First trimester screening for congenital defects

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isuog.org

GUIDELINES

ISUOG Practice Guidelines: performance of first-trimester fetal ultrasound scan

When should a 1st trimester fetal US scan be performed?

- Not simply to confirm an ongoing early pregnancy in the absence of any specific indications.
- Ideally between 11 and 13+6 weeks' gestation to:

  - Confirm viability
  - Establish gestational age accurately
  - Determine the number of viable fetuses
  - Evaluate fetal gross anatomy and (if requested)
  - Risk of aneuploidy4–20.

Before the examination, a healthcare provider should Inform the woman/couple regarding the potential benefits and limitations of the first-trimester ultrasound scan.

First trimester US :

1. Dating of Pregnancy
2. Survey of fetal anatomy
3. Screening for Aneuploidies
4. Screening for Structural anomalies
5. Screening for Pre-eclampsia
6. and a lot more…….(premature labour, macrosomia etc.)

= Early pregnancy global risk assessment
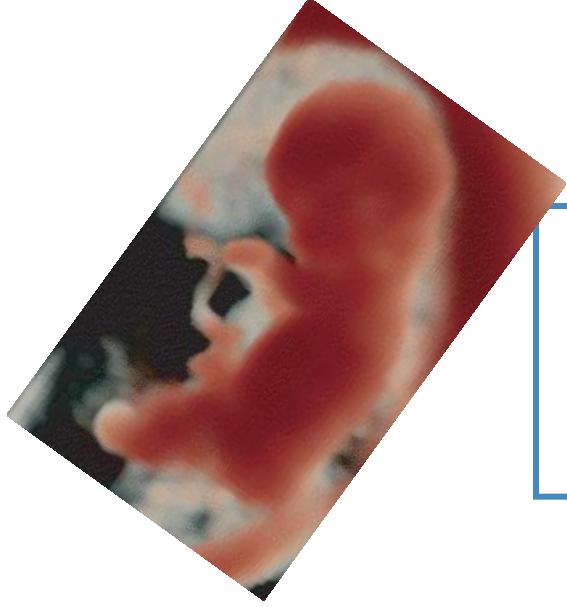
Accurate dating of pregnancy by a "good" CRL measurement

Good magnification
The fetus fills almost the entire screen.

Mid-sagittal section
The profile, spine and rump are visible.

Neutral position
There is fluid visible between the chin and the chest of the fetus

Fetus is horizontal
Almost 90° to the ultrasound beam

Crown and rump are clearly seen

Callipers are placed correctly
The intersection of the callipers should be placed on the outer borders of the skin over the head and rump.

Optimal dating by CRL between 10 and 12+6 wks (CRL 33- 68 mm)

$(8.052* .(CRL*1.037)+23.73):702-10.11$

Robinson HP, Fleming JEE. A critical evaluation of sonar crown-rump length measurements. Br JObstet Gynaecol 1975;82

The LMP is taken into account, but **the due date is calculated based on US measurements, even when there is 1 day difference.** Two recent UK reviews* indicate the corrected CRL curve of Robinson en Fleming as the best one.

*Bottomley C, Bourne T. Dating and growth in the first trimester. Best Pract Res Clin Obstet Gynaecol 2009
*Loughna P, Chitty L, Evans T, Chudleigh T. Fetal size and dating: charts recommended for clinical obstetric practice. Ultrasound 2009

Beyond a CRL of 64 mm BPD and HC become more accurate
They should be used for pregnancy dating

Early anatomy survey

1st trimester screening for congenital anomalies

(Syngelaki et al. 2011)

Author	Total	L Scan route	GA (weeks)	Fetal anomalies			Detected
				Total	CyHy	Aneuploidy	**Detected**
Hernadi and Torocsic, 1997	3991	TA, TV	11–14	**49 (1.2%)**	7 (14.3%)	4 (8.2%)	**20 (40.8%)**
D'Ottavio et al., 1998	4078	TV	13–15	**88 (2.2%)**	30 (34.1%)	19 (21.6%)	**54 (61.4%)**
Bilardo et al., 1998	1690	TA, TV	10–14	**23 (1.4%)**	3 (13.0%)	—	**10 (43.5%)**
Hafner et al., 1998	4233	TA	10–13	**56 (1.3%)**	—	—	**7 (12.5%)**
Whitlow et al., 1999a	6443	TA, TV	11–14	**63 (1.0%)**	14 (22.2%)	14 (22.2%)	**37 (58.7%)**
Guariglia and Rosati, 2000	3478	TV	10–16b	**57 (1.6%)**	15 (26.3%)	8 (14.0%)	**33 (57.9%)**
Taipale et al., 2004	4789	TV	10–16c	**33 (0.7%)**	—	4 (12.1%)	**6 (18.2%)**
Chen et al., 2004	1609	TA, TV	12–14	**26 (1.6%)**	1 (3.8%)	11 (42.3%)	**14 (53.8%)**
Becker and Wegner, 2006	3094	TA, TV	11–13	**86 (2.8%)**	—	56 (65.1%)	**72 (83.7%)**
Cedergren and Selbing, 2006	2708	TA	11–14d	**32 (1.2%)**	3 (9.4%)	1 (3.1%)	**13 (40.6%)**
Saltvedt et al., 2006	18053	TA	11–14	**371 (2.1%)**	Not stated	—	**74 (19.9%)**
Dane et al., 2007	1290	TA	11–14	**24 (1.9%)**	3 (12.5%)	5 (20.8%)	**17 (70.8%)**
Chen et al., 2008	7642	TA	10–14	**127 (1.7%)**	30 (23.6%)	32 (25.2%)	**51 (40.2%)**
Oztekin et al., 2009	1805	TA	11–14	**21 (1.2%)**	3 (14.3%)	—	**14 (66.7%)**
Ebrashy et al., 2010	2876	TA, TV	13–14	**31 (1.1%)**	7 (22.6%)	—	**21 (67.7%)**
Total	67779	—	10–16	**1087 (1.6%)**	116 (10.7%)	154 (14.2%)	**443 (40.8%)**

Fetal abnormality   NT>P95th   11 – 13wks   *Syngelaki et al 2011*

	NT>P95th	11 – 13 wks
Neural tube		
Acrania/iniencephaly	27%	100%
Open spina bifida	5%	14%
Brain		
Alobar holopros.		100%
Face		
Facial cleft	15%	5%
Lungs		
Diaphragmatic hernia	37%	50%
Exomphalos only bowel	18%	100%
+ liver	10%	100%
Gastroschisis		11%
Renal		
Bladder length<16 mm	19%	100%
Bladder length>16 mm	50%	100%
Renal agenesis unilateral	17%	
Inf. Pol kidneys		33%
Skeleton		
Lethal skeletal dysplasia	50%	50%
Talipes unilateral	4%	
Talipes bilateral	25%	
Short long bone unilateral		50%
Absent hand/or foot		78%
Polydactyly		60%
Other		
Body stalk anomaly	100%	100%
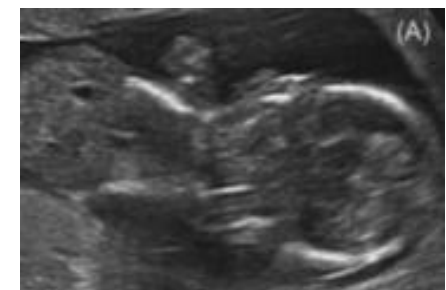
Mid-sagittal section
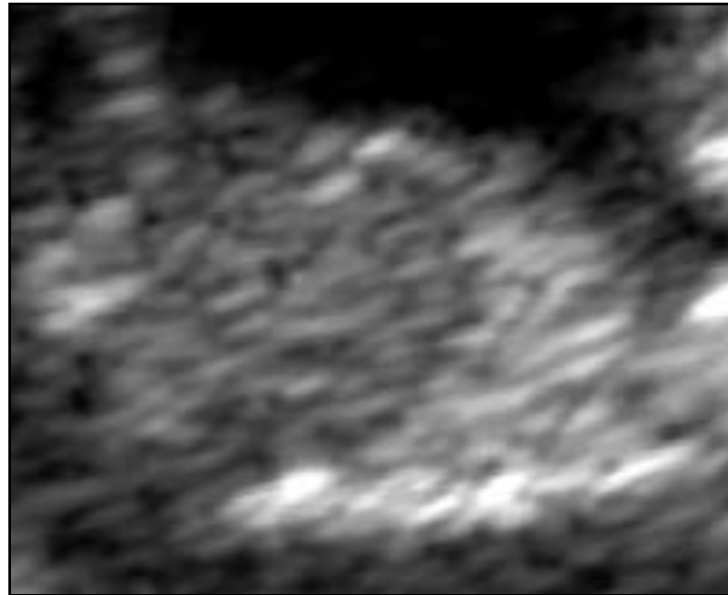
Diaphragm

Bladder:

10 wks visible in 50%
11 wks visible in 80%
12 wks visible in 90%
13 wks visible in all

Rosati et al 1996
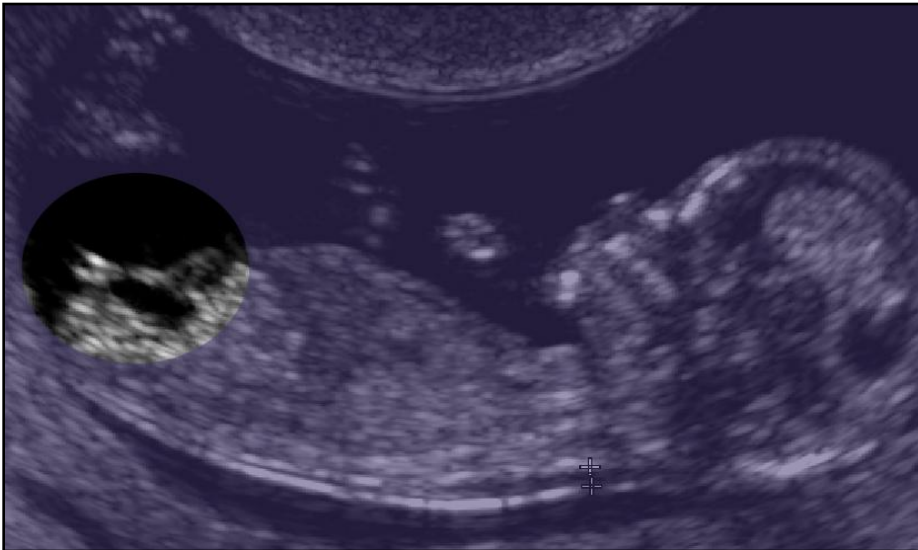
Gender:
is the angle that counts!
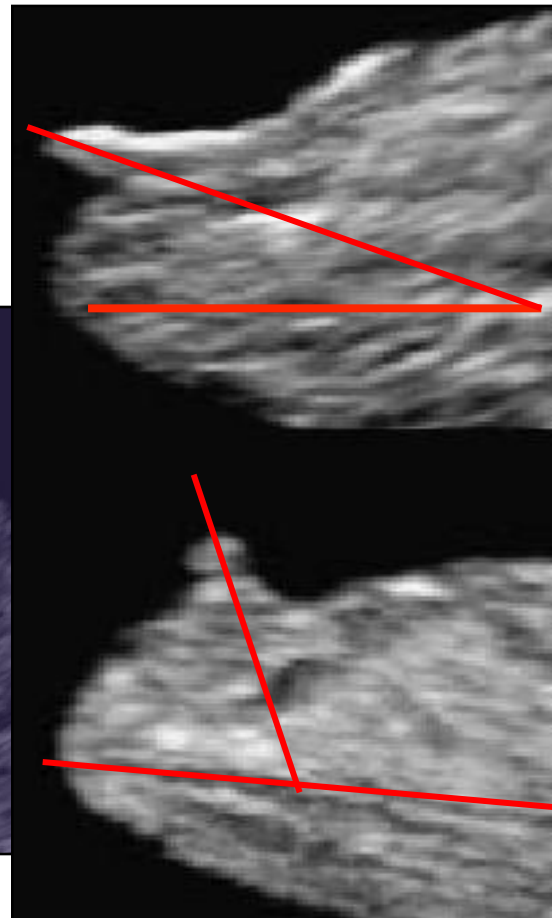
☐ *Midline sagittal plane*
☐ *Tubercle/spine angle*
☐ *From 13th week*
(32mm DBP, 100% accuracy)
(Effrat, Mazza)

☐ *Angle > 27° = XY*
(Youssef et al. 2011)

Megacystis

35 of 57,119 singleton pregnancies

Kagan et al 2010

Transverse Section
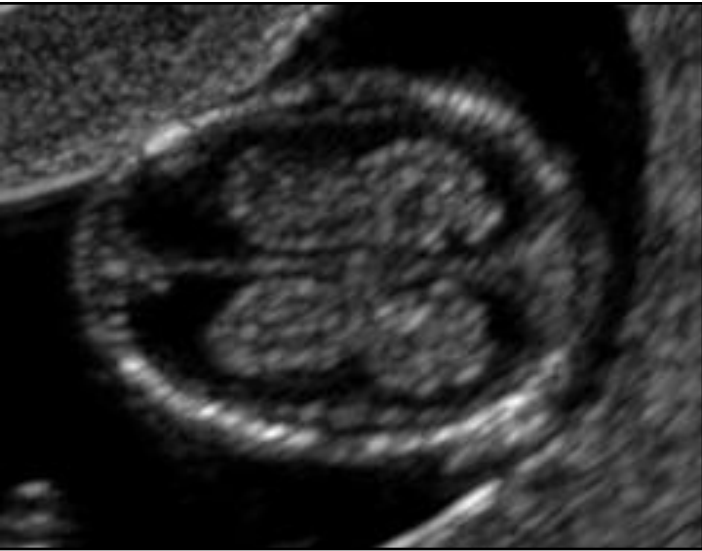
Skull

Midline

Ventricles / choroid plexus
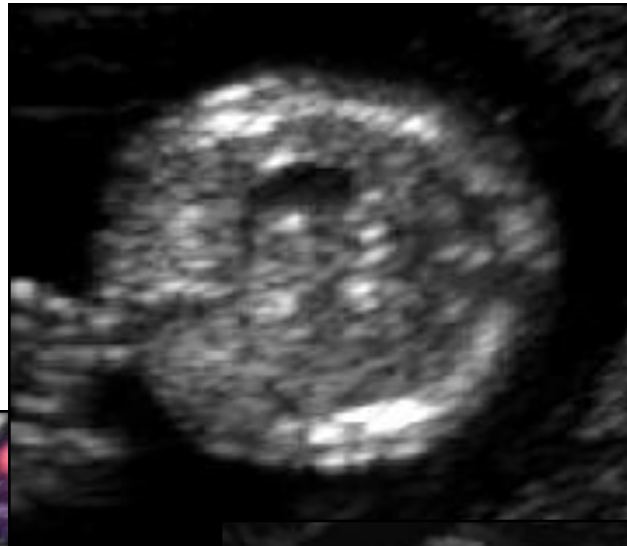
Can be visualized from 8-9wks

Always from 12-13 wks
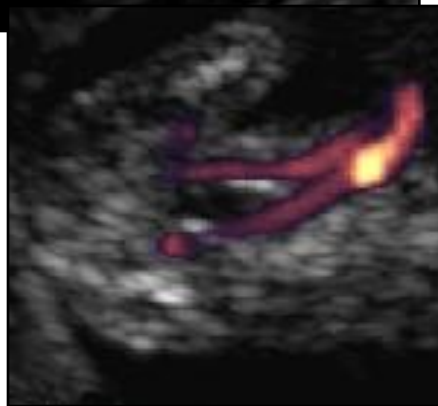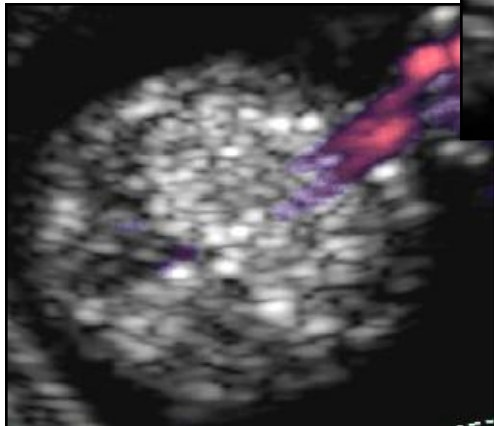
Skull

calcification completed by 11 weeks
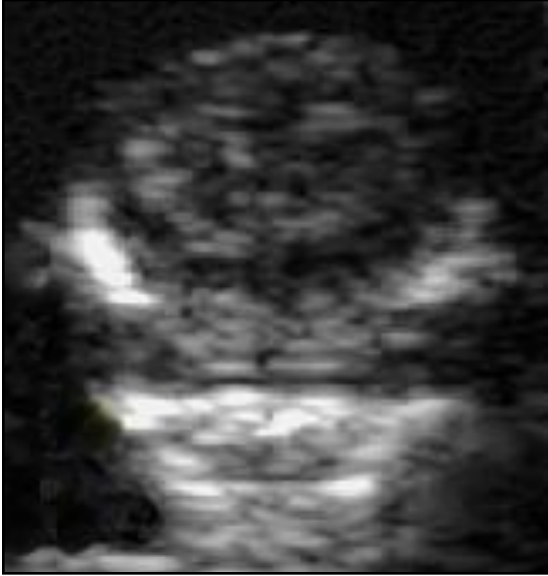
Eyes, Lips, palate
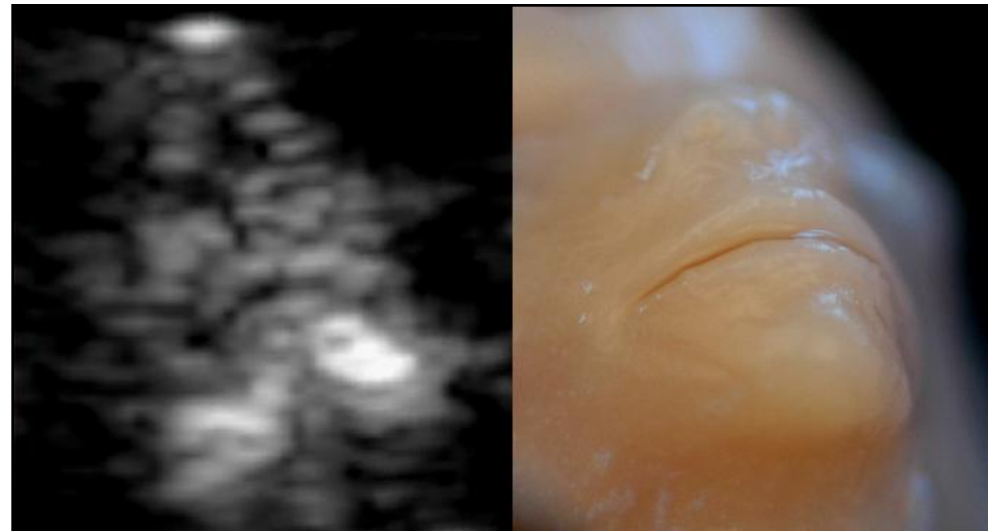
Retronasal triangle: a sonographic landmark for the screening of cleft palate in the first trimester

W. SEPULVEDA*†, A. E. WONG*, P. MARTINEZ-TEN‡ and J. PEREZ-PEDREGOSA‡

Normal fetus
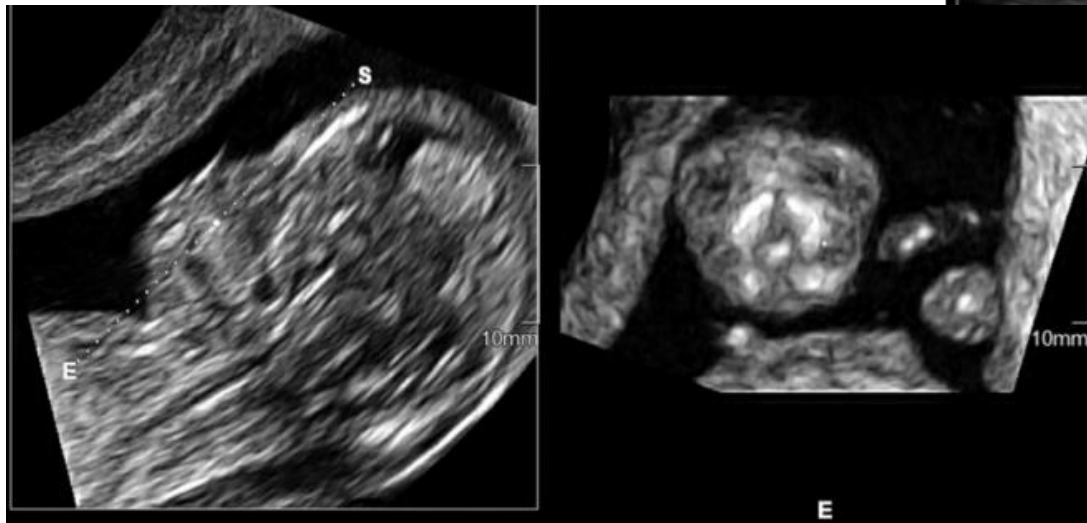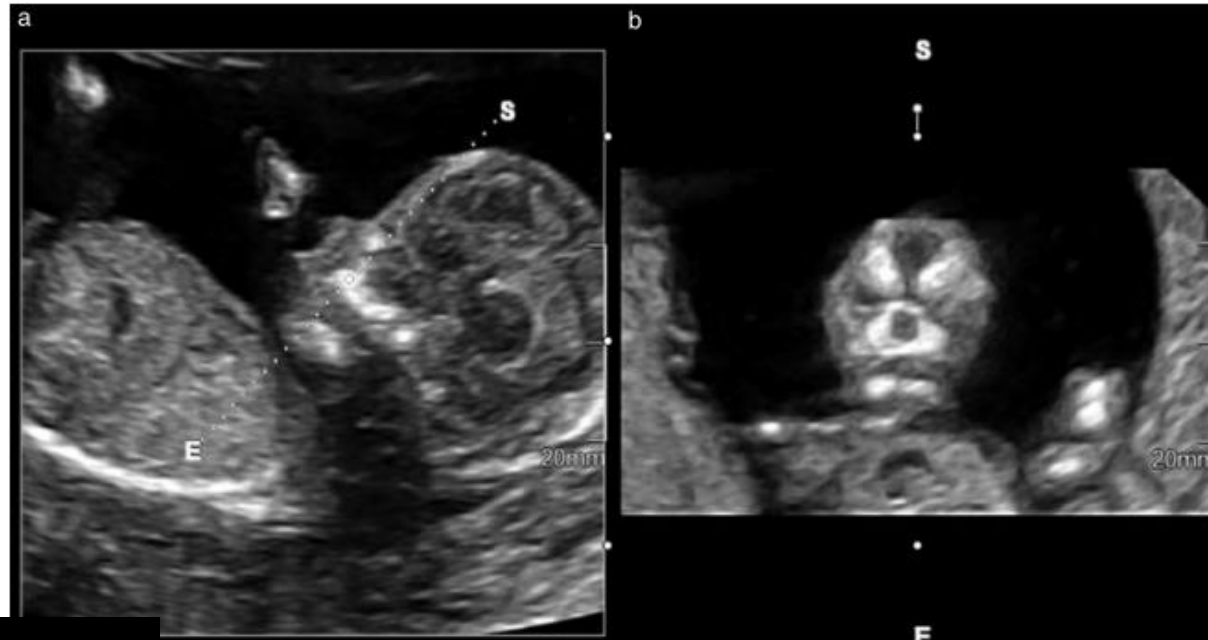
Fetus with cleft palate holoprosencephaly

First-trimester diagnosis of cleft lip and palate using three-dimensional ultrasound

P. MARTINEZ-TEN*, B. ADIEGO*, T. ILLESCAS*, C. BERMEJO†, A. E. WONG‡
and W. SEPULVEDA‡

Normal fetus
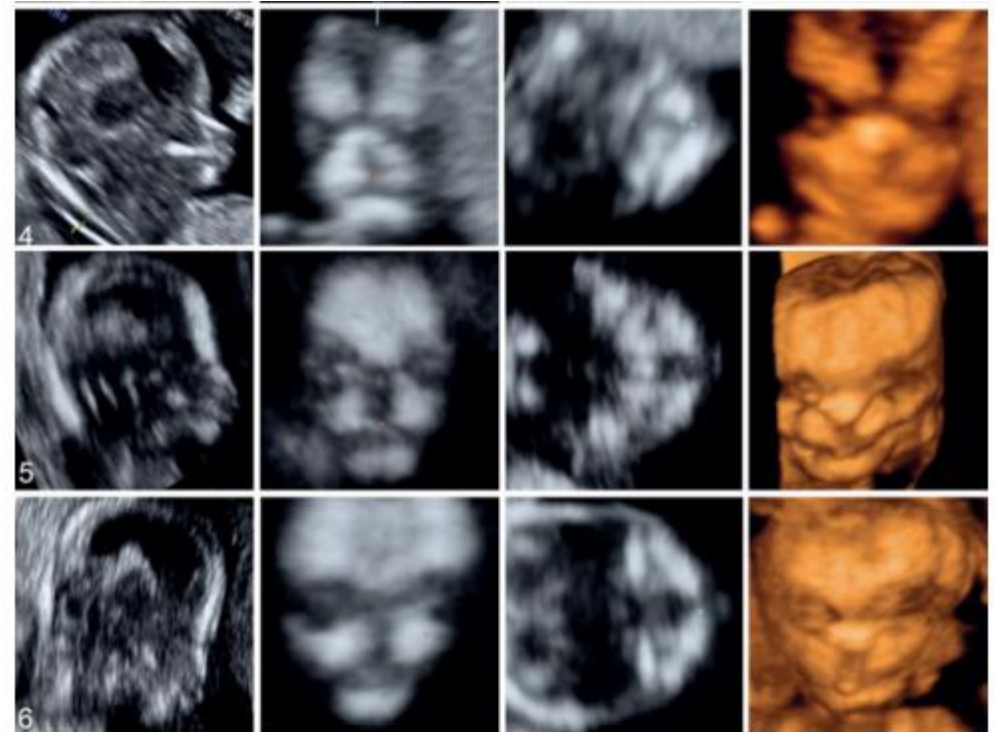
Cleft lip and palate

Mandibular gap and retro-micrognathia

Absent mandibular gap in the retronasal triangle view: a clue to the diagnosis of micrognathia in the first trimester

W. SEPULVEDA*, A. E. WONG*, F. VIÑALS†, E. ANDREEVA‡, N. ADZEHOVA‡ and
P. MARTINEZ-TEN§
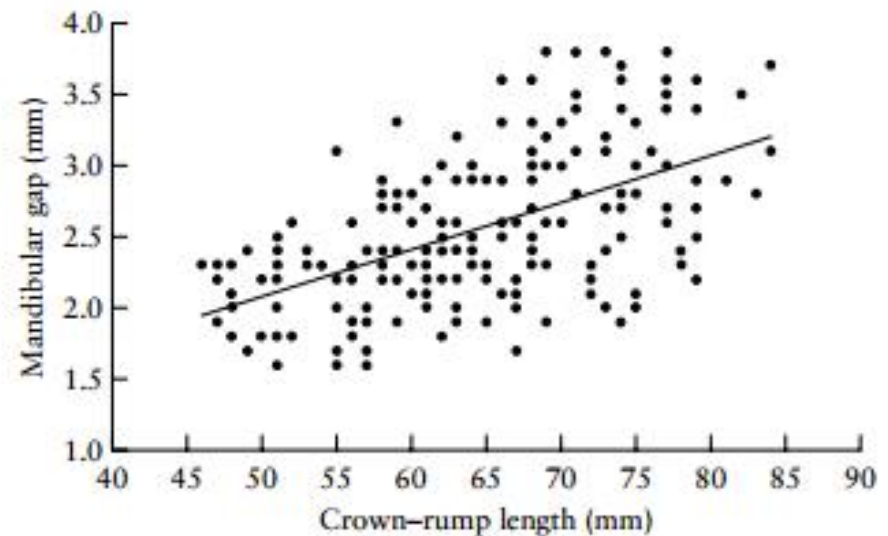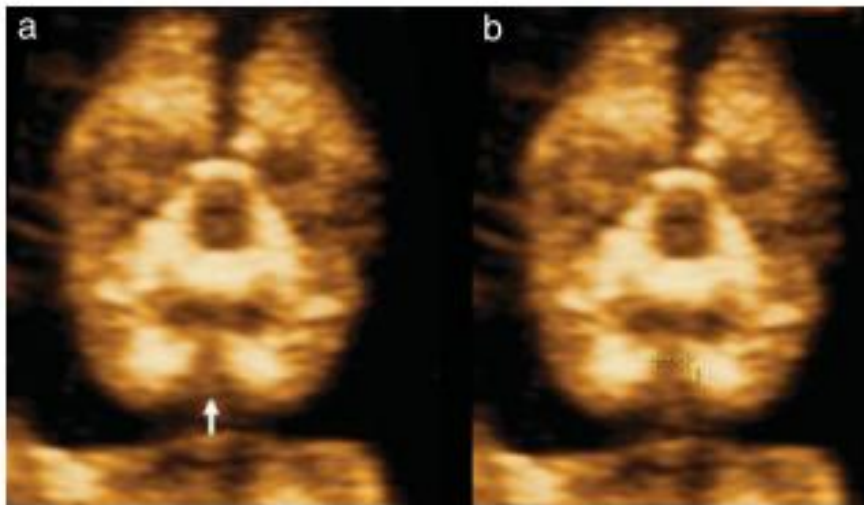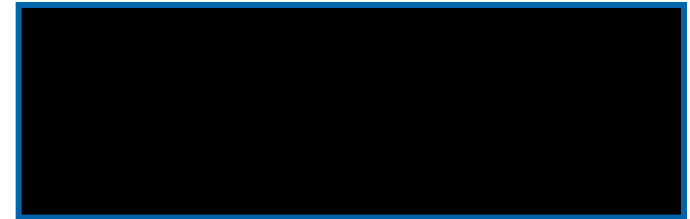
Holoprosencephaly

44 of 57,119 singleton pregnancies
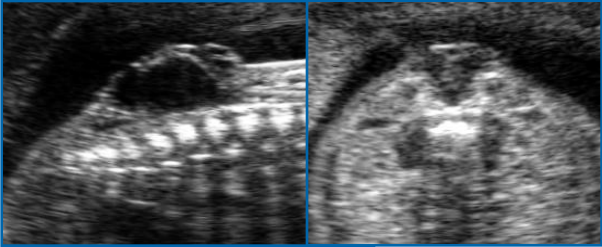
Kagan et al 2010

Encephalocele

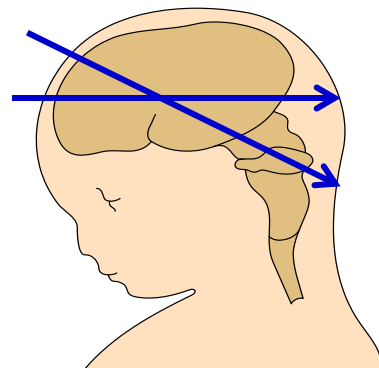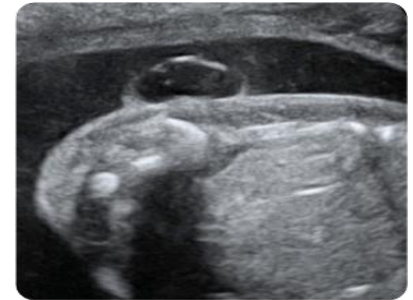# Early detection of fetal defects: Spina Bifida

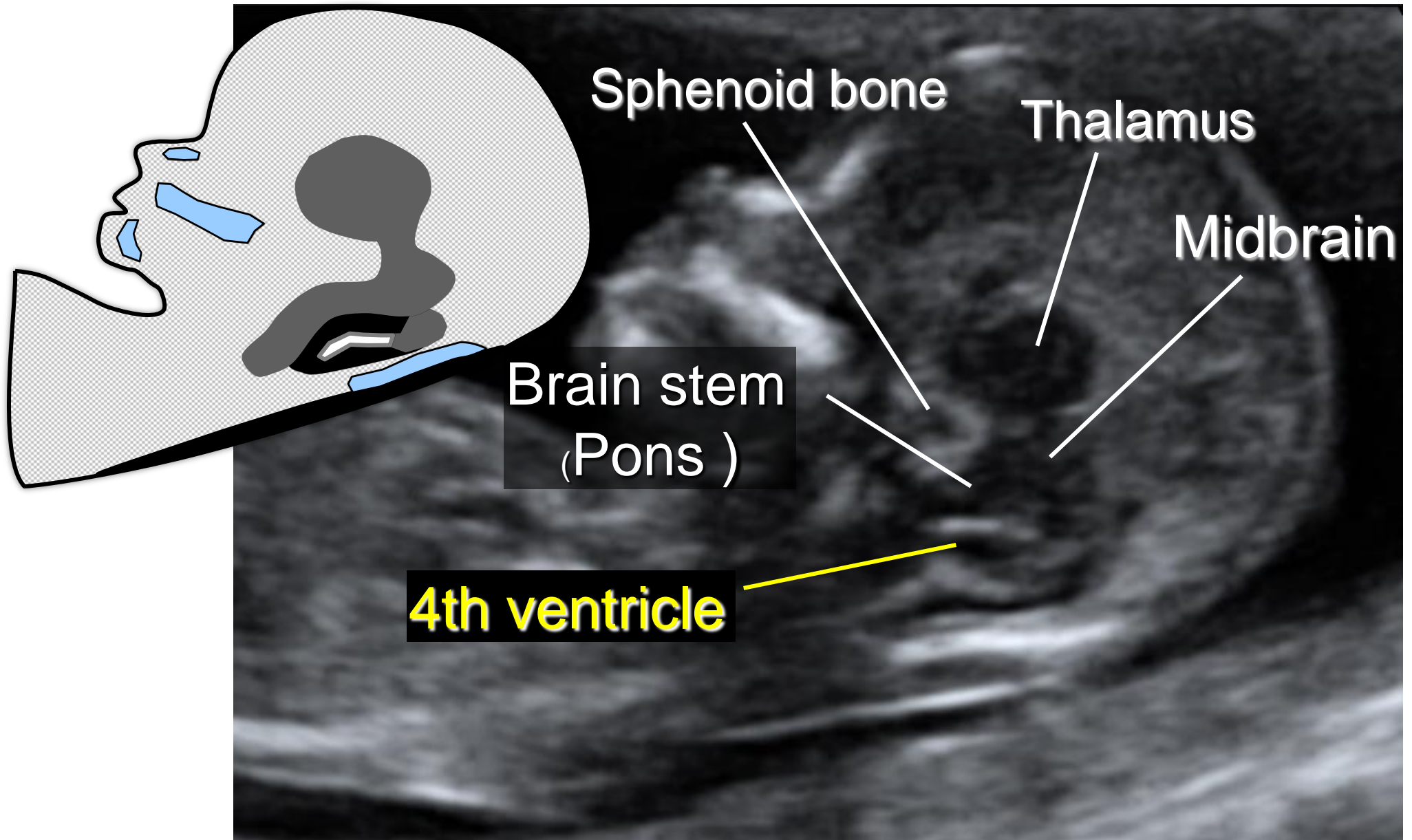| Spina bifida	DR
History	5%
MS AFP	75%
Ultrasound	98%
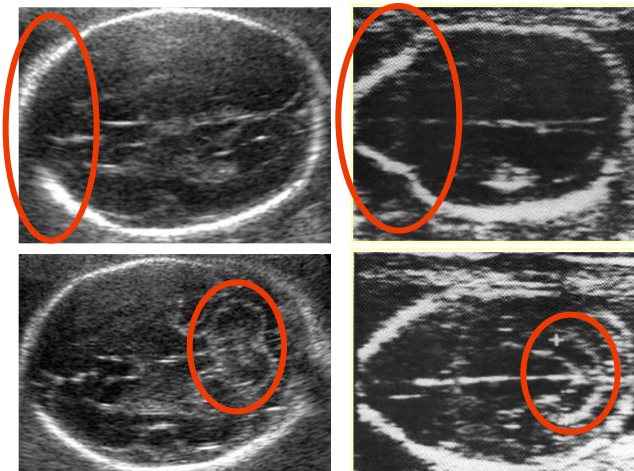
Assessment of intracranial translucency (IT) in the detection of spina bifida at the 11–13-week scan

R. CHAOUI*, B. BENOIT†, H. MITKOWSKA-WOZNIAK‡, K. S. HELING*
and K. H. NICOLAIDES§

The brain @ 11-14 weeks scan

Sphenoid bone

Thalamus

Midbrain

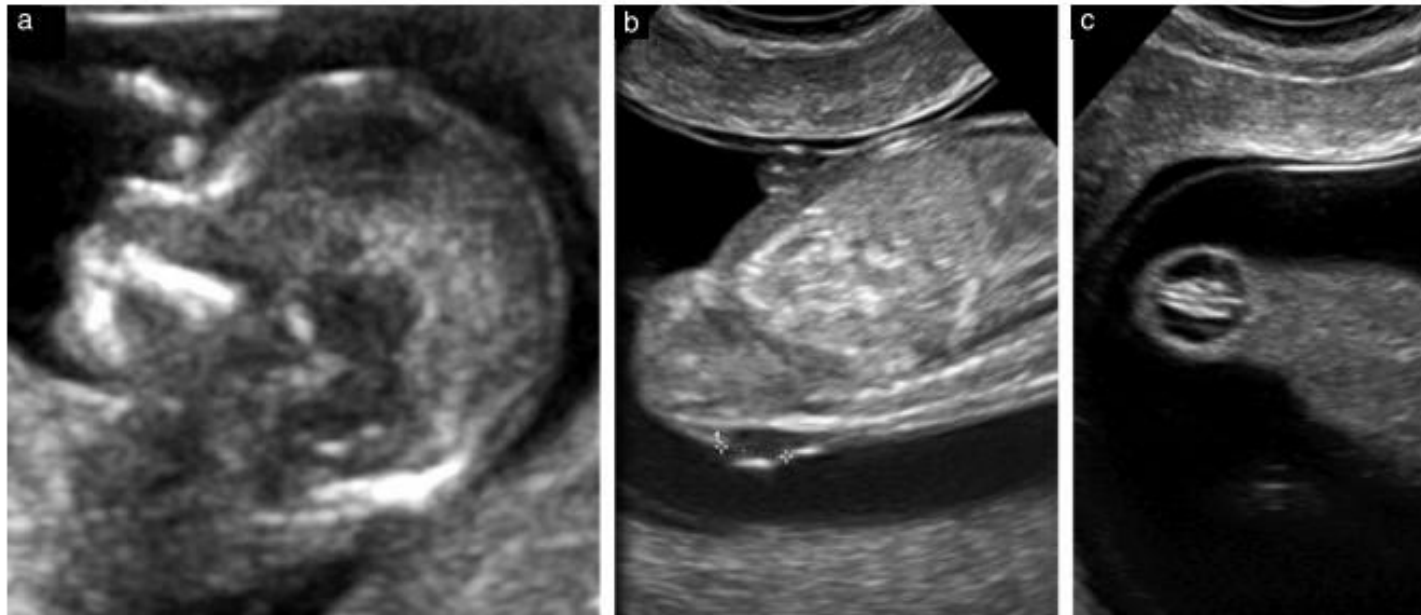Brain stem
(Pons)

4th ventricle

Open SB at 13 weeks

Figure 2 Case 1. (a) In the mid-sagittal view of the fetal face, obtained by transabdominal ultrasound, no typical intracranial translucency with clear borders is identifiable. The brainstem is thickened and the distance from the posterior border of the brainstem to the occipital bone is shorter than the brainstem diameter. (b) Subsequent examination of the spine, performed transvaginally, revealed a small lumbosacral spina bifida 5 mm in diameter (calipers) and, once the fetus had turned (c), the small myelomeningocele could be visualized in a coronal tangential plane.

R. Chaoui UOG 2011

Could additional measurements help in increasing the accuracy of detecting open NTD @ the 11-14 w scan ?
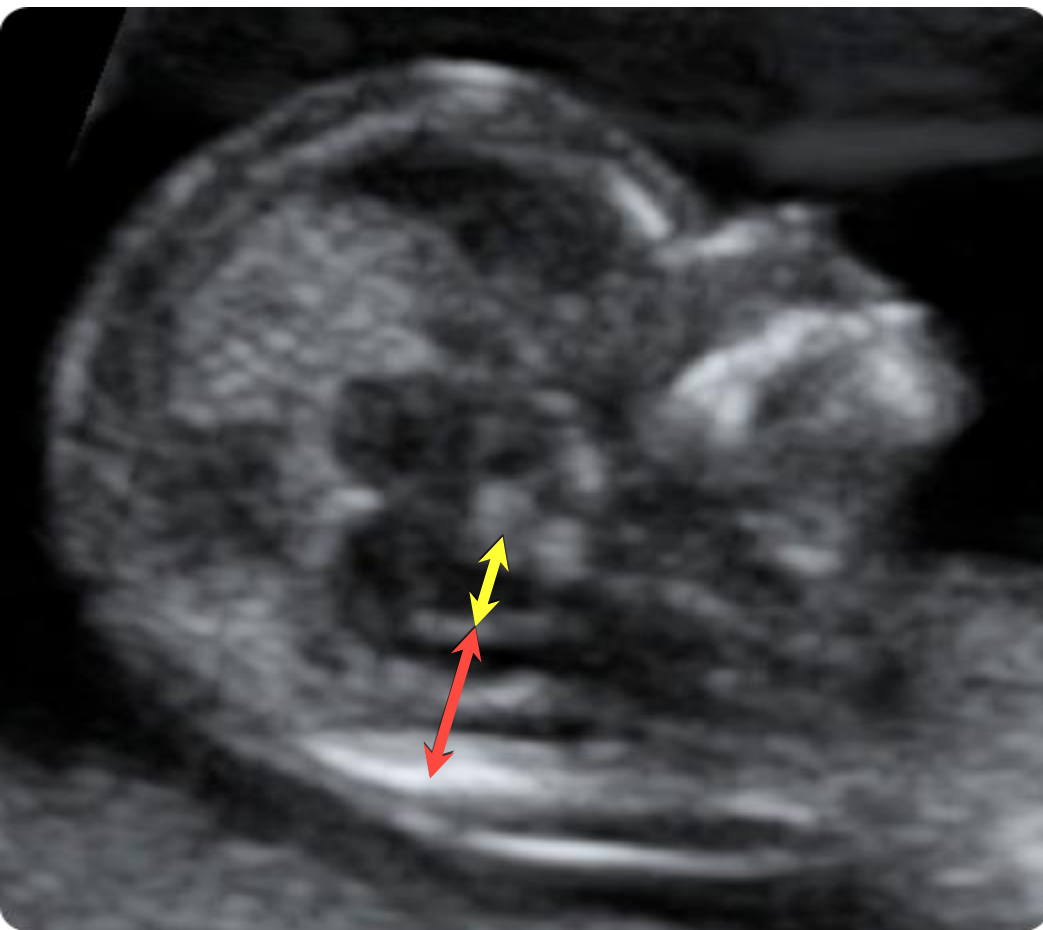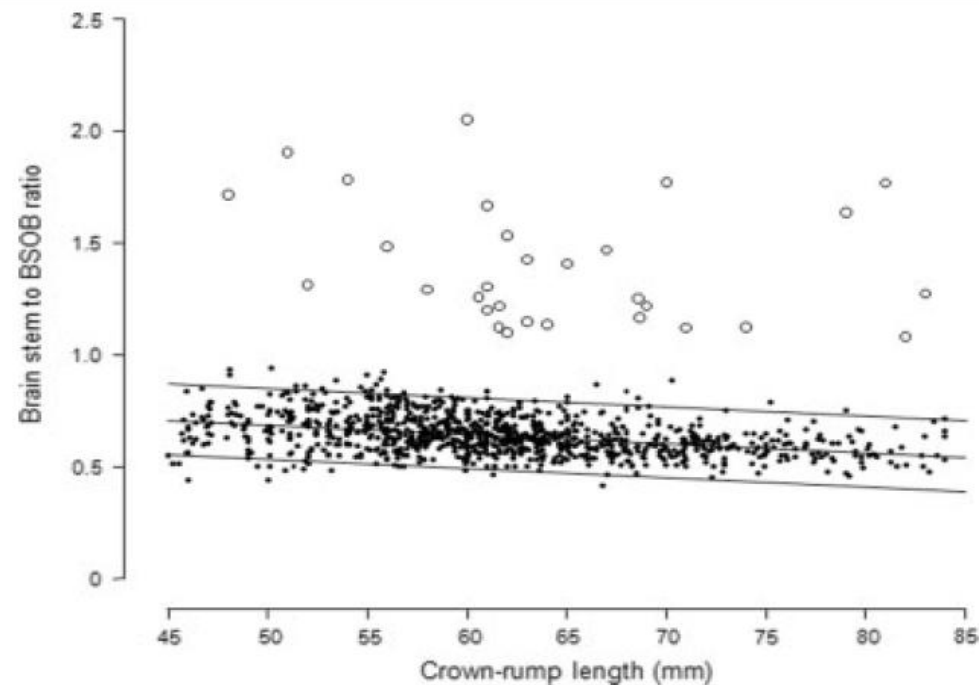

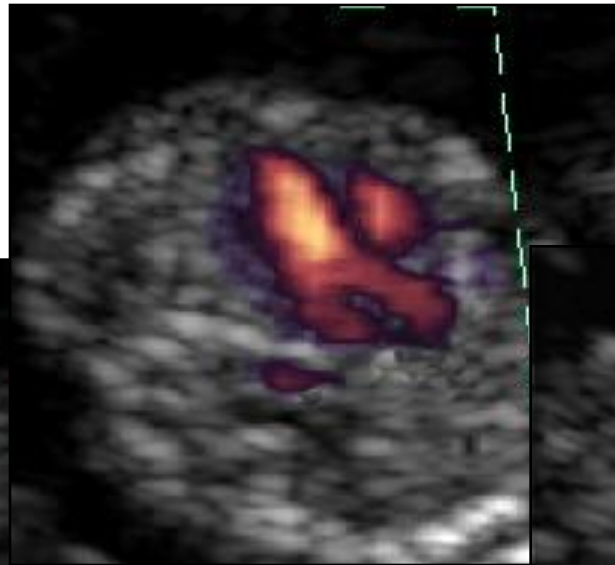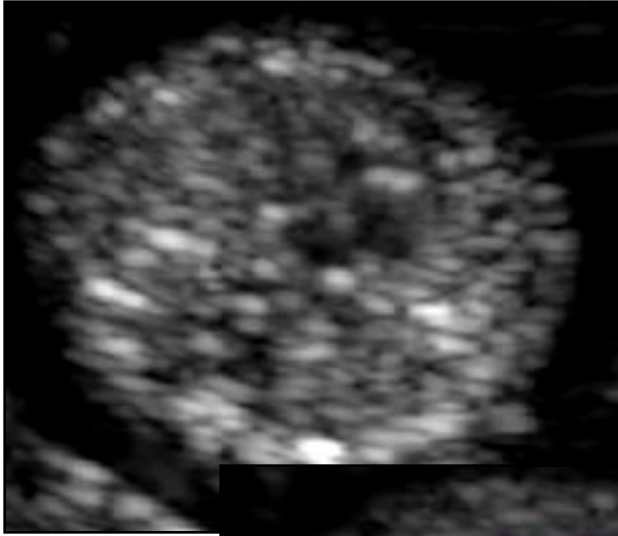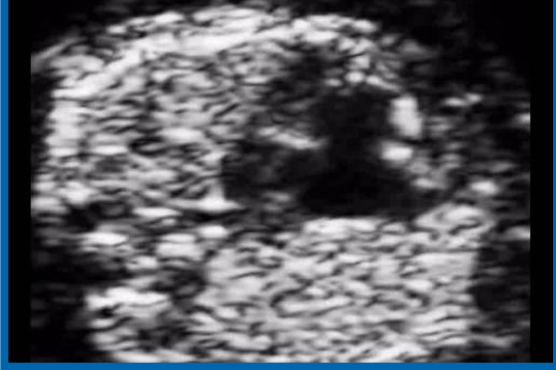### Ratio of Brainstem/ Posterior Fossa

Figure 4—Individual measurements of the ratio between the brain stem diameter and BSOB diameter in fetuses with OSB (open circles) and normal controls (closed circles) plotted on the reference range for CRL (median, 5th and 95th percentiles)

R. Lachmann, R. Chaoui, J.Moratalla, G.Picciarelli, K.H. Nicolaides, Prenatal Diagnosis 2011

Heart

4 chamber view - outflow tract

Early detection of CHD
(106/488= 21%)

| Defect	N	NT>P95
Coarctation (CoA)	15	2 (50%)
Tetralogy of Fallot (TOF)	10	2 (67%)
Hypoplastic left heart	10	2 (40%)
Atriov.sept. def. (AVSD)	9	3 (100%)
Double outlet (DORV)	7	3 (75%)
Ebstein anomaly	5	
Transposition (TGV)	5	
Pulmonary stenosis	5	
Pulmonary atresia (PA)	3	1 (100%)
Others	32	
Total	106	64%

↑ NT in 35% of the major CHD

Syngelaki et al. Pren. Diagnosis 2011

Early detection of fetal defects
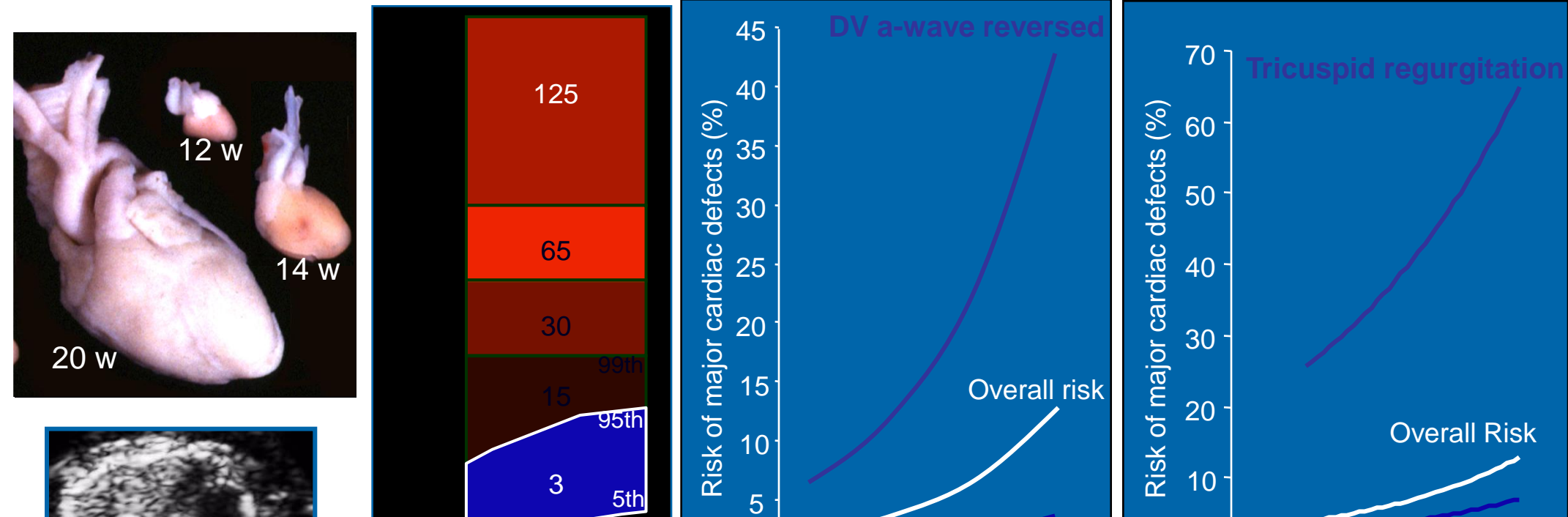
Major cardiac defects
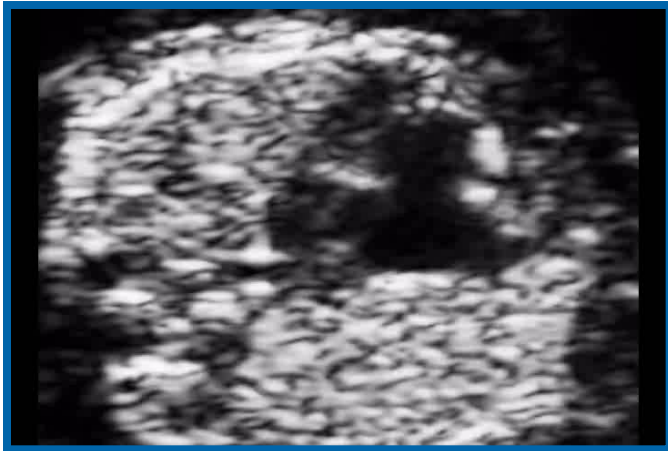
AVSD
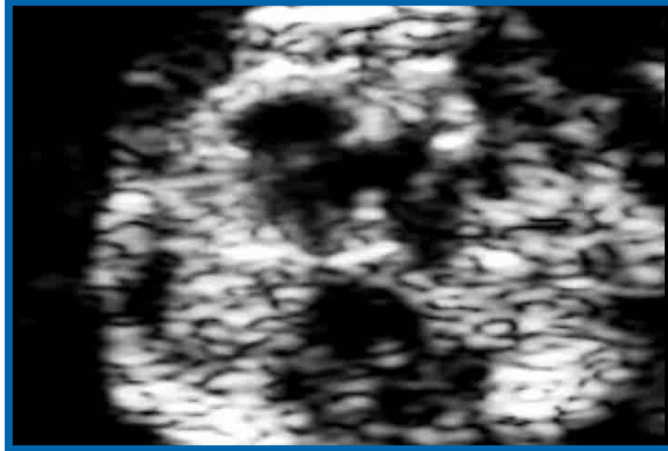
Hypoplastic L heart

Tetralogy of Fallot

Early detection of fetal defects

Major cardiac defects

Enlarged NT and abnormal DV or TR detects 65% of CHD
Abnormal 4chv detects 50% of CHD
Abnormal 4Chv + abn DV / TR detects 75%
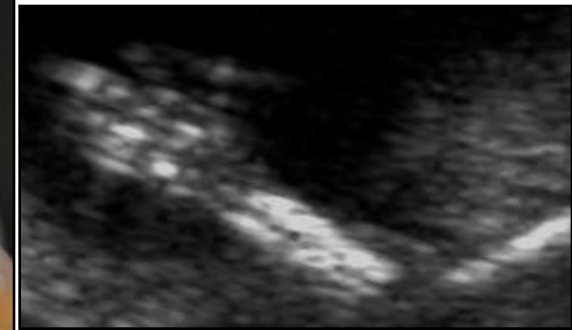
Exomphalos

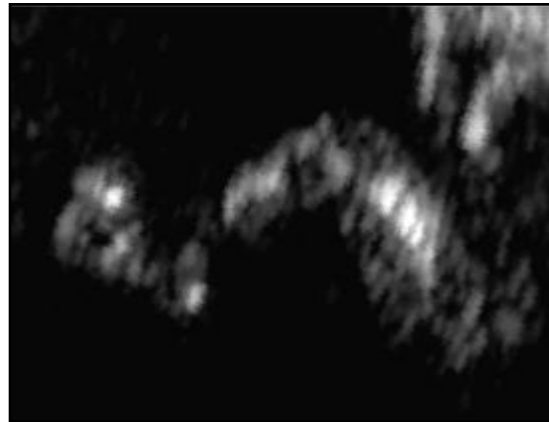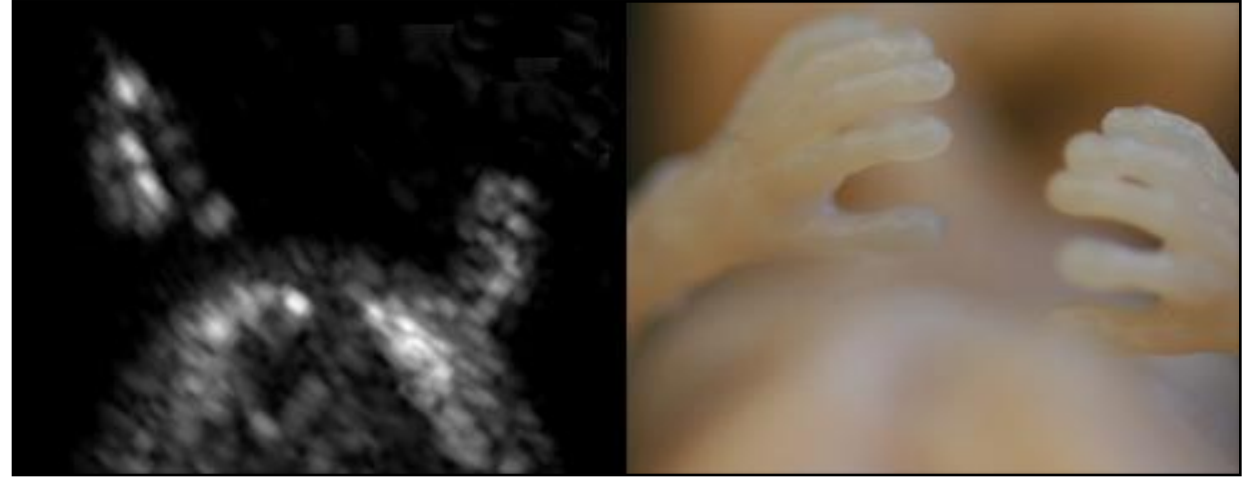- Prevalence: 1 in 380
  Bowel only
  CRL of 45-54.9 mm: 1 in   100
  CRL of 55-64.9 mm: 1 in   800
  CRL of 65-84.0 mm: 1 in 2100
  Liver: 1 in 3400

- Trisomy 18 or 13 (55%)
- Resolution by 20 wks
  Bowel: 90%
  Liver: 0%

150 of 57,119 singleton pregnancies

Kagan et al 2010

Extremities

Stephens et al 1983
Bronshtein et al 1992
Dimaio et al 1993
Macrydimas et al 1996

Limb shortening / fractures may be noted in the First Trimester

Skeletal Dysplasias
(Majority presents with enlarged NT)

Achondrogenesis I
Achondrogenesis II
Ellis van Creveld
Osteogenesis imperfecta II
Thanatophoric dysplasia
Campomelic dysplasia
Congenital hypophosphatasia
Spondyloepiphyseal dysplasia congenita
Jeune asphyxiating thoracic dystrophy
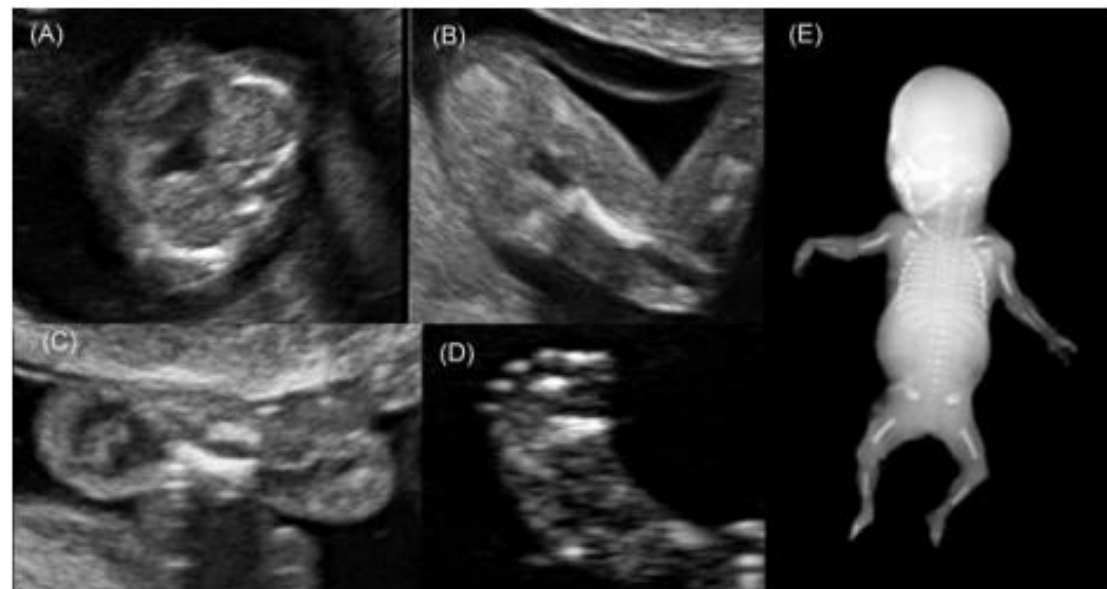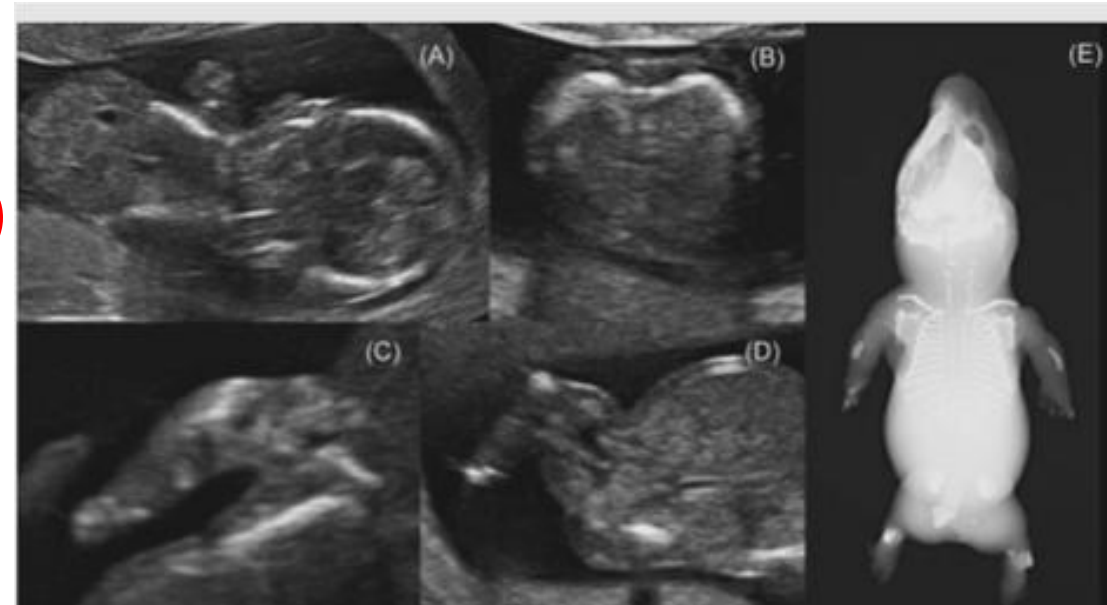**SRPS II (Majewski)**
SRPS III (Verma-Naumoff syndrome)
SRPS IV (Beemer Langer)
Blomstrand dysplasia
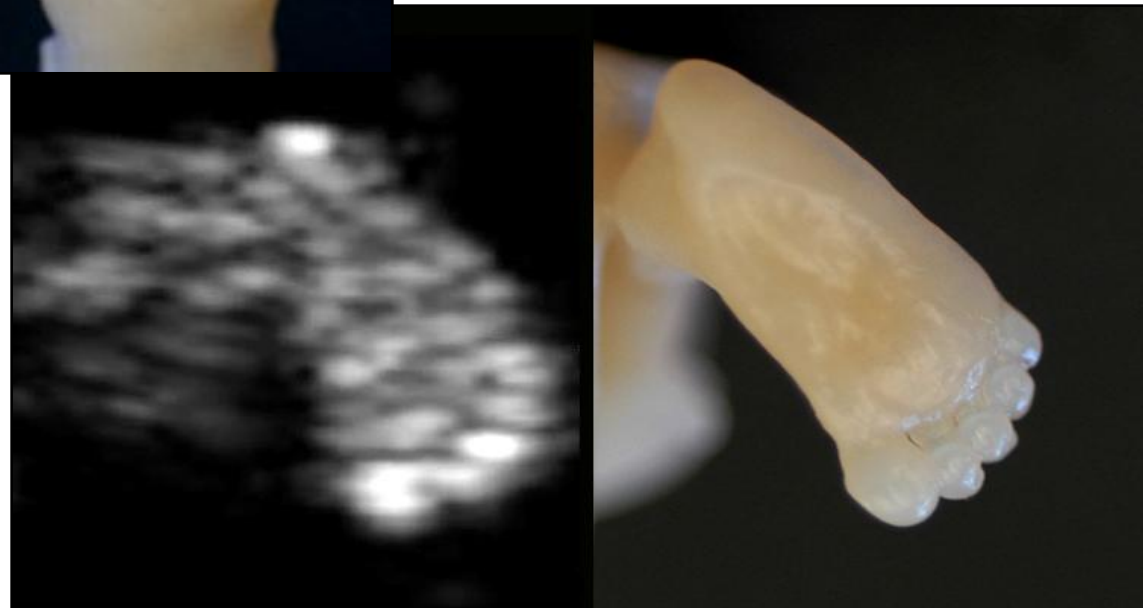**Roberts syndrome**
Schneckenbecken dysplasia
Cleidocranial dysplasia

Khalil et al.2011

Early detection of fetal defects

Skeletal dysplasia

Assessment of digits

Polydactyly

2 3 4 5
1
6
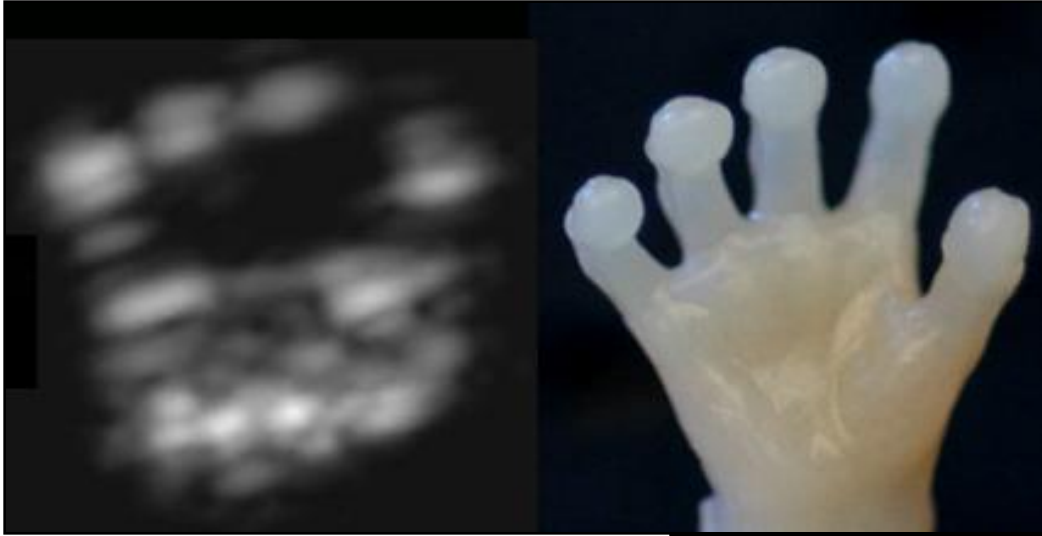
Early detection of fetal defects

Hand and foot abnormalities
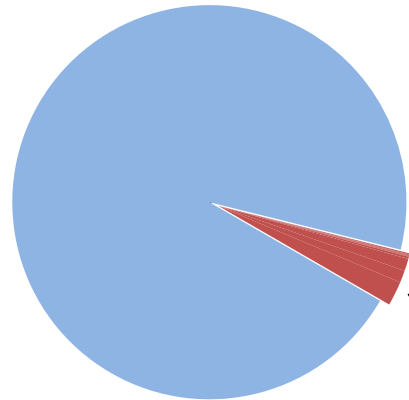
Conclusions:

- 1st trimester Ultrasound is essential for:

  – Accurate pregnancy dating

  – **Diagnosis of major structural anomalies**

  – Screening for chromosomal anomalies

Time to reconsider the importance of the role of the 12-13 weeks scan in the NIPT era
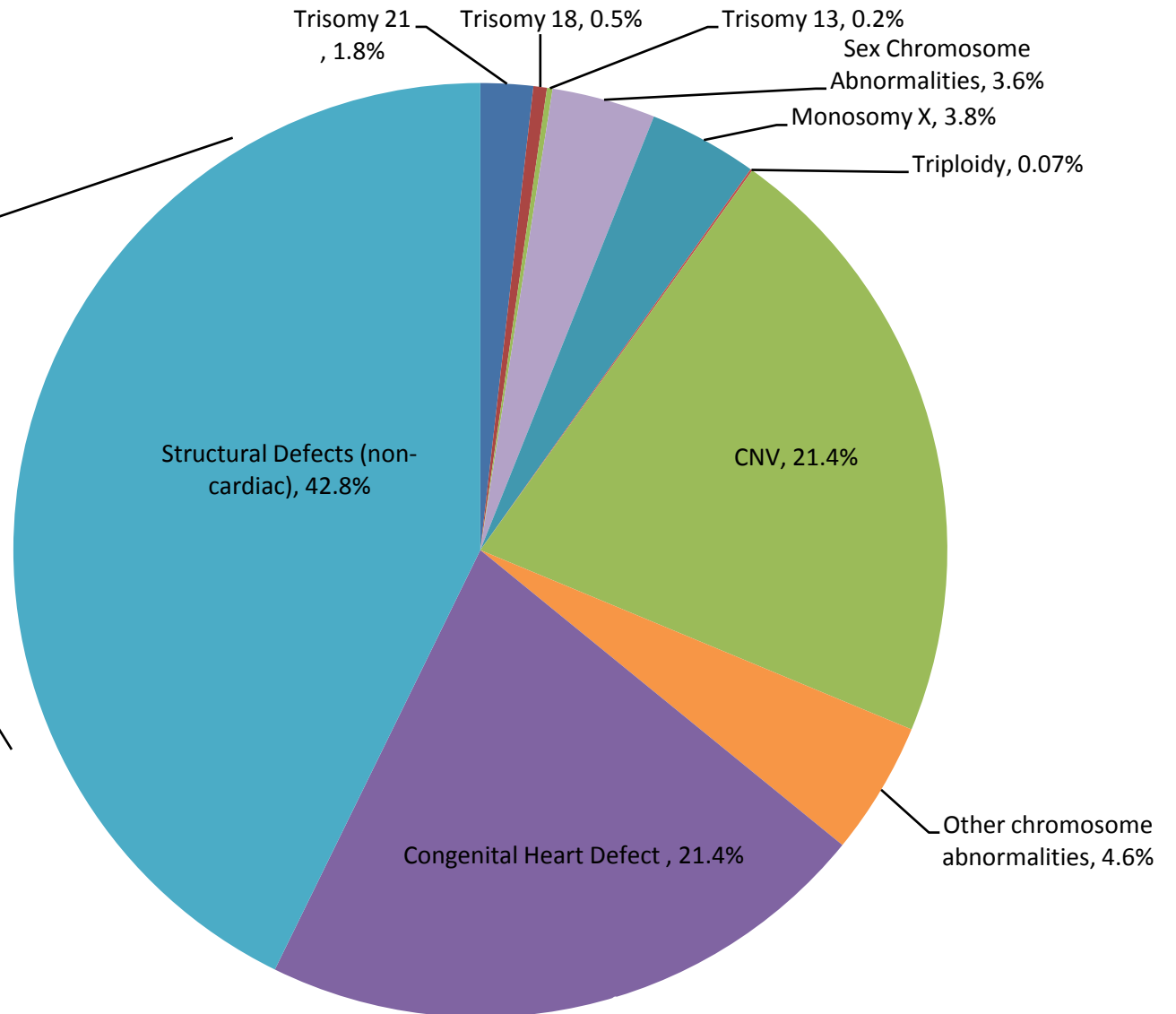
Abnormalities at 13 Weeks Gestational Age
Maternal Age: <25

Normal: 95.3%

Abnormal: 4.7%

Trisomy 21, 1.8%
Trisomy 18, 0.5%
Trisomy 13, 0.2%
Sex Chromosome Abnormalities, 3.6%
Monosomy X, 3.8%
Triploidy, 0.07%
CNV, 21.4%
Other chromosome abnormalities, 4.6%
Congenital Heart Defect, 21.4%
Structural Defects (non-cardiac), 42.8%

Thanks!