Antenatal diagnosis of renal tract abnormalities and what I tell my patients

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Referral groups

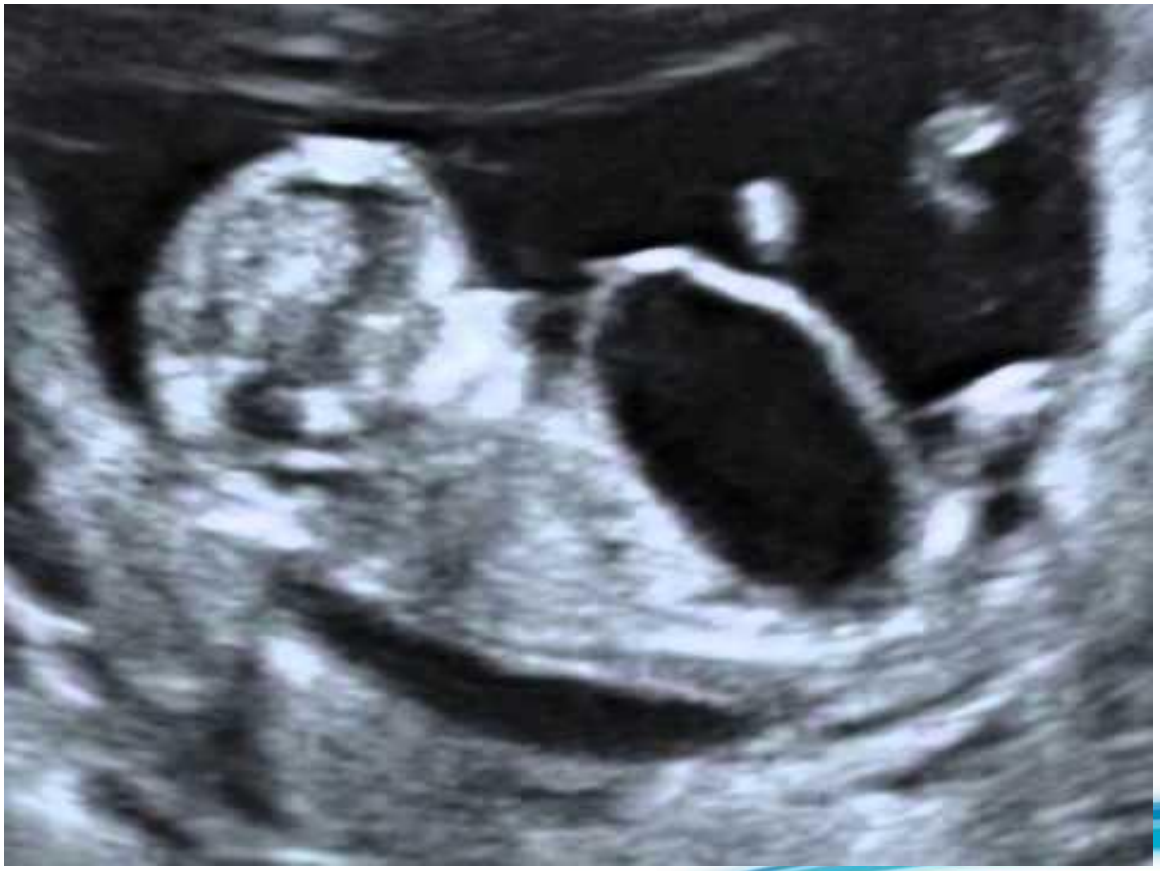
- Previously affected pregnancy
  - Almost anything!
- Family history
  - Dysplasia
  - Cystic disease
  - Sometimes severe reflux
- Scan findings (largest group)
  - Can be anything at any gestation from 11 weeks when the first scan is usually performed

What is visible and when?

- Fetal kidneys begin to function after 12 weeks
- Bladder usually visible from 12 weeks and major obstruction can be visible at this stage
- By 14 weeks urine output takes over amniotic fluid production
- By 16 weeks urine output is such that upper renal tract obstruction can begin to cause problems and be visible
- Nephrogenesis continues to term, so dysplasia can worsen during fetal life
- Posterior urethral valves can present late

12-14 weeks

- Bladder outflow obstruction/megacystis



Lower urinary tract obstruction

- Associations
  - Associated anomalies are common and include:
    - posterior urethral valves
    - Urethral agenesis
    - chromosomal anomalies
- At 10-14 weeks
    - if the longitudinal bladder diameter is 7-15 mm risk of chromosomal defects ~25%

- microcolon intestinal hypoperistalsis (MMIH) syndrome (Berdon syndrome)
- megacystis megaureter syndrome
- prune belly syndrome

Treatment and prognosis: What do I tell patients?

- A karyotype should be considered (CVS)
- Prognosis can be variable. It can completely resolve or lead to progressive obstruction
- A follow-up ultrasound is necessary
- If the fetus is chromosomally normal
  - spontaneous resolution in about 90% if he bladder diameter is 7-15 mm
  - if the bladder diameter is >15 mm there is a very high likelihood of progressive obstructive uropathy
- Management will depend on the whole clinical picture
  - Liquor volume
  - Other findings
- Vesicoamniotic shunting may improve survival in severe cases, but survival with normal renal function is rare.

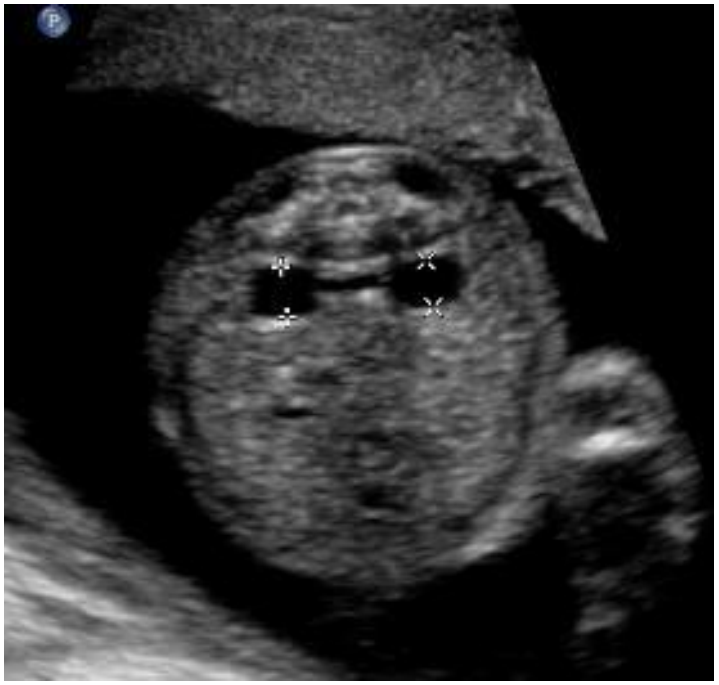
Second Trimester Diagnoses

- Renal pelvis dilatation
- Cystic kidney disease
- Renal agenesis

Renal pelvis dilatation

- Common
- Mild/moderate 7-15mm
- Severe >15mm
- Look for cortical changes

- Often resolves
- Commoner in boys
- Must always look for other anomalies (VATER etc)
- Can be caused by
  - Obstruction
  - Reflux
  - Other kidney problems such as duplex/horseshoe kidney

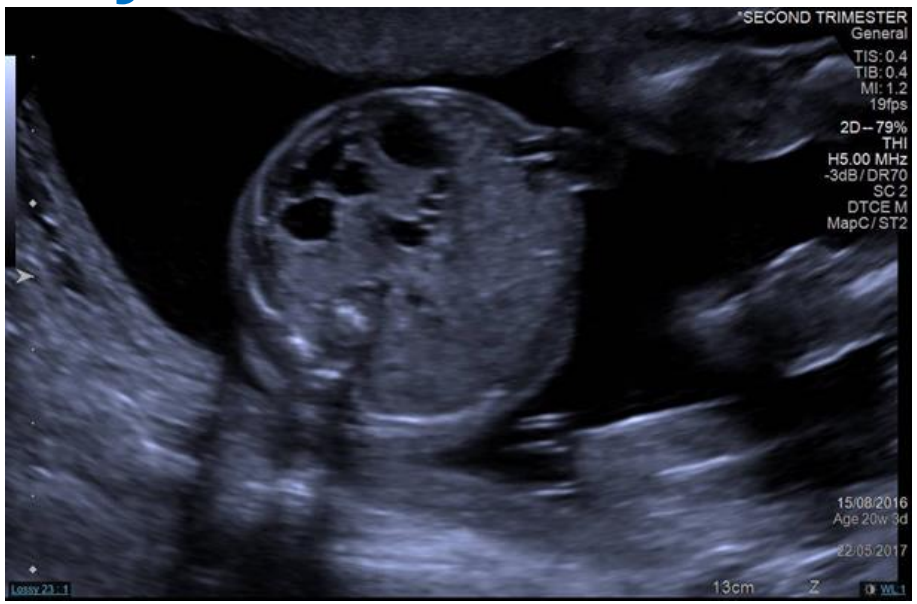


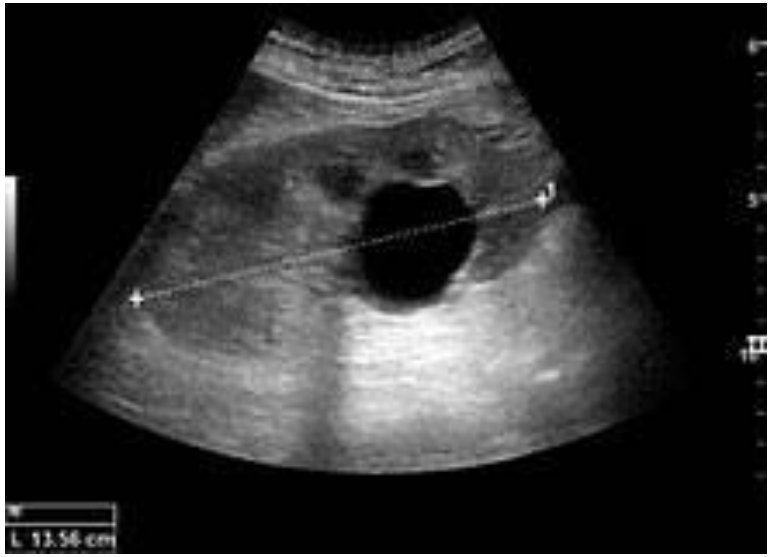
Renal Pelvis dilatation: what I tell the parents

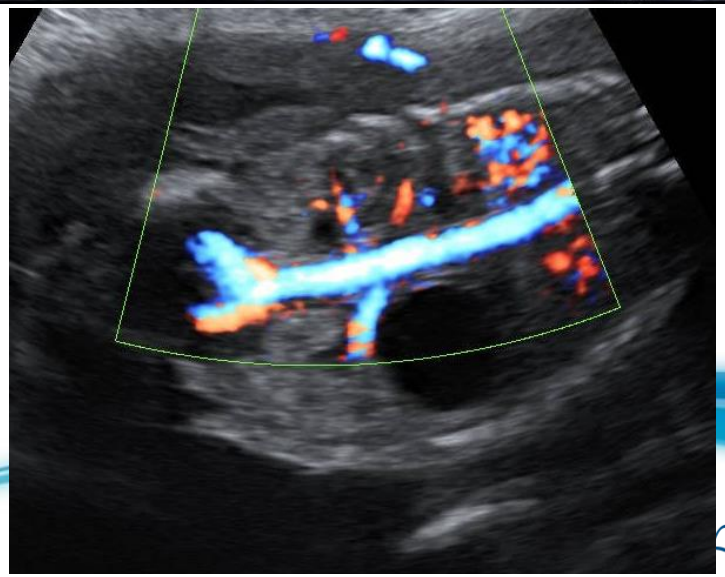
– Plan for pregnancy
  - Watch for progression (generally doesn't)
  - Can resolve
  - Watch liquor volume
  - Doesn't alter timing of or method of delivery
  - Postnatal plan for ultrasound at a few weeks
  - Refer severe cases or if evidence of cortical extension

Cystic kidney disease

– Large cysts
  - Single
  - Multiple
  - Sometimes not kidney







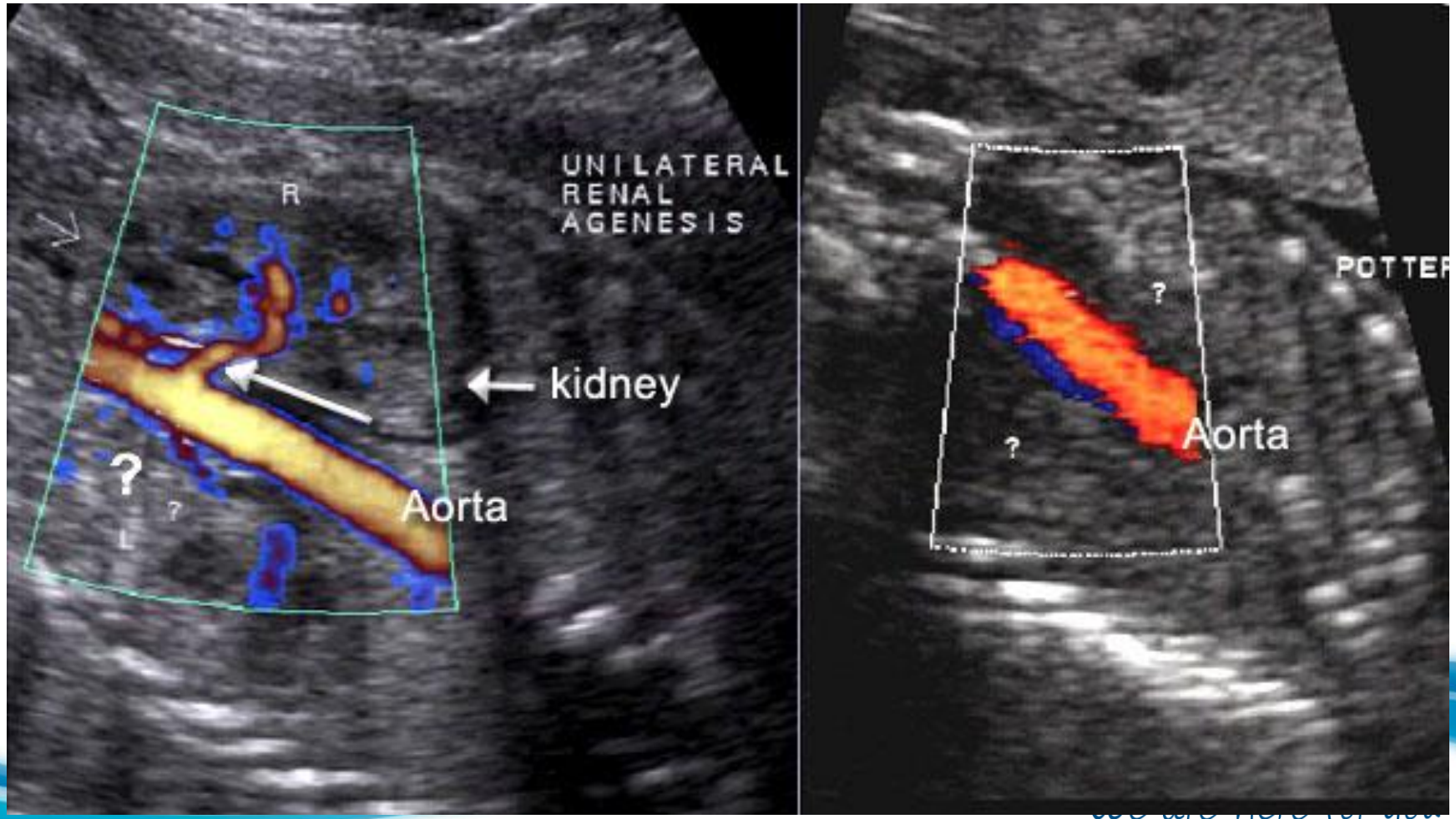
Renal cysts: what I tell the parents

- Single Cysts
  - Often resolve
  - Watch for compression
  - Not usually a problem
- Multiple cysts
  - Usually represent a multi-cystic kidney (though sometimes can be more than one simple cyst)
  - Usually that kidney will not work (gets bigger) though sometimes can get only part affected
  - Watch the other kidney
  - Assess for other abnormalities
  - Doesn't influence method or timing of delivery
  - Complex counselling if bilateral

Renal agenesis

- Uni or bilateral (beware a difficult to find kidney)
- Significant genetic influence
- Associated with a number of syndromes and chromosomal abnormalities
- Careful look for associated features
- Scan parents
- Sometimes MRI can help if a kidney is difficult to locate

Renal agenesis



Renal agenesis: what I tell the parents

– Single missing kidney
  - Not usually a problem as long as remaining kidney healthy
  - Watch for progress of single kidney (higher risk of dysplasia)
  - Sometimes associated with other problems
  - Can have a genetic element
  - Scan parents and refer to genetics if either has a renal anomaly

– Bilateral renal agenesis
  - Will not result in a baby that survives
  - Pulmonary hypoplasia is the immediate threat to life at birth
  - Scan parents (at some point)
  - Refer to genetics if either parent has a renal anomaly or if any other anatomical abnormality

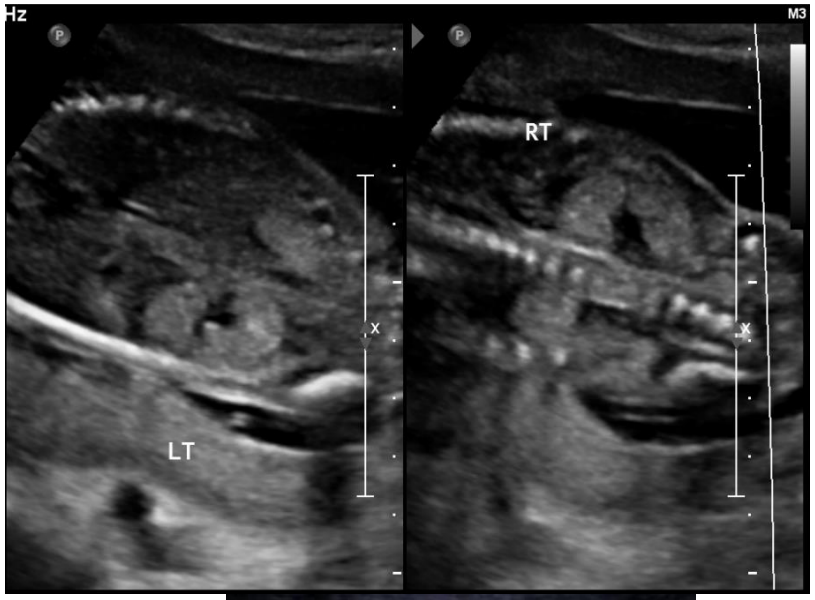
Bright/echogenic kidneys

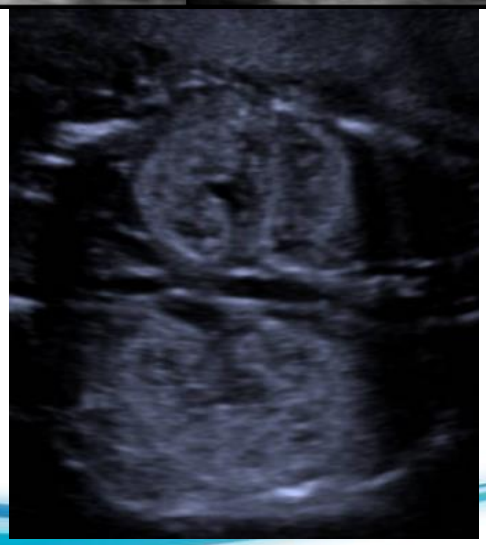
- Usually seen at anomaly scan
- Considerations:
  - Are the kidneys large?
  - Is the liquor volume normal?
  - Is there a family history of kidney problems?
  - Any illnesses during pregnancy?
  - Any other abnormalities?

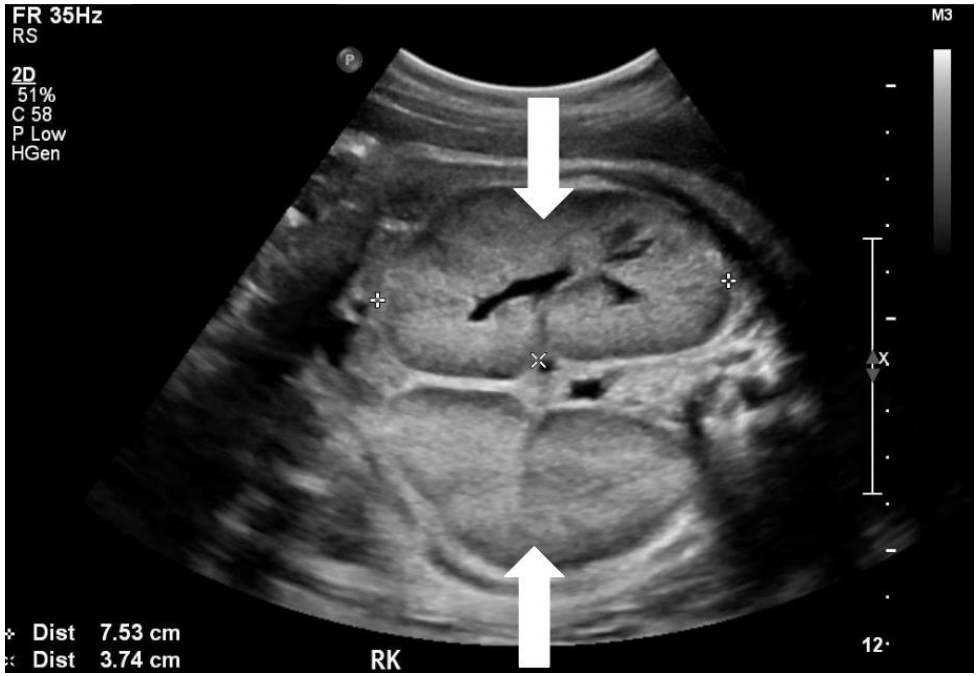
Bright Kidneys

- Physiologic variation
- Intrinsic renal disease
  - » Autosomal dominant polycystic renal disease
  - » Autosomal recessive polycystic renal disease
- Obstructive uropathy
- Aneuploidy
  - » Trisomy 13
  - » Trisomy 18
- Infection
  - » Cytomegalovirus
  - » Candida
- Overgrowth Syndromes
  - » Beckwith-Wiedemann
  - » Perlman
  - » Simpson-Golabi-Behmael
  - – Meckel-Gruber
  - – Bardet-Biedl syndrome
  - – Renal vein thrombosis
  - – Congenital nephrotic syndrome

Bright/echogenic kidneys





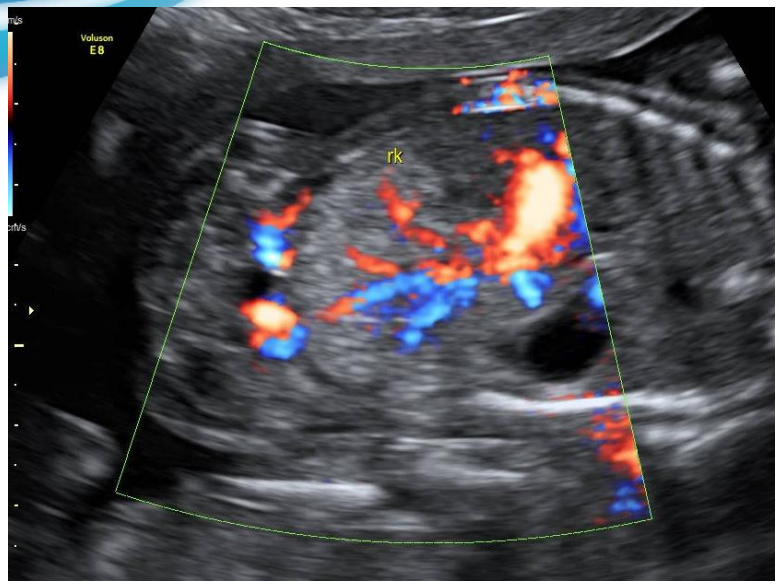


Bright kidneys: what I tell the parents

- Differential diagnosis is broad
  - Test for CMV
  - Karyotype if any other anomalies
  - Scan parents
  - Offer testing for ARPKD (quicker if CVS/amniocentesis but can test parents)
  - Outcome dependent on diagnosis (which can be difficult)
  - If ARPKD outcome usually poor (pulmonary hypoplasia, sometimes associated liver disease)

Familial Renal disease

- Usually referred because there is an affected family member
- Broad range of conditions
- Some will have visible features in-utero
- Some will have available PND
- Beware variable progression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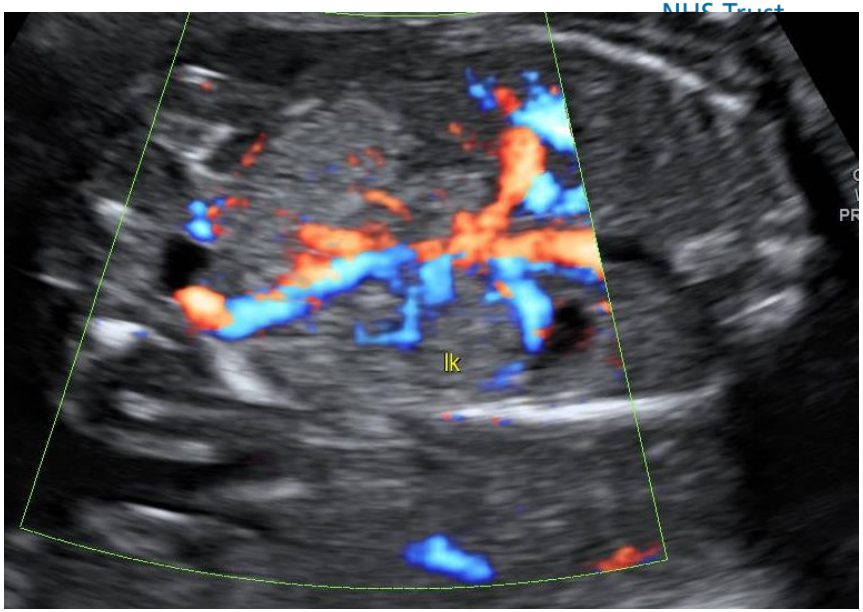
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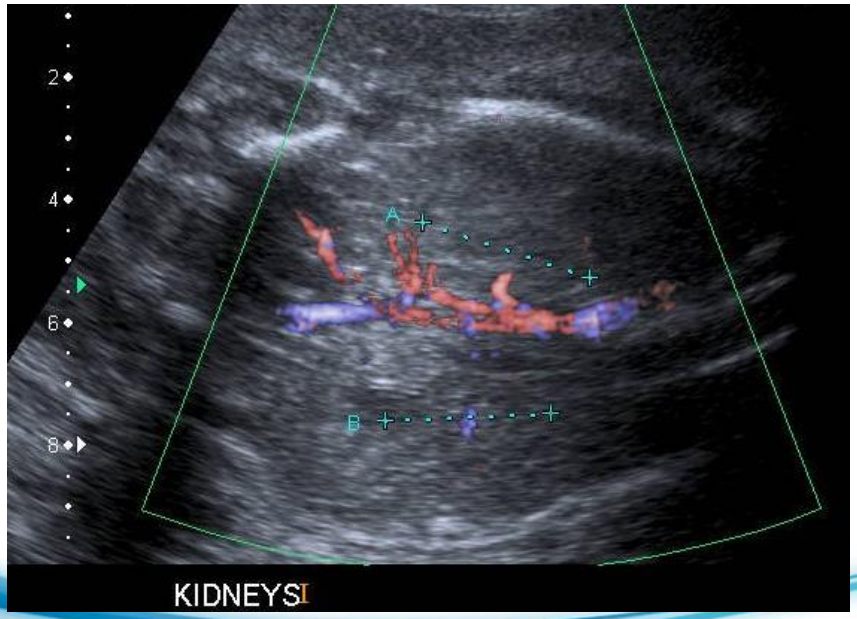
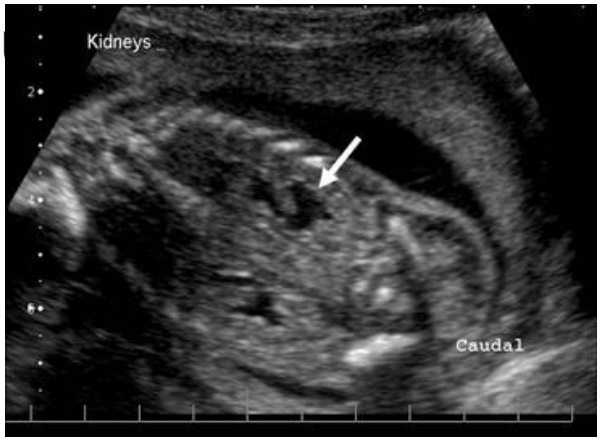
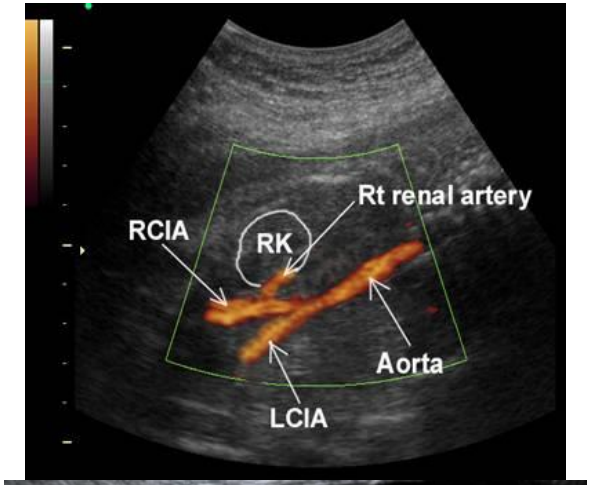
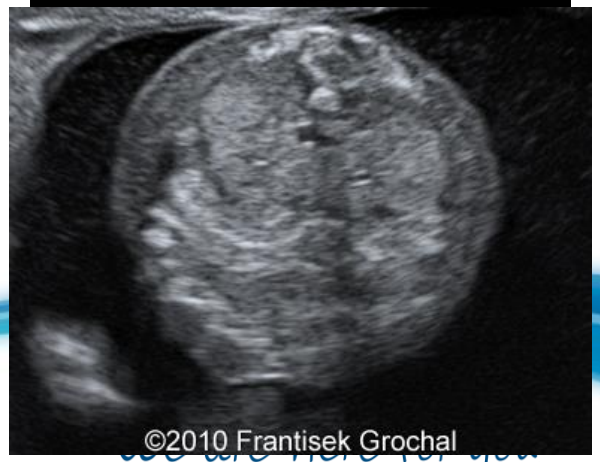

2
4
6
8
A
B
KIDNEYS

Other anomalies

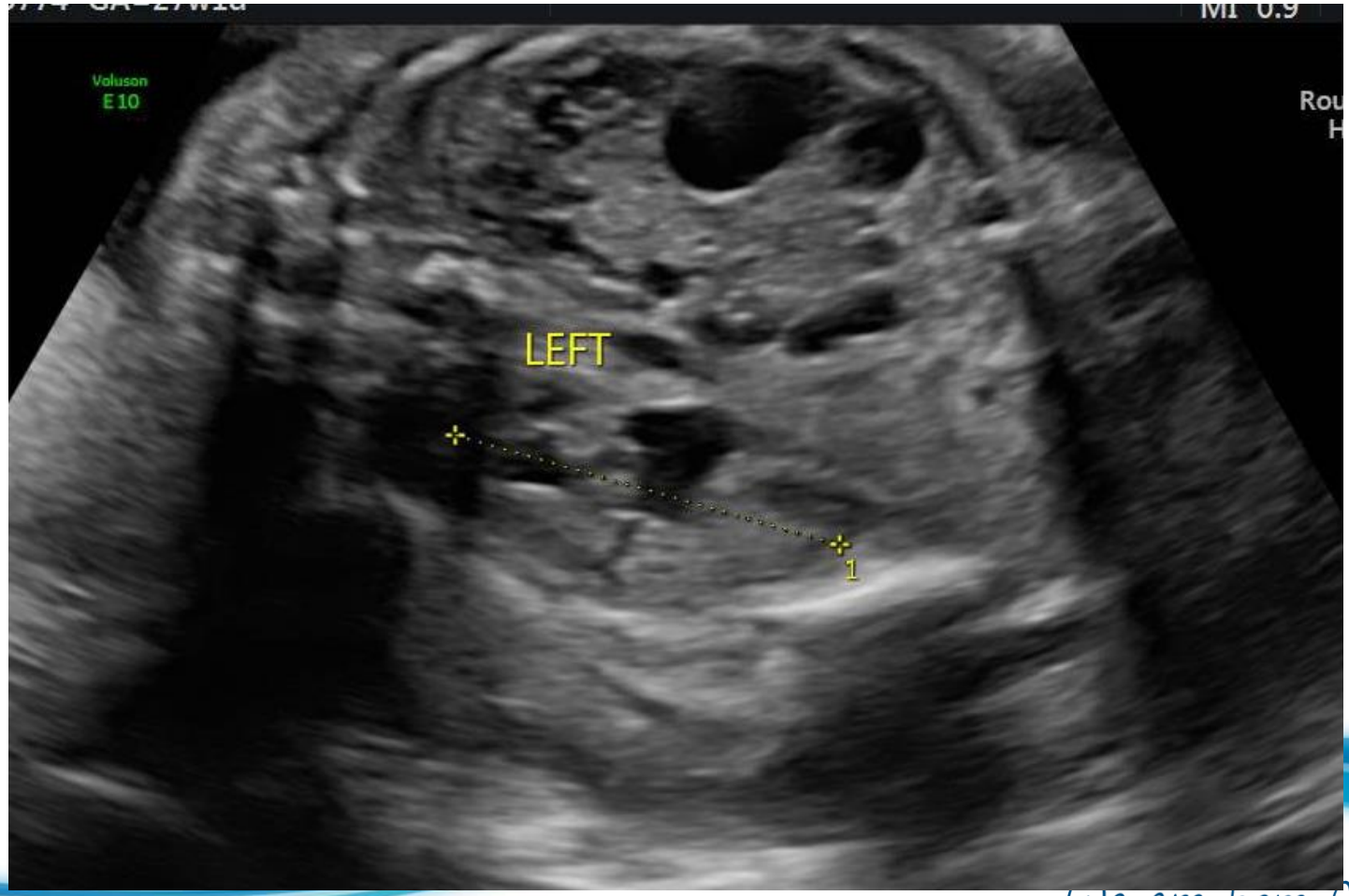
- Duplex
  - Usually seen
  - Sometimes seen with some obstruction
- Ectopic kidneys
  - Often when you can't see kidney in normal place.
- Horseshoe kidney
  - Often missed as view at hilum is usually normal
    - Can be associated with trisomies and monosomy X







More than one abnormality is not uncommon



Questions?