Overview

- Topics: echogenic intracardiac focus, choroid plexus cyst, echogenic bowel, renal pelviectasis, single umbilical artery. If time permits: ventriculomegaly, clubfoot, cleft lip
  - What it is
  - What it means (if anything)
  - What to do about it antenatally and postnatally
  - What to say to your patient
- Helpful websites for you and your patient

Goals

- Review 8 ultrasound findings: EIF, CPC, echogenic bowel, renal pelviectasis, SUA, ventriculomegaly, clubfoot, cleft lip
- Review briefly the implications of these findings and the workup recommended, if any
- Be able to discuss these findings with your patients either comprehensively or as a “bridge” to MFM consultation

Echogenic Intracardiac Focus
**Echogenic Intracardiac Focus**

- **What is it?**
  - An echogenic (bright) spot in the left cardiac ventricle
  - Correlates to mineralization (calcification) of the papillary muscle

- **What does it mean?**
  - Present in 3-4% of normal fetuses
  - Can be associated with trisomy 21: approximately 18% of fetuses with trisomy 21 have an EIF
  - Therefore, in a high-risk population, does increase the risk of chromosomal abnormalities
  - In a low-risk population, the increase in risk is usually not substantial and the finding is a “normal variant”

**What to do about it: Prenatal**

- Confirm the finding truly exists: false positives are possible
- Look for other markers or malformations associated with chromosomal abnormalities and review screening test results
- If other findings are seen, or if the patient is considered high-risk for chromosomal abnormalities, refer to genetics/prenatal diagnosis

**What to do about it: Postnatal**

- Nothing: of no significance

**What to say to your patient**

- EIF is an ultrasound finding that is usually normal and of no significance. It does not mean there is anything wrong with the heart and in fact, we do not recommend doing anything about it even after birth.
- In women who are at a higher risk for having a baby with trisomy 21 especially if other findings are seen on ultrasound, we do like to sit down and discuss what this finding may mean for them.

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**Choroid Plexus Cysts**

- [Image of ultrasound showing choroid plexus cysts]
Choroid Plexus Cysts

- **What is it?**
  - Cystic areas seen in the lateral ventricles of the brain within the choroid plexus. They can be unilateral, bilateral, single or multiple.

- **What does it mean?**
  - Present in 0.3-3.6% of normal fetuses
  - Can be associated with trisomy 18: approximately 30-50% of fetuses with trisomy 18 have CPC
  - Fetuses with trisomy 18 often have other findings on ultrasound. Isolated CPC therefore are more often seen with a normal fetus.

- **What to do about it: Prenatal**
  - Look for other markers or malformations associated with trisomy 18 (clenched fist, strawberry shaped skull, cardiac defect) and review screening test results
  - If other findings are seen, or if the patient is considered high-risk for trisomy 18, refer to genetics/prenatal diagnosis
  - Usually not seen after 23 weeks gestation

- **What to do about it: Postnatal**
  - Nothing: of no significance

What to say to your patient

- CPC is an ultrasound finding that is usually normal and of no significance. It does not mean there is anything wrong with the brain and in fact, we do not recommend doing anything about it even after birth.
- In women who are at a higher risk for having a baby with trisomy 18, especially if other findings are seen on ultrasound, we do like to sit down and discuss what this finding may mean for them. If no other findings are seen on ultrasound and the previous screening tests have been favorable, we consider CPC of no clinical significance.

Echogenic Bowel
**Echogenic Bowel**

- **What is it?**
  - A remarkably echogenic (bright) appearance to the fetal intestines.
  - The entire intestine or a discrete segment may appear echogenic.
- **What does it mean?**
  - Present in 0.5% of normal fetuses.
  - Can be associated with trisomy 21: approximately 7-12% of fetuses with trisomy 21 have echogenic bowel.
  - Can be associated with congenital infections including CMV, Toxo, HSV, parvovirus.
  - Can be associated with cystic fibrosis (meconium ileus) in up to 20% of fetuses.
  - Can be associated with bowel pathology including bowel obstruction, atresia and malformations.
  - Can be a benign finding possibly related to maternal bleeding and fetal swallowing of blood.

- **What to do about it: Prenatal**
  - Confirm the finding truly exists: false positives are common.
  - Look for other markers or malformations associated with chromosomal abnormalities and review screening test results.
  - Do refer to genetics/prenatal diagnosis.
  - Send maternal blood for CMV and Toxo. Consider testing as well for HSV and parvovirus.
  - Send maternal blood for cystic fibrosis carrier status.
  - If amniocentesis is performed, fluid should be sent for karyotype, infection, CF.
  - If workup is negative, do consider follow-up ultrasound in the third trimester for growth and evaluation of the bowel.

- **What to do about it: Postnatal**
  - Depends on findings of workup. If unrevealing, notify NICU at delivery.

**What to say to your patient**

- Echogenic bowel is an ultrasound finding that is often normal and of no significance. However, it can be associated with some chromosomal abnormalities, some infections, cystic fibrosis and bowel problems.
- The finding is very subjective and therefore it is often worth an evaluation by an experienced sonologist and experienced providers in prenatal diagnosis.
- In order to make the most of that second opinion, we do recommend sending some blood tests prior to the visit.

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**Renal Pelviectasis**
Renal Pelviectasis

- What is it?
  - Dilation of the pelvis or central region of the kidney.
  - Also called renal pyelectasis.
- What does it mean?
  - Present in 2% of normal fetuses
  - Can be associated with trisomy 21: approximately 18% of fetuses with trisomy 21 have renal pelviectasis
  - Can be associated with other pathology including ureteropelvic obstruction, vesicoureteric reflux, kidney duplication and bladder outlet obstruction (posterior urethral valves)
  - In cases of mild dilation, the finding is usually in an otherwise normal fetus with little implication

What to do about it: Prenatal

- Look for other markers or malformations associated with chromosomal abnormalities and review screening test results
- Look for other signs indicating possible urinary obstruction: bladder size and appearance, amniotic fluid level, dilation of other parts of the urinary system
- Refer high-risk patients or those with other sonographic abnormalities of the urinary system to genetics/prenatal diagnosis
- For isolated mild pelviectasis, repeat ultrasound at 28-32 weeks is indicated
- More severe cases require serial ultrasound and multidisciplinary counseling

What to do about it: Postnatal

- Depends on suspected etiology.
- Crucial to notify NICU of the finding and the suspected etiology.

What to say to your patient

- Renal pelviectasis is generally an ultrasound finding that needs to be followed but usually is of little significance.
- On ultrasound, we usually see that the plumbing of the urinary system appears a little “backed up”. If this worsens, we may have to follow the fluid around the fetus. In addition, at birth, we will notify the pediatricians who may need to put the baby on antibiotics and do other studies to follow the plumbing.
- Because this can be seen in fetuses with trisomy 21, we do like to review the results of previous screening tests. However, almost all fetuses with this finding do not have trisomy 21.

Renal Pelviectasis

What to say to your patient

Single Umbilical Artery
Single Umbilical Artery

- **What is it?**
  - The umbilical cord, which normally has 2 arteries and one vein, is missing an artery.
  - Also called two vessel (umbilical) cord
- **What does it mean?**
  - Present in 1% of normal singletons, 4.6% of twins
  - In fetuses with isolated SUA, likely no increased risk of chromosomal abnormalities. However, before it is determined to be isolated, likely a 17% and 20% risk of chromosomal abnormalities and anomalies, respectively.
  - Can be associated with Trisomy 18 (seen in 10-50%), Trisomy 13 (seen in 10-50%) and other chromosomal abnormalities.
  - Can be associated with other congenital anomalies.

- **What to do about it: Prenatal**
  - Look for other markers or malformations associated with chromosomal abnormalities, review screening test results
  - Refer to genetics/prenatal diagnosis
  - Some advocate performing fetal echo as part of screening due to an association with congenital cardiac disease.
  - Some advocate an ultrasound for growth at 32 weeks to screen for growth restriction
- **What to do about it: Postnatal**
  - If isolated, nothing needs to be done and no complications postnatally are expected.

What to say to your patient:
- Some fetuses have an umbilical cord that is slightly different than others and is missing a vessel. This is of no consequence to the fetus and does not cause any problems.
- Rarely, this finding can be a sign of a chromosomal problem or a sign that other anomalies exist. Because of this, a more detailed ultrasound and consultation at a prenatal diagnosis center are indicated.

Ventriculomegaly
**Ventriculomegaly**

- **What is it?**
  - Enlargement of the lateral ventricles of the brain.
  - Mild=10-12mm, Moderate=12.1-15mm, Severe=>15mm
  - Can be unilateral or bilateral

- **What does it mean?**
  - Mild ventriculomegaly can be a normal variant especially in male fetuses
  - Can be associated with chromosomal abnormalities, especially trisomy 21
  - Can be associated with other intracranial pathology of many etiologies including problems in development (developmental delay), injury and infection

**What to do about it: Prenatal**

- Look for other markers or malformations associated with chromosomal abnormalities, look for other cranial abnormalities, review screening test results
- Refer to genetics/prenatal diagnosis for genetic and multidisciplinary counseling
- Consider fetal MRI
- Serial ultrasounds during the pregnancy are indicated
- Term, vaginal delivery can be considered

**What to do about it: Postnatal**

- Depends on findings of workup
- Postnatal ultrasound and MRI are indicated
- Notify NICU at delivery

**What to say to your patient**

- Ventriculomegaly is an ultrasound finding that can be of great concern though it can also be considered normal. It can be associated with some chromosomal abnormalities, as well as other brain abnormalities.
- An evaluation by an experienced sonologist and experienced providers in prenatal diagnosis is indicated. They may recommend evaluation with MRI and will provide you with information specific to your situation.
- With mild ventriculomegaly and no other problems on the work-up, a baby may be normal. However, if the ventriculomegaly is more severe, babies may have more concerning outcomes and require much more evaluation after birth.

**Clubfoot**
Clubfoot

- **What it is**
  - Malformation of the bones of the ankle and foot causing the foot to be turned in
  - Occurs in 1 in a 1,000 births

- **What it means**
  - The diagnosis itself depends on how well the sonologist can see the extremity. The finding may be quite certain at a second trimester scan, but may be more difficult to ascertain later in pregnancy when fetal positioning and size make visualization more challenging.
  - The implication is debated. Some investigators have shown a 6% risk of chromosomal abnormalities (Trisomy 21, 18 and others), others have shown no increased risk if the finding is truly isolated.

- **What to do about it: Prenatal**
  - Look for other markers or malformations associated with chromosomal abnormalities, review screening test results
  - Refer to genetics/prenatal diagnosis for genetic and multidisciplinary counseling
  - Some patients may choose amniocentesis for “isolated” clubfoot given the small risk that other anomalies exist but are not detectable by ultrasound.

- **What to do about it: Postnatal**
  - Refer to pediatric orthopedics
  - The severity of the deformity determines the treatment needed:
    - Postural only: no treatment
    - True clubfoot: casting and possible surgery required usually with an excellent outcome

- **What to say to your patient:**
  - Sometimes on ultrasound, the foot and ankle can appear to be “clubbed” or turned inward. It is often difficult to say for certain, though, until birth. If the foot/feet are clubbed, the baby will be followed by an orthopedic doctor who may recommend a cast or even surgery to correct the problem. Sometimes this condition runs in families.
  - This finding does not usually indicate that there are other problems as well (for example chromosomal abnormalities), but we like to make certain of that by having a detailed ultrasound and a review of previous screening tests. Even when all of these things are normal, some parents do choose to do an amniocentesis just to be certain.

Cleft Lip
Cleft lip

- **What it is**
  - The typical cleft lip is a linear defect from one side of the lip to the nostril.
  - Can be unilateral, bilateral and involve a cleft palate as well.
  - Isolated cleft palate (without cleft lip) is a separate entity.
  - The incidence depends on ethnicity: seen in 0.3-3.6 births out of 1,000.

- **What it means**
  - If truly isolated, cleft lip (without cleft palate) does not increase the risk of chromosomal abnormalities.
  - If unilateral cleft lip and palate are seen, 5-20% risk of aneuploidy. If bilateral CL/CP, 15-30% risk of aneuploidy. These risks are higher if further anomalies are seen.
  - The finding of cleft lip with normal chromosomes can be associated with other congenital anomalies: 5% risk with unilateral cleft lip, 11% risk if unilateral CL/CP, 18% risk if bilateral CL/CP.

- **What to do about it: Prenatal**
  - Look for other markers or malformations associated with chromosomal abnormalities, review screening test results.
  - Look for other anomalies; consider fetal echo.
  - Refer to genetics/prenatal diagnosis for genetic and multidisciplinary counseling.
  - Some patients may choose amniocentesis for “isolated” cleft lip given the small risk that other anomalies exist but are not detectable by ultrasound.

- **What to do about it: Postnatal**
  - Refer to appropriate center (craniofacial clinic typically).
  - The severity of the cleft lip and palate determine the treatment needed:
    - Mild clefts or soft palate only: no treatment
    - Larger defects and hard palate defects: surgical correction and multidisciplinary care.

- **What to say to your patient:**
  - A cleft lip is an abnormality in the formation of the lip. It can also be seen with a defect in the palate (or roof of the mouth). Usually, this is not seen with other problems (chromosomal abnormalities or other anomalies) and is correctable after birth. It sometimes runs in families.
  - This finding does not usually indicate that there are other problems as well (for example chromosomal abnormalities), but we like to make certain of that by having a detailed ultrasound and a review of previous screening tests. Even when all of these things are normal, some parents do choose to do an amniocentesis just to be certain.

Key Points

- Consider discussing the value and limitations of ultrasound prior to the patient's second trimester scan.
- When relaying results of the ultrasound to the patient, know what is simply a marker (EIF, CPC) versus what is a abnormality/malformation (cleft lip, clubfoot).
- Before relaying the results of the ultrasound, do a careful review of the patient's history and screening results. This will help you frame the significance of the ultrasound findings.
Helpful Resources for Patients and Providers

- March of Dimes: http://www.marchofdimes.com
- UCSF Fetal Treatment Center: http://fetus.ucsfmedicalcenter.org
- Northwestern Children’s Memorial Hospital: http://www.childrensmemorial.org/depts/fetalhealth/overview.aspx
- UCSF Prenatal Diagnosis Center: 415-476-4080 and UCSF Fetal Treatment Center: 415-476-0445

References

- UpToDate: www.uptodate.com
- March of Dimes Resource Center