CARDIAC FINDINGS ASSOCIATED WITH DOWN SYNDROME MANAGEMENT OF THE FETUS AFTER DIAGNOSIS

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How and when is a Down syndrome fetus diagnosed?

Screening for Down syndrome in pregnancy

- Combined screening:
  - 1st trimester 10 weeks + 0 days to 14 weeks + 1
- Quadruple testing:
  - Second trimester 14+0 to 20+0
- Privately available NIPD

Cardiac defects: Timing and Patient Group

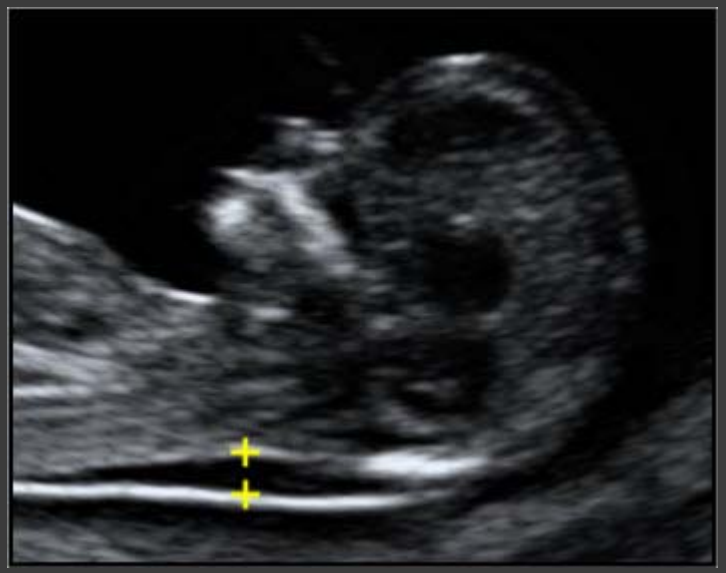
- Women with a high risk screen who have declined invasive testing
- Women who have declined screening
- Women who have a diagnosis after screening and are continuing pregnancy
- Women with a low risk screen
- Women presenting too late for screening

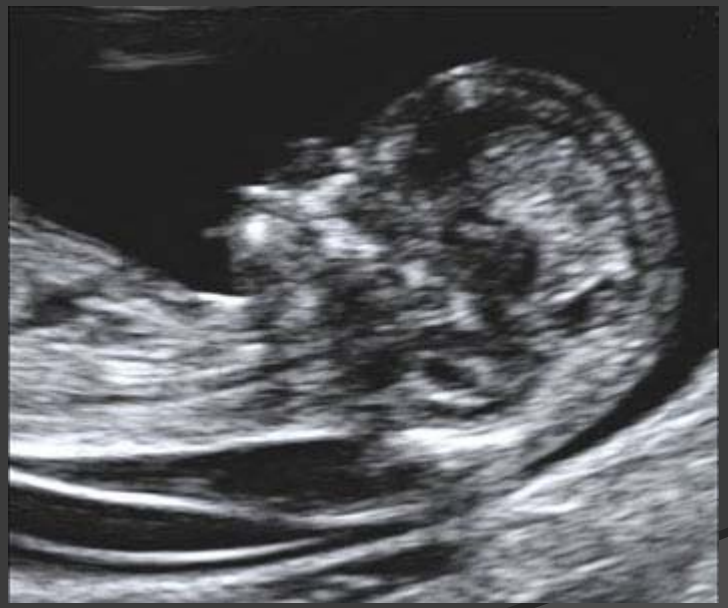
Timing and diagnosis

- First trimester: FASP recommend referral to tertiary centre if NT ≥ 3.5mm
- Many have a scan at 16 weeks' if raised NT (>3.5mm)
- Detailed anomaly scanning at 18-21 weeks'

Combined Screening: Ultrasound component

- Crown rump length (45-84mm)
- Nuchal translucency





Interesting facts:

- AVSD is more common in female fetuses with Down syndrome
- There is no difference in NT in Down Fetuses with or without CHD
- Fetuses with Down syndrome with functional defects have greater NT measurements

First Trimester echocardiography

- Can be performed for
  - Raised NT
  - Previous affected baby
- 4-chamber view
- Outflow tracts
- Mostly can be done transabdominally
- up to 75% detection rate in skilled hands (and thin women)

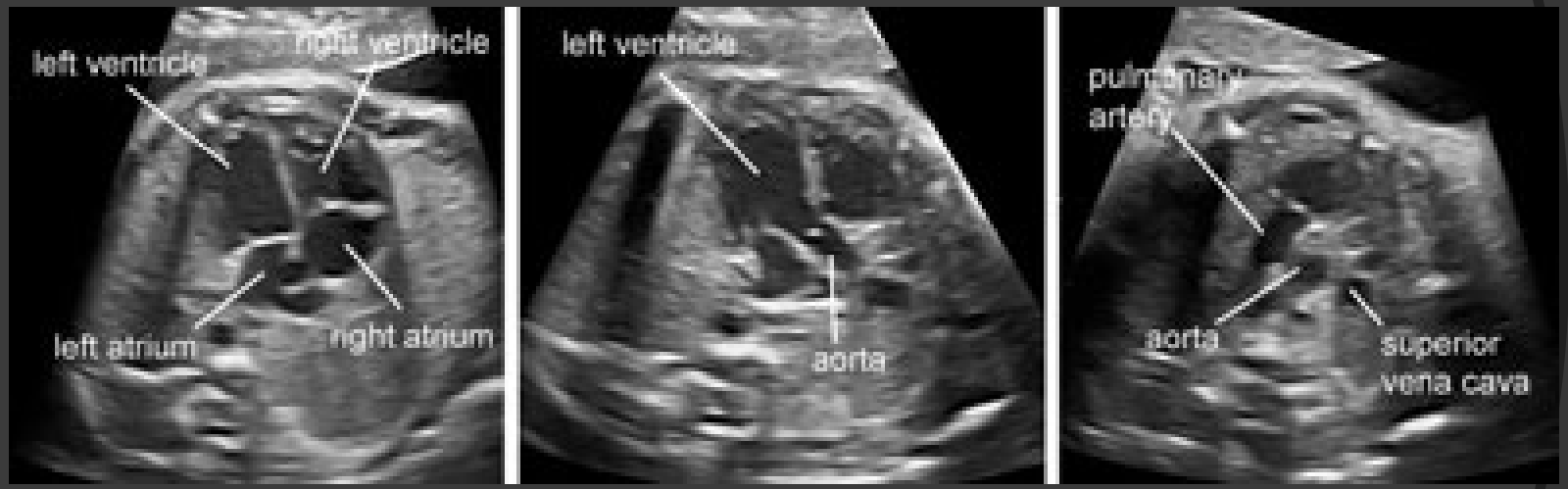
Second trimester Ultrasound screening

- 4-chamber view
- Outflow tracts
- Extended views (three vessel and trachea views)
- Ultrasound may not detect small VSD's or ASD's
  - Equal pressures
  - Presence of the foramen ovale
- Ductus is open in-utero

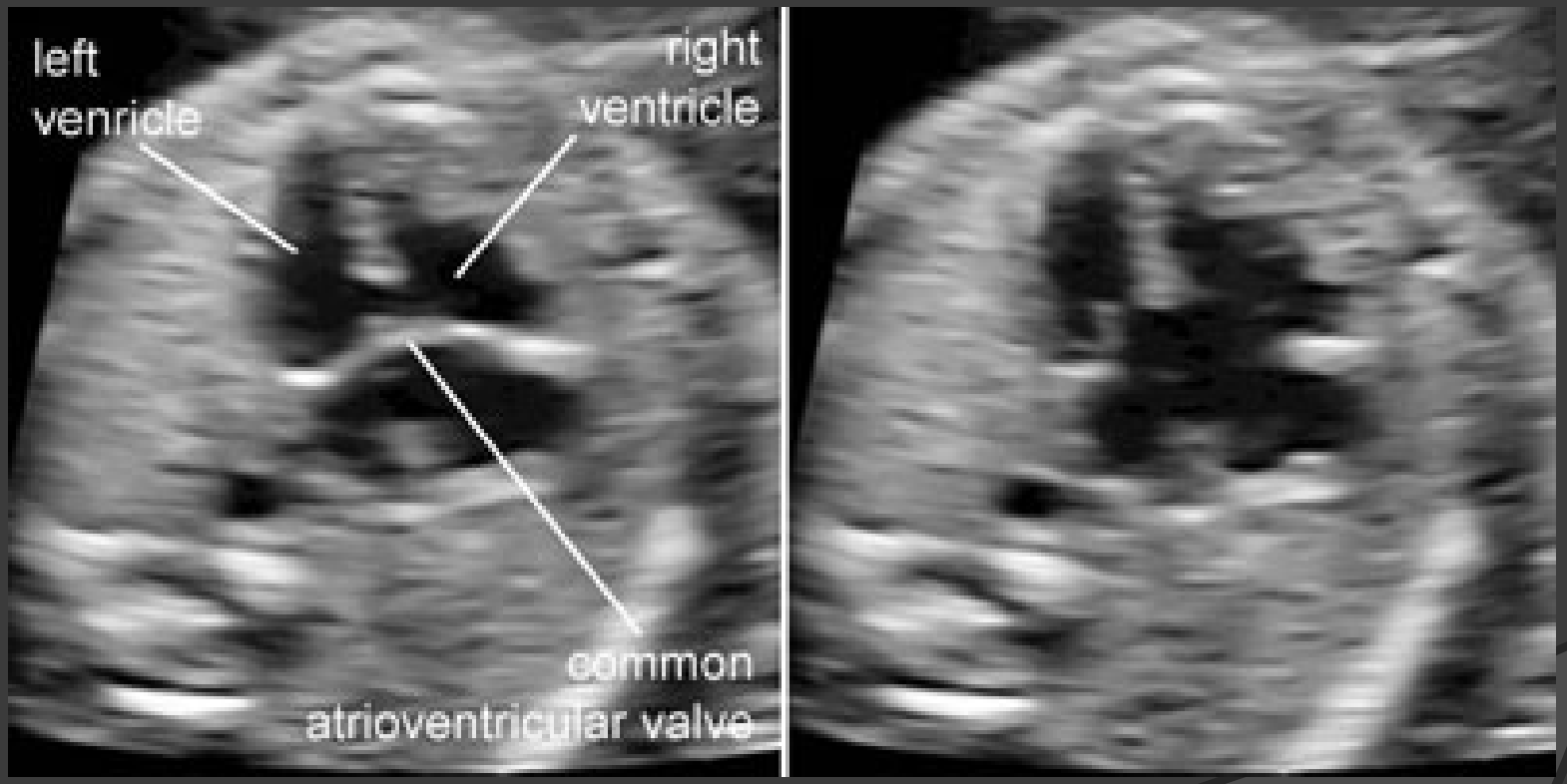
Commoner Cardiac diagnoses (33% in utero 40-60% postnatally)

- AVSD
  - Whole spectrum of AVSD including just absence of offsetting of AV valves
- VSD
  - Usually moderate or large as small not seen
- ASD
  - Hard to confirm in-utero
- Tetralogy of Fallot sometimes with a concurrent AVSD
- Aberrant right subclavian artery
- Postnatally: patent ductus arteriosus

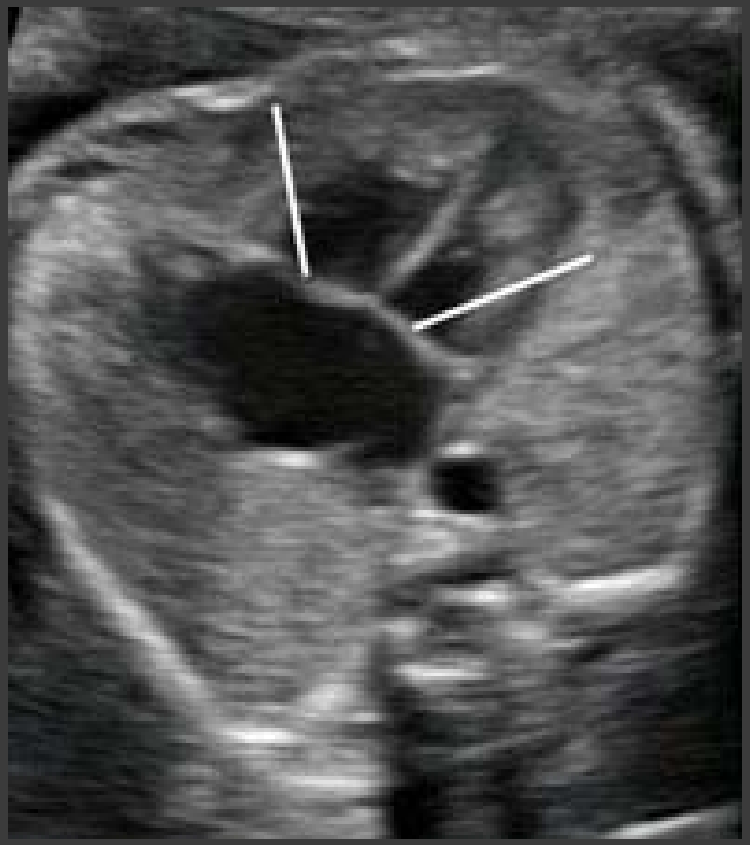
Normal Heart



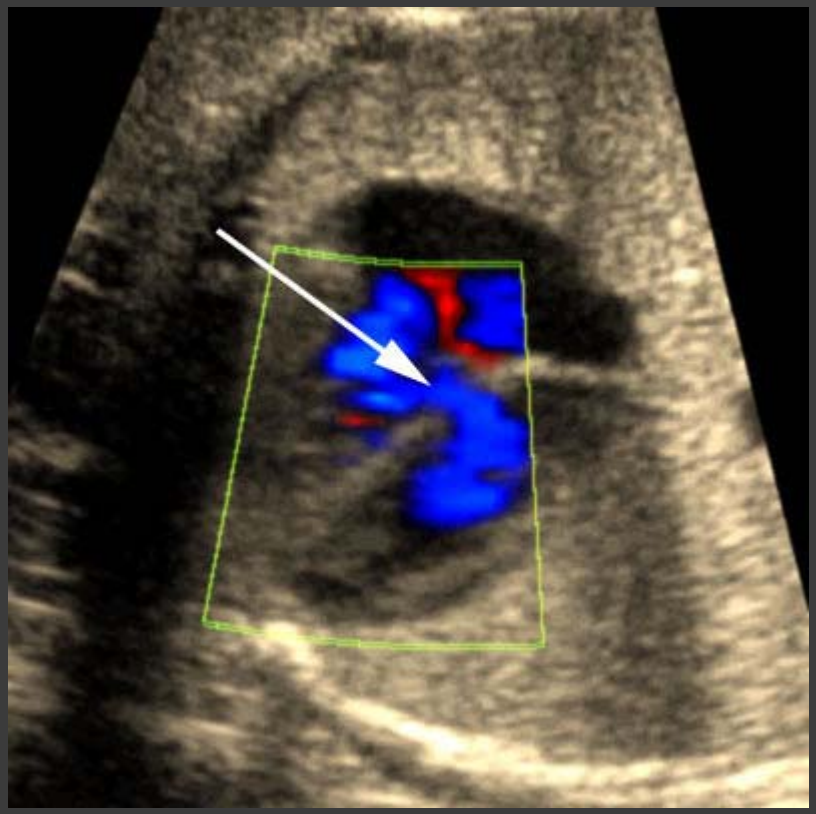
Complete AVSD



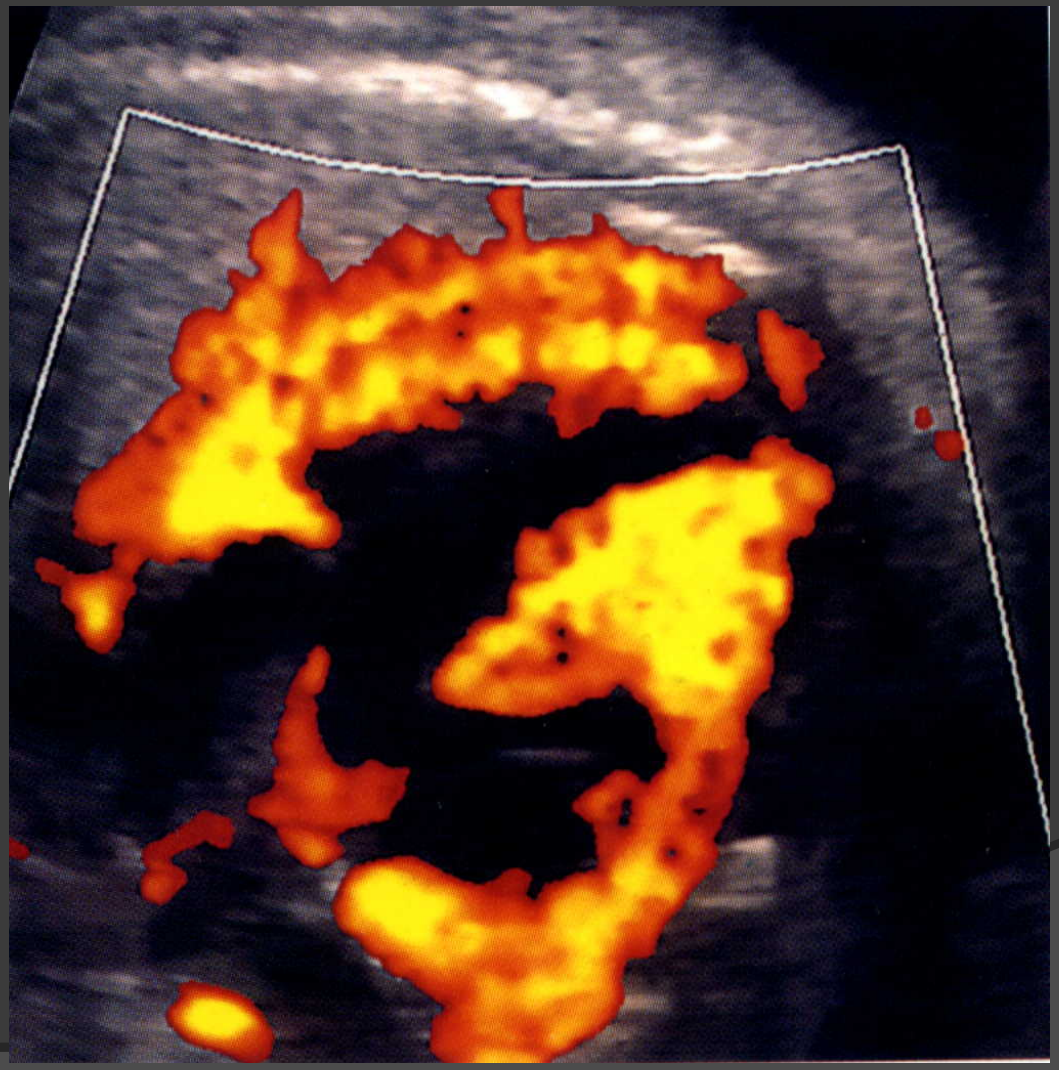
Partial AVSD



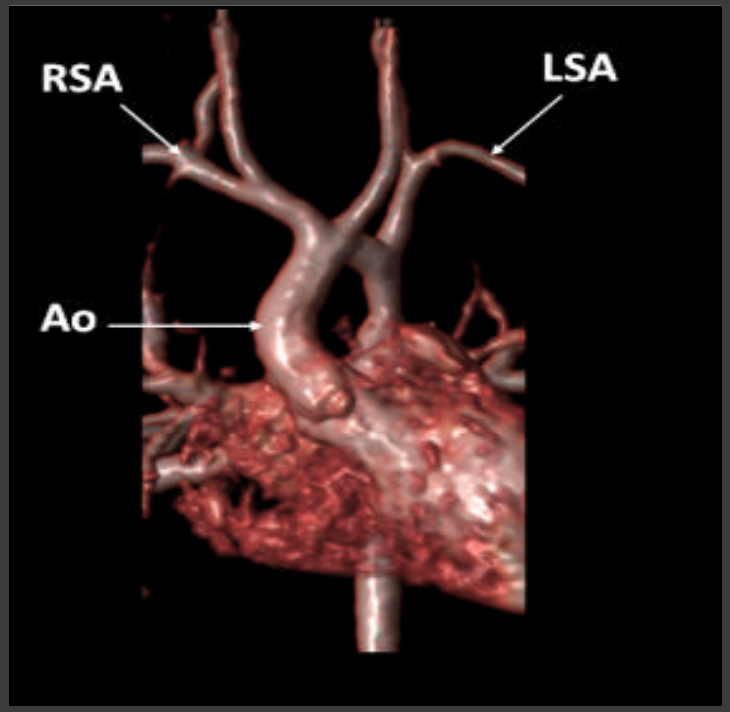
Ventricular Septal defects

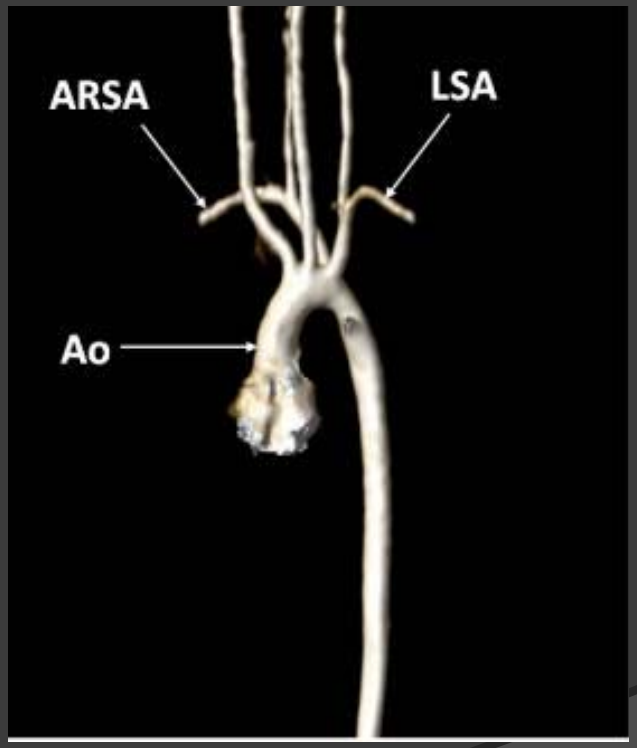


VSD on Colour flow mapping



Aberrant Right Subclavian Artery





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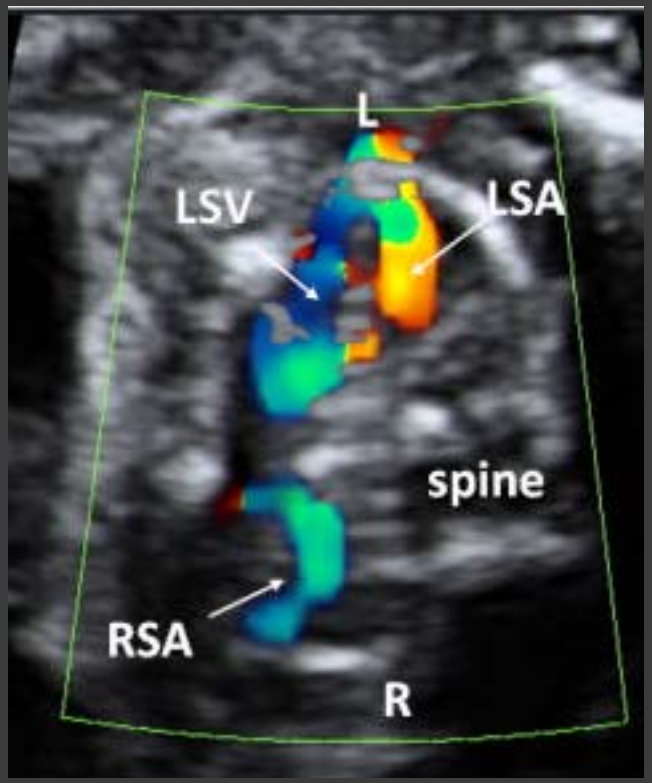
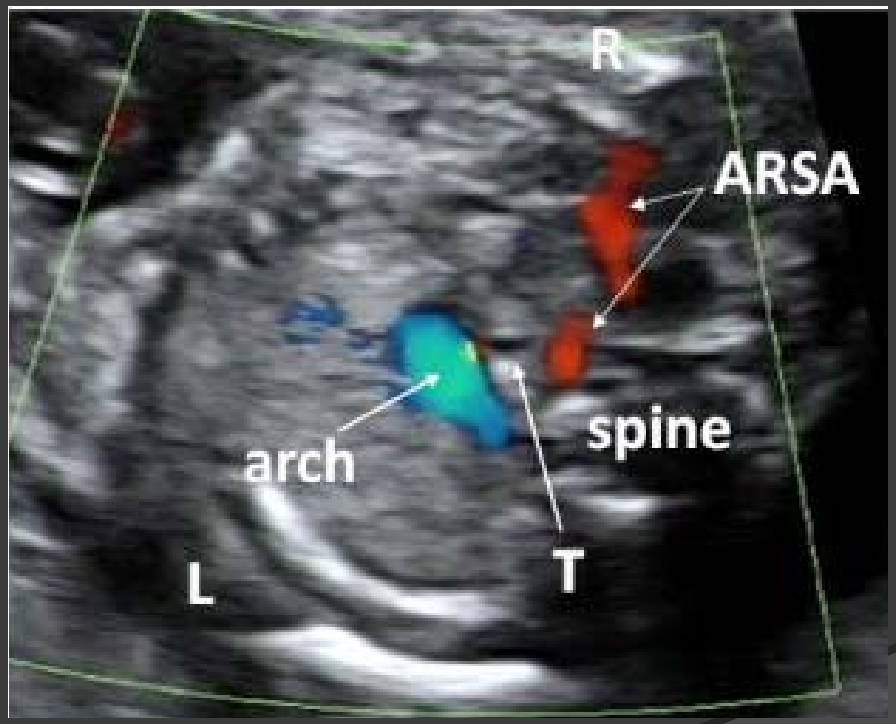




Rarer defects

- Coarctation of aorta
- Tricuspid dysplasia
- Ebstein's anomaly
- Double inlet left ventricle
- Persistent left superior vena cava

Functional heart abnormalities on late first trimester scanning (37%)

- Tricuspid regurgitation +/- mitral regurgitation +/- ventricular disproportion
- Isolated mitral regurgitation
- Isolated ventricular disproportion

Diagnostic testing options

- Chorionic villus sampling from 11 weeks
- Amniocentesis from 15 weeks
  - Both CVS and Amnio can be done any time up to birth
  - Late diagnosis can present issues regarding termination of pregnancy
- NIPD can exclude T21/13/18
  - Advantages:
    - Avoids risk and offers accurate screening for major aneuploidies

Management of pregnancy following a diagnosis of Down Syndrome

- Surveillance during pregnancy
- Planning and preparation for birth

Surveillance

- Structural anomalies
- Hydrops
- Growth
- The reasons why more Down syndrome fetuses are lost is unclear

Structural anomalies

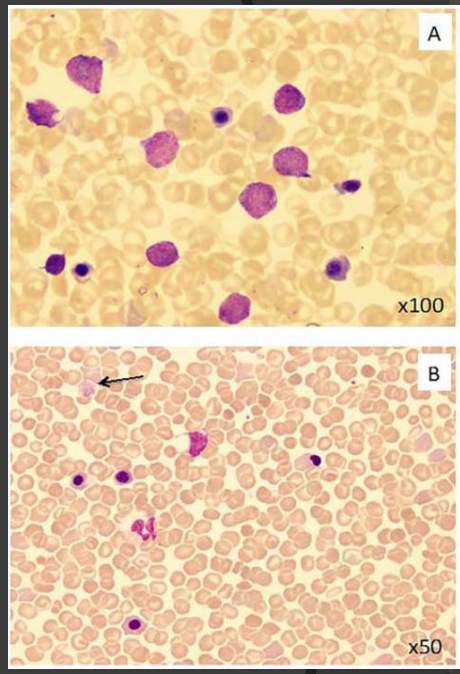
- Heart
  - Rarely severe enough to cause problems in-utero
- Renal
  - Surveillance for obstructive uropathy
- Duodenal atresia
  - Polyhydramnios management
    - Reducing the risk of prematurity

Hydrops

- Consider whether secondary to Transient Abnormal Myelopoiesis
- MCA Dopplers can help determine whether anaemia is underlying cause
- Therapies have included transfusion and drainage of pericardial effusions
- TAM may lead to fetal death but can spontaneously resolve

Transient Abnormal Myelopoiesis

- Can affect 10% fetuses
- Acquired GATA-1 mutation in blasts
  - key haematopoietic transcription factor associated with Down syndrome
- Usually suspected in third trimester
- Can resolve spontaneously



Common features of TAM in-utero

- Pericardial effusion
- Hepatomegaly/splenomegaly
- Ascites
- Pleural effusion
- Peripheral oedema
- Increased liquor volume
- Generalised hydrops

Prognosis for TAM in utero

- If generalised hydrops + hepatomegaly
  - >90% fetal/early neonatal demise
- Even without hydrops loss rates may be >50%
- Treatment has included transfusion and pericardial drainage

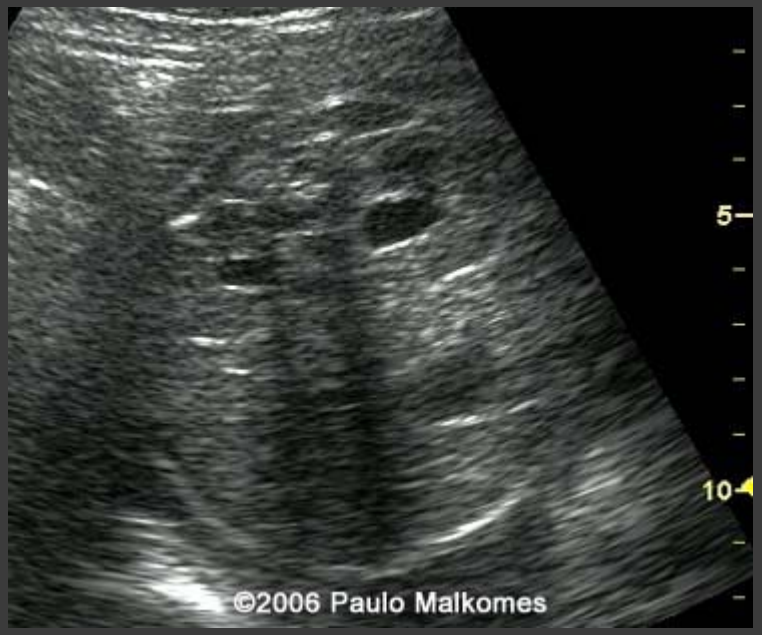
Growth surveillance

- Growth restriction is more common
  - Especially closer to term
  - Fetal loss rates are higher at all gestations
  - Declining fetal activity should prompt increased surveillance
- Where growth is tailing off surveillance in line with normal guidelines is required
  - Biometry
  - Umbilical artery Dopplers
  - MCA Dopplers
  - Venous Dopplers

Surveillance with regard to other abnormailties

- Renal pelvis dilatation (25%)
- Duodenal atresia (8%)
- Diaphragmatic hernia
- Cerebral ventriculomegaly (5%)

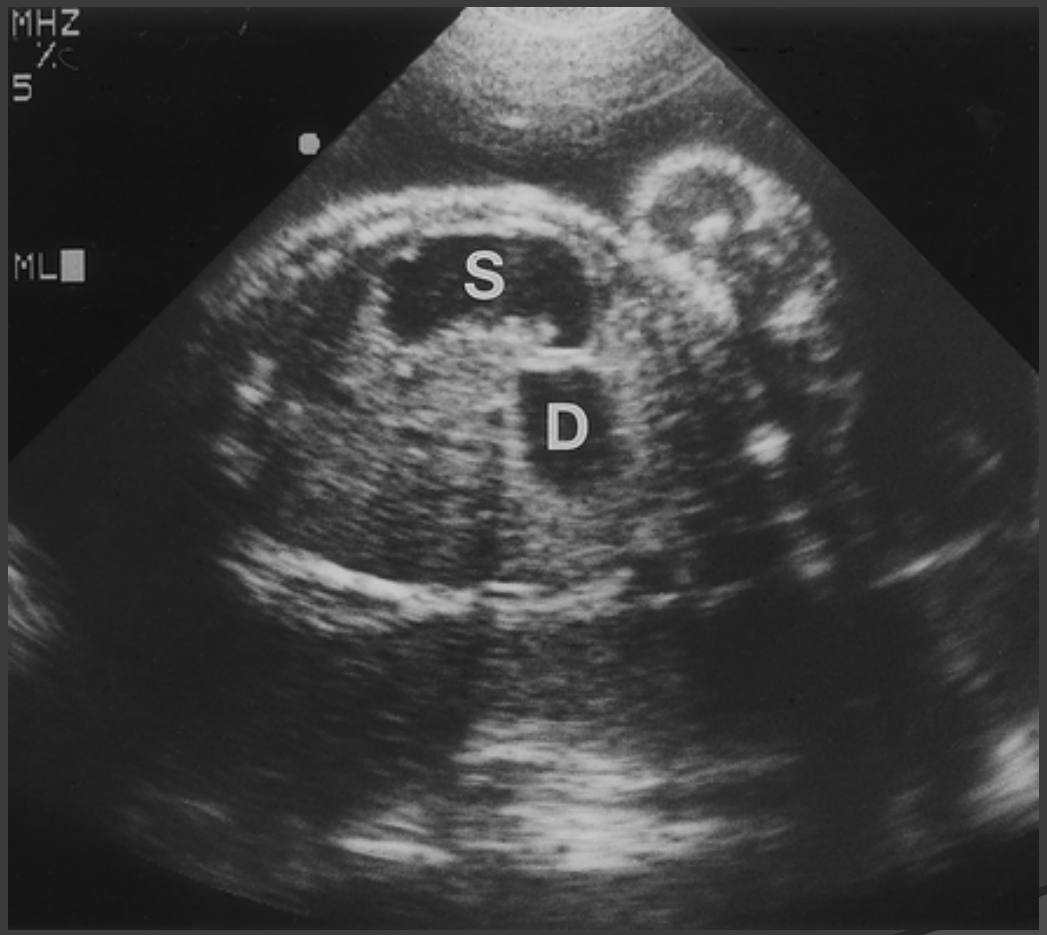
Mild renal pelvis dilatation



Mild renal pelvis dilatation

- Affects about 25% Down syndrome fetuses
- Can spontaneously regress
- Should prompt postnatal imaging along standard guidelines

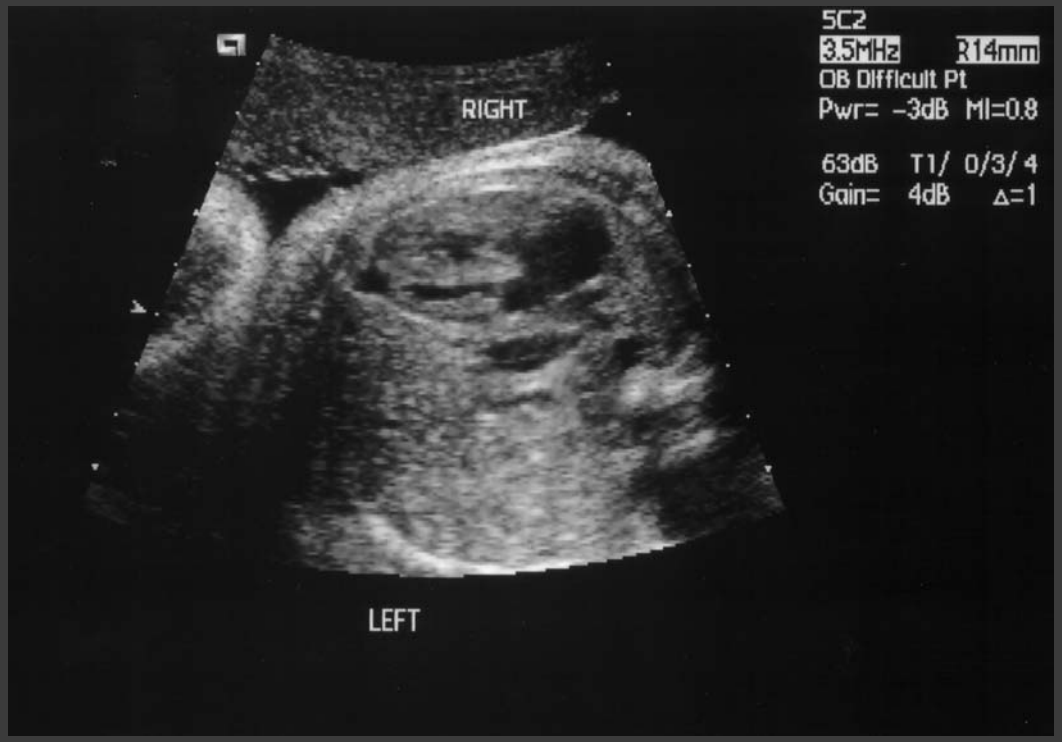
Duodenal atresia



Duodenal/jejunal atresia: main Problems

- Polyhdramnios
- Preterm labour
- Growth restriction

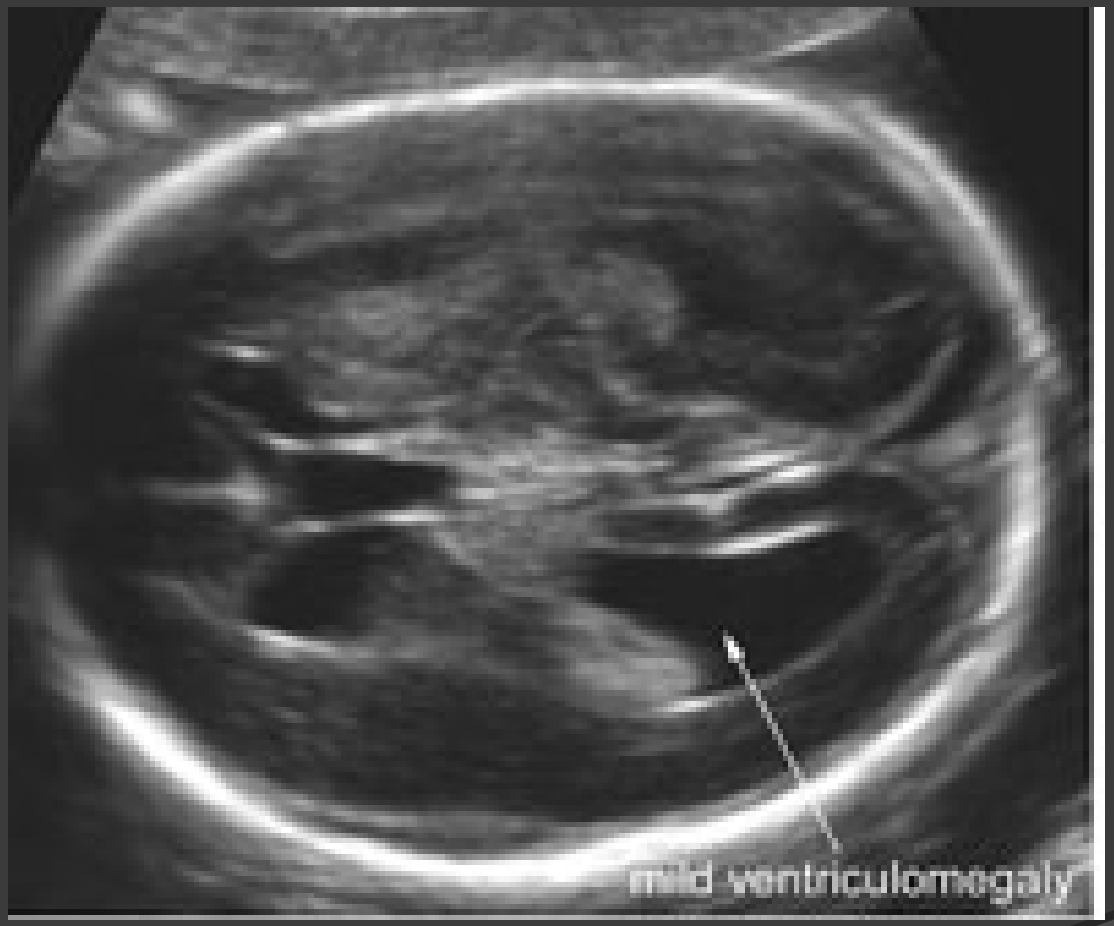
Diaphragmatic hernia



Diaphragmatic hernia: Main problems

- Polyhydramnios
- Preterm labour

Mild Ventriculomegaly



Birth Planning

- Place of birth
- Method of delivery
  - Dependent on growth, presentation, obstetric history
- Surveillance in labour
- Post birth plan for anomalies
- Post birth planning for follow-up and support

Holistic Approach

- Care of the fetus
- Planning for birth
- Care of the parents

Thank you

- Any questions?