Early pregnancy anomaly scan (EPAS)

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1st trimester: Structural development of the fetus & early detection of anomalies

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UCL HOSPITALS

Clinical / Research / Training Interest

- ✓ Early Pregnancy Anomaly Scan (11-13 wks)
  - Echocardiography
  - Neurosonography
  - Early detection of spina bifida
  - Face, skeleton, kidneys….
- ✓ Screening Fetal Echocardiography
- ✓ Medical use of 3D / 4D
  - Face
  - Brain
  - Syndromes…

fetalechocardiography.com

fetalechocardiodraphy.com

London School of Ultrasound

Early Fetal Echocardiography at 11-13 weeks

11th November 2017

Early Fetal Neurosonography at 11-13 weeks

10th March 2018

Fetus at 11-13 weeks

- Majority of severe anomalies are already present
- Appropriate dimensions to TVS scanning
- Mobile fetus
  - ✓ spontaneous movement
  - ✓ +/-manipulations
- Relatively large amount of amniotic fluid
- Incomplete ossification

The instruments: use of different transducers

- It is important to know your scanner and pros and cons of every transducer
- Different transducers can be used during examination in order to get best available quality of imaging

Different transducers: quality of the image is difficult to predict without trying

Similar quality and resolution of the image

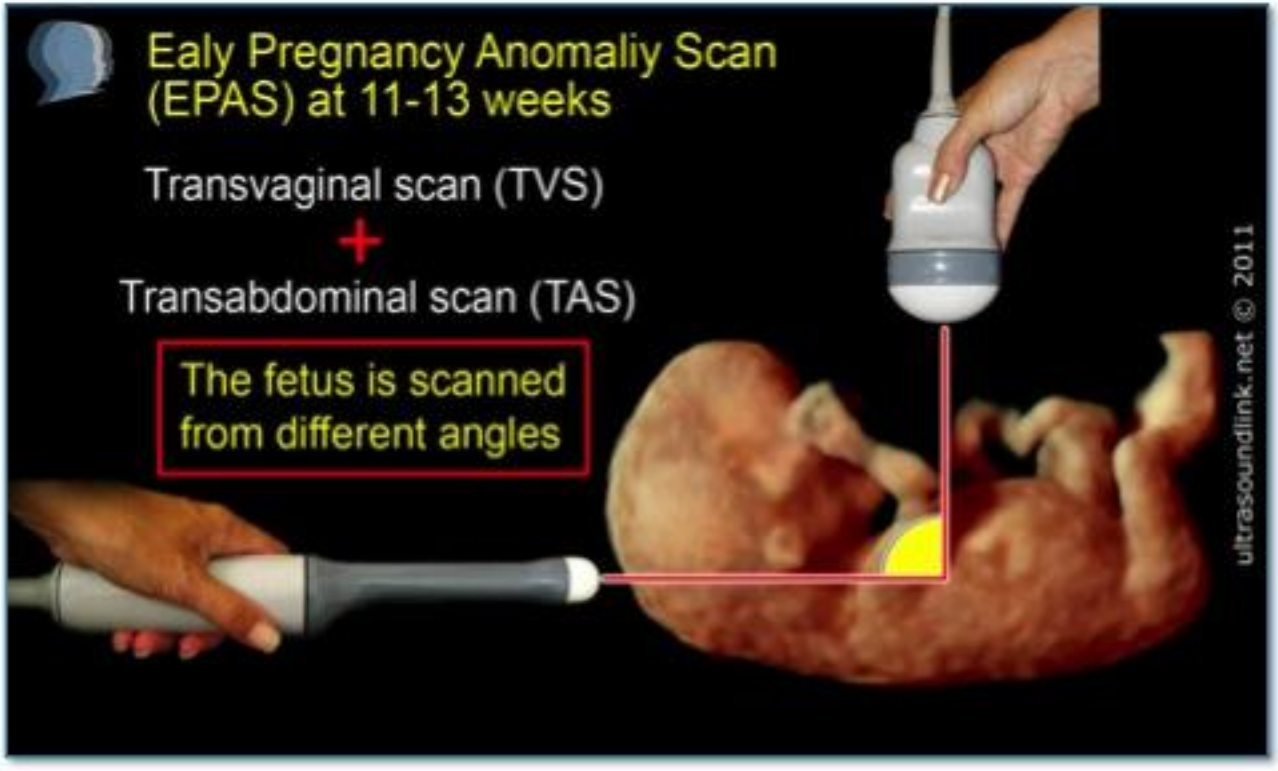
Matrix 6 MHz TA probe

9 MHz TV probe

Linear 9 MHz TA probe

Different structures visible better by different probes

Advantages of combined approaches: angle



Transabdominal vs transvaginal: angle of scanning

TAS:
Sagittal view

TVS:
Transverse (axial) view

Retroverted uterus: indication for TVS examination

TAS

TVS

11-13 weeks TAS basic anomaly scan

~ 45,000 pregnant women

PRENATAL DIAGNOSIS
Prenat Diagn 2011; **31**: 90–102.
Published online in Wiley Online Library
(wileyonlinelibrary.com) **DOI:** 10.1002/pd.2642

Challenges in the diagnosis of fetal non-chromosomal abnormalities at 11–13 weeks

Argyro Syngelaki[1,2,3], Teodora Chelemen[1,2], Themistoklis Dagklis[1], Lindsey Allan[1] and Kypros H. Nicolaides[1,2,3]*

[1]*Harris Birthright Research Centre of Fetal Medicine, King's College Hospital, London, UK*
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NHS Fetal Anomaly Screening Programme

18+0 to 20+6 Weeks Fetal Anomaly Scan National Standards and Guidance for England

The conditions that should be screened for as a minimum from 2010 in the NHS for England are:

Condition	Detection rate (%)	11-13 wk detection rate (%) *Syngelaki et al 2011*
Anencephaly	98	100
Open spina bifida	90	14
Cleft lip	75	5
Diaphragmatic hernia	60	50
Gastroschisis	98	100
Exomphalos	80	100
Serious cardiac abnormalities	50	26
Bilateral renal agenesis	84	0
Lethal skeletal dysplasia	60	50
Edwards' syndrome (Trisomy 18)	95	90
Patau's syndrome (Trisomy 13)	95	95

Anomalies detection rate at 11-13 wk *Syngelaki et al 2011*

100% detection

- body stalk anomaly
- anencephaly
- alobar holoprosencephaly
- exomphalos
- gastroschisis
- megacystis

Potentially detectable

- posterior fossa defects
- spina bifida: ~100%
- facial cleft
- **cardiac defects**
- renal defects
- absent hands / feet

+

- diapragmatic hernia
- encephalocele
- lethal skeletal dysplasias
- hemivertebra

0% detection

- microcephaly
- agenesis of the corpus callosum
- **ventriculomegaly**
- fetal tumors
- echogenic lung lesions

+

- amniotic band syndrome
- lethal arthrogryposis
- bladder exstrophy
- cloacal anomaly
- anal atresia
- many other…

ISUOG: 2013

Ultrasound Obstet Gynecol 2013; **41**: 102–113
Published online in Wiley Online Library (wileyonlinelibrary.com). **DOI**: 10.1002/uog.12342

isuog.org

GUIDELINES

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

isuog.org GUIDELINES

Early pregnancy anomaly scan

11-13+6 wks

Table 2 Suggested anatomical assessment at time of 11 to 13 + 6-week scan

Organ/anatomical area	*Present and/or normal?*
Head	Present
	Cranial bones
	Midline falx
	Choroid-plexus-filled ventricles
Neck	Normal appearance
	Nuchal translucency thickness (if accepted after informed consent and trained/certified operator available)*
Face	Eyes with lens*
	Nasal bone*
	Normal profile/mandible*
	Intact lips*
Spine	Vertebrae (longitudinal and axial)*
	Intact overlying skin*
Chest	Symmetrical lung fields
	No effusions or masses
Heart	Cardiac regular activity
	Four symmetrical chambers*
Abdomen	Stomach present in left upper quadrant
	Bladder*
	Kidneys*
Abdominal wall	Normal cord insertion
	No umbilical defects
Extremities	Four limbs each with three segments
	Hands and feet with normal orientation*
Placenta	Size and texture
Cord	Three-vessel cord*

*Optional structures. Modified from Fong *et al.*[28], McAuliffe *et al.*[87], Taipale *et al.*[60] and von Kaisenberg *et al.*[88].

20 wks

Table 1 Recommended minimum requirements for basic mid-trimester fetal anatomical survey

Head	Intact cranium
	Cavum septi pellucidi
	Midline falx
	Thalami
	Cerebral ventricles
	Cerebellum
	Cisterna magna
Face	Both orbits present
	Median facial profile*
	Mouth present
	Upper lip intact
Neck	Absence of masses (e.g. cystic hygroma)
Chest/Heart	Normal appearing shape/size of chest and lungs
	Heart activity present
	Four-chamber view of heart in normal position
	Aortic and pulmonary outflow tracts*
	No evidence of diaphragmatic hernia
Abdomen	Stomach in normal position
	Bowel not dilated
	Both kidneys present
	Cord insertion site
Skeletal	No spinal defects or masses (transverse and sagittal views)
	Arms and hands present, normal relationships
	Legs and feet present, normal relationships
Placenta	Position
	No masses present
	Accessory lobe
Umbilical cord	Three-vessel cord*
Genitalia	Male or female*

*Optional component of checklist: can be evaluated if technically feasible.

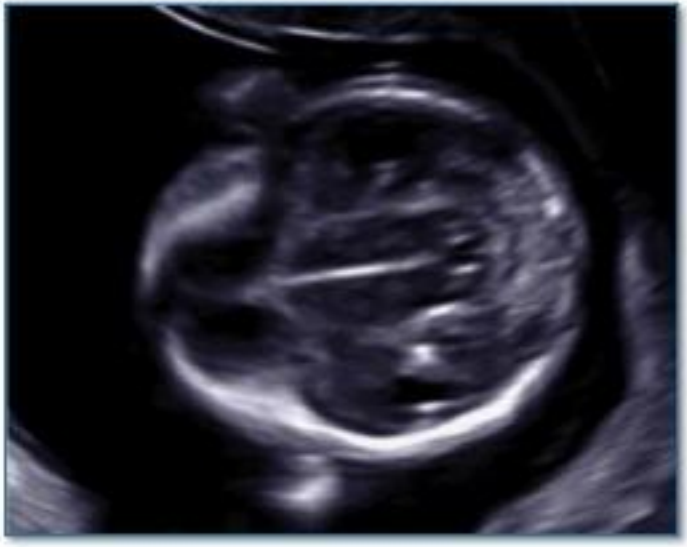
CRL: pregnancy dating

Transvaginal (TVS)

Transabdominal (TAS)

Brain: transverse sweep

Transvaginal (TVS)



High frequency probe

Fetal profile (midsagittal plane)

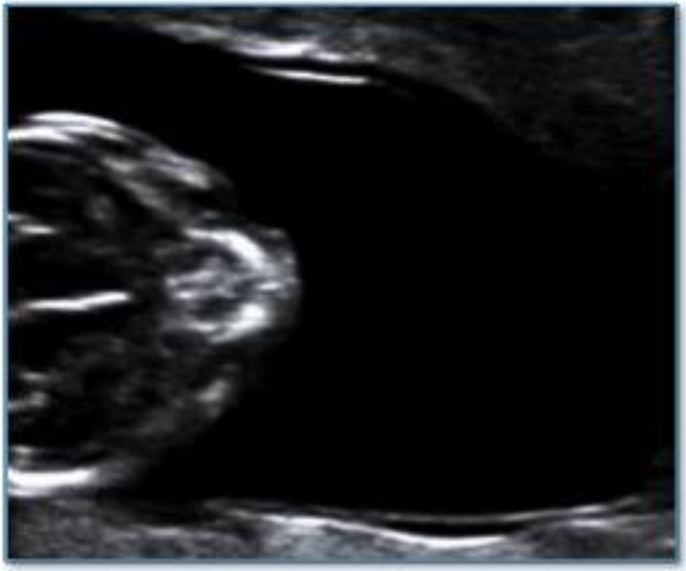
Transvaginal (TVS)

High frequency probe

Face: transverse sweep

Transvaginal (TVS)

High frequency probe



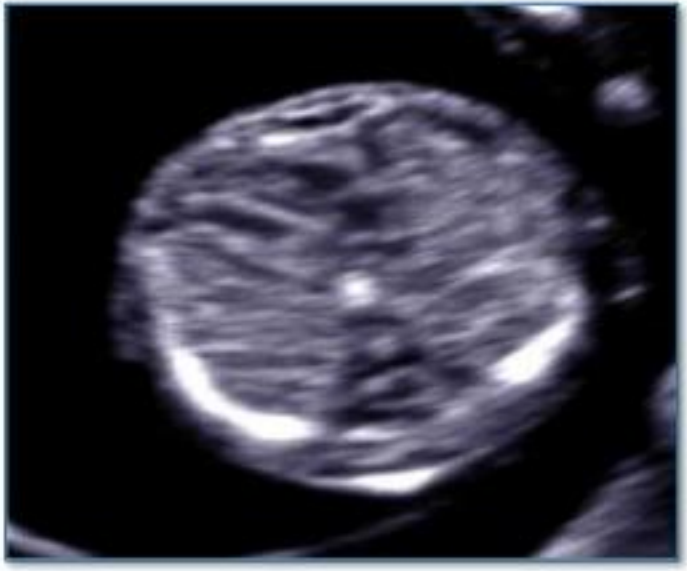
Face: transverse sweep

Transvaginal (TVS)

High frequency probe

Heart: septal view

Transvaginal (TVS)



High frequency probe

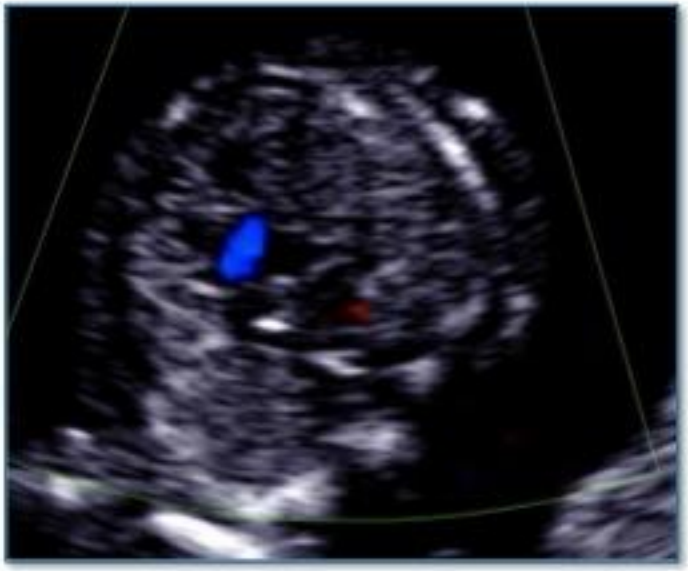
Heart: apical view

Transvaginal (TVS)

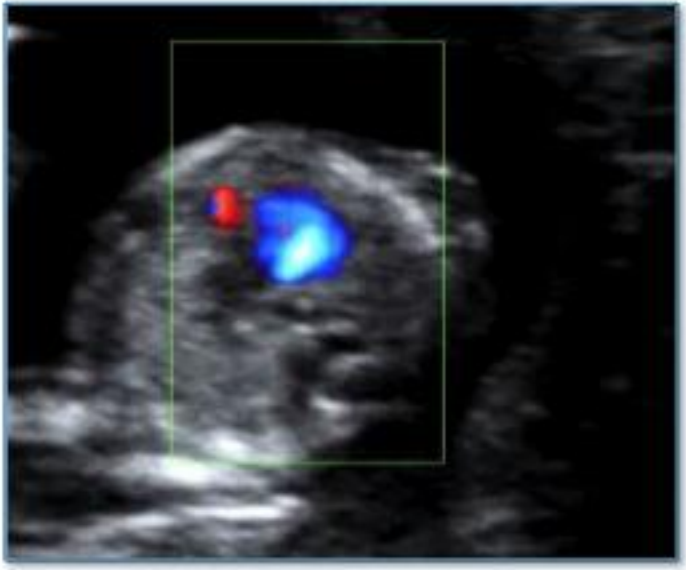
High frequency probe

Heart: colour Doppler

Transvaginal (TVS)



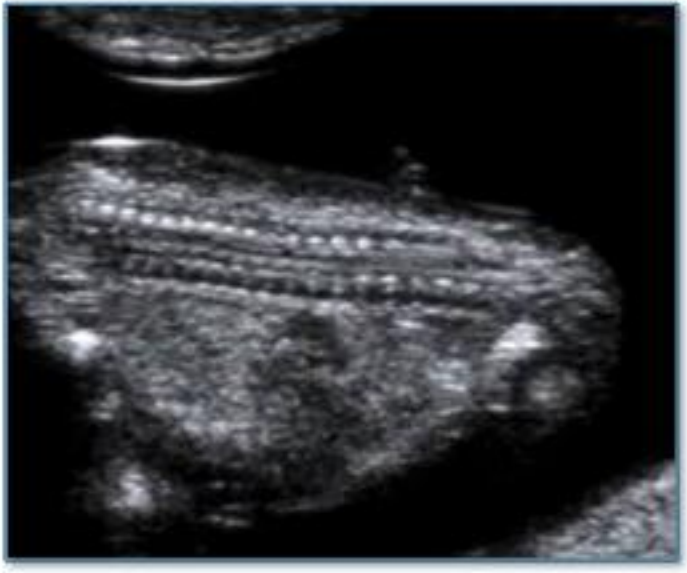
High frequency probe



Spine

Transvaginal (TVS)

High frequency probe



Stomach

Transvaginal (TVS)

High frequency probe

Diaphragm

Transvaginal (TVS)

High frequency probe

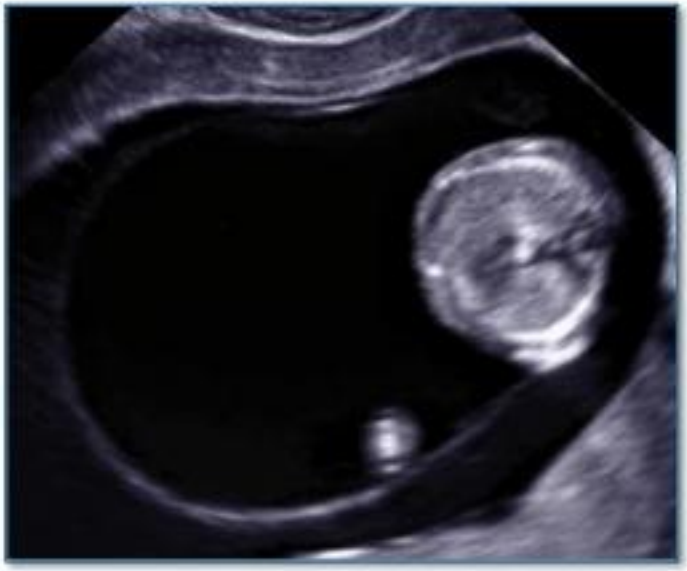
Kidneys

Transvaginal (TVS)

High frequency probe

Umbilical cord insertion

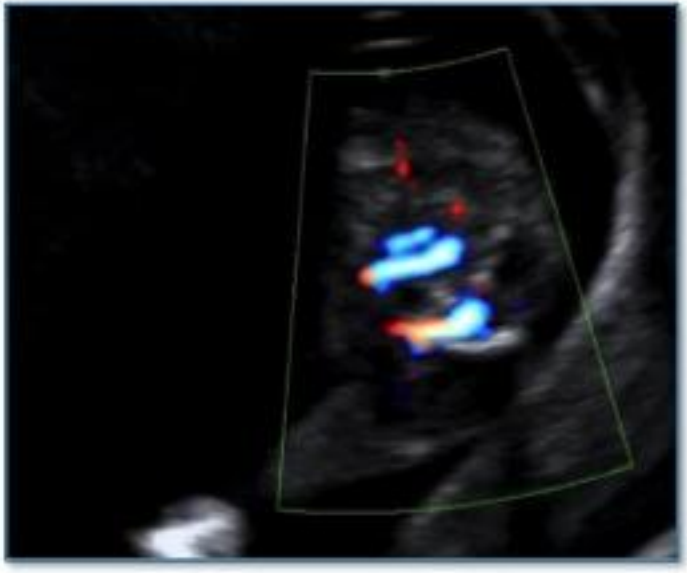
Transvaginal (TVS)



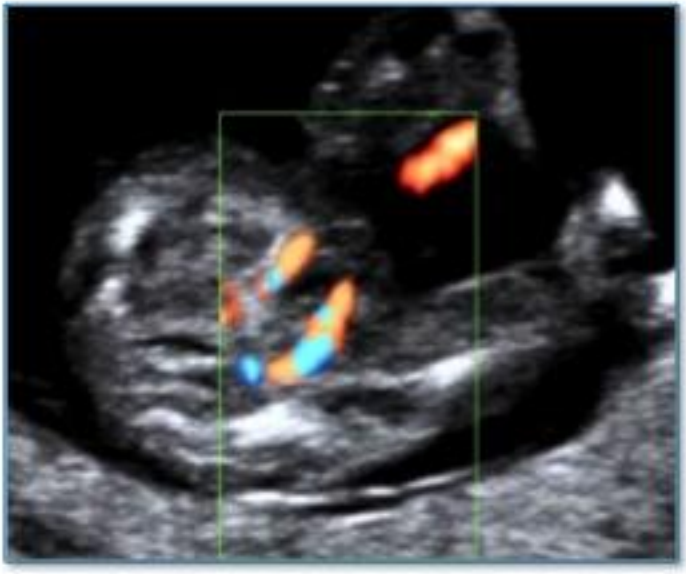
High frequency probe

Bladder and umbilical arteries

Transvaginal (TVS)



High frequency probe



'No flow' if the vessel is 90° to US beam

HD flow, PRF = 0.9 kHz

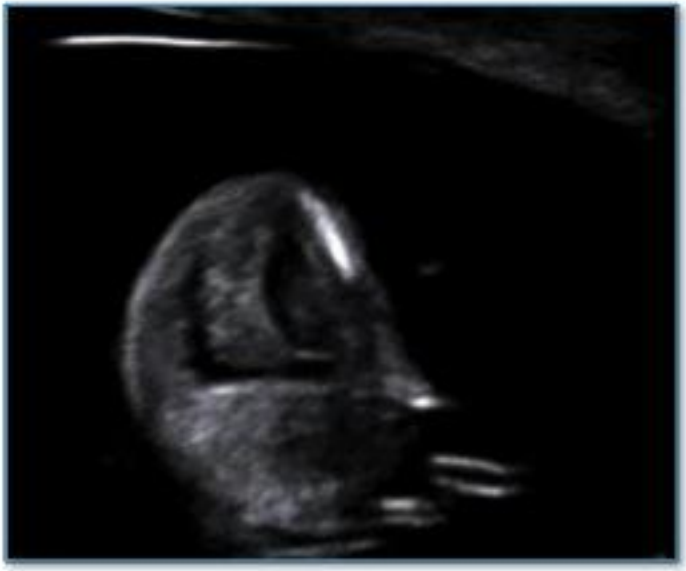
False single umbilical artery

Two UA after angle correction

Upper extremities

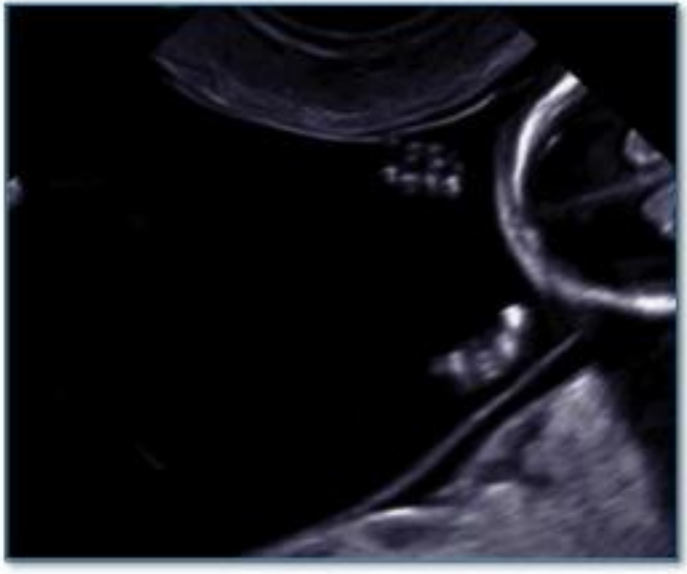
Transvaginal (TVS)

High frequency probe



Hands

Transvaginal (TVS)

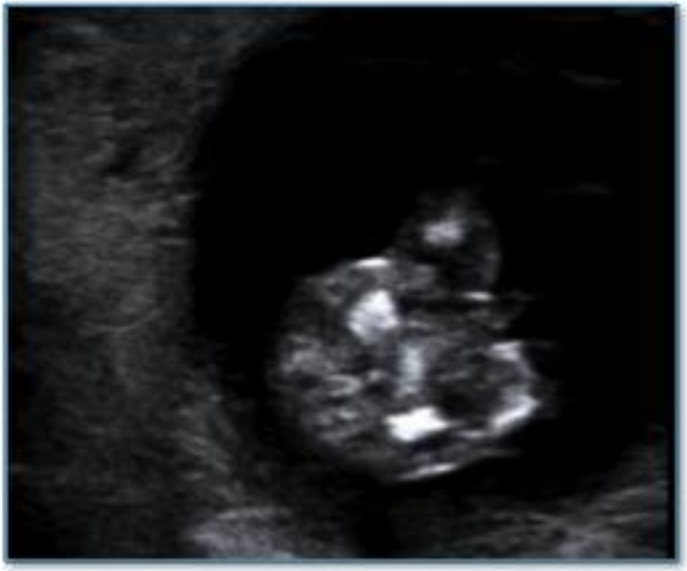


High frequency probe

Lower extremities

Transvaginal (TVS)

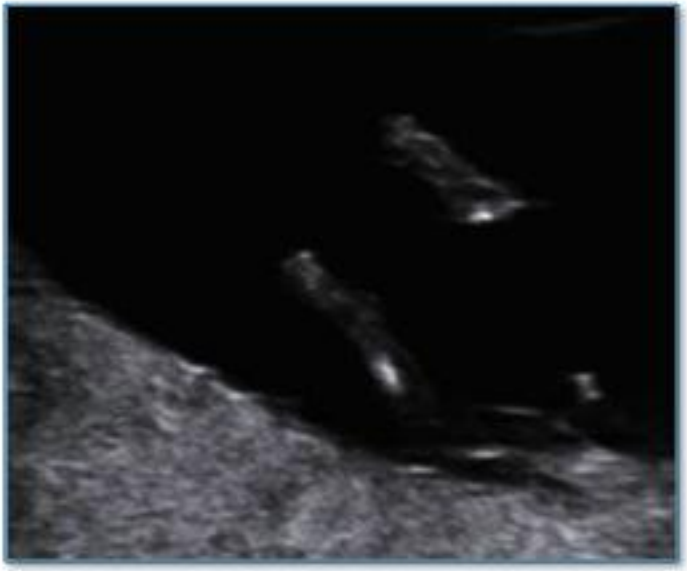
High frequency probe



Lower extremities

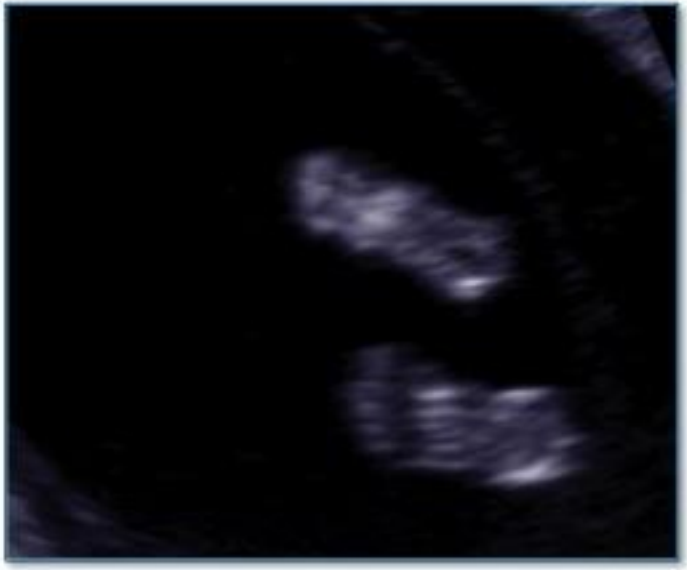
Transvaginal (TVS)

High frequency probe



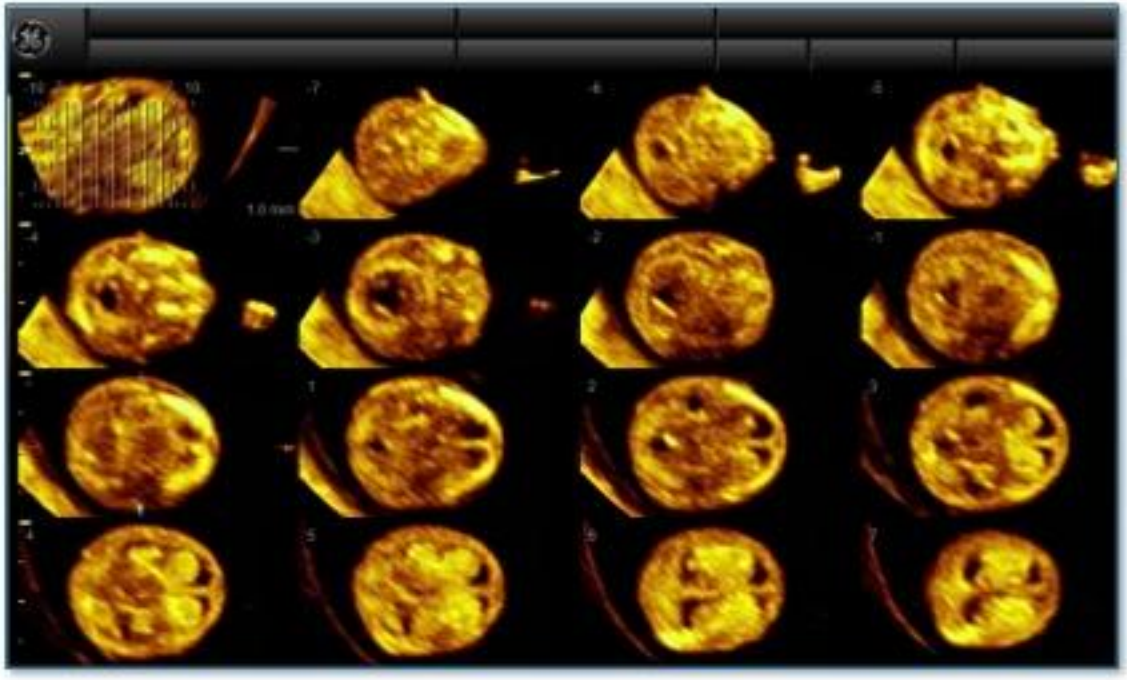
Feet

Transvaginal (TVS)



High frequency probe

How to do 3D neurosonography at 11 wks



Fred Ushakov

CRL = 33.7 mm (10+1wk) 'hydropic baby'

Referred to FMU (scan in 7 days)

FMU: 11 wks → no 'hydrops' # ?spina bifida

CRL = 45.4 mm

NT = 1.0 mm

?No intracaranial translucency (IT) ?Brainstem

FMU: 11 wks → no 'hydrops' # ?spina bifida

How to check the brain at 11 wks (CRL=45.4 mm)?

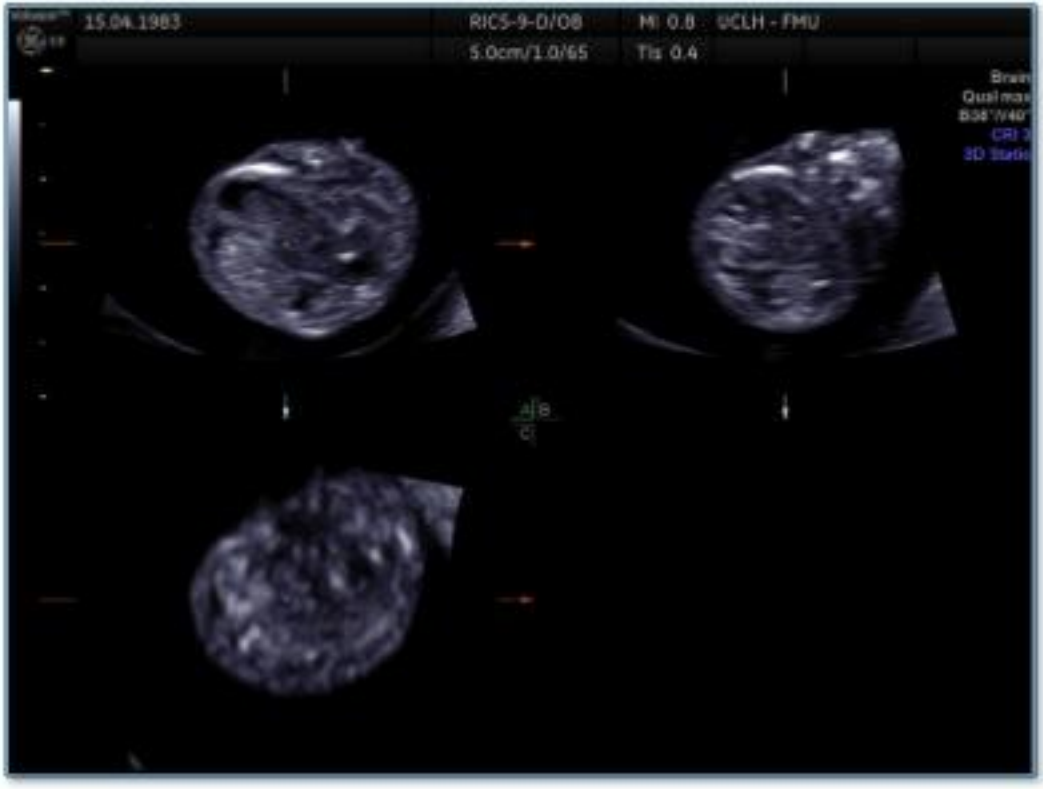
It is easy to exclude serious CHD at 11 wks by TAS

TVS → to check brain → baby 'stands' on its head

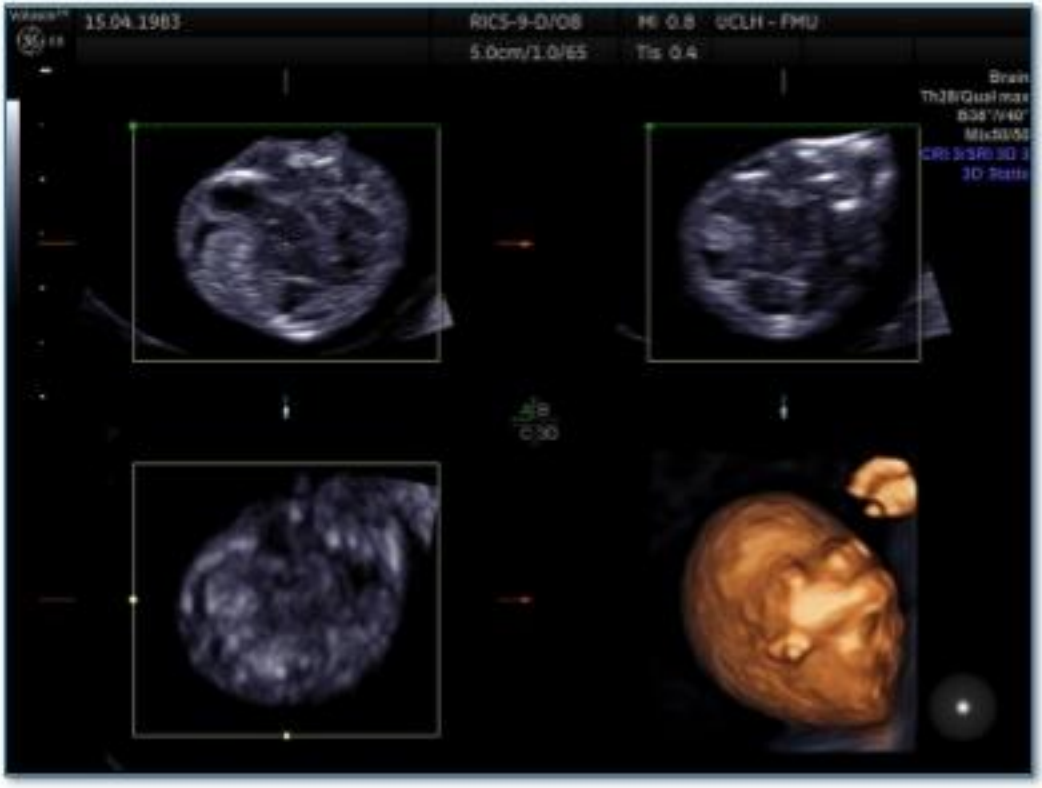
TVS → to check brain → baby 'stands' on its head

3D neurosonography: ROI (region of interest)

3D neurosonography: multiplanar mode

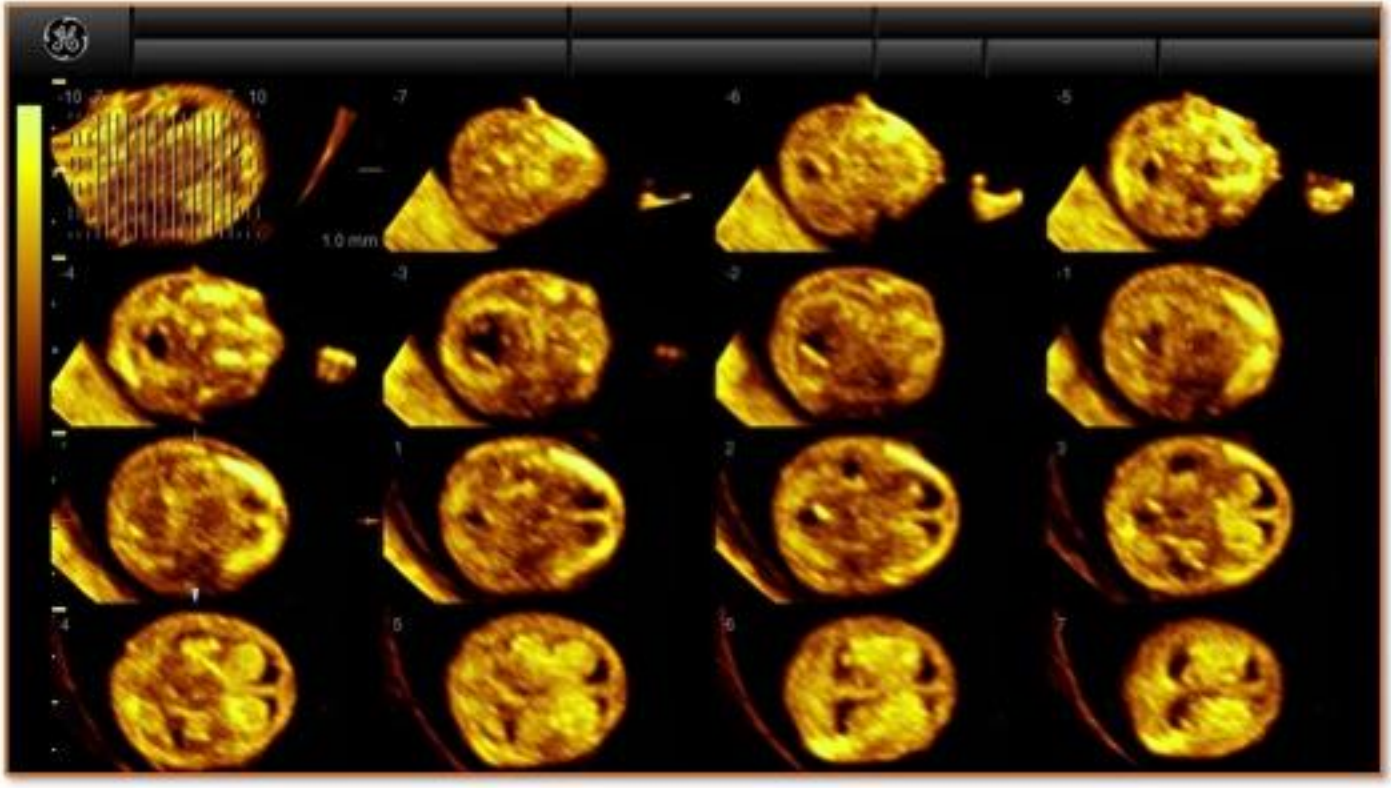


3D neurosonography: surface rendering

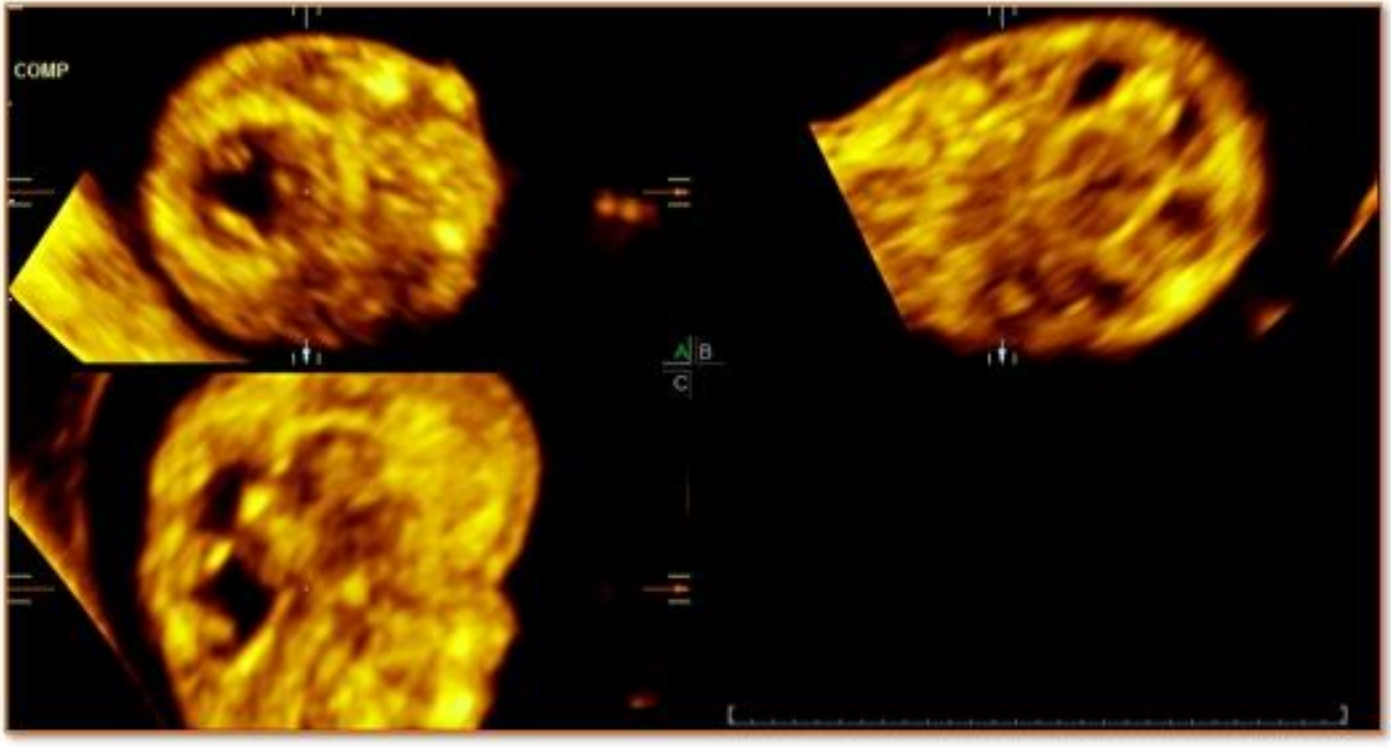


3D neurosonography: surface rendering

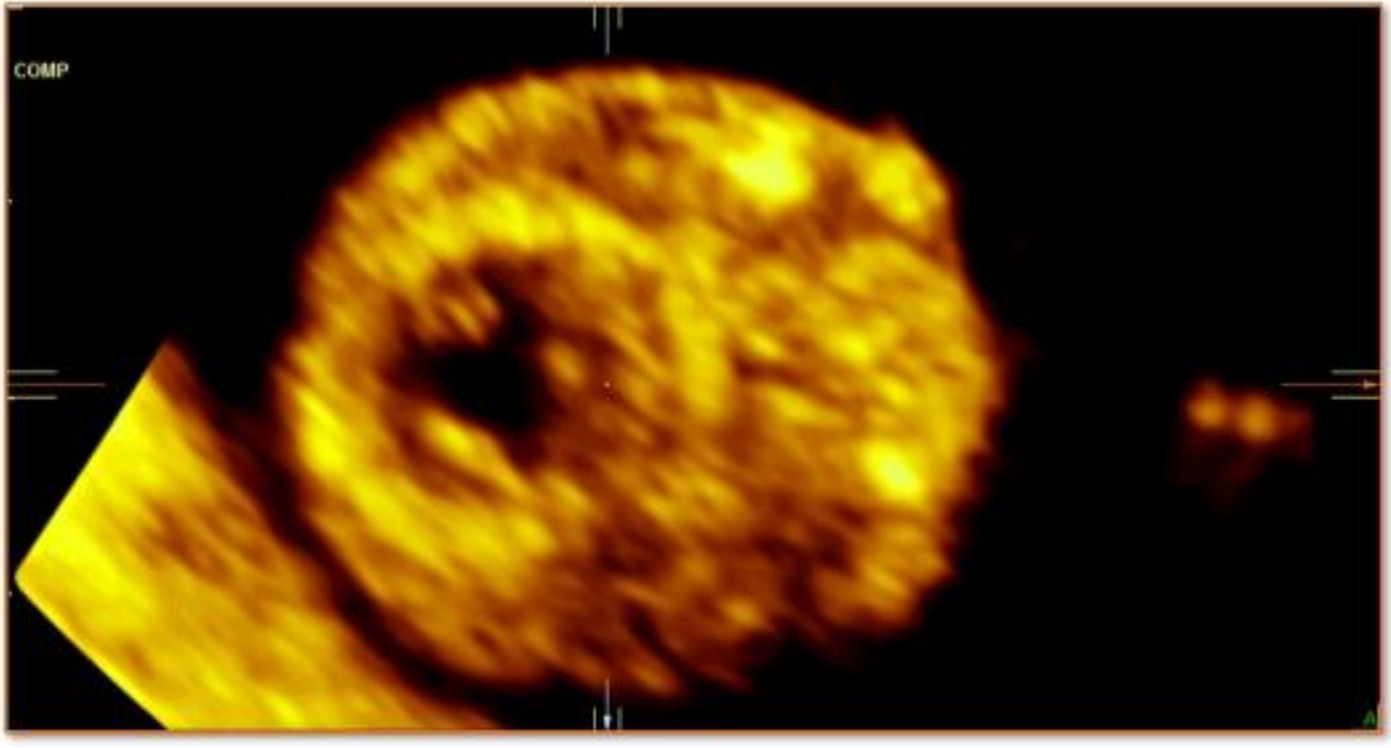
3D neurosonography: TUI



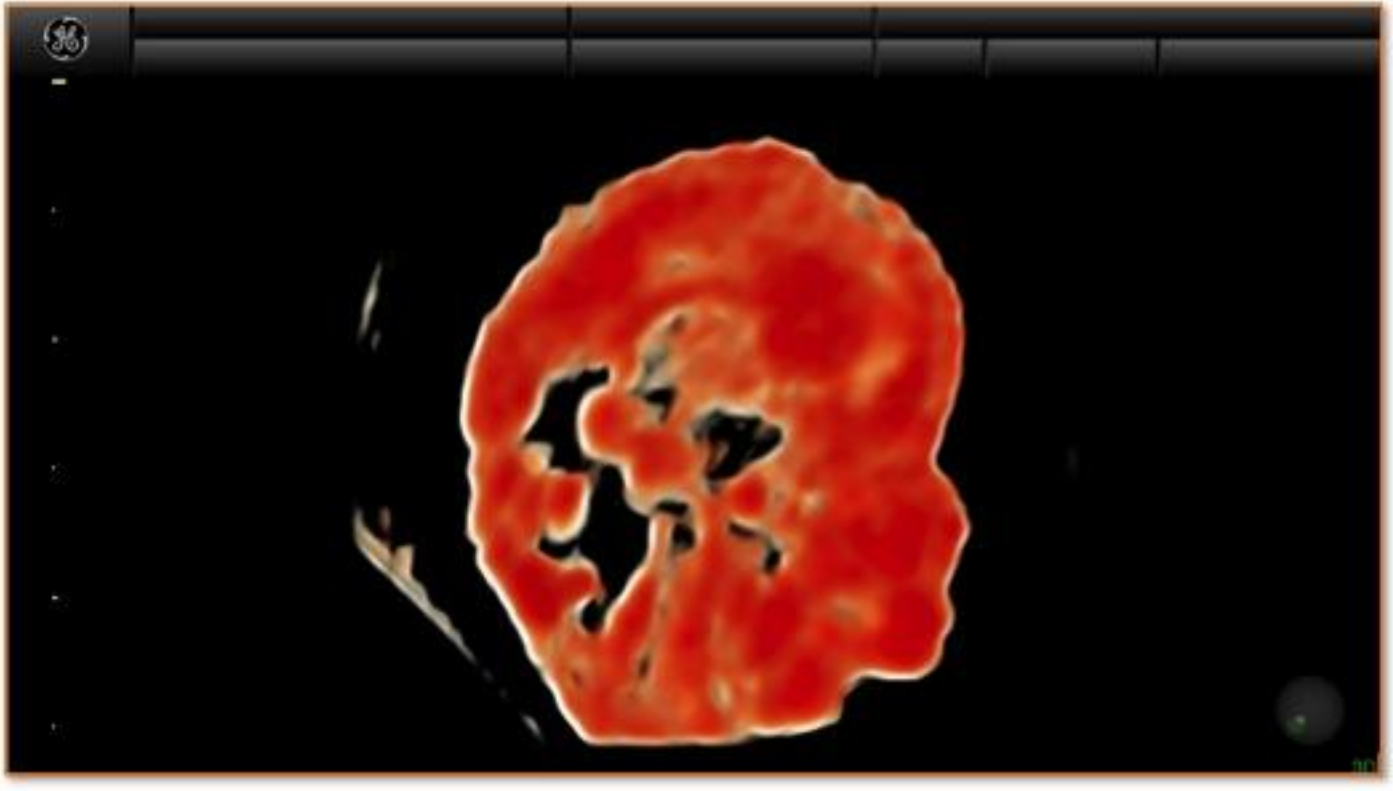
‘Store and scroll’ technique



‘Store and scroll’ technique

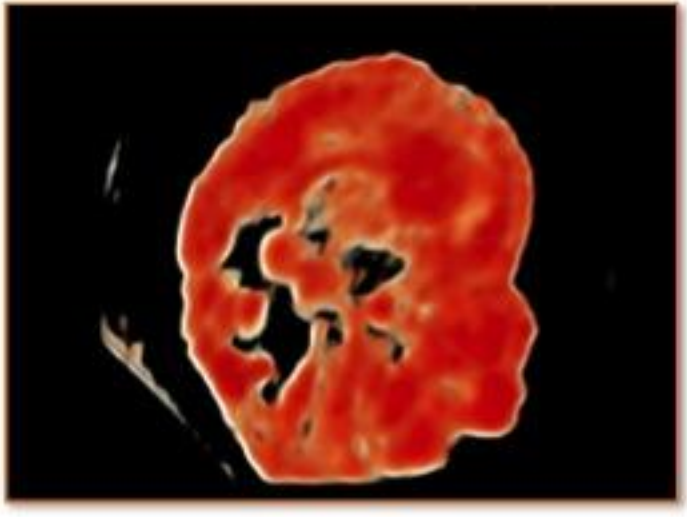


3D mid-sagittal reconstruction ('C' plane)

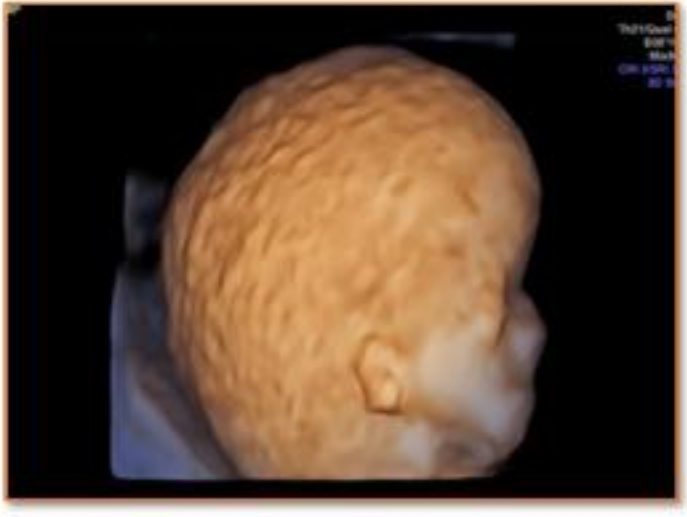


3D mid-sagittal reconstruction ('C' plane)

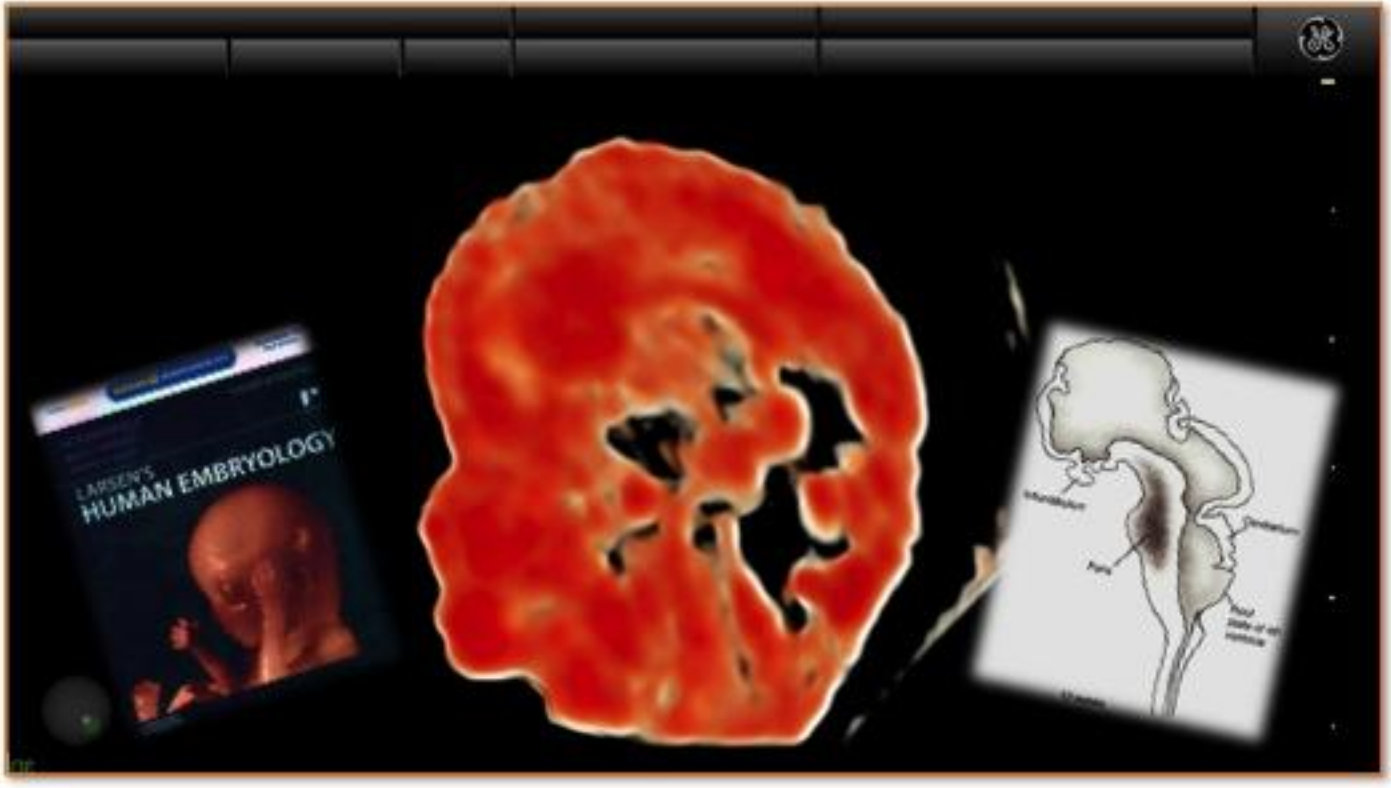
HD*live* Surface rendering



Surface rendering



3D sononeuroembryology



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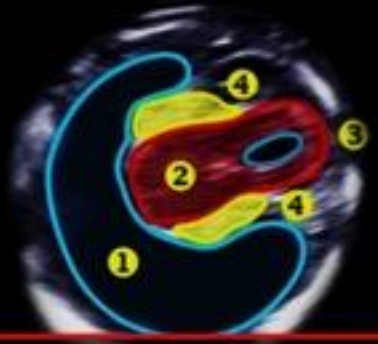


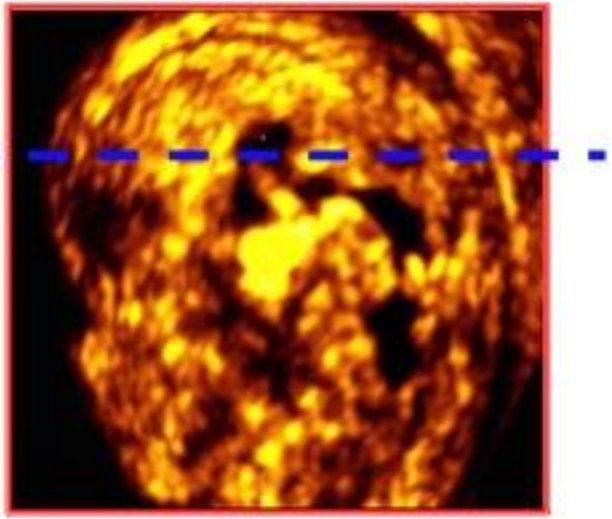
EARLY NEUROSONOGRAPHY: BRAIN, SPINE AND FACE AT 11-13 WEEKS

Early Fetal Neurosonography at 11-13 weeks

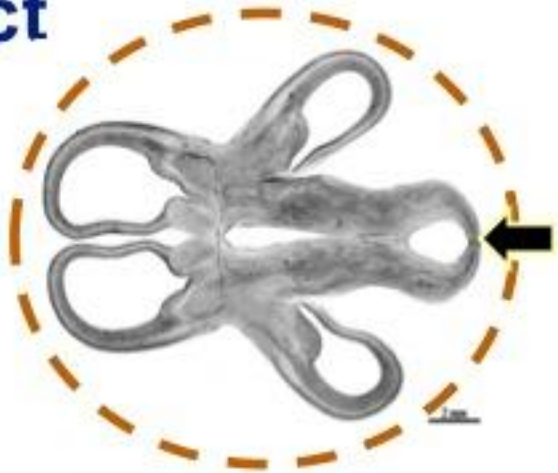
10th March 2018

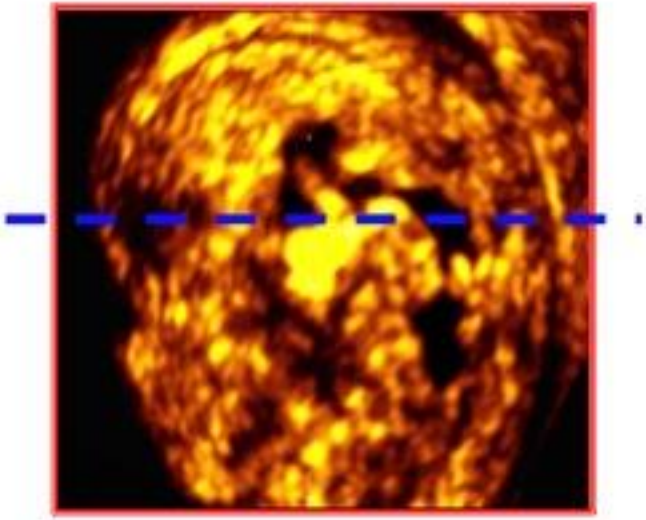
Normal brain: 3rd ventricle

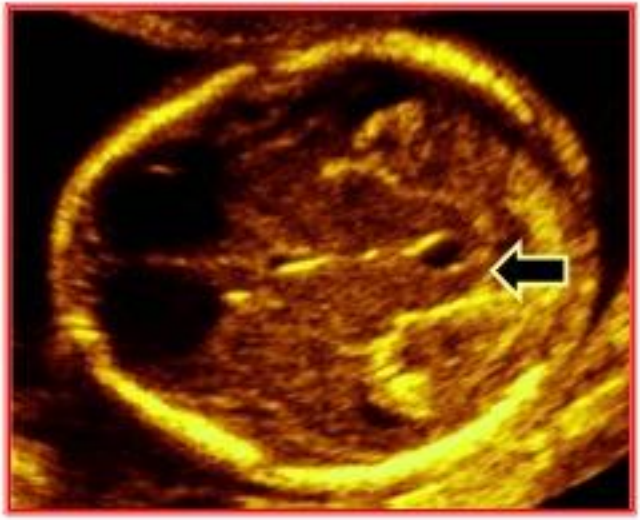
Roof of the 3rd ventricle



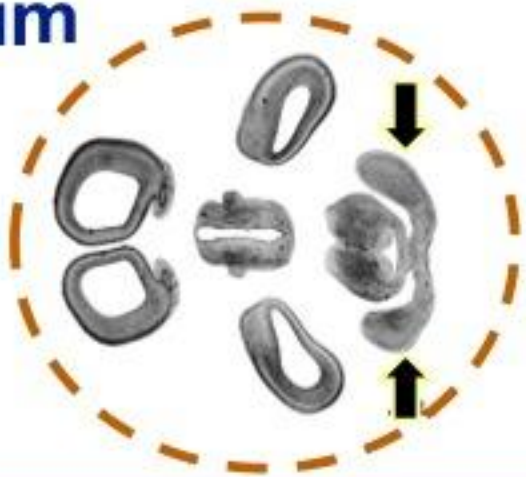
Normal brain: Aqueduct

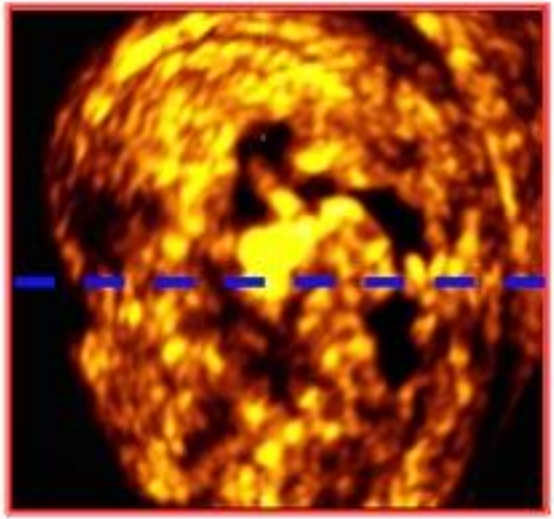






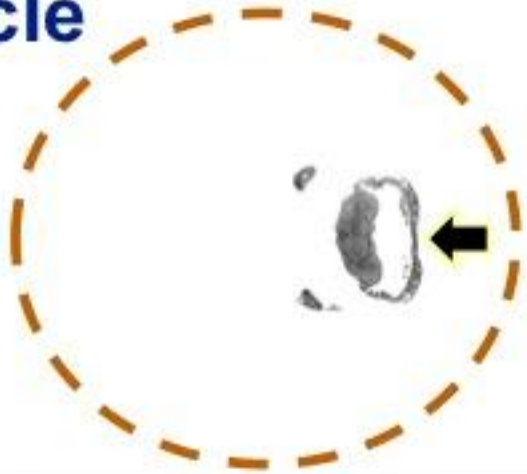
Normal brain: Cerebellum

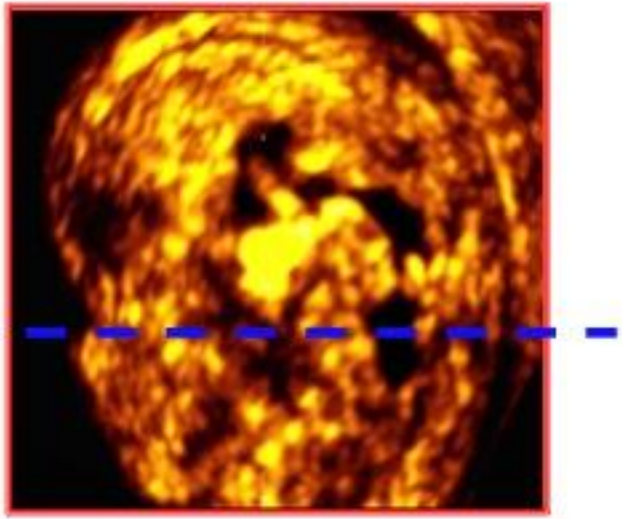




Normal brain: 4th ventricle

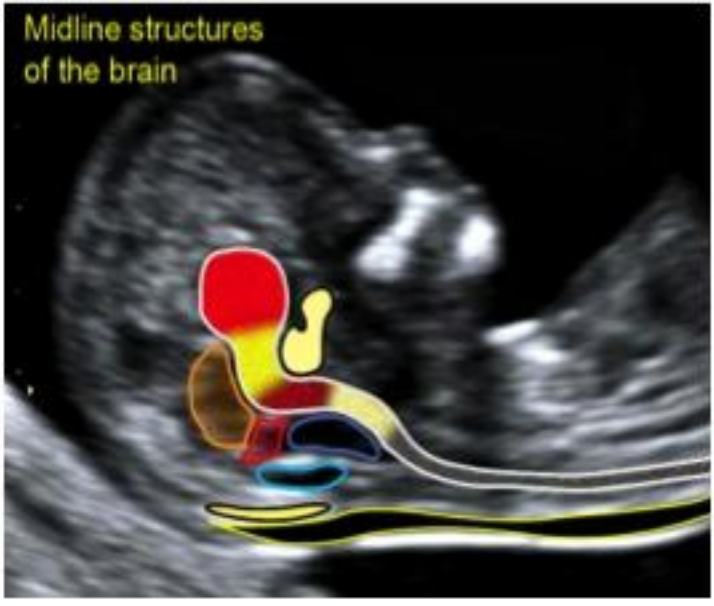
Lateral Pool of
Rhombencephalic
Superventricle



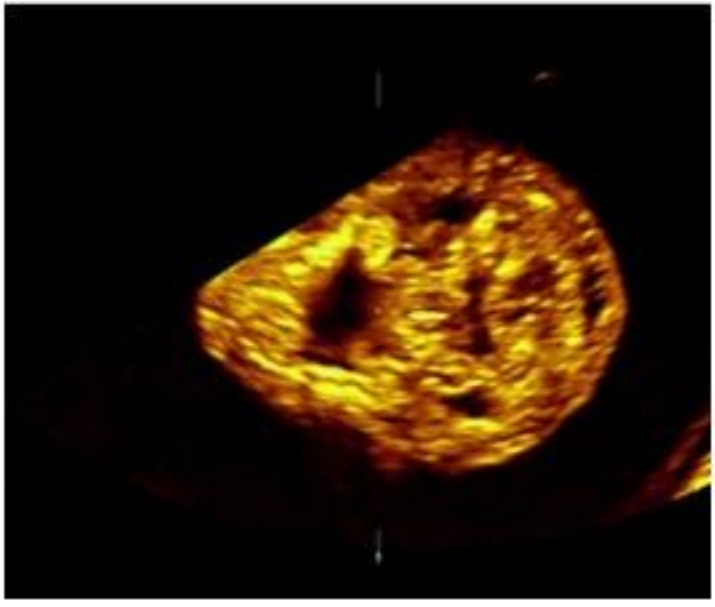


New in 11-13wk neurosonography

Midsagittal (NT) view - TAS



3D "Store & Scroll" - TVS



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New in 11-13wk neurosonography

Midsagittal view – X-rays

3D "Store & Scroll" - CT

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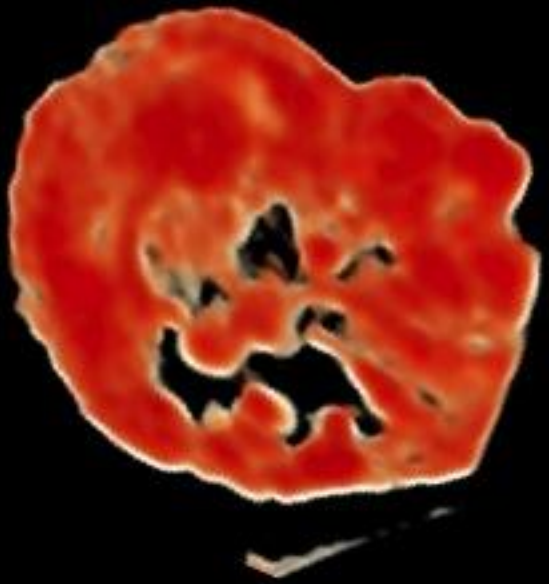
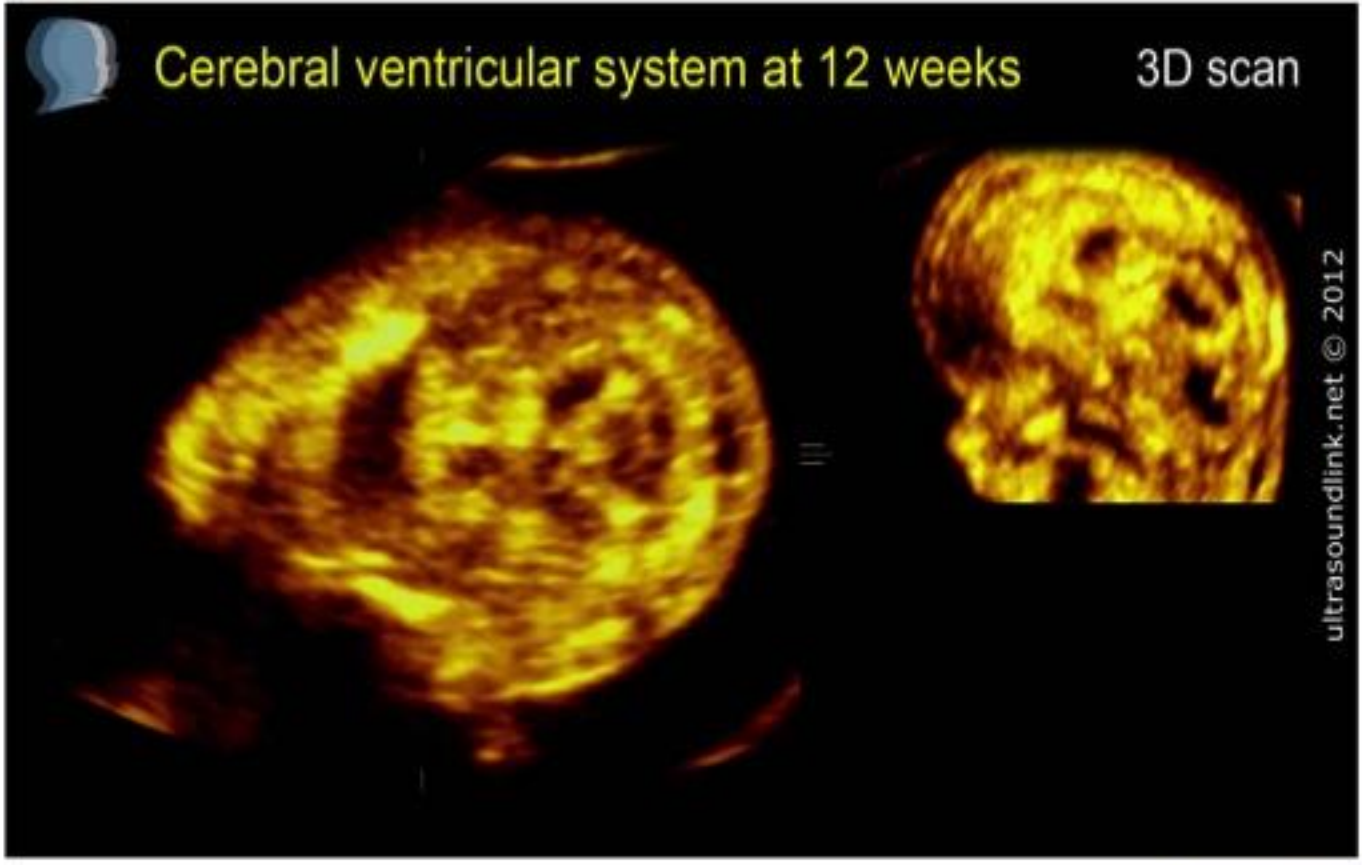
Please learn how to perform 3D neurosonography ☺

Mid-sagittal

Reconstructive sagittal



3D Neurosonography 11-13 weeks



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Common and/or important anomalies at 11-13 wks

- **CNS**
  - ✓ Acrania
  - ✓ Holoprosencephaly
  - ✓ Spina bifida
- **Heart**
  - ✓ Transposition of the great arteries (TGA)
  - ✓ Tetralogy of Fallot (TOF)
  - ✓ Atrioventricular septal defect (AVSD)
  - ✓ Hypoplastic left heart syndrome (HLHS)
- **Cystic hygroma**
- **Chest**
  - ✓ Congenital diaphragmatic hernia (CDH)
- **Abdomen**
  - ✓ Body stalk anomaly
  - ✓ Omphalocele
  - ✓ Gastroschisis
- **Renal**
  - ✓ Megacystis
- **Exteremities**
  - ✓ Polydactyly
  - ✓ Transverse defects
- **Early fetal growth retardation** (FGR)

Acrania – thought to be NTD

"Milky" amniotic luid

Absent cranium, disintegration of the brain structures

Alobar holoprosencephaly (HPE)

12 wks (trisomy 13)

Sagittal view

Axial view

Holoprosencephaly prevalence

Study group	Prevalence	Source
Conceptuses	1:250	Matsunaga E, Shiota K, 1977
11-13 wks scan	**1:1300**	Kagan K, *et al*, 2010
Birth	1:8000 live births	Leoncini E, *et al*, 2008

Majority of embryos/fetuses with HPE die *in utero* or TOP

Holoprosencephaly at 11-13 wks: Diagnosis & outcome

Ultrasound Obstet Gynecol 2010; 36: 10–14
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2010

The 11–13-week scan: diagnosis and outcome of holoprosencephaly, exomphalos and megacystis

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- prospective screening
- 57,199 pregnancies
- 11+0 to 13+6 wks
- TAS
- Prevalence of HPE

  1:1298

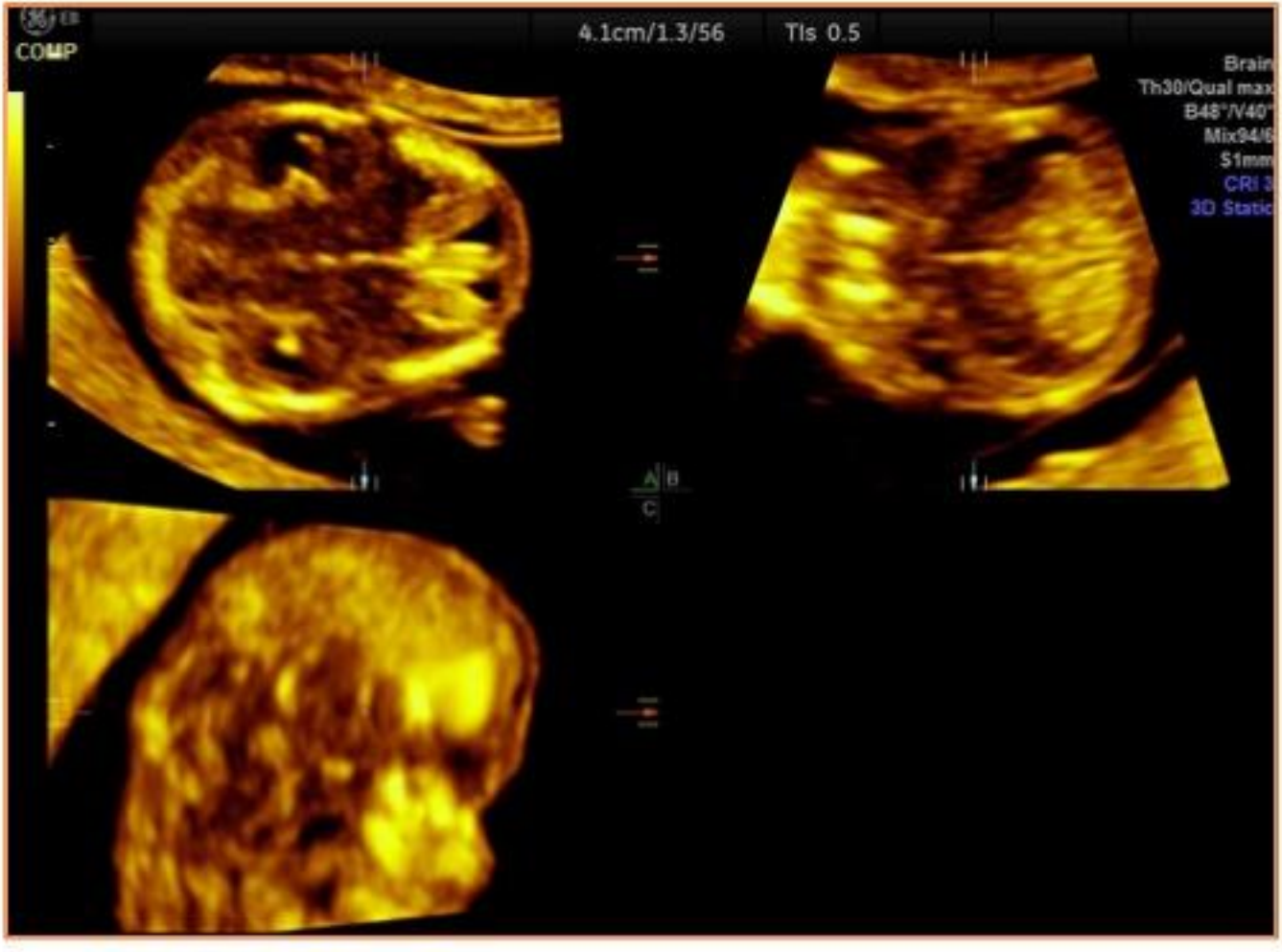
Chromosomal anomalies:

65.9%

- ✓ **Trisomy 13 – 86%**
- ✓ Triploidy – 6%
- ✓ Trisomy18 – 4%

Diagnosis of spna bifida at 11-13 wks

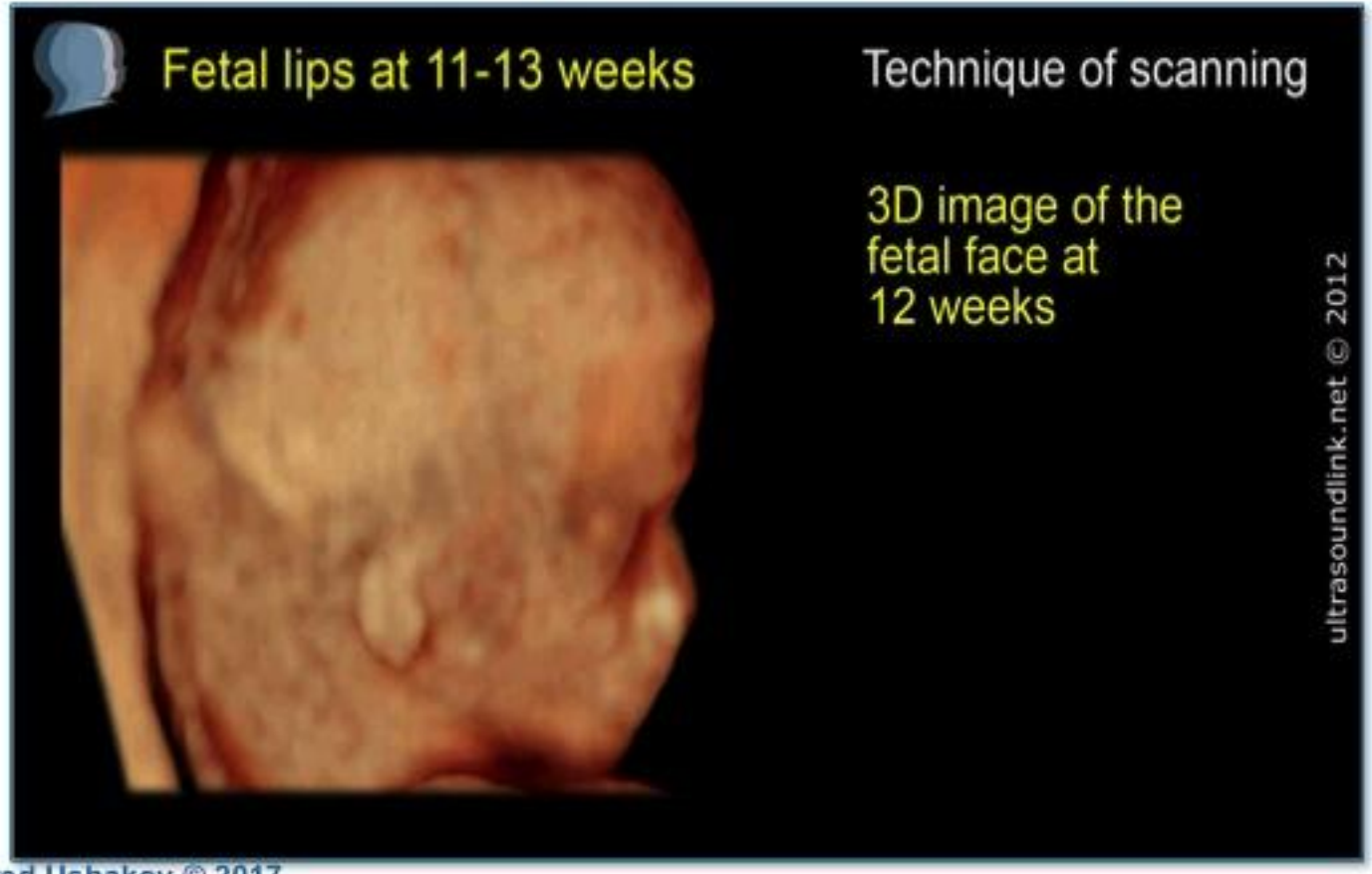
Crash sign and dried-up brain: 11 wks


COMP
4.1cm/1.3/56
TIs 0.5
Brain
3D Static

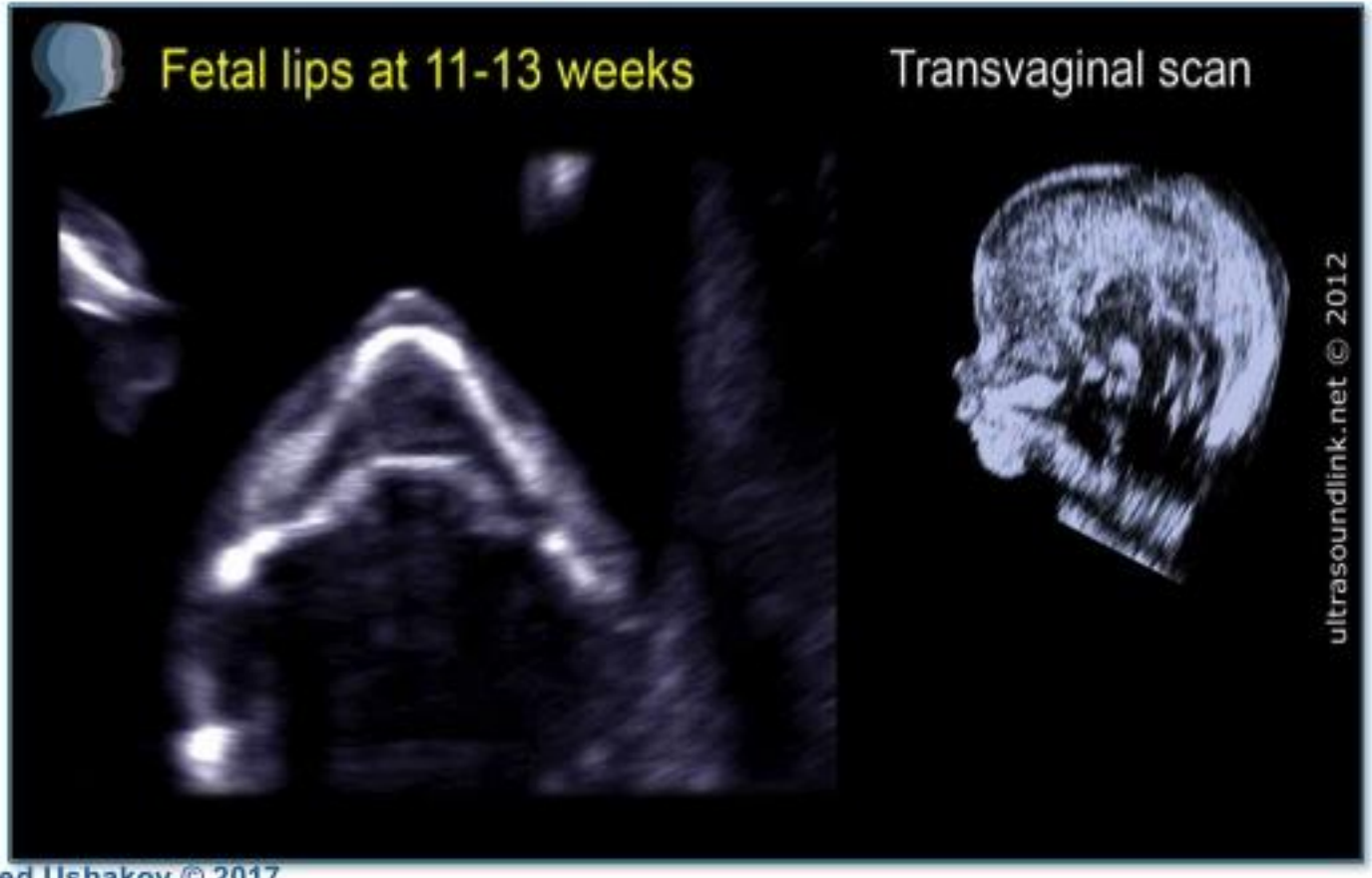
Spina bifida confirmation (follow-up at 13 wks)

Lumbo-sacral meningomyelocele

How to check the lips: transverse sweep



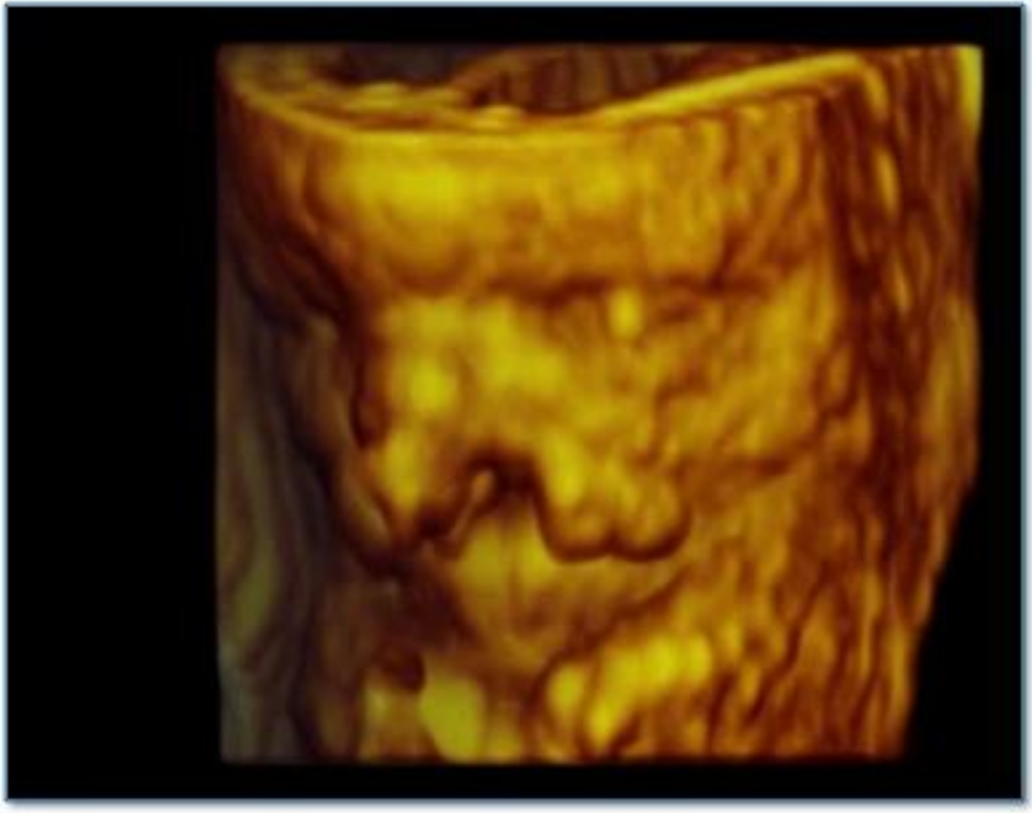
How to check the lips: transverse sweep



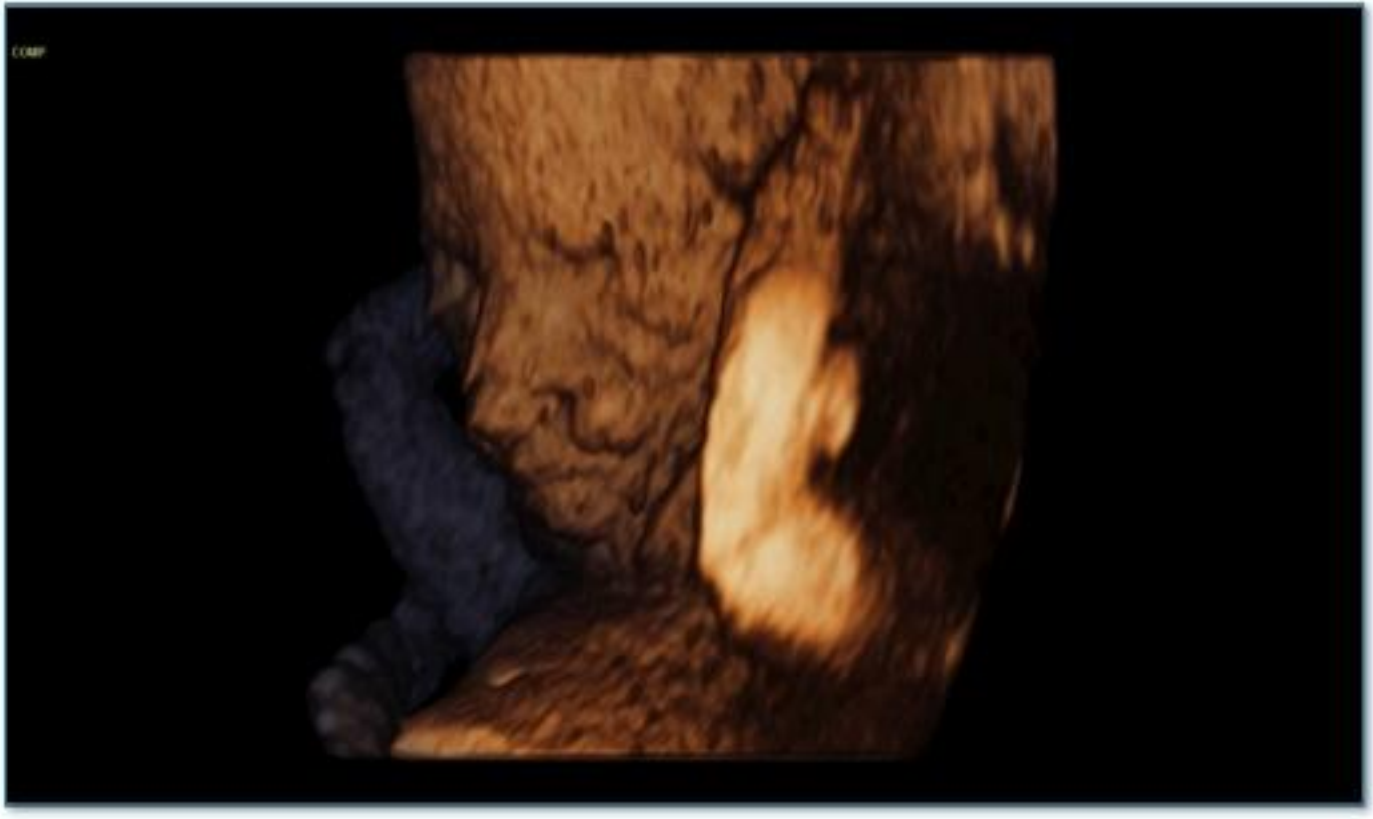
Midsagittal plane: bilateral cleft

Transverse sweep: bilateral cleft

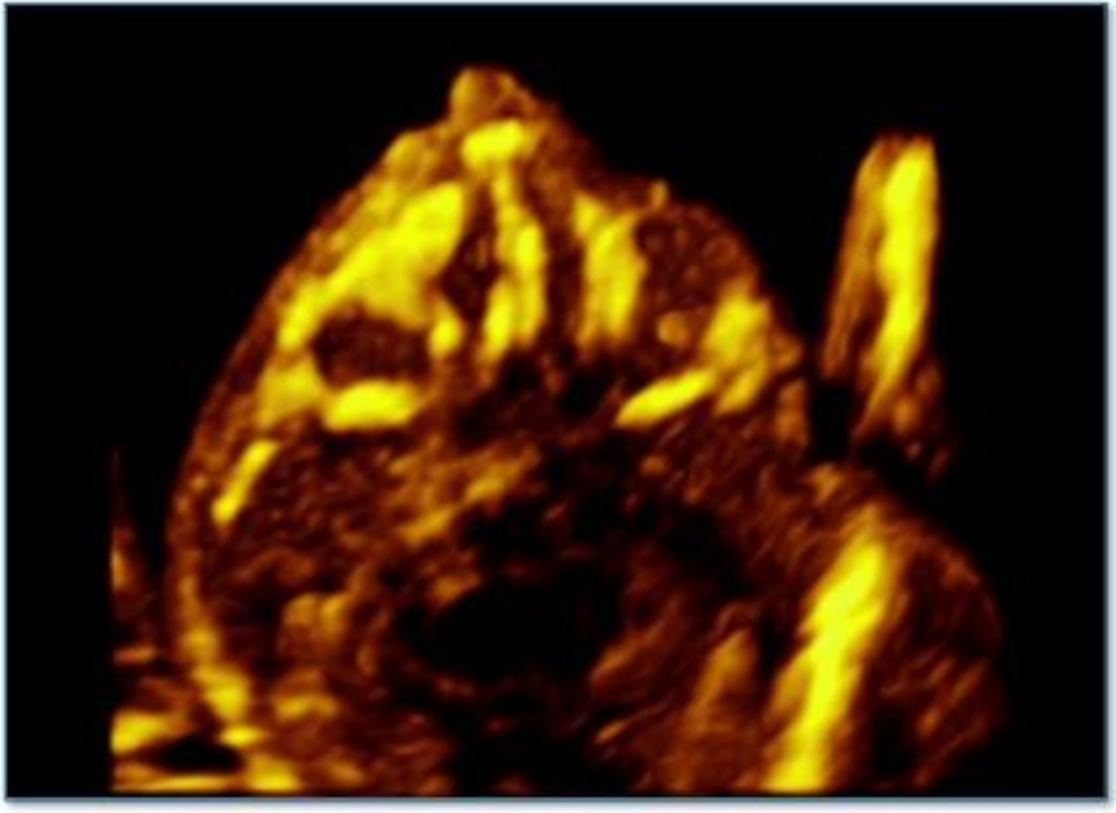
3D rendering; bilateral cleft



Bilateral cleft lip/palate – 13 wks: Normal karyotype

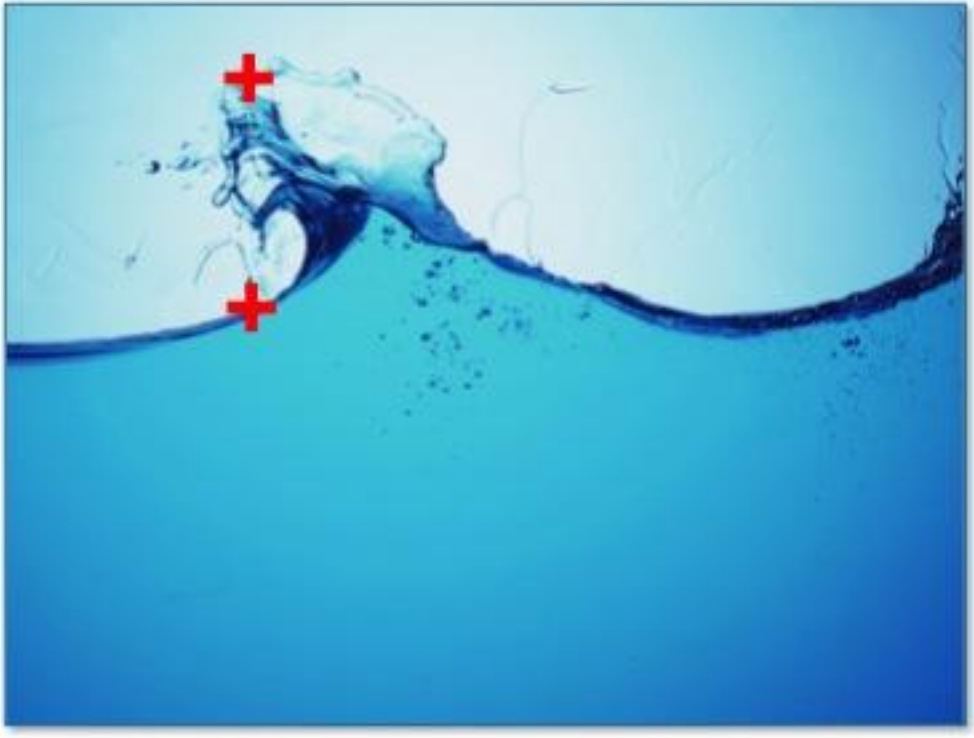


Bilateral cleft lip/palate at 13 wks



Crisis of nuchal translucency (NT)

Dr Fred Ushakov



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NT: Future

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October 17, 2011

The future is the end?

www.news.com.au/lifestyle/parenting/the-chosen-ones-australias-shrinking-number-of-down-syndrome-kids/story-fnet08ui-1226598360762

parenting kids

Is this the beginning of the end for Down syndrome?

This story was published: 4 YEARS AGO | MARCH 17, 2013 1:15PM

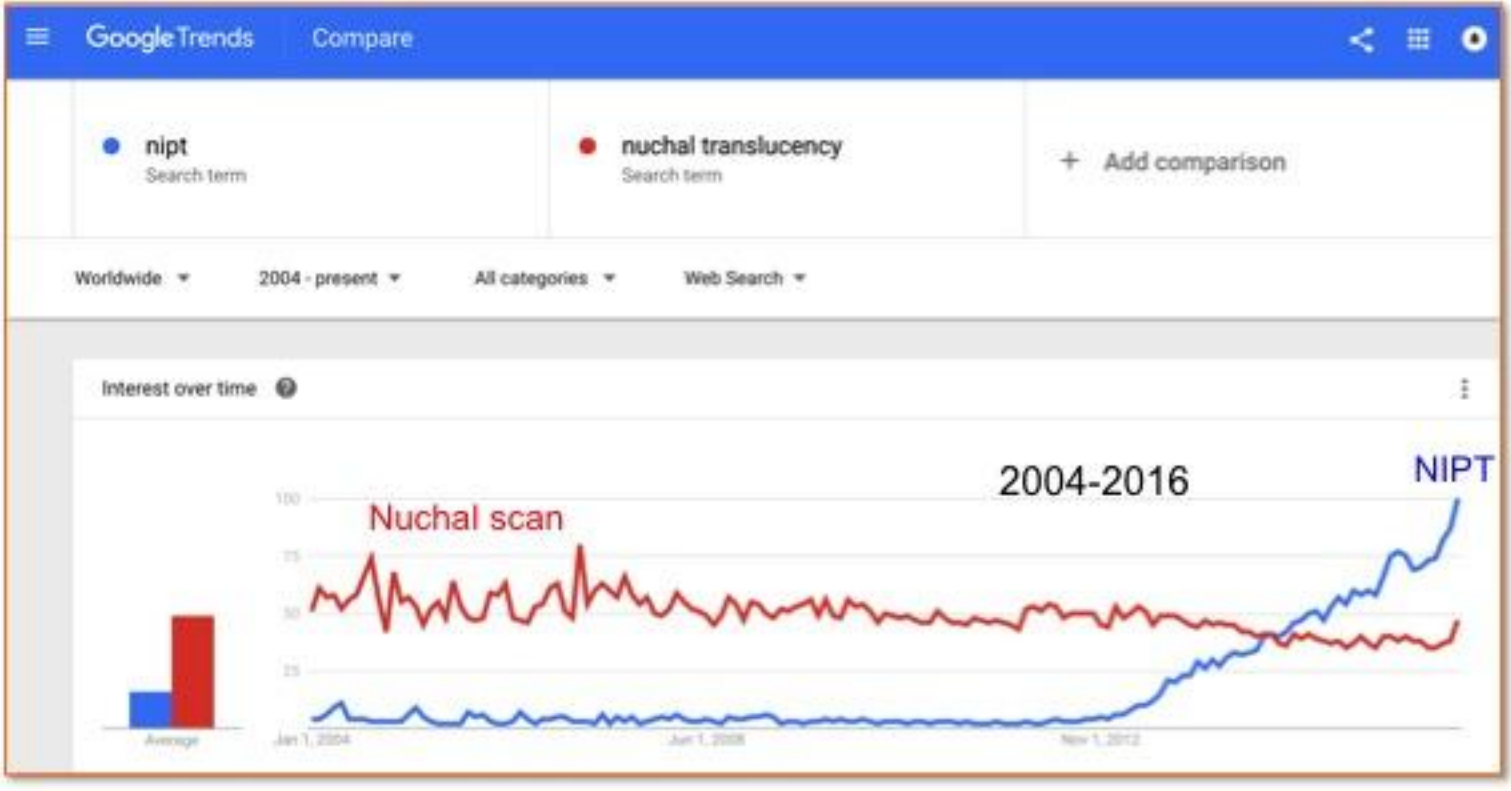
Nicholas Love. Photo: Russell Shakespeare Source: The Courier-Mail

STORY BY

By Leisa Scott

Courier-Mail

Google → Nuchal scan *vs* NIPT



11-13 wks screening: trisomy 21 vs CHD

	Trisomy 21	CHD (severe)
% of all anomalies	8%	8% (31% for all CHD)
Main problem	Extra chromosome 21	Abnormal structure of the heart
Diagnosis	Karyotype	Echocardiography
Screening past	Structural / biochemical markers	To check the structure of the heart
Screening future	Search for extra chromosome cfDNA = ffDNA = NIPT	To train the specialists to check the heart
Screening base	Genotype	Phenotype
Screening aims	TOP	To improve outcome TOP

Down's syndrome is only 8.6% of congenital anomalies

Anomaly	Live birth (LB) / 10,000 births	LB+IUD +TOP / 10,000 births	%TOP	% of all anomalies (LB+IUD+TOP)
All Anomalies	176.3		15%	100%
Congenital heart defects	55.4			31%
Severe CHD	**16.8**	**17.41**	**15%**	**8.3%**
• Transposition of great vessels	3.31			
• Tetralogy of Fallot	3.29			
• Atrioventricular septal defect	2.74			
• Hypoplastic left heart	1.29		36%	
• Coarctation of aorta	3.31			
Ventricular septal defect	26.5			14%
Chromosomal	13.6			14%
Down syndrome	**8.82**	**17.88**	**46%**	**8.6%**
Edwards syndrome/trisomy 18	0.86			
Patau syndrome/trisomy 13	0.37			
Turner syndrome**	0.63			

Detection of CHD at 11-13 weeks

CHD are:

- Common
- Severe
- As a group very variable
- Associated with other anomalies
- Generally poorly detectable
- Can be detected at 11-13 weeks
- Prenatal diagnosis improves the outcome
- Training of sonographers improves detection
- Have devastating impact on the family
- Ignored by medical society

The 12 wks Cardiac Scan: transabdominal

1. Heart scan is not 'taking pictures' → dynamic examination of normal pattern of cardiac anatomy
2. Same scanning planes as for 20-22 wks
3. Directional power (or color) Doppler

Stomach - situs

4 chamber view

LVOT

RVOT+ V-sign

Congenital heart disease at 11-13 wks

4 common & clinically important anomalies: detectable!

1. Transposition of the great arteries (TGA)
2. Tetralogy of Fallot (TOF)
3. Atrioventricular septal defect (AVSD)
4. Hypoplastic left heart syndrome (HLHS)

Transposition of the great arteries (TGA) - 13 wks

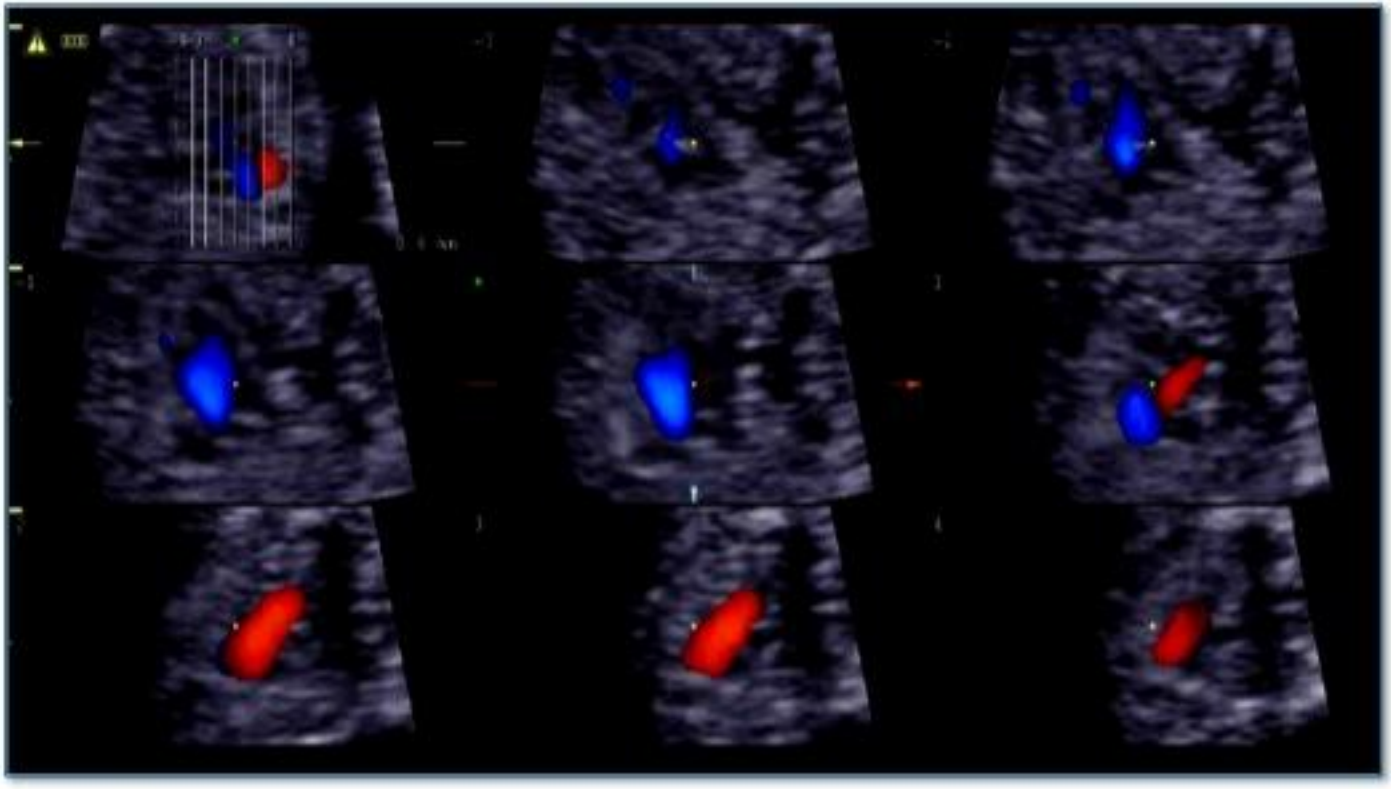
Parallel arteries

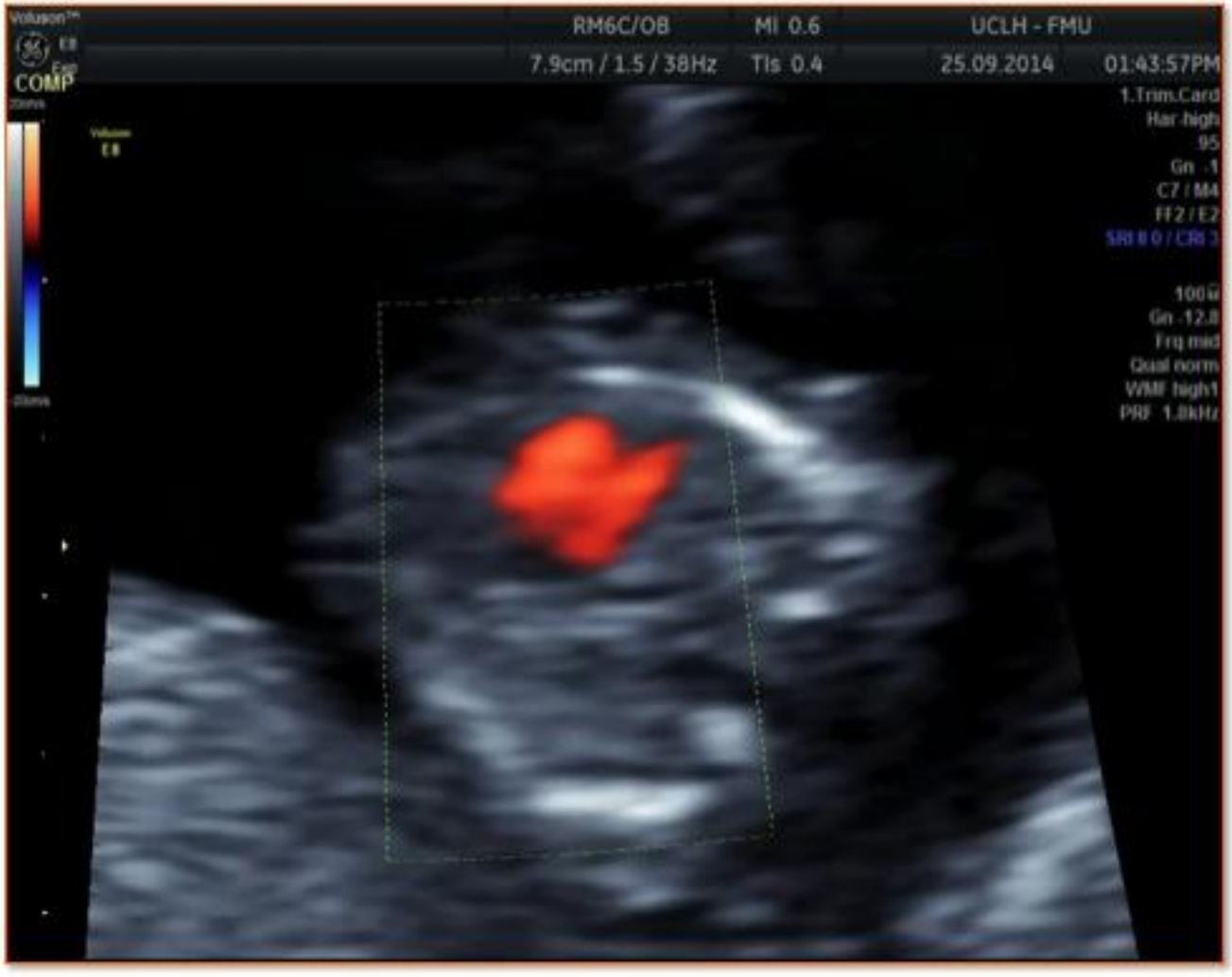
Normal 4CV, no crossing

TGA at 12 wks → fantastic diagnosis in 5 cardiac cycles!

Dr Anna Knafel, Krakow, Poland

Tetralogy of Fallot (TOF) at 11wks (first in 2009!)




COMP
RM6C/OB
MI 0.6
UCLH - FMU
7.9cm / 1.5 / 38Hz
TIs 0.4
25.09.2014
01:43:57PM
1.Trim.Card
Har high
95
Gn -1
C7 / M4
Ff2 / E2
100
Gn -12.8
Frq mid
Qual norm
WMF high1
PRF 1.8kHz

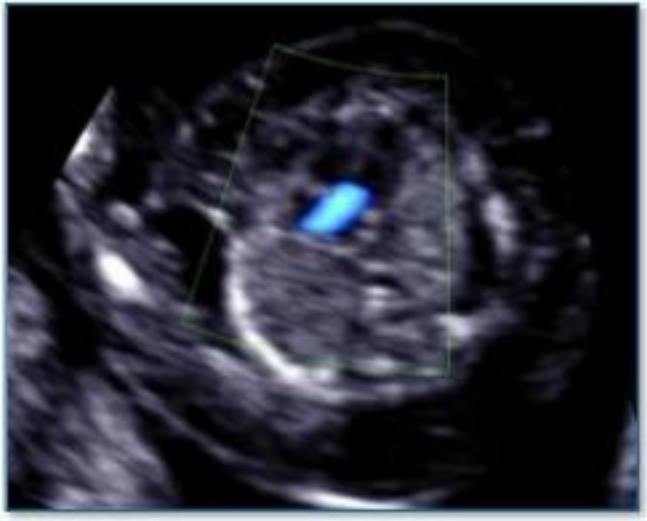
Conotruncal anomaly pattern (simplified as TOF)

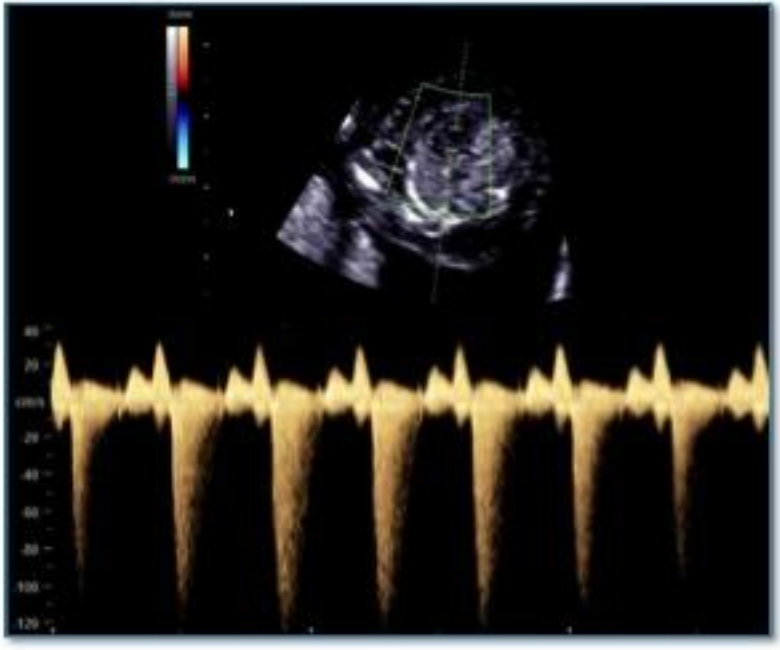
Normal 4CV & single visible large overriding vessel forming aortic arch

DD: Possibilities:

- Tetralogy of Fallot
  - ✓ PA is not visible (small)
- DORV
- Truncus arteriosus
- Pulmonary atresia

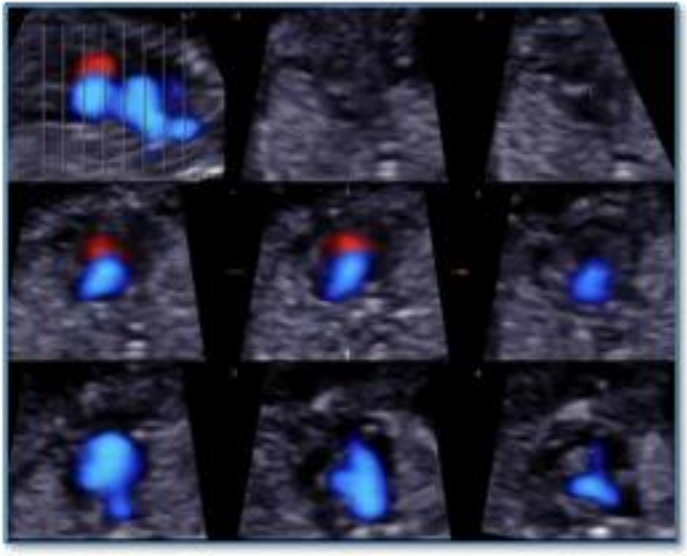
AVSD: common AV valve regurgitation





Trisomy 21

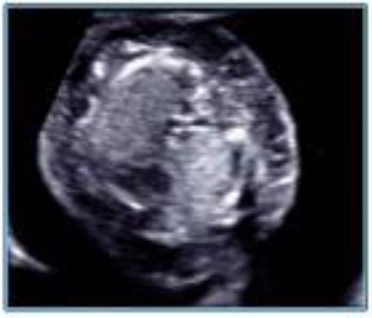
D.G. AVSD at 13 wks



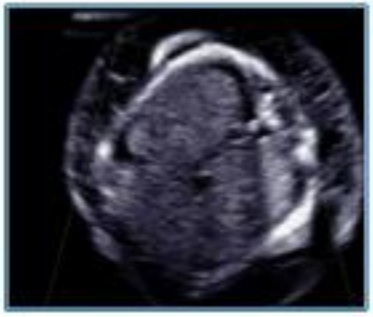
Trisomy 21

Hypoplastic left heart syndrome at 12 wks

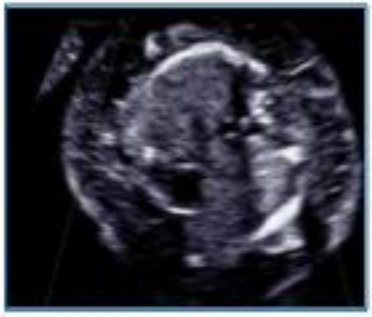
4 chamber view
1. Right ventricle big
2. Left ventricle small



Left outflow tract
1. Aortic atresia



Right outflow tract
1. Big pulmonary artery



Arterial arches
1. Ductus arteriosus
2. Transverse aorta
3. Retrograde flow in aorta

fetalechocardiodraphy.com

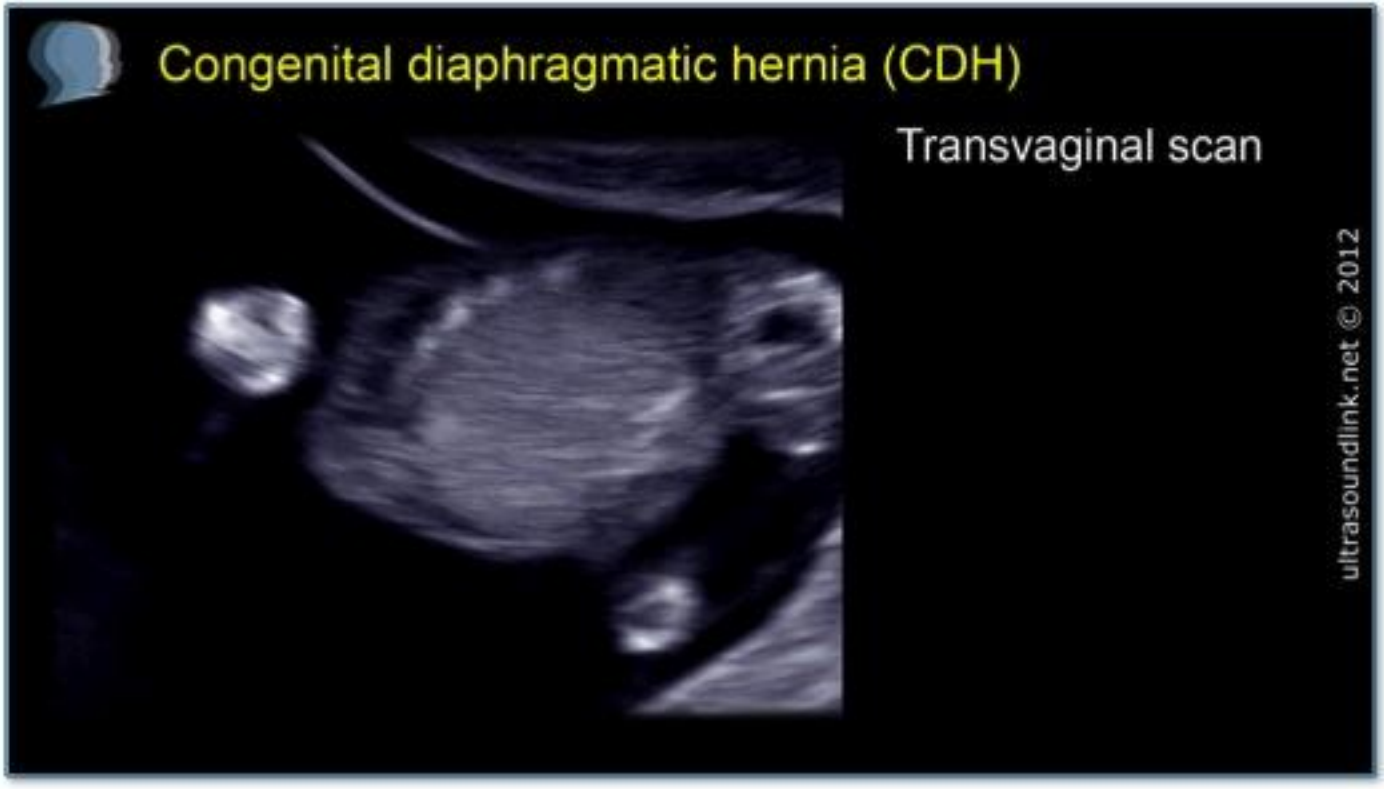
London School of Ultrasound

Early Fetal Echocardiography at 11-13 weeks

11th November 2017

Congenital diaphragmatic hernia (CDH) at 12 wks (TAS)

CDH at 11+6 wks



Anterior wall defects: D/D at 11-13 wks

- Physiological bowel herniation
- Omphalocele (exomphalos)
  - ✓ Pentalogy of Cantrell
    - upper mid-line omphalocele, anterior diaphragmatic hernia, sternal cleft, pericardial defect and intracardiac defects
- Ruptured omphalocele(!??)
- Gastroschisis
- Body-stalk anomaly (BSA)
- Limb-body wall complex (LBWC)
- Amniotic band syndrome
- Bladder or cloacal extrophy
- Ectopia Cordis

Anterior abdominal wall defects: D/D

Defect	Herniated viscera	Herniation site	Umbilical cord	Amniotic membrane	Celomic space	Fetal mobility	Spine	
Exomphalos	Liver, bowel	Base of umbilical cord	Free-floating	Continuous, fused with chorion	Obliterated	Normal	Normal/ Kyphoscoliosis	
Gastroschisis	Bowel	Amniotic cavity	Free-floating	Continuous, fused with chorion	Obliterated	Normal	Normal	
Pentalogy of Cantrell	Heart, liver, bowel	Amniotic cavity	Free-floating	Continuous, fused with chorion	Obliterated	Normal	Normal	
Cloacal extrophy/OEIS complex	Cloaca	Amniotic cavity	Free-floating	Continuous, fused with chorion	Obliterated	Normal	Normal/ Kyphoscoliosis	
Body stalk anomaly	Liver, bowel	Celomic cavity	Absent	Interrupted at the level of herniated abdominal organs	Contains abdominal organs	Stuck through abdominal-placental attachment	Kyphoscoliosis	
Abdominoschisis with amniotic bands	Liver, bowel	Amniotic cavity	Free-floating	Ruptured	Obliterated	Normal	Normal	

Body-stalk anomaly (BSA) at 12 wks

Body-stalk anomaly: extraamniotic viscera

Body-stalk anomaly

(?) Type of defect at 12 wks

Gastroschisis

D/D???

(?) Gastroschisis

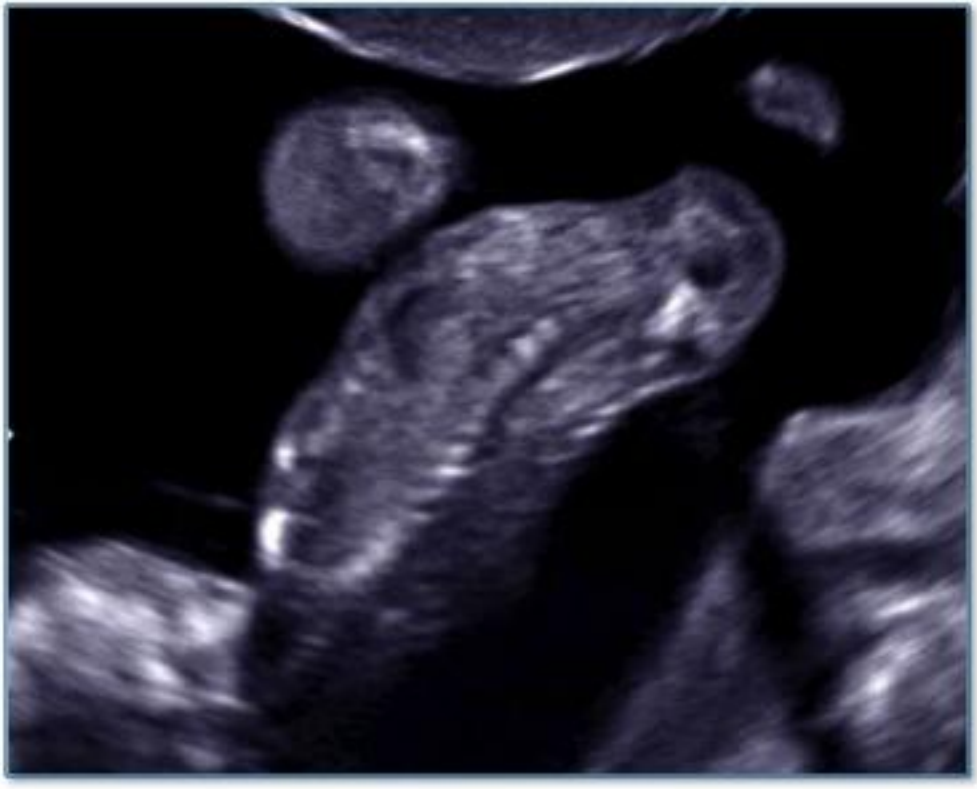
Omphalocele

D/D

Gastroschisis

Omphalocele

Omphalocele (exomphalos) with liver



Exomphalos with liver at 12 wks

TAS before CVS

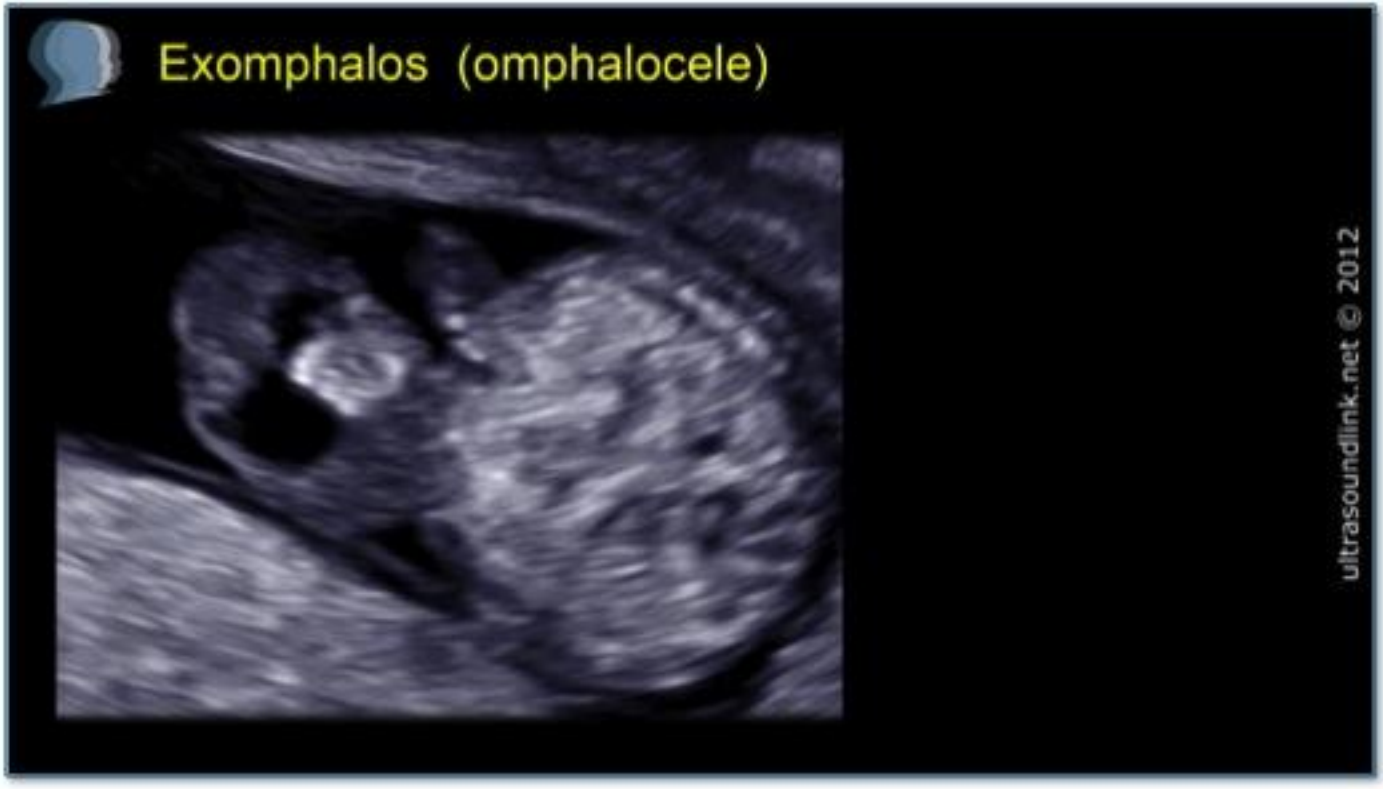
Exomphalos: liver + bowel

Suspicious great arteries

Omphalocele with or without liver at 12 wks (?)

Exomphalos: liver or bowel ?

Omphalocele bowel only

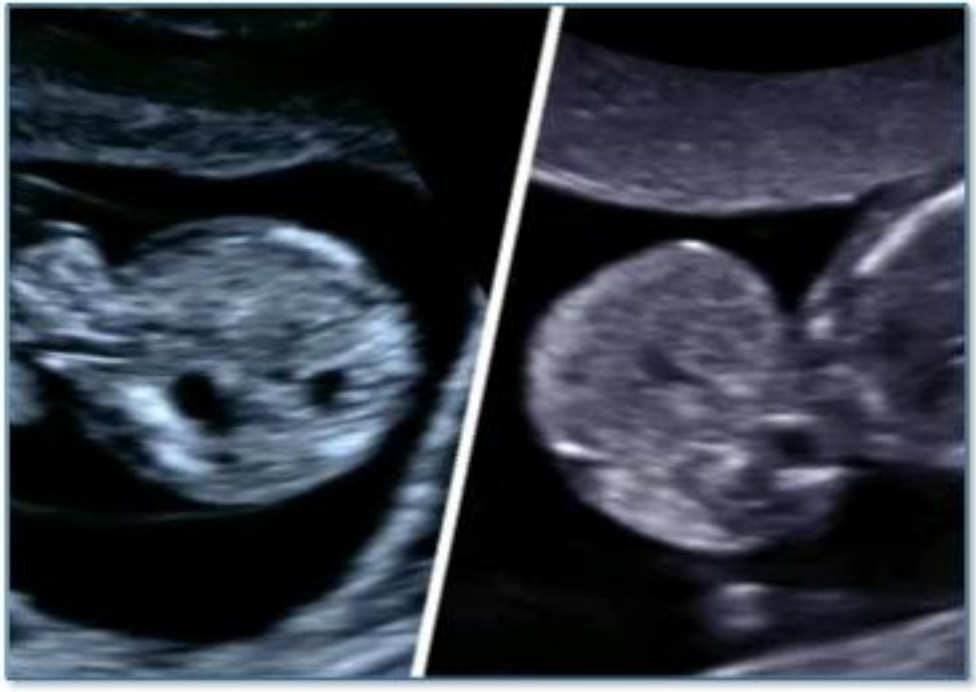


Omphalocele – 13 wk

Different omphalocele

Bowel

Liver



Exomphalos with liver at 13 wks; normal PCR

Small narrow chest

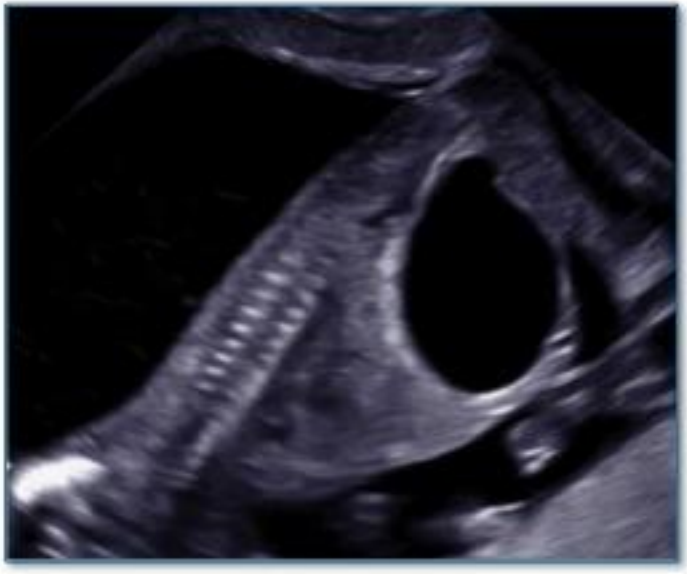
1/3 will die from pulmonary complications

Megacystis at 12 wks

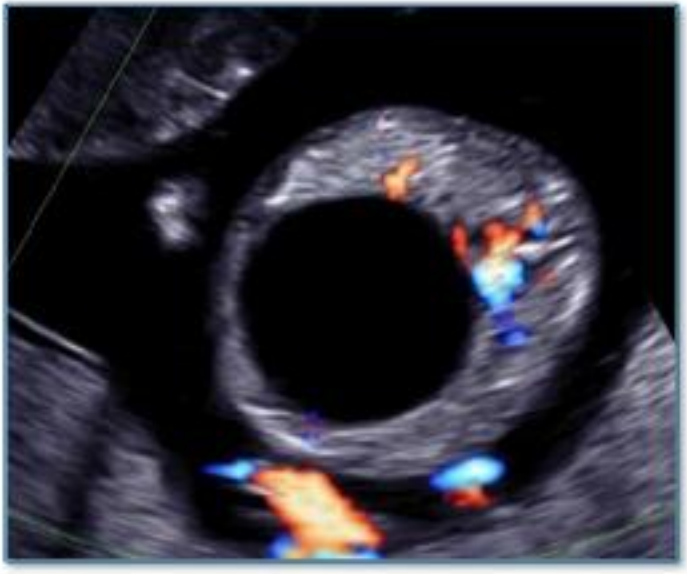
Spontaneous resolution

IUD after CVS

Massive megacystis: follow-up for low urinary tract obstruction (LUTO)



?Keyhole sign



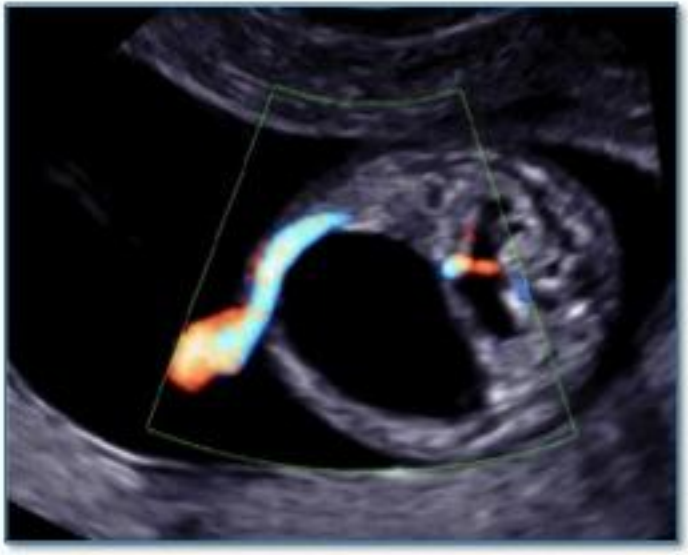
Two umbilical arteries

Megacystis (?) at 13 wks → To perform TVS!!!

TAS

Horseshoe kidney + hydronephrosis at 13 wks

Development of oligohydramnios at 16 wks



Single umbilical artery at 13 wks

Horseshoe kidney at 16 wks

Arms/hands defects in chromosomal anomalies

Aneuploidy	Typical defects
Trisomy 21	Normal hands
Trisomy 18	**Radial aplasia, poor visible/overlapping fingers**
Trisomy 13	**Postaxial polydactyly**
45X0	Normal hands
Triploidy	**Syndactyly**

Polydactyly → easier to detect in 1st than in 2nd trimester

3D rendering is very useful

Polydactyly → easier to detect in 1st than in 2nd trimester

Main question: isolated or not?

30-40 genetically well known syndromes with postaxial polydactyly

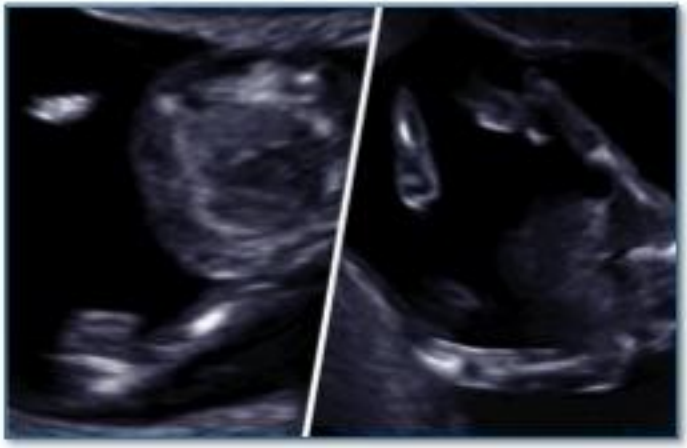
- **Trisomy 13**
- **Ciliopathies:**
  - ✓ **Meckel-Gruber**
  - ✓ **Ellis Van Creveld**
  - ✓ **Short-rib polydactyly**
  - ✓ **Bardet-Biedl**
  - ✓ **McKusick-Kaufman**
- **Smith-Lemli-Opits ,ets**

Overlapping/poor visible fingers

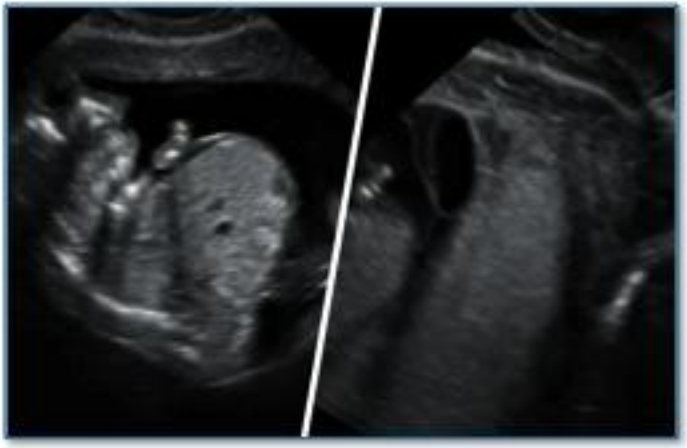
Associated with trisomy 18

Radial aplasia

- Usually part of aneuploidy, syndrome or association
- Isolated unilateral radial lesions have good prognosis



Trisomy 18 & TAR syndrome



(!) Amniotic bands

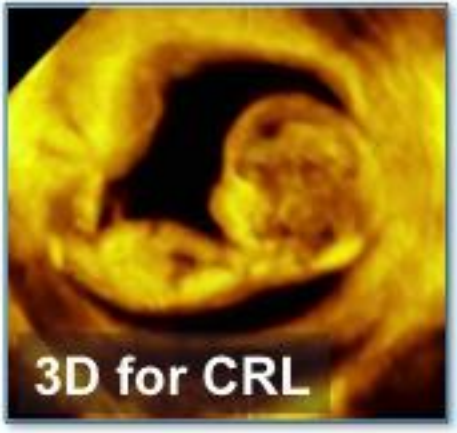
12(14) weeks fetus

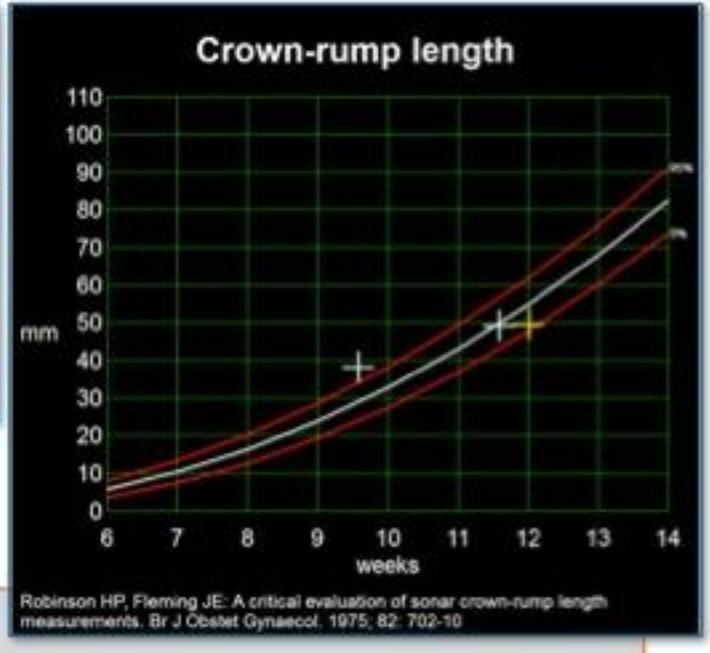
Not possible to measure CRL

TAS

TVS

Early 11-13 wks FGR → (?) Diagnosis





Possible triploidy:

1. Early pregnancy anomaly scan
2. Levels of PAPP-A & hCG
3. CVS (karyotype!, not microarrays)

Take home message

Early pregnancy anomaly scan (EPAS): 11-13 wk

- Examination of anatomy at 11-13 wks is essential part of the scan
- Majority of severe anomalies are detectable
- TAS & TVS are adjunctive approaches providing different information
  - ✓ TAS better for heart & screening
  - ✓ TVS for brain & expert examination
- TVS is essential for CRL < 55 mm