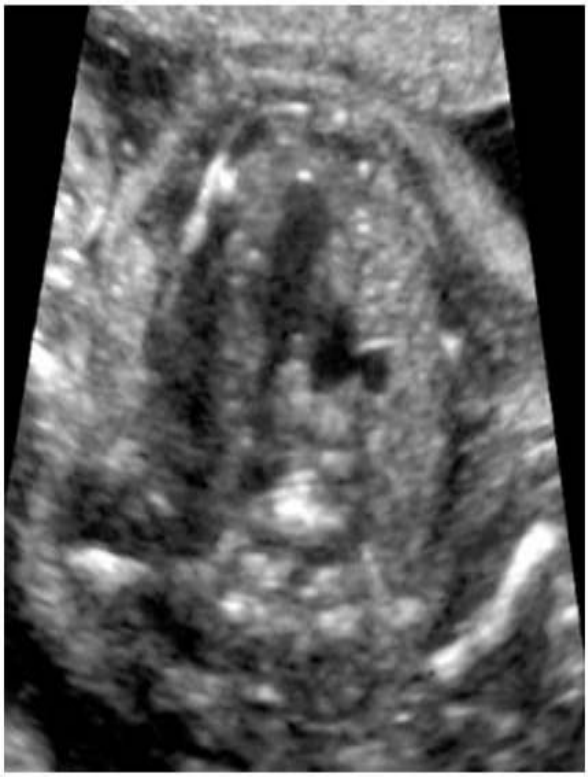
How to acquire 3VV –

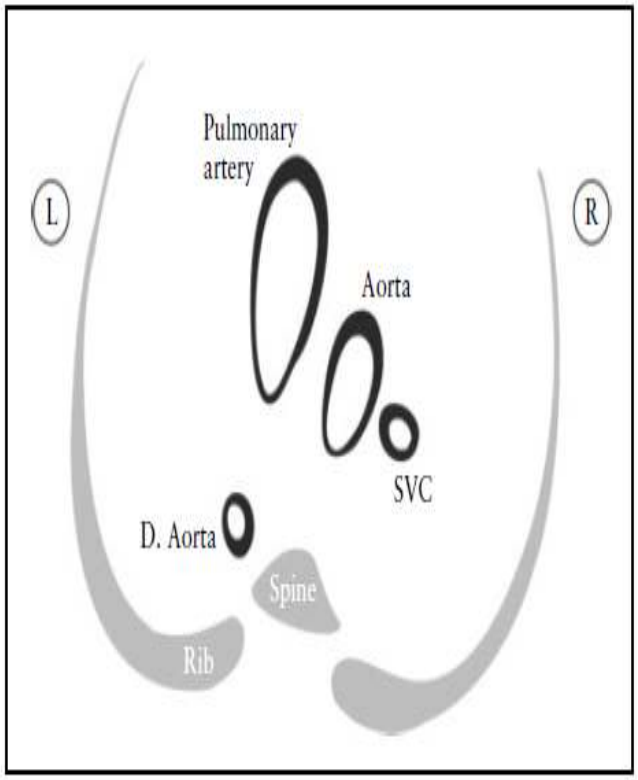
- By sliding the transducer more cranially from RVOT view & with slight manipulation 3VV is obtained.

3 VESSEL VIEW – check list :

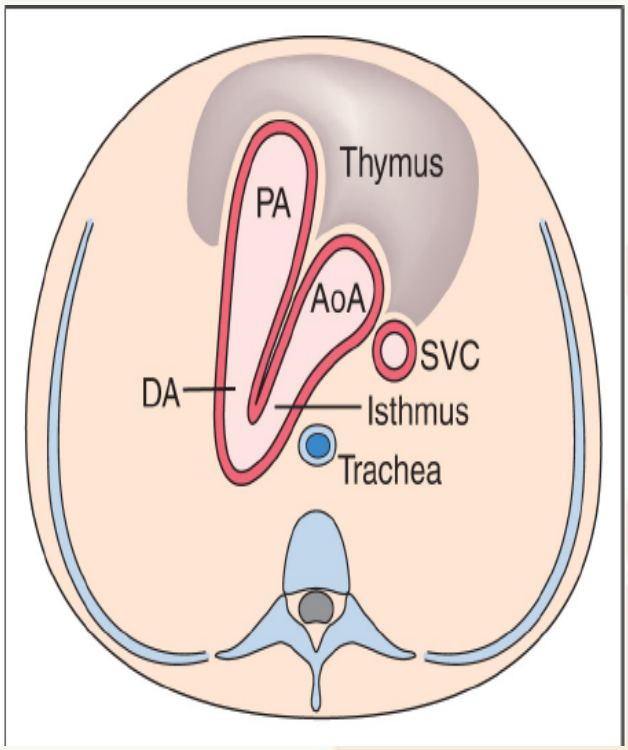
- 3VV includes – from left to right Pulmonary artery , Ascending Aorta & Superior vena cava
- Normally size of the vessel decreases from lt. to rt. – with PA being largest & SVC smallest
- PA is the most anterior vessel & SVC is the most posterior one
- 3VV is important for – assessment of vessel number, size, alignment & arrangement
- Certain abnormalities associated with normal 4 chm view but abnormal vessel size, number, alignment & arrangement are picked up on 3 vessel view easily

NORMAL 3 VESSEL VIEW :





3 VESSEL TRACHEA VIEW (3VT) :

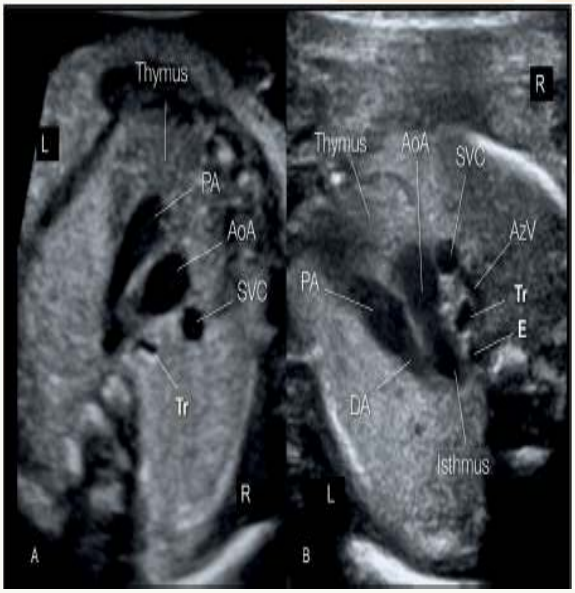


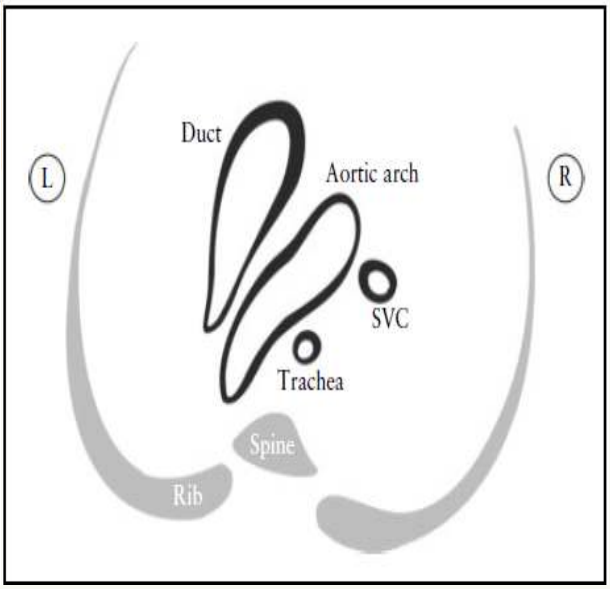
HOW TO ACQUIRE 3VT VIEW

- From the 3 vessel view, the 3VT view is obtained by slightly angulating the transducer caudally, towards fetal left side until fusion of aortic & ductal arches are seen.

3VT VIEW check list

- Both the aortic & the ductal arch together form a 'V' shape as they join the descending aorta to the left of the trachea.
- SVC is seen in a cross section to the rt. of aortic arch.
- Ductal arch, the largest of the 3, and more anterior in position; the aortic arch (AoA), slightly smaller than the ductal arch, in the middle; and the SVC, the smallest of all.
- Thymus is visualised as hypoechoic structure , anterior to 3VT & behind the anterior chest wall.





ABNORMALITIES OF 3VV & 3VT VIEW

Abnormality	Condition asssociated
Abnormal vessel number	• **2 vessels** (one great vessel & one SVC)/ Single great vessel– complete transposition of great arteries (TGA), Common arterial trunk, DORV • **4 vessels**- persistent left superior vena cava (PLSVC)
Abnormal vessel size	• **Small aorta/aortic arch**- coarctation of aorta, interruption of aortic arch ,critical Aortic stenosis, aortic atresia • **Small PA/absent PA**- Pulmonary stenosis or atresia, TOF, Tricuspid valve dysplasia • **Enlarged SVC**- Interrupted IVC with azygous continuation • **Dilated PA** – Isolated pul.valve stenosis, TOF with absent pul.valve
Abnormal vessel alignment & arrangement	• Corrected transposition of great arteries, DORV
Aortic arch to the rt. of trachea	• **Right aortic arch** with left duct ('U' shape is seen instead of a normal V on 3VT), Double aortic arch, Rt. Aortic arch with rt.duct (V to the right of trachea instead of left)
Connection between both great vessels	• **Aorto -pulmonary window**

CONCLUSION :

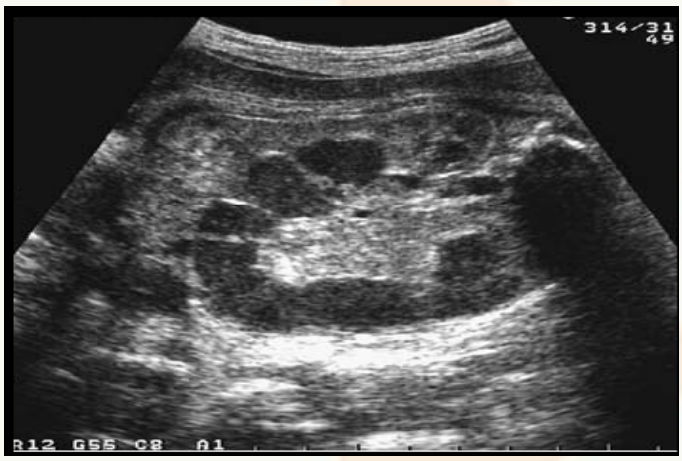
- Application of above described guidelines for fetal heart screening helps in maximum detection of CHD.
- Allows an obstetrician to give timely reference to a Fetal-medicine expert.
- Helps in counselling of patients & to engage in multi-disciplinary approach involving Obstetrician, Fetal medicine expert, Geneticist, Pediatric cardiologist.
- Also aids in identifying patients requiring Fetal echocardiography .

FETAL ABDOMEN- GI TRACT,ABDOMINAL WALL & KIDNEYS

- DR.NEHA MUNIYAR PUNIYANI

INTRODUCTION

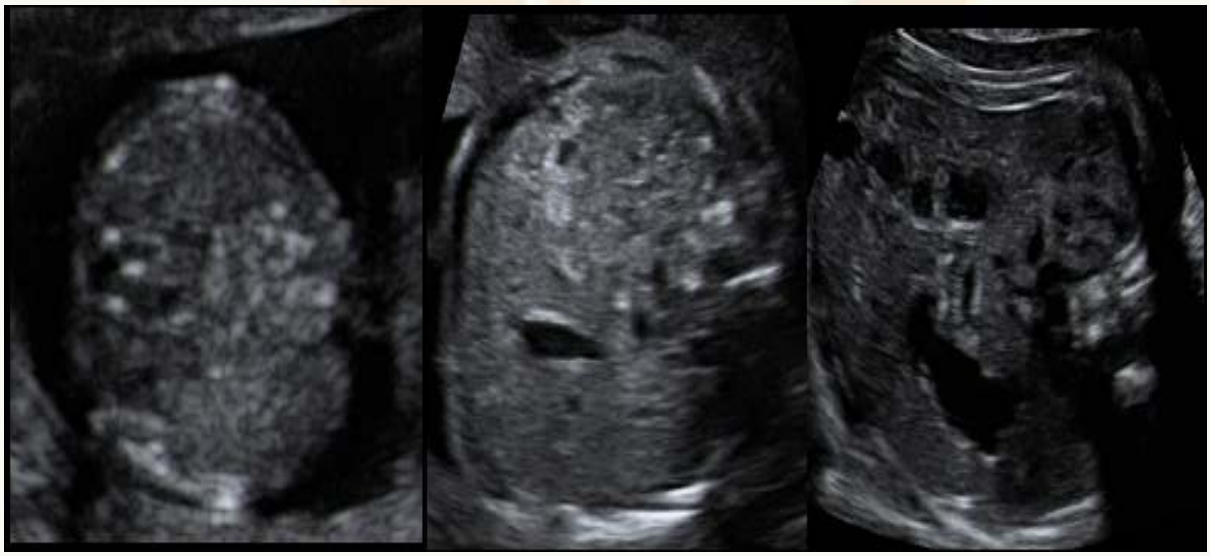
- Detection of gastro-intestinal (GI) tract abnormality can become particularly challenging due to overlap between appearance of normal & abnormal fetal bowel.
- Antenatal diagnosis of GI tract anomalies is quite difficult due to absence of any sonographic signs before the 3rd trimester.
- Another main differential feature of the GI tract is that its ultrasound (US) appearance varies significantly during pregnancy and also during the course of same ultrasound examination, due to the physiology of swallowing, stomach emptying, & intestinal peristalsis.



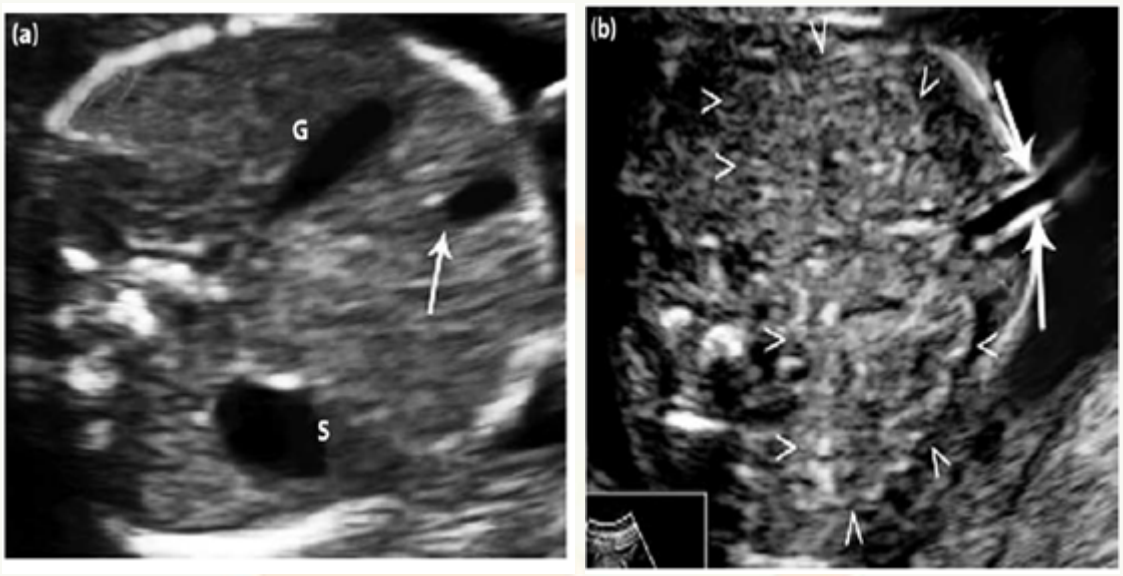
Axial view of abdomen in 35 weeks old fetus. Note the dilatation of colon with haustra. This finding may be indicative of an obstruction or may be normal

Varied US appearance of GI tract during the course of pregnancy

18 wks 24 wks 36 wks

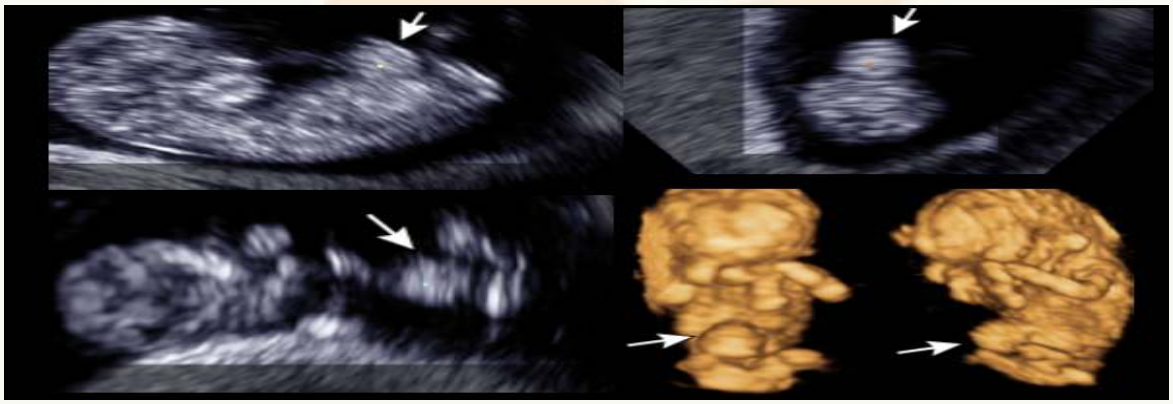


NORMAL APPEARANCE OF GI TRACT

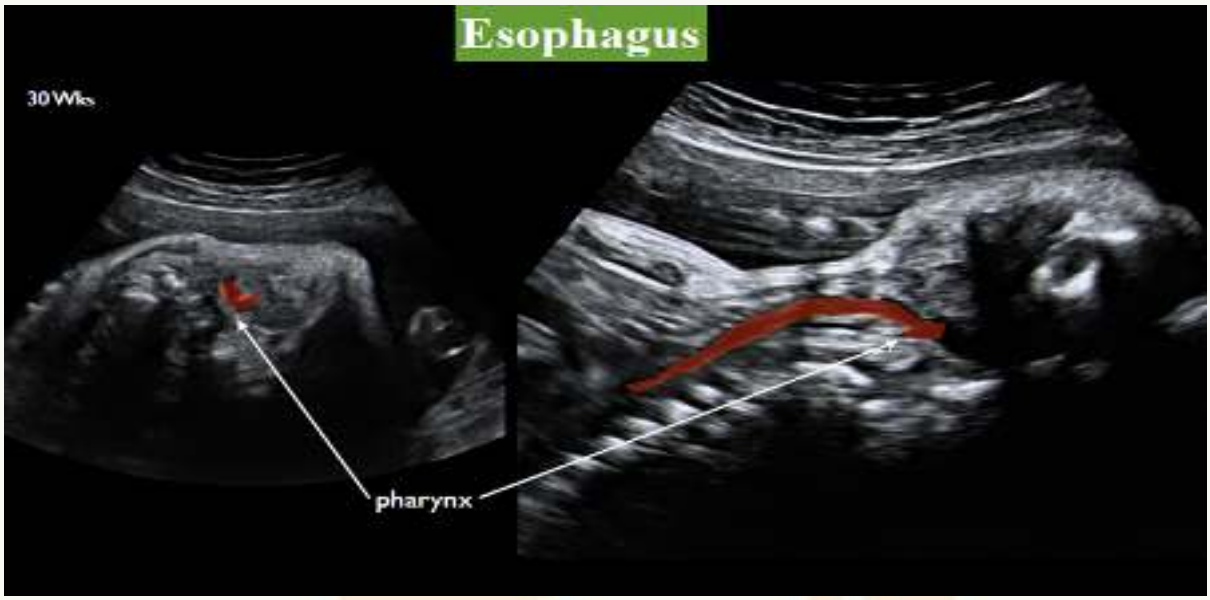


- Sonographically, fetal stomach (S) is visible as early as 9 weeks of gestation as a sonolucent cystic structure.
- Bowel is normally uniformly echogenic until 3rd trimester, when prominent meconium filled bowel loops of large bowel are commonly seen.
- Liver comprises most of the upper abdomen.
- Normal cord insertion (CI) can be visualised on mid-sagittal or axial section of abdomen.
- Gall bladder (G) is seen as a cystic ovoid structure to the right & below the intra-hepatic part of umbilical vein.

PHYSIOLOGIC MID-GUT HERNIATION

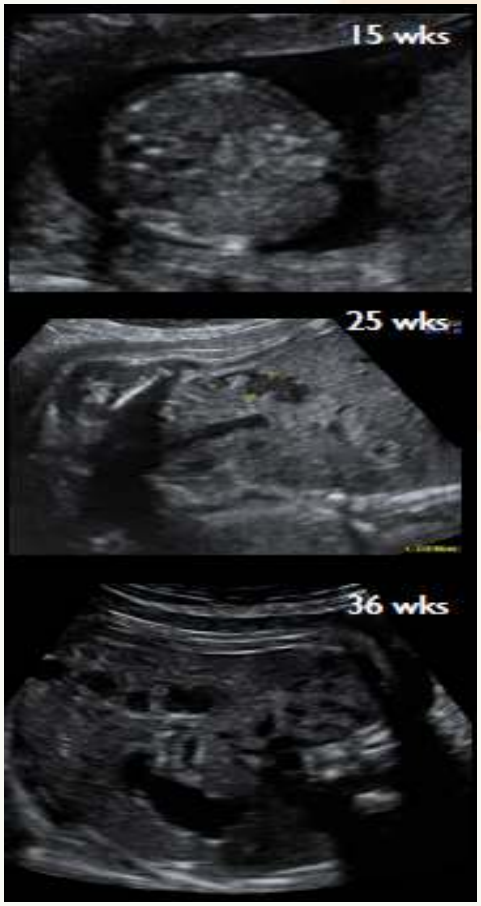


Physiologic Midgut Herination Between 9-11th Week due to rapid growth of the Intestine & Liver beyond the capacity Of the abdominal cavity, reduced by 11 weeks 6 days



- The fetal esophagus is normally collapsed & typically not visualized.
- Swallowing occurs by 11 To 14 weeks of gestation.
- Phases of swallowing can occur at 20-30 minutes

NORMAL ULTRASOUND APPEARANCE OF BOWEL



- In First And Second Trimesters:

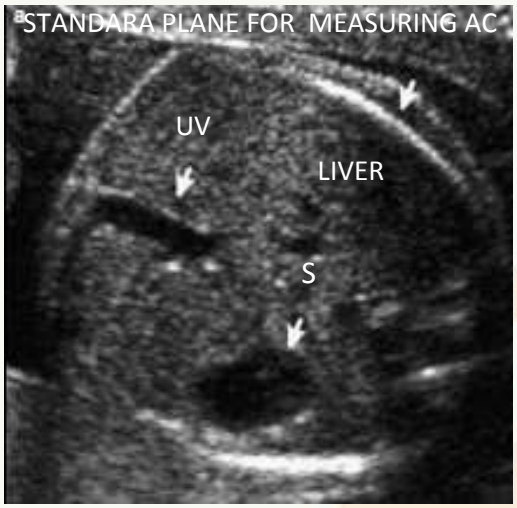
- Fluid in the lumen fills after 13 weeks

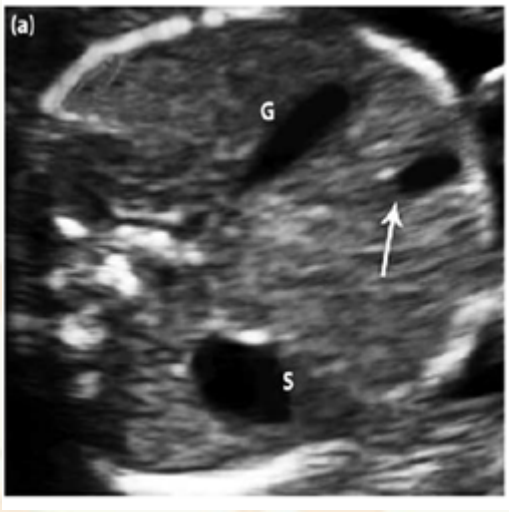
- Peristalsis can be observed as early as 18 weeks

- Colon is best visualized after 24 weeks as hypoechoic regions along the periphery of the abdomen.

- **Late Second and Third Trimesters**:
- Increased echogenicity with accumulation of meconium.
- Normal small bowel loops do not exceed 7 mm in diameter or 15 mm in length.
- The large bowel/colon can achieve a diameters up to 23 mm at term

NORMAL AXIAL VIEW OF UPPER ABDOMEN



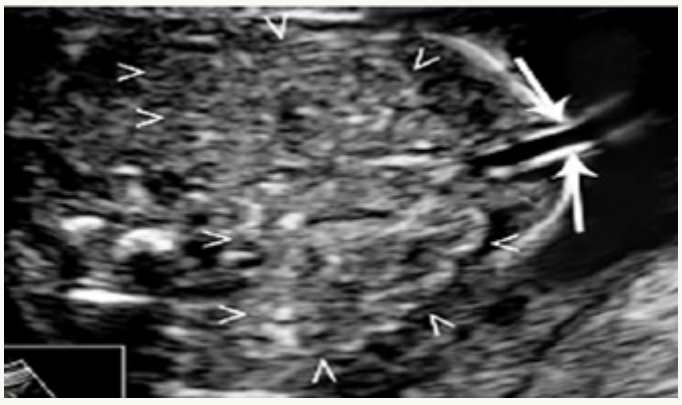


- On the left, the gastric bubble/stomach (S) is seen as well-defined , anechoic, round or oval area .
- On the right, most of the liver, seen as weakly hyper echogenic structure & the intrahepatic tract of the UV (anechoic).
- This the standard plane for measuring abdominal circumference (AC)
- On the right of the UV, the gallbladder(G) can also be seen in the right upper quadrant next to the liver.

GI ANOMALIES RECOGNISED ON AXIAL VIEW OF UPPER ABDOMEN

- **Oesophageal atresia** – non visualisation of gastric bubble
- **Duodenal atresia/stenosis** – double bubble
- Choledochal cyst- An anechoic cystic structure just below the liver
- Hepatomegaly - Increased liver volume
- Hepatic calcifications – hyperechogenic spots within the liver

NORMAL AXIAL VIEW OF LOWER ABDOMEN



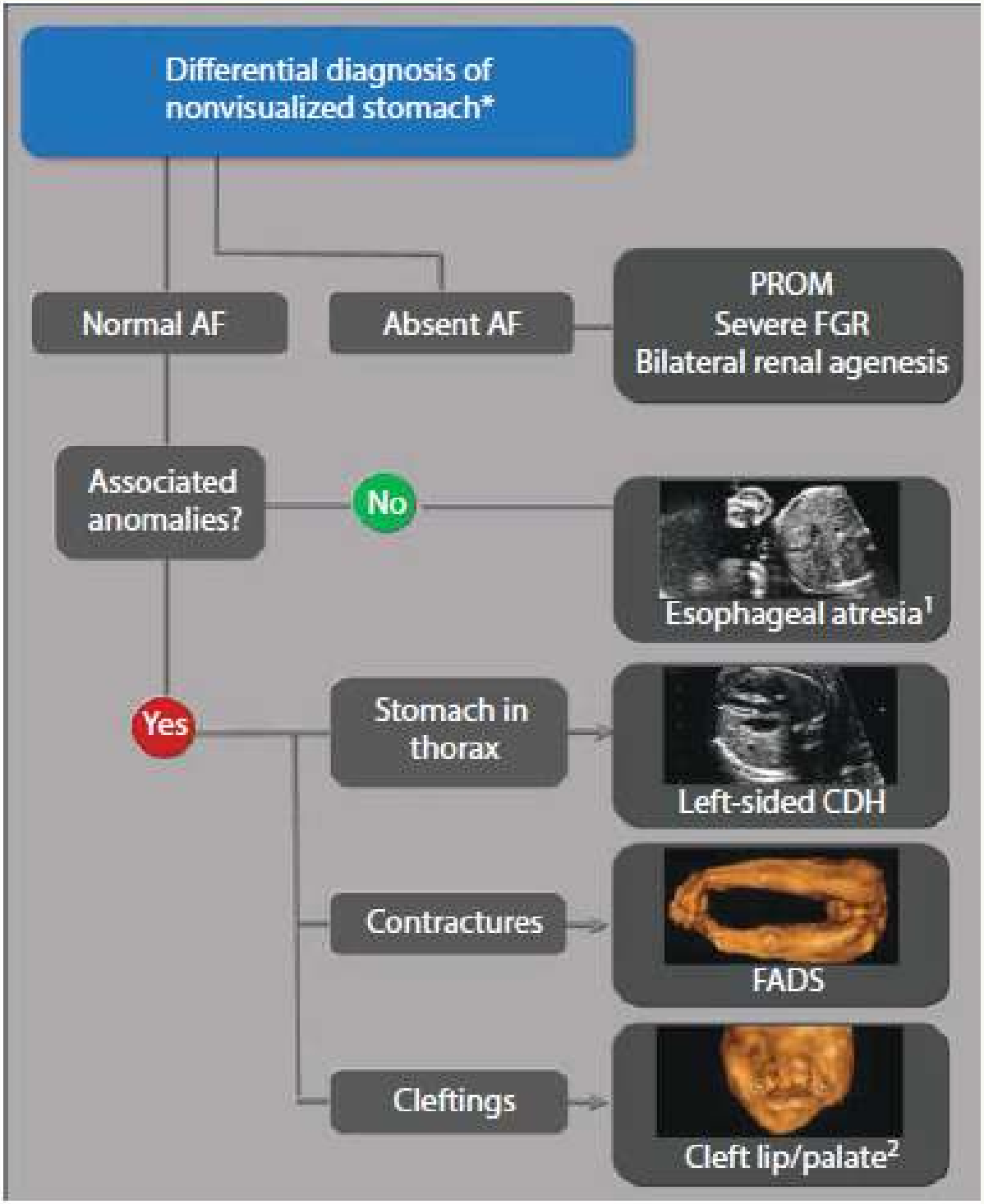
- This view is parallel & caudal to axial view of upper abdomen
- By sliding the probe caudally from the AC plane axial view of lower abdomen is obtained.
- On this view - small bowel (jejunum & ileum, arrow heads in the picture), transverse colon & the cord insertion (big arrows) can be recognized.
- The ileal loops appear weakly hyperechoic in comparison with the relatively hypoechoic colon.

ANOMALIES DETECTED ON AXIAL VIEW OF LOWER ABDOMEN

- Omphalocele- defect of the anterior abdominal wall, presenting as a sac containing bowel and/or liver, which bulges out from the cord insertion area.
- Gastroschisis – bowel loops floating freely in the amniotic fluid
- Small bowel atresia- severe dilation of ileal loops proximal to the atretic tract
- Meconium ileus - Diffuse hyperechogenicities and calcifications within the intestinal lumen, sometimes associated with small-bowel obstruction.

ULTRASOUND SIGNS SUGGESTIVE OF GI ANOMALIES

- Non-visualization of the gastric bubble.
- Cystic lesions
- Dilated small bowel
- Dilated large bowel
- Echogenic bowel
- Large liver
- Abdominal wall defects


Differential diagnosis of nonvisualized stomach*
Normal AF
Absent AF
PROM
Severe FGR
Bilateral renal agenesis
Associated anomalies?
No
Esophageal atresia[1]
Yes
Stomach in thorax
Left-sided CDH
Contractures
FADS
Cleftings
Cleft lip/palate[2]

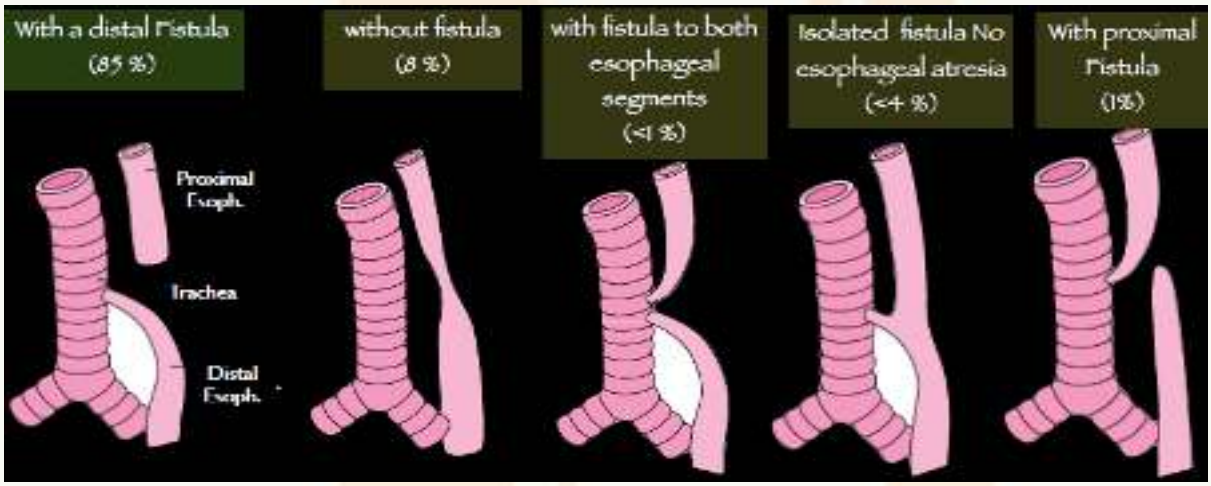
OESOPHAGEAL ATRESIA :

- **Incidence** : 1/2500 – 1 /4000 live births.
- Etiology: failure of division of the primitive foregut into the ventral tracheobronchial part & the dorsal digestive part around about 8 weeks of gestation
- **Associated Anomalies**:

✦Chromosomal anomalies: (20–44%):Trisomy 21 & to a lesser extent 18.

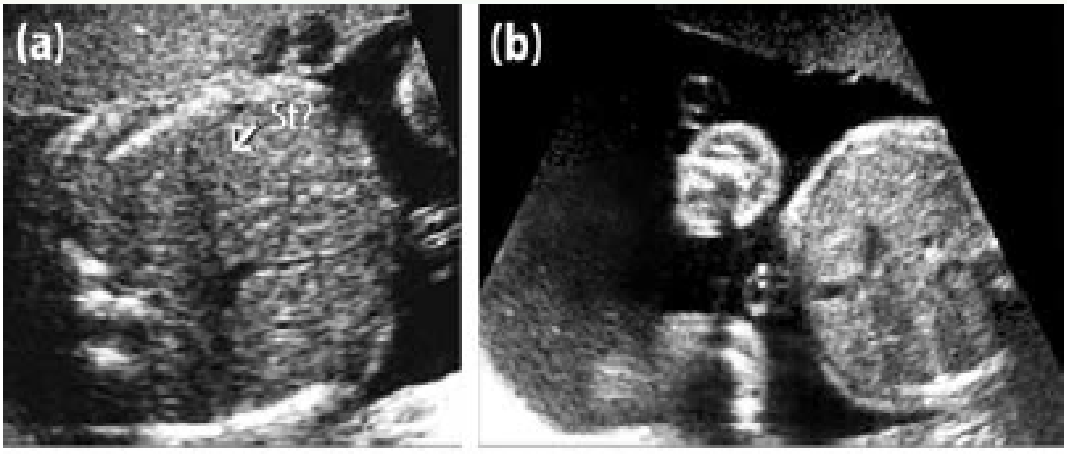
✦Non-chromosomal Syndromes: 50 % have additional anomalies, mainly cardiac defects & also as a part of VACTERL anomaly (vertebral defect, anal atresia, cardiac defects, tracheoesophageal fistula, renal anomalies, single umbilical artery).

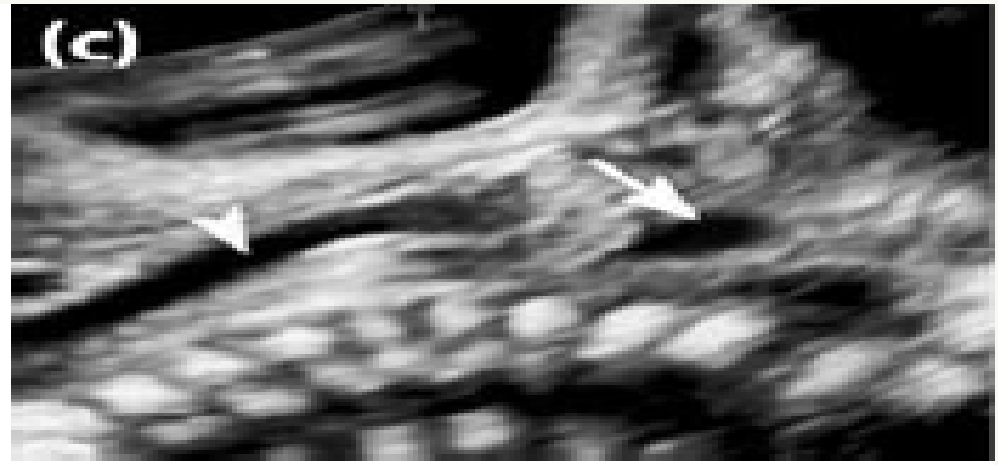
5 TYPES OF OESOPHAGEAL ATRESIA



- Small gastric bubble may be seen due to normal gastric secretions & in the presence of TE fistula due to secretions from the fistula.
- Presence of TE fistula - responsible for poor prenatal diagnosis of Esophageal atresia.

ULTRASOUND FINDINGS



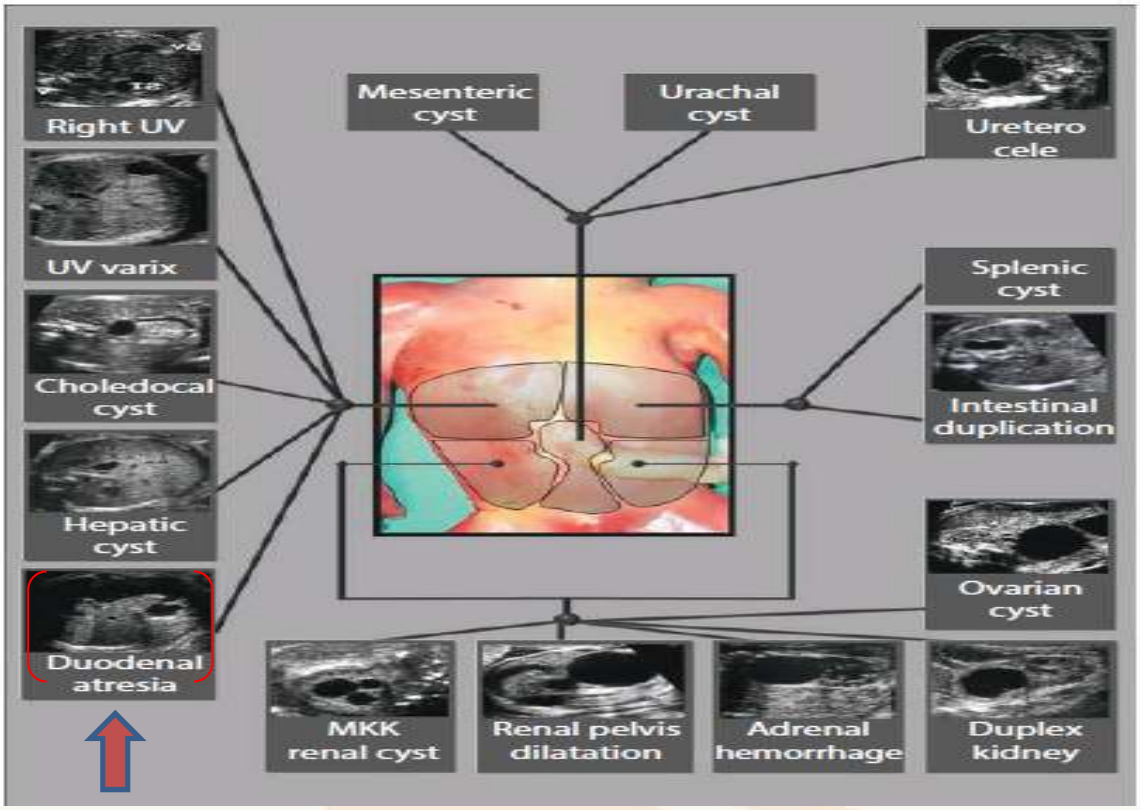


- **Diagnostic triad** (8-10 % cases):
- ✓ **Polyhydramnios** (image b**)**: becomes evident in late 2nd trimester
- ✓ **Absent/Small Stomach** (image a) :in 85% cases stomach is seen
- ✓ **"Pouch Sign"** (image c): dilated proximal oesophageal during swallowing (arrow)
- Overall detection rate of oesophageal atresia considering all possible signs of ranges from -24 – 42%

OBSTETRIC MANAGEMENT

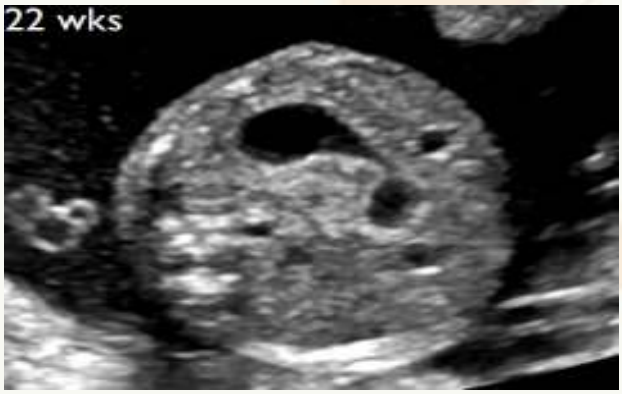
- Assess for associated anomalies
- Genetic amniocentesis
- Delivery at tertiary care centre.
- Esophageal abnormalities alone are not an indication for altering the route of delivery
- **Outcome depends on**:
  - Extent of the atretic tract
  - Associated anomalies

DUODENAL ATRESIA :



- **Duodenal atresia is one of differential diagnosis of abdominal cystic lesions**

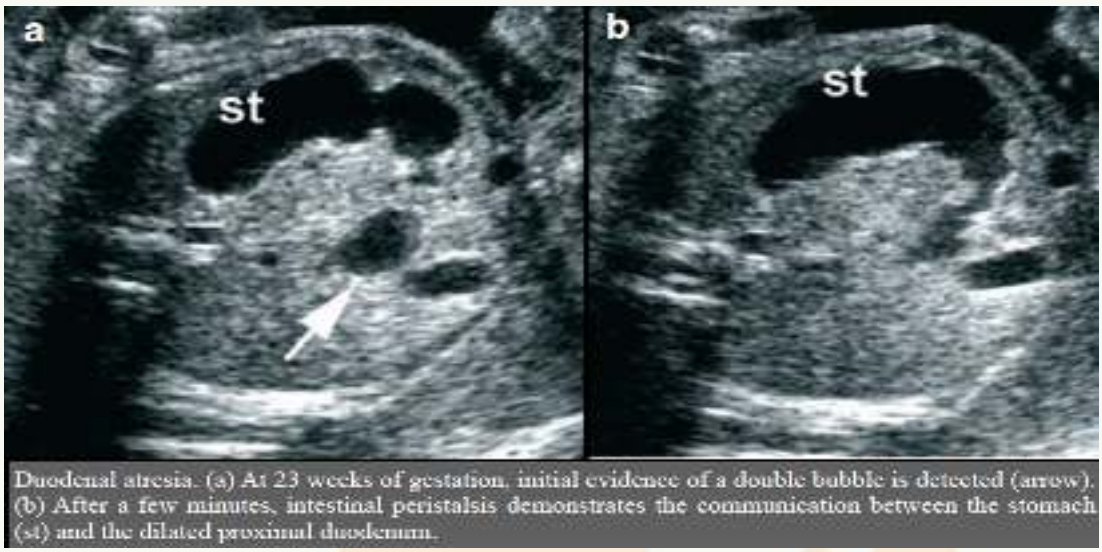
DUODENAL ATRESIA :



- **Definition**: condition where tract between the proximal & distal portions of the duodenum is atretic
- **Incidence**: 1/2500 - 1/10,000 life births
- **Associated Anomalies**:

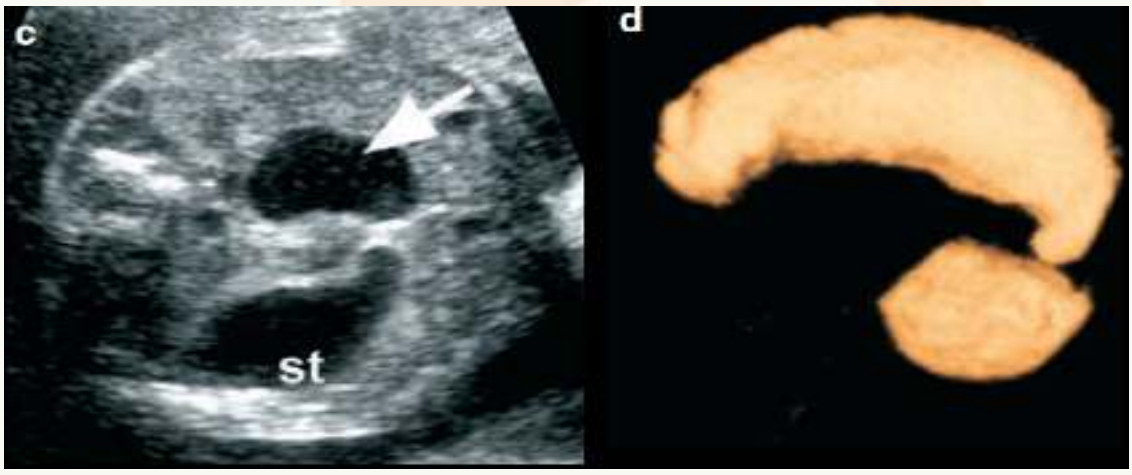
- Chromosomal anomalies (50%): 40% association with Trisomy 21
- Non Chromosomal Anomalies:40–50% cases (GI tract anomalies, skeletal defects , cardiac & renal defects)

ULTRASOUND DIAGNOSIS:



Duodenal atresia. (a) At 23 weeks of gestation, initial evidence of a double bubble is detected (arrow). (b) After a few minutes, intestinal peristalsis demonstrates the communication between the stomach (st) and the dilated proximal duodenum.

- **'Double bubble**' sign: of dilated stomach & proximal duodenum with communication between the two
- **Polyhydraminos** : seen in late 2nd & early 3rd trimester
- In the absence of communication between stomach & duodenum : DD of other upper abdominal cysts should be considered



(c) Later in gestation, a clear double bubble (arrow) has developed, confirming the suspicion of duodenal atresia.

(d) Three-dimensional ultrasound with inversion mode rendering: the site of the obstruction is clearly visible

- Diagnosis may be suspected late in pregnancy because of constantly dilated stomach with evidence of the pylorus in late gestation

OBSTETRIC MANAGEMENT :

- Karyotyping for amniocentesis
- Search for associated malformations (including fetal echocardiography)
- Measure against risk of preterm delivery because of severe polyhydramnios
- Delivery in a tertiary care centre

Prognosis:

- Isolated cases have overall survival of about 90%
- Late- onset sequelae may develop: e.g. mega duodenum , gastro oesophageal reflux, and peptic ulcers.

SMALL BOWEL ATRESIA (ILEAL & JEJUNAL) :

- Common cause for dilated bowel on ultrasound
- **Incidence:** 1 /2500 – 1 /5000 live births

Ultrasound diagnosis:

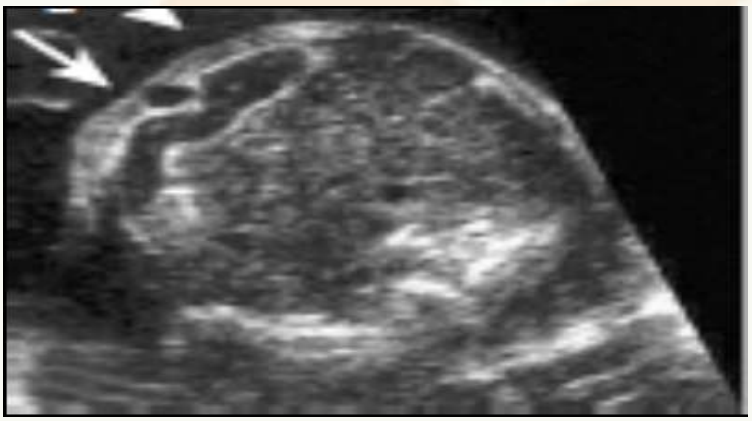
- Late- onset severe **dilatation (diameter > 7mm) of the ileal loops** proximal to the obstruction.
- **Polyhydramnios**- late onset
- Other signs – contributing to diagnosis are:

- central abdominal location of the affected loop
- its hyperechoic walls
- increased peristalsis & presence of abdominal calcifications

Associated Anomalies:

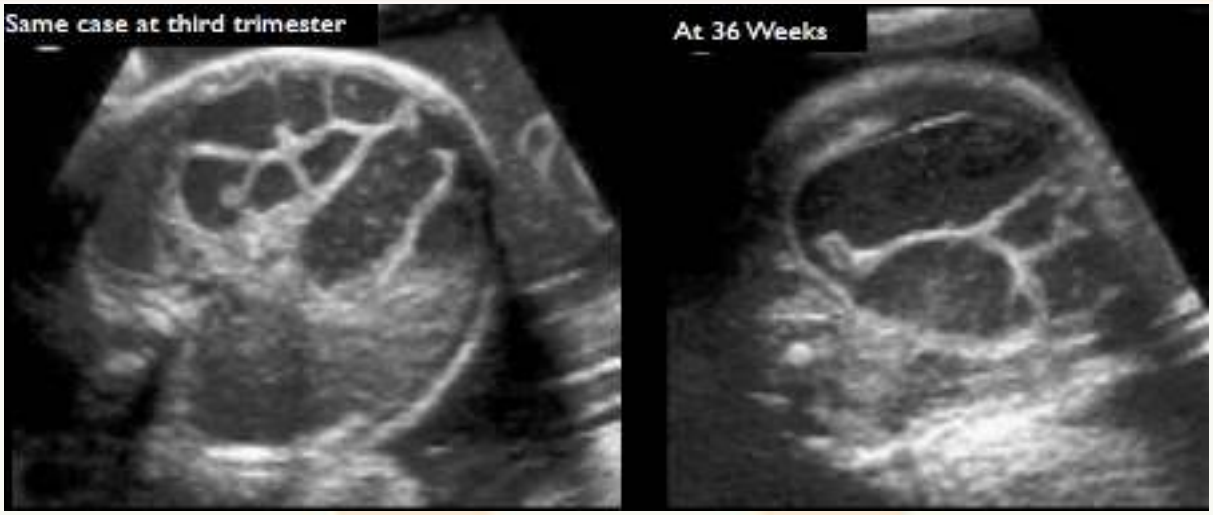
- **Low risk** of both chromosomal & non-chromosomal anomalies

ULTRASOUND FINDINGS : 24 Weeks Suspected Abnormal Dilatation



- Axial midlevel abdominal scan at 24 weeks showing doubtful sign of atresia

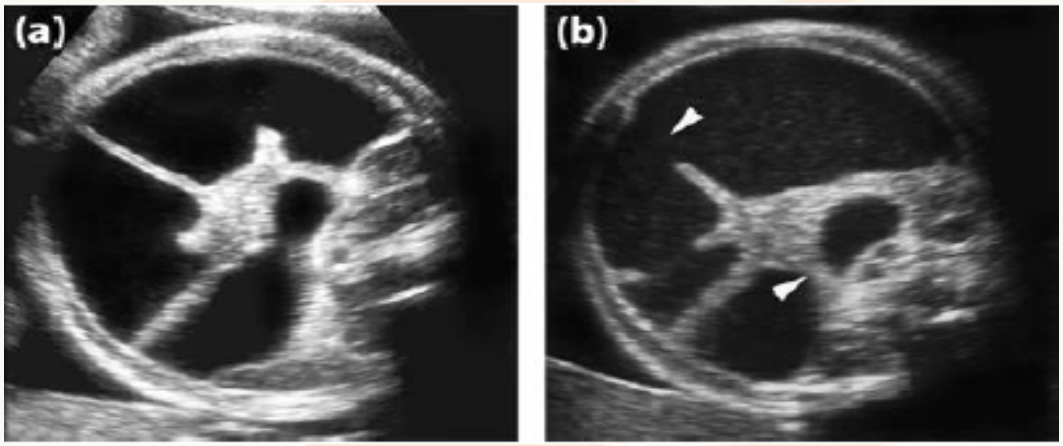
Moderate dilatation (>7mm) of a single Ileal / jejunal loop & hyperechoic bowel walls



-The obstruction becomes evident, with moderately severe dilatation of various loops.

-The communication between the various dilated segments (the maximum transverse diameter of the loops was 23 mm).

JEJUNAL ATRESIA ULTRASOUND FINDINGS :



Jejunal atresia (37 weeks of gestation). Note the extremely severe dilation without evidence of perforation (absence of meconium peritonitis). The arrowheads indicate the site of the peristaltic wave, opening and closing the communication between adjacent loops from (a) to (b).

- **Differentiation between ileal Or jujenal atresia** is difficult
- Only points are - evidence of intestinal perforation (ascites with particulate matter and/or calcifications) for the ileal & extreme dilatation without perforation for the jejunal.

OBSTETRIC MANAGEMENT :

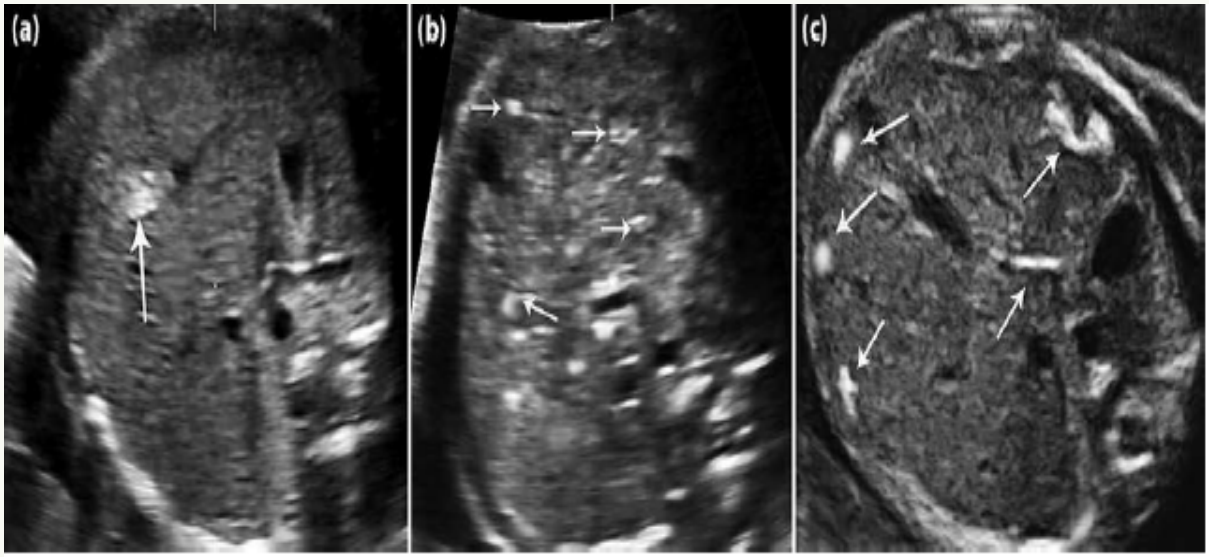
- Karyotyping : not recommended due to low risk of chromosomal anomalies
- Increased risk of pre-term delivery – due to polyhydraminos
- Delivery should be planned in tertiary referral centre

MECONIUM PERITONITIS :

- Condition that occurs as a result of antenatal bowel perforation
- **Incidence**- 1/35,000 live births (rare), but higher in utero
- **Ultrasound diagnosis** - peritoneal calcifications can appear as a continuous line on the border of the peritoneum, as scattered calcifications throughout the peritoneum, or as a focal calcification with shadowing
- **Risk of chromosomal anomalies**- low
- **Risk of non chromosomal syndromes**: risk of cystic fibrosis is high in fetus (10%) & in neonates(15-40%)
- **Outcome**-In the presence of simple peritonitis, generally the outcome is good and surgical intervention is not necessary. In cases of complex peritonitis, the outcome is more guarded.

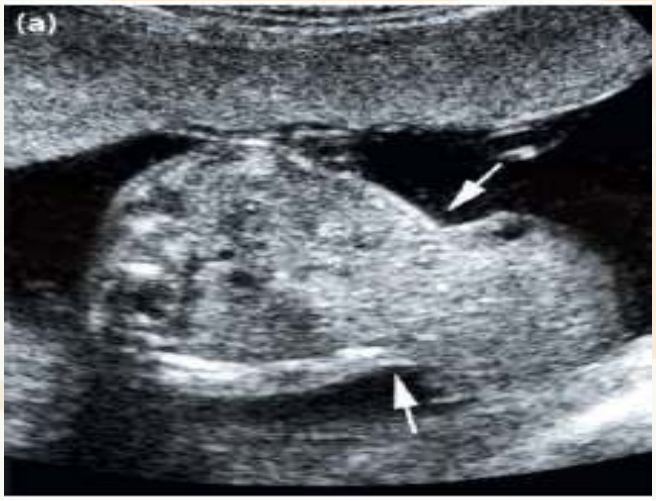
HEPATIC CALCIFICATIONS :

- **Incidence** – infrequent
- Calcifications can be divided as- peritoneal, parenchymal & vascular
- **Etiology**- **Peritoneal** calcifications may be due to meconium peritonitis,
  Parenchymal due to intrauterine infections(rubella,CMV,parvovirus),
  Vascular due to thrombo -embolism of hepatic &portal veins
- **Ultrasound diagnosis**- one or more calcified foci can be visualized, or even a diffuse calcification of the liver or of its peritoneum
- Risk **of both chromosomal & non-chromosomal anomalies - low** (if calcifications are isolated)
- **Outcome** - good if the maternal serology is negative, the fetal karyotype is normal, and no other fetal anomalies are associated



Hepatic calcifications- Axial view of the fetal abdomen showing (a) an isolated intrahepatic calcification (arrow); (b) multiple intra-parenchymal calcifications (arrows) due to a varicella infection; and (c) multiple calcifications (arrows) along the liver capsule due to meconium peritonitis.

OMPHALOCELE :

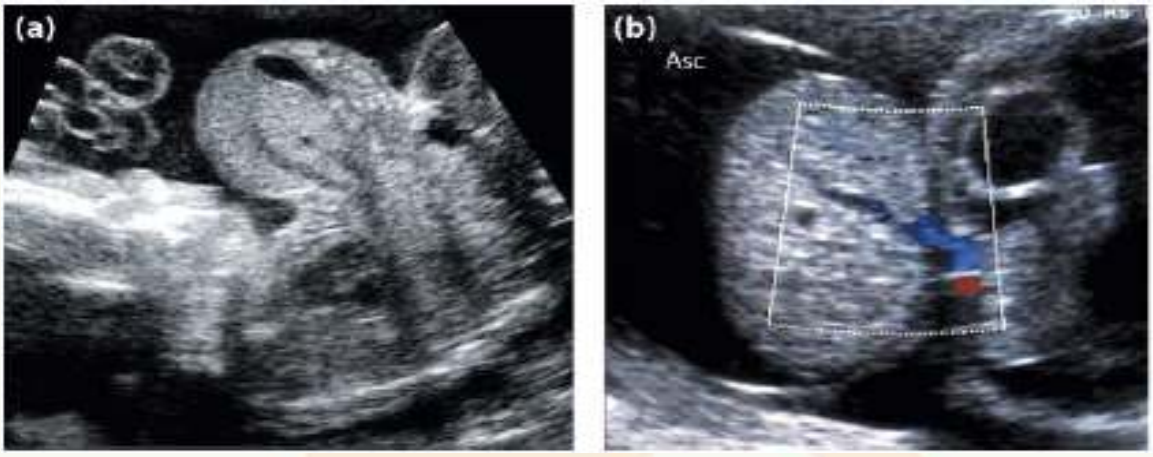


(a) At 23 weeks of gestation, the axial view of the abdomen demonstrates a large omphalocele containing the liver (the arrows indicate the large wall defect)

- Omphalocele is a defect in the closure of the abdominal wall that also involves the cord insertion
- **Incidence**- relatively infrequent, 1/3000 live births, but higher in utero
- **Usg diagnosis-** on usg it is seen as a bulging structure that (on axial & midsagittal view)
  - arises from the anterior abdominal wall

- contains some abdominal viscera (liver and/or bowel)
- defect is covered by membrane
- cord inserts on the apex/top of the sac

- Risk of **chromosomal anomalies- high** (15% to 40%): trisomy 18 ,13 & triploidy
- Risk of **nonchromosomal syndromes**- relatively high: Beckwith–Wiedmann syndrome & pentalogy of Cantrell (PC)
- **Outcome**- - Good if the lesion is isolated and the liver is not completely herniated.
  - Very poor in the case of associated malformations and/or chromosomal Aberrations



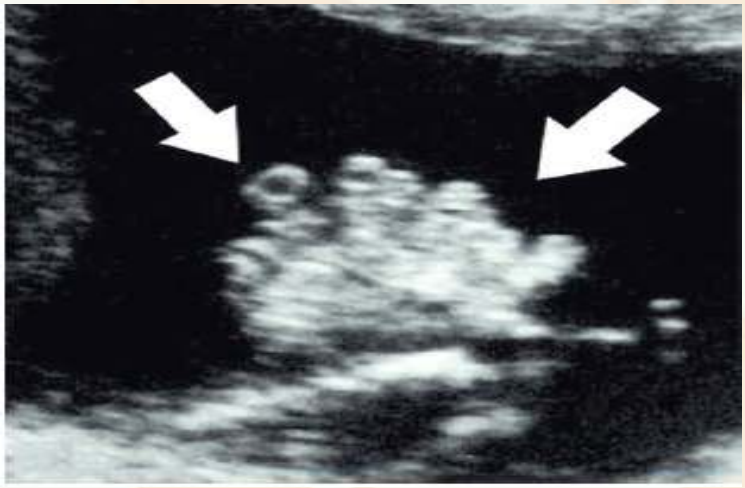
(a)Midsagittal view of the abdomen: a case of omphalocele containing the liver at 29 weeks of gestation (b) Rarely, ascites can be associated with the omphalocele and can be detected in the sac (Asc); color Doppler
shows the umbilical vein.

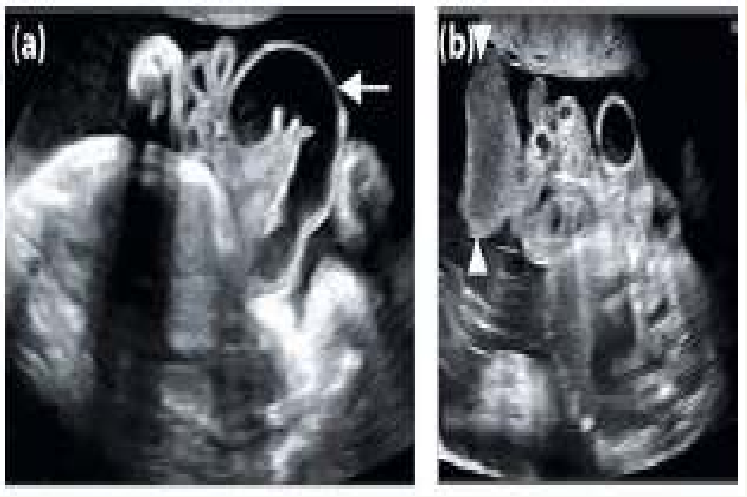
(c) The still born fetus: omphalocele (arrowhead), with the cord insertion

GASTROCHISIS :

- Gastroschisis is characterized by **a para-umbilical defect** of the abdominal wall through which bowel loops herniate to float freely in the amniotic fluid
- **Incidence**- 1/4000 births

- **Ultrasound findings** :
  - normal cord insertion
  - herniated bowel /intestinal loops floating freely in amniotic fluid
  - para-umbilical wall defect (generally on the right side)
  - defect is not covered by membrane
- Since bowel loops are directly exposed to the environment through a small defect – chances of perforation, infraction & infection of bowel are more.
- Risk of **chromosomal anomalies** - very low
- Risk **of non chromosomal syndromes** – low (10-30%)
- **Outcome**.-Very good, unless rare complications including perforation, infarction, or infection of the herniated loops occur.
- Image below showing bowel loops floating freely in the amniotic fluid



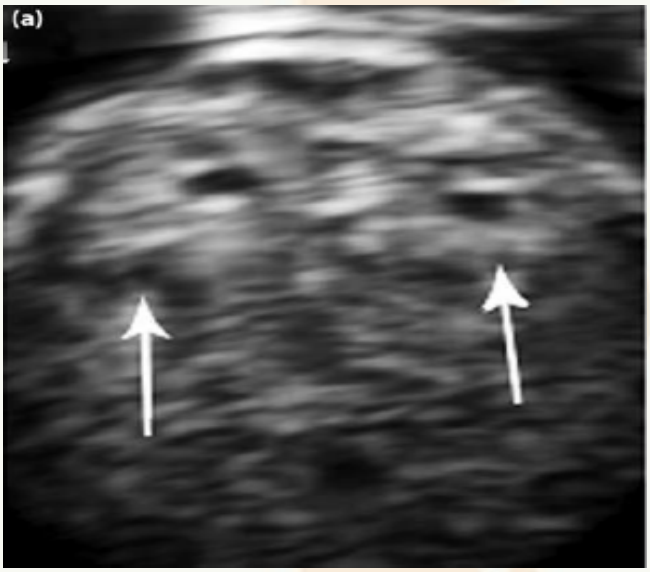


(a) Sagittal view showing some normally sized loops & one severly dilated tract (arrow)
(b) Axial view showing meconium blocked in dilated loop (arrowhead)

CONCLUSION :

- Diagnosis of GI tract anomalies remains challenging.
- Majority of the abnormalities are detected in late 2nd or 3rd trimester, this should be discussed with the couple.
- Therefore, in case of any doubt on ultrasound patient should be referred to an expert dealing in fetal ultrasound for further work-up & counselling.
- Due to late diagnosis in majority of cases, counselling becomes difficult.

FETAL KIDNEYS & BLADDER :



Normal fetal kidneys at 13 weeks of gestation. At this stage, the kidneys appear as bilateral hyperechoic structures in the paravertebral regions (arrows)

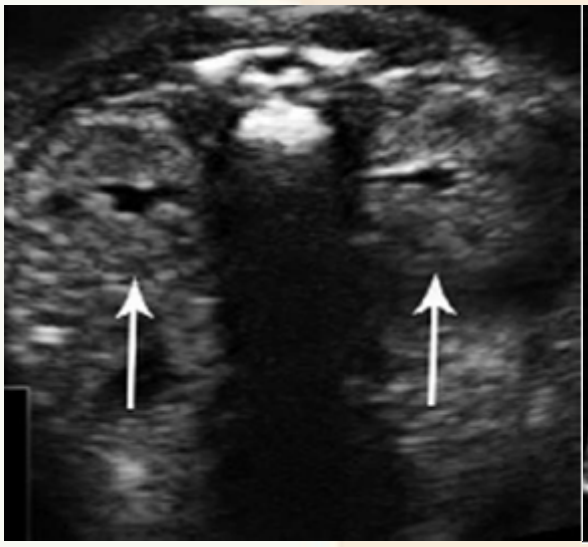
Ultrasound anatomy of fetal kidneys :

- Fetal kidneys can be visualized on ultrasound in most cases by 11-12 weeks.
- Two scanning planes are used: axial & longitudinal.
- On axial views, the kidneys appear as two round paravertebral structures with pelvis oriented toward the midline.
- On longitudinal views, they appear elliptic
- The adrenal glands are located just cranial to kidneys
- On axial view : width, renal pelvis & thickness of kidneys can be measured(picture)
- On longitudinal view : length of kidneys can be measured

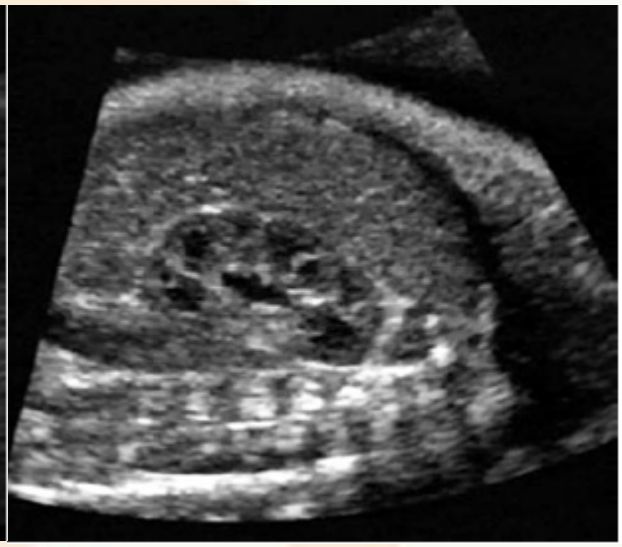
-At 11-12 weeks, they appear as two hyperechoic paravertebral structures; in the 2nd trimester, they lose their hyperechoic appearance & by 3rd trimester it is possible to distinguish between the cortex and the medulla

-The cortex is peripheral to the medulla & is slightly more echogenic.

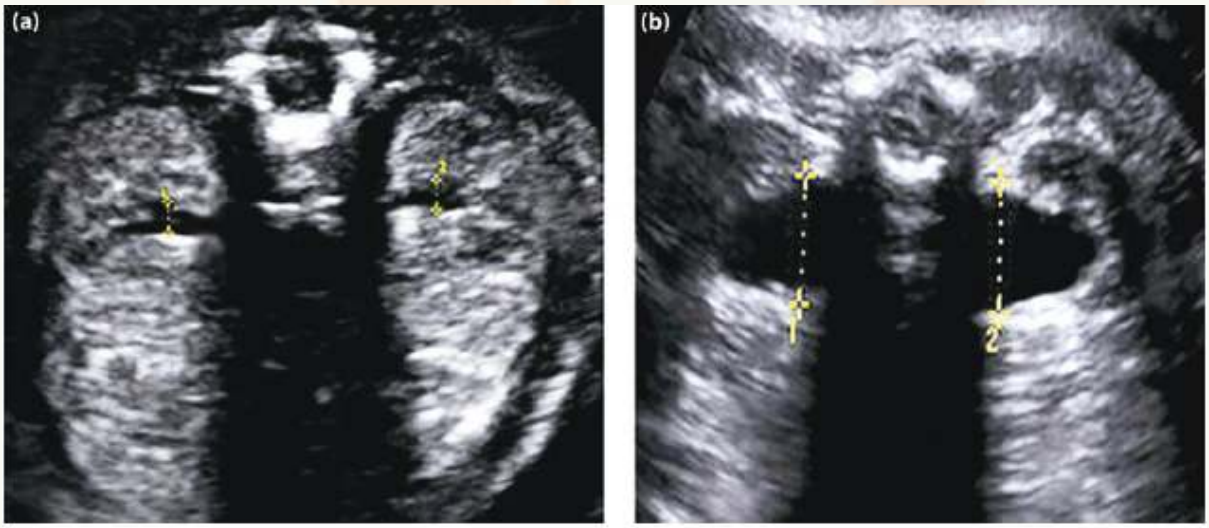
-Size of renal pelvis on axial section should not be more than, 4mm before 29 weeks & not more than 7mm from 29 weeks till term



Axial view at 22 weeks- kidneys appear less Echoic(arrows) than in 1st trimester



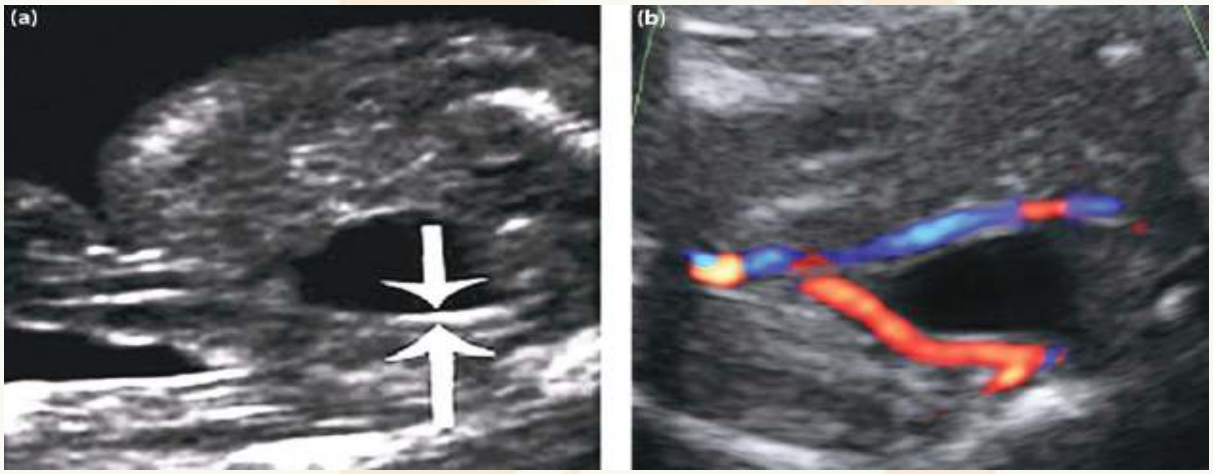
Longitudinal view at 28 weeks



Axial scan through the fetal abdomen, showing measurement of the normal (a) and enlarged (b) renal pelvises

BLADDERS & URETERS :

- On US, the bladder can always be visualized by 12 weeks in the middle of the fetal pelvis as a circular anechoic structure with echoic walls.
- The thickness of its walls should never exceed 2–3 mm.
- Perivesical arteries run laterally to the bladder, are easily seen on color Doppler.
- Presence of the these arteries surrounding the bladder on either side serve as important landmark to identify bladder & differentiate it from any other cysts seen in fetal pelvis.
- Ureters & urethra : Both of these structures are not visible prenatally, except in cases of disease.



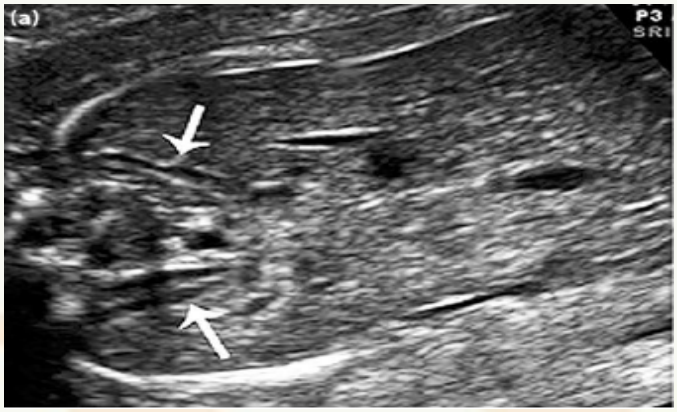
(a) Axial scan through the fetal pelvis showing the bladder wall (arrows). (b) Color flow Doppler image showing both perivesical arteries separating around the bladder (arrow)

RENAL ANOMALIES -

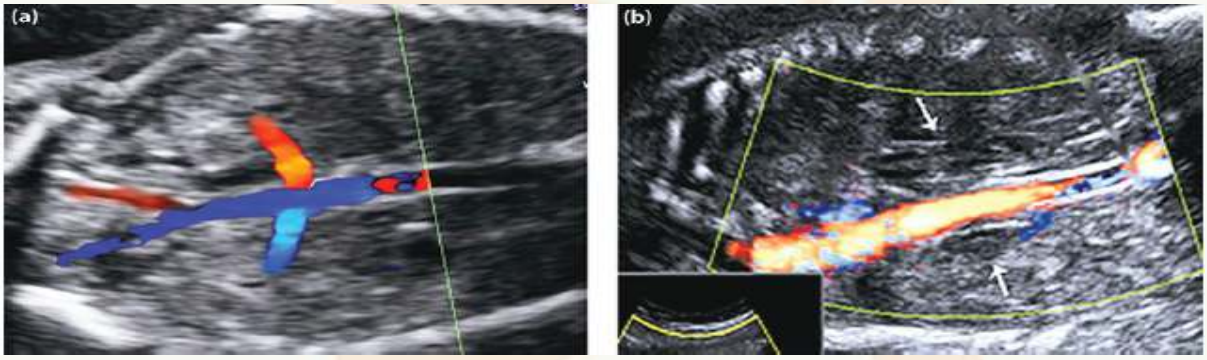
RENAL AGENESIS :

- **Incidence**-1–2/1000 for the unilateral form & 1/3000–4000 for the bilateral form.
- **US diagnosis -Bilateral form**: lack of visualization of the kidneys & bladder associated with severe oligohydramnios (after the 16th week). **Unilateral form**: lack of visualization of one kidney, with normal bladder & amniotic fluid
- **Risk of chromosomal anomalies**: Low risk in isolated unilateral forms (<1%); slightly higher risk in isolated bilateral renal agenesis.
- **Risk of nonchromosomal syndromes**. High: 20%–25%.

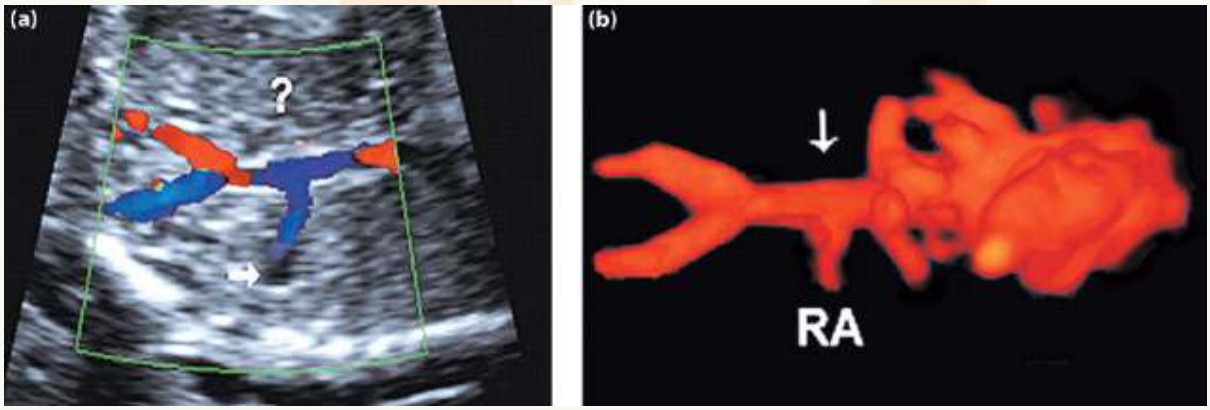
- **Outcome - Bilateral form**: uniformly fatal**. Unilateral form**: good, if isolated



Bilateral renal agenesis. (a) The absence of both kidneys is evident despite the associated oligohydramnios; the arrows indicate both adrenal glands in the paraspinal regions.



(a) Color flow Doppler shows both renal arteries in a 22-week fetus. (b) Absence of the renal arteries is noted in a case of bilateral renal agenesis, while the aorta is clearly shown; the arrows indicate the adrenal glands

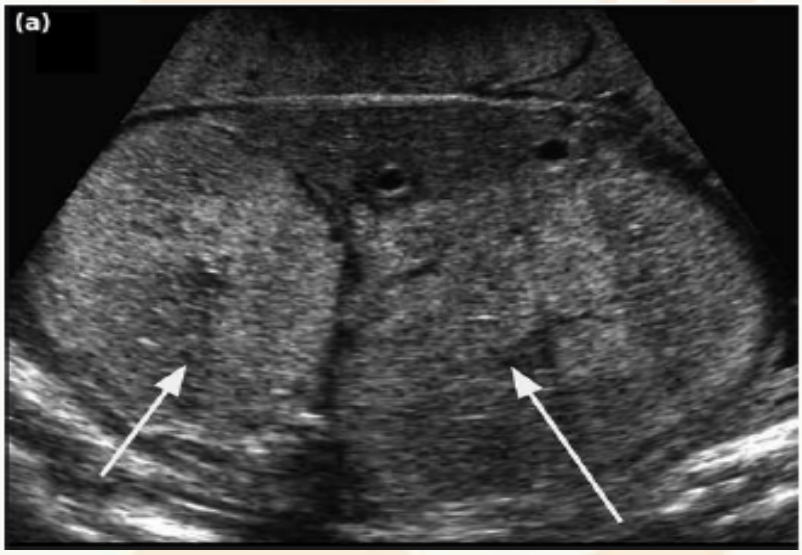


Unilateral renal agenesis. (a) Color Doppler image demonstrating a single renal artery. The arrow indicates the pelvis of the single kidney; ? indicates the empty contralateral renal area. (b) 3D power Doppler of the same case, showing a single renal artery (RA) branching off the abdominal aorta; the arrow indicates the absence of a contralateral renal artery

ECHOGENIC DYSPLASTIC KIDNEYS

AUTOSOMAL RECESSIVE POLYCYSTIC KIDNEY DISEASE (ARPKD) :

- **Incidence** -1/20,000–40,000 newborns.
- **Ultrasound diagnosis**:
  - Grossly enlarged kidneys(>95th centile)
  - Hyperechogenic with loss of corticomedullary(CMD) differentiation
  - Non-visualisation of bladder and severe oligohydramnios
- **Risk of chromosomal anomalies**: It's a single gene disorder, generally not associated with karyotypic alterations
- **Risk of non-chromosomal syndromes**: High, if the polycystic aspect (and not the genetic cause) is considered.
- **Outcome** : Unfavourable, due to associated severe pulmonary hypoplasia (severe oligo) .



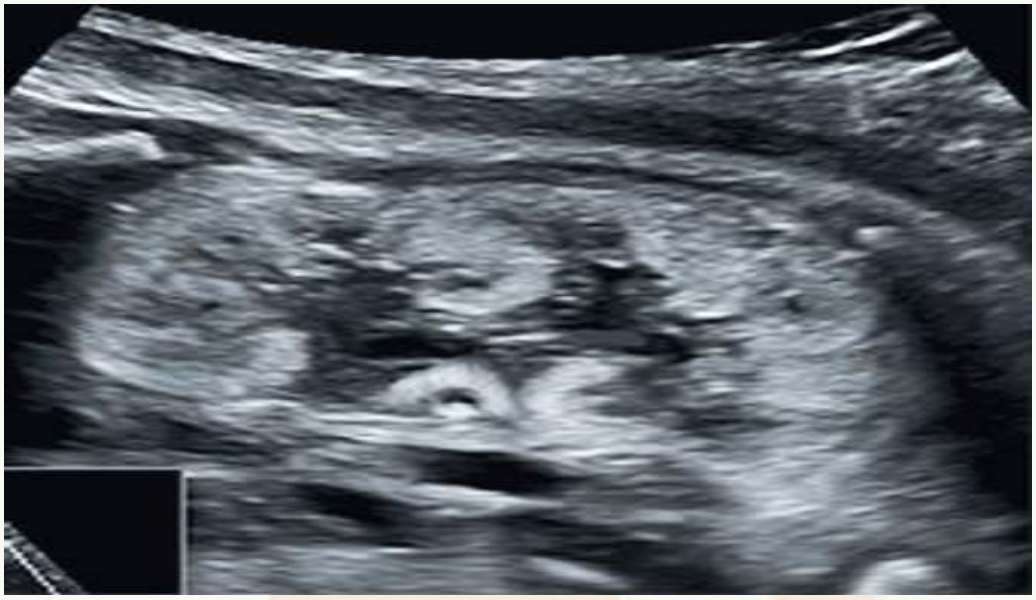
ARPKD - (a) Axial scan through the fetal abdomen showing the enlarged hyperechoic kidneys (arrows).

AUTOSOMAL DOMINANT POLYCYSTIC KIDNEY DISEASE :

- **Incidence** -1/1000 live births
- **US diagnosis**:

- Kidneys are moderately enlarged
- Hyperechoic (often the cortex/periphery only – due to microcysts) with visible pelvis
- CMD is exaggerated
- Bladder can be visualized & amniotic fluid is normal or slightly reduced

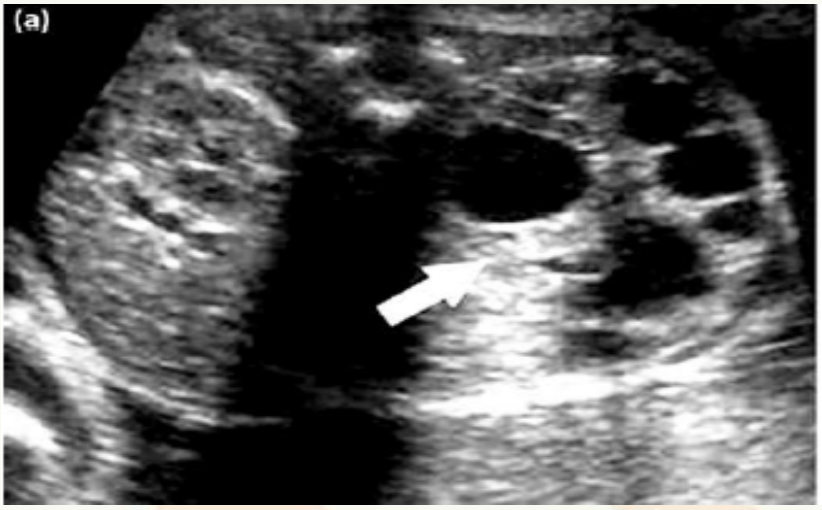
- **Risk of chromosomal anomalies**- It's a monogenic disorder
- **Risk of non-chromosomal syndromes**- Relatively high, if the polycystic aspect (and not its cause) is considered.
- **Outcome** - although the symptoms usually occur in the 3rd to 5th decades of life, the prognosis is apparently worse in the prenatally detected forms.
- Parents will have similar kidneys & should be checked.



ADPKD: Ultrasound image showing an enlarged kidney with increased differentiation between the cortex and the medulla.

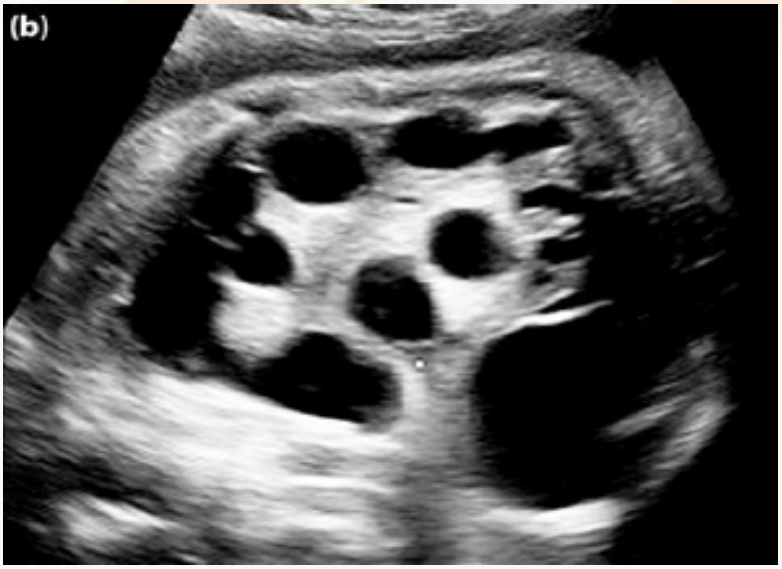
MULTICYSTIC DYSPLASTIC KIDNEYS :

- **MCDK** : Is a non-hereditary condition, characterised by an enlarged kidney whose parenchyma is replaced by multiple, non communicating macro cysts of variable size and number
- **Incidence**- 1/1000–5000 live births
- Unilateral in approximately 75%–80% of cases.
- **Ultrasound diagnosis**- Unilateral: the kidney is enlarged, with multiple non communicating cysts of variable size; the parenchyma is hyperechoic; there is a normal amount of amniotic fluid and bladder is visualized. Bilateral: same as above + severe oligohydramnios and inability to visualize bladder**.**



(a) Axial scan through the fetal abdomen showing an enlarged right kidney (arrow) with multiple cysts within the hyperechoic parenchyma; the contralateral kidney is normal

- **Risk of chromosomal anomalies-** relatively low in isolated unilateral forms (2%–4%); reaches 15%–18% in bilateral forms & 25%–28% when associated with other anomalies
- **Risk of non chromosomal syndromes-** 5%–10% (VACTREL ,Meckel- gruber syndrome)
- **Outcome**- **Unilateral form**: involution of the kidney, resulting in hypoplastic kidney in a significant percentage of cases within the first two years of life.
  Bilateral form: unfavourable prognosis

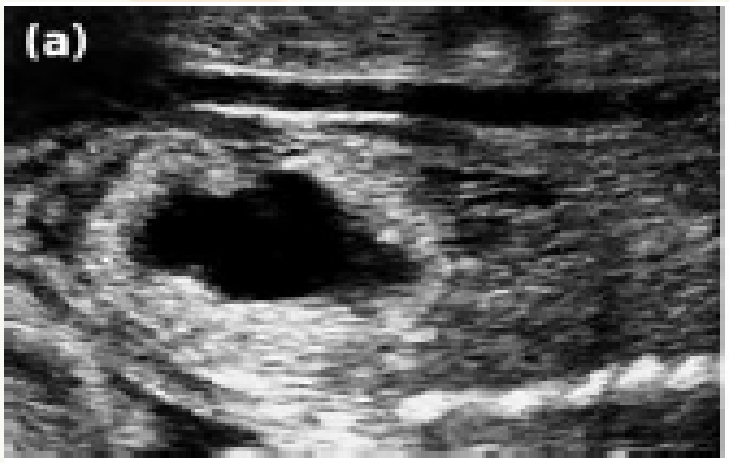


(b) Coronal scan through the fetal abdomen showing an enlarged kidney with increased echogenicity and numerous non communicating cysts

OBSTRUCTIVE CYSTIC DYSPLASIA :

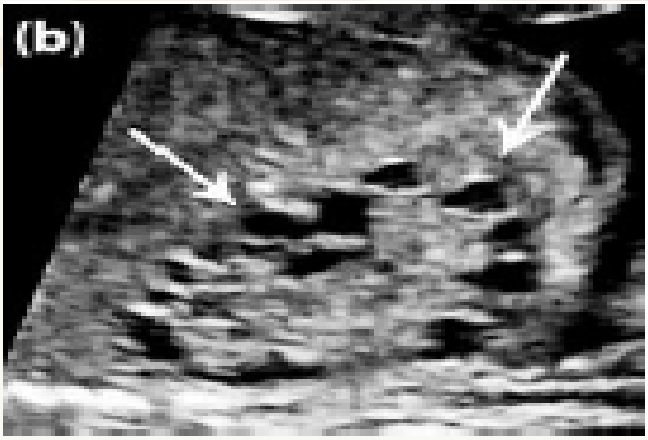
- Occurs due to early & severe obstruction of the collecting system causing pressure changes with formation of cysts mainly in the cortical region; kidney volume is often decreased
- **Incidence**- Bilateral form is more frequent
- **Ultrasound diagnosis**-

  - Kidney(s) of normal or reduced dimensions with increased echogenicity of the parenchyma & presence of cysts in variable positions (frequently pericortical)
  - If bilateral, oligohydramnios & a dilated bladder with thick walls are present



(a) Longitudinal scan through the fetal abdomen showing the hydronephrotic kidney with echogenic parenchyma.

- **Risk of chromosomal anomalies –**5%–10% in isolated forms; higher if associated with other anomalies (15%– 25%)
- **Risk of non chromosomal syndromes** - 3%–6% in isolated forms
- **Outcome** - In isolated forms, this depends on the severity of the obstruction and the extent of the dysplasia.



(b) Ultrasound image of the cystic kidney showing several subcortical small cysts(arrows)

OBSTRUCTIVE KIDNEY DISEASES :

- Can be due to-
  - Upper urinary tract obstruction OR
  - Lower urinary tract obstruction

Causes of urinary tract dilatation on antenatal Usg:

- Transient/physiologic	50-70 %
- Uretero pelvic junction (UPJ) obstruction	10-30 %
- Vesico ureteric reflux(VUR)	10-40 %
- Uretero-vesical junction obstruction	5-15 %
- MCDK	2-5 %
- Posterior urethral valve	1-5 %

TRANSIENT & PHYSIOLOGIC HYDRONEPHROSIS :

- Have normal postnatal evaluation
- **Etiology**
  - Increased fetal urinary output
  - Fetal ureteric folds
  - Vesicoureteral reflux
- In general, the fetuses with transient dilation tend to have AP diameters of less than 6 mm in the second trimester and less than 8 mm in the third trimester.

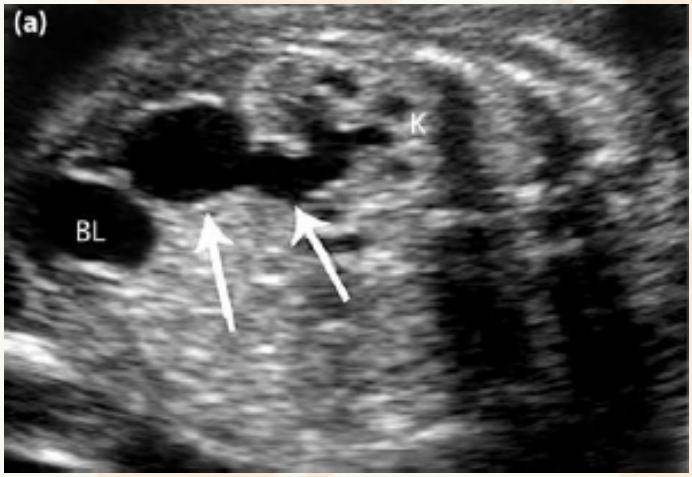
URETERO PELVIC JUNCTION OBSTRUCTION :

- **Incidence**: 1 in 2000 births
- Male to female ratio: 3:1 , Bilateral in 20-25%
- **Diagnosis**:
  - Dilation of renal pelvis
  - Dilation of calyces
  - No dilation of ureter & normal liqour volume
- Associated renal/ extra-renal anomalies – 12 to 25%
- **Outcome** -Usually good prognosis
  -19-25% baby may require some form of surgical intervention postnatally

URETEROVESICAL JUNCTION OBSTRUCTION :

- **Incidence**- 5 to 15%
- Bilateral in 25% cases
- **Etiology**

  - Primary megaureter
  - Ureter stricture or atresia
  - Vascular obstruction
- **Diagnosis:**
- Dilated ureter and renal pelvis(hydronephrosis & hydroureter)
- Normal bladder
- Peristaltic wave of ureter can be seen
- **Prognosis**: generally good, dilation Increases in utero, but decreases after birth.



(a) Hydronephrosis and dilation of the ureter (arrows) are present. K: kidney; BL: bladder.

VESICO-URETERIC REFLUX :

- **Incidence** - 10 to 40% of antenatal hydronephrosis

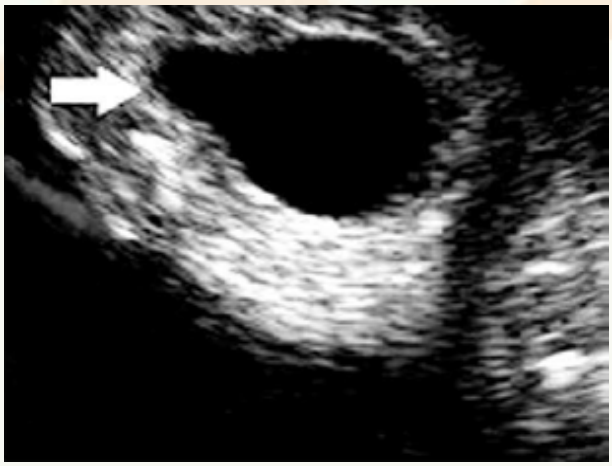
Etiology

- Transient bladder outlet obstruction that resolves prior to delivery
- High voiding pressure resulting in distortion of the vesicoureteric junction
- Delayed maturation of the vesicoureteric junction
- Genetic cause in some cases
- Diagnosis:
- Renal pelvic dilation
- Unilateral/bilateral ureteral dilations

- Variability in the diameter of renal pelvis during one single exam
- The **diagnosis** is confirmed with a postnatal voiding cysto-urethrogram

LOWER URINARY TRACT OBSTRUCTION :

- Most common cause is posterior urethral valves in males and urethral atresia in females
- Rarer causes- anterior urethral valves, prolapsed ureterocoele, congenital megalourethra
- **Posterior urethral valve (PUV):**
- **Incidence** : 1:8000 to 1: 25000 boys
- Occurs exclusively in males
- Associated anomalies (40%)- cardiac malformations, GI malformations, aneuploidy(5-8%)
- **Etiology**: membranes develop within the posterior urethra causing bladder outlet obstruction & can lead to back pressure changes like VUR, cyctic renal dysplasia.
- **Diagnosis**:
  - megacystis (markedly enlarged bladder)
  - Dilated posterior urethra **(keyhole sign**)
  - thickened bladder wall
  - severe oligoamnios
  - bilateral hydronephrosis,
  - hydroureter
  - Renal dysplasia features (small echogenic kidneys, cystic dysplasia)



-Significant dilatation of the bladder and of the proximal part of urethra (arrow) is clearly seen.

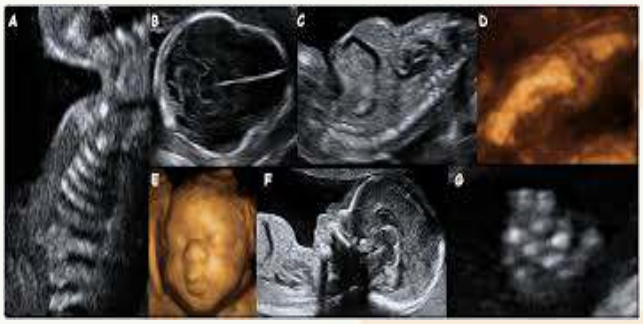
- Perinatal mortality is about 50% even with fetal intervention
- Mortality is mainly due to pulmonary hypoplasia & renal failure
- Antenatal intervention- vesicoamniotic shunt & fetal cystoscopy can be done.

FETAL SKELETAL DYSPLASIA

- DR UNNATI SHENDE

FETAL SKELETAL DYSPLASIA

- Incidence – 1:4000 births
- 25 % are stillbirth and 30 % neonatal death



ETIOLOGY OF FETAL SKELETAL DYSPLASIA

- Complex
- Chromosomal abnormalities
- Genetic defects
- Intrauterine factors
- Maternal disorders
- Exposure to teratogens
- Environmental factors

CURRENT PROTOCOL FOR SCREENING

- Assessment of fetal limbs – during first trimester nuchal translucency scan (NT scan) and second trimester (anomaly scan)- Evaluation of all three segments of limb –proximal , mid and distal segments

- Third trimester- fetal dimensions and movements

APPROACH TO PRENATAL DIAGNOSIS

- There is a wide range of skeletal dysplasia, each with a specific recurrence risk, dysmorphic expressions and implications for neonatal survival, quality of life.
- The incidental discovery of a skeletal dysplasia on routine ultrasound screening in a low risk pregnancy necessitates a systemic examination of limbs, head, thorax, spine and chest to arrive at the correct diagnosis.

LIST OF COMMON LETHAL SKELETAL DYSPLASIA

- Thanatophoric dysplasia
- Osteogenesis imperfecta
- Achondrogenesis
- Atelosteogenesis
- Hypophosphatasia
- Short rib polydactyly syndrome
- Camptomelic dysplasia
- Diastrophic dysplasia

TYPES OF LIMB SHORTENING

Limb segments term	Proximal segment (humerus / femur)	Midsegment (radius,ulna,tibia,fibula)	Distal segment (hand and foot)
Rhizomelia	short	normal	normal
Mesomelia	normal	short	normal
Acromelia	normal	Normal	Short

Micromelia	Short	Short	Short

WORKSHEET FOR SUSPECTED SKELETAL DYSPLASIA

Long bones – **All long bones to be measured**

Bones		Measurement (mm)	Standard measurement for _____ wks	Mineralization	Curvature	Fractures
Femur	Right					
	Left					
Tibia	Right					
	Left					
Fibula	Right					
	Left					
Humerus	Right					
	Left					
Ulna	Right					
	Left					
Radius	Right					
	Left					

Bones	
	Absent
	Hypoplastic
	Any malformation

Thorax

Chest circumference to be obtained at the level of the four chamber view of the heart

	Measurement	Normal measurement for _____ wks
Chest circumference		
Chest Cir/ Abdominal Cir		

Clavicle

		Measurement	Normal measurement of clavicles at _____ wks
Clavicles	Right		
	Left		

Hands and feet

Foot measurement

	Measurement	Normal measurement of foot at _____ wks
Foot		
Femur/foot ratio		Normal = 1

Shape of thorax –
Bell Shaped – Yes / No

Any bones missing?
Feet – Yes / No
Hands –Yes / No
Polydactyly
Preaxial
Postaxial
Syndactyly Yes / No
Postural deformities
Clubfeet Yes / No
Clubhand Yes / No

Micrognathin: Yes / No
Short upper lips: Yes / No
Abnormally shaped ears: Yes / No

Fetal Motion – Normal / Decreased
AFI – Normal / Decreased / Increased

Skull and Face:

Mucrocranin: Yes / No
Mineralization of skull bones: Normal / Decreased
Frontal bossing: Yes / No

Spine

Relative length – Normal / Decreased
Mineralization of vertebral bodies – Normal / Decreased
Vertebral Height – Normal / Decreased

Biorbital diameter – Distance between the inner margins of the orbits

	Measurement	Normal measurement of biorbital diameter at __ wks
Biorbital diameter		

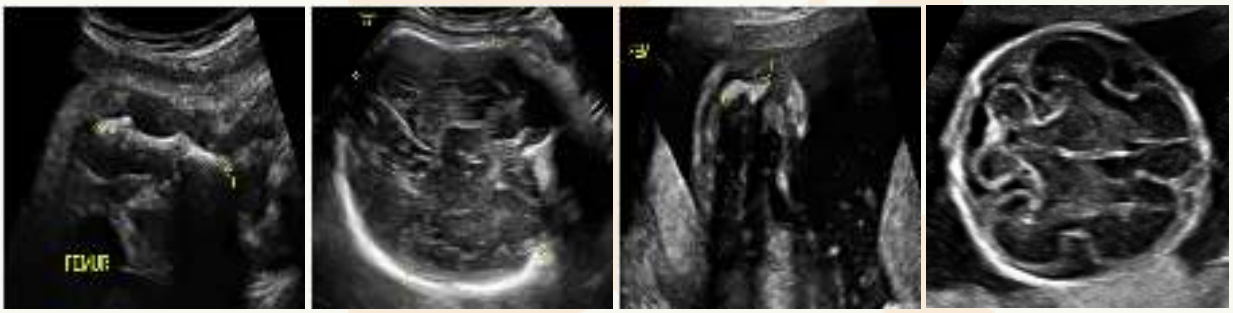
Any other organ abnormality:

3D dataset of the following to be acquired:

1. **Face (profile view to look at the facial features)**
2. **Chest (transverse and sagittal 3D dataset from anterior aspect preferably to calculate lung volumes for pulmonary hypoplasia)**
3. **Hand (one only)**
4. **Foot (one only)**
5. **Pelvis (transverse 3D dataset from anterior aspect for iliac flaring)**
6. **Spine (sagittal 3D dataset to look at the vertebral height)**

QUALITATIVE ASSESSMENT OF LONG BONES

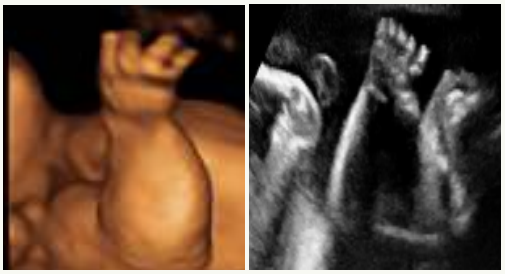
- Mineralization
- Shape
- Echogenicity
- Contour / any fracture
- Metaphyseal flaring/ spikes / irregularities
- Absence of an extremity or any particular segment of limb



EVALUATION OF HANDS AND FEET

Severe skeletal dysplasias are associated with alternations of hand and feet

- Polydactyly- presence of more than 5 digits (postaxial/ preaxial)
- Syndactyly- soft tissue or bony fusion of adjacent digits
- Clinodactyly- deviation of a finger(s)
- Disproportion- between hands and feet and the other parts of extremity may also be a sign of skeletal dysplasia
- Positional deformity



EVALUATION OF OTHER BONES

EVALUATION OF CRANIUM

- Macrocrania
- Frontal bossing
- Cloverleaf skull
- Hypertelorism / hypotelorism
- Severe skeletal dysplasias are associated with reduced ossification of the skull bones

EXAMINATION OF SPINE

EXAMINE FACE

- Cleft lip/ cleft palate
- The face should be examined for the diagnosis of micrognathia,retrognathia, hypertelorism ,ear ,lip and palate defect

EXAMINATION OF FETAL MOVEMENTS

- Arthrogryposis and multiple pterygium syndrome are characterized by limitation of flexion and extension of the limbs

THORACIC CIRCUMFERENCE

- Gestational age independent

- Measured - At the level of 4 chamber of heart

  Only one rib on either side of chest

- Less than 5th centile - lethal

TC/ AC ratio - Rule out pulmonary hypoplasia (Normal range – 0.89 ± 0.06)

FINDINGS MORE SUGGESTIVE OF SKELETAL DYSPLASIA ARE –

FL below 4SD

FL / foot length – below 0.8

FL / AC less than 0.16

LETHAL VS NON LETHAL LIMB SHORTENING

Diagnosis	Down Syndrome	Achondroplasia	Skeletal Dysplasias/Syndromes	Intrauterine Growth Restriction	Constitutional
Detailed anomaly scan	Markers of aneuploidy (echogenic intracardiac focus, pyelectasis, choroid plexus cysts, duodenal atresia, nuchal translucency, etc)	Frontal bossing, short fingers, trident hand short arms, narrow chest, macrocephaly	Abnormalities of other tubular bones, bowing, metaphyseal changes, abnormal posture/movements, thoracic circumference important	Small biometric measurements, possible abnormal Doppler findings	No obvious changes
Femur growth pattern	Often about 5th centile from early pregnancy on	Normal until 25 weeks, then falls in centiles	Severely short at second trimester or before	Variable onset, might begin with short FL weeks before small abdominal circumference	Often near 5th percentile, growth velocity is normal
Diagnosis	Down syndrome screening tests, karyotype analysis if > 1 soft marker	Molecular testing (Family history may be positive, de novo mutations common)	Depends on diagnosis and inheritance pattern. Postnatal diagnosis is possible. . Cell culture and DNA banking must be offered.	Karyotype analysis if > 1 soft marker, growth restriction develops during follow-up	Mother and /or father has short stature. History of short stature

LIST OF COMMON SKELETAL DYSPLASIA

SKELETAL DYSPLASIA	ETIOLOGY	PROGNOSIS	FEATURES
Thanatophoric dysplasia	Mostly sporadic	Lethal	Micromelia ,normal mineralization, narrow thorax , large head with prominent forehead, platyspondyly, curved femur in type I , clover leaf skull shape in type II

Osteogenesis imperfecta	Autosomal dominant	Lethal	Micromelia with hypomineralisation, fragile bones ,multiple fractures,narrow thorax
Achondrogenesis	Autosomal recessive/ sporadic	Lethal	Micromelia with hypomineralization, narrow thorax ± rib fracture, short trunk ,large head with prominent forehead, micrognathia , space suit
Achondroplasia	Autosomal dominant	Normal	short limbs after 22 weeks, large head with prominent forehead, lumbar lordosis
asphyxiating thoracic dystrophy	Autosomal recessive	variable	Limbs short > 22 weeks ,rhizomelic shortening ,narrow and short thorax,polydactyly, renal anomalies may be seen
hypophosphatasia	Autosomal recessive	Lethal	Micromelia with hypomineralisation, narrow thorax , clavicle sparing
Ellis -van creveld syndrome	Autosomal recessive	variable	Acromelic and mesomelic shortening,narrow and short thorax,post axial polydactyly, heart defect in >50%
SKELETAL DYSPLASIA	**ETIOLOGY**	**PROGNOSIS**	**FEATURES**
diastrophic dysplasia	Autosomal recessive	Normal	Short and bowed limbs,flexion contracture,talipes,scoliosis, micrognathia,hitchhiker thumb
Jarcho-levin syndrome	Autosomal recessive	variable	normal limbs,short narrow thorax,short trunk,fused vertebral bodies and ribs

Short rib polydactyly	Autosomal recessive	lethal	Micromelia with hypomineralisation, polydactyly ,congenital heart defect Severe thoracic hypoplasia
campomelic dysplasia	Autosomal recessive	Lethal	Short and bowed leg bones, , narrow thorax , hypoplastic scapulae, large head with small face,micrognathia,

- The prognosis can be variable dependent on the presence of associated anomalies
- Despite detailed sonographic evaluation, some of the syndromes cannot be ruled out and postnatal clinical genetic evaluation of fetuses or newborns should be considered