Fetal Echocardiography, Fetal and Placental MR – Imaging Techniques to Optimize the Screening, Analysis and Consultation of the Obstetrical Patient and Abnormal Fetus.

Daniel J. Cohen, M.D.
HUDSON VALLEY RADIOLOGY ASSOCIATES

- HVRA is the only regional provider of fetal echocardiography accredited by the American Institute of Ultrasound in Medicine (AIUM)

- Last year HVRA performed 530 Obstetrical MR studies for the tri state Obstetrical and MFM community.

What is Universal Fetal Echocardiography and how does it –

optimize the safe delivery of healthy newborns in a community hospital setting?

minimize medical-legal exposure in cases of stillbirth the second most common cause of obstetrical litigation?

optimize the risk assessment of unscreened pregnancies and pregnancies at increased risk for chromosomal and non- chromosomal syndromes?

Fetal MR is underutilized by MFMs and Obstetricians

- when and why should the primary OB know to order a fetal MR
- in ' wrongful birth' litigation, the question of whether or not a fetal MR should have been performed is increasingly being asked.

- Case presentations will demonstrate the unique, problem solving attributes of fetal MR across all organ systems.

- Case presentations will highlight the detection of abnormalities – not seen on ultrasound that alter patient management, consultation and/or site of delivery.

Universal Fetal Cardiac Ultrasound-
At the Heart of Newborn Well-Being
HUDSON VALLEY RADIOLOGY ASSOCIATES

Universal Fetal Cardiac Ultrasound – At the Heart of Newborn Well-being

Optimizes detection of congenital heart disease (chd) in the general low risk obstetrical population

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Universal fetal cardiac ultrasound and the Cardio Vascular Genetic/Level 2 Obstetrical Ultrasound Exam

Optimizes detection of CHD and in doing so optimizes risk assessment for chromosomal & non chromosomal syndromes.

From the patients perspective – why do we perform antenatal ultrasound?

Is my baby (fetus) okay?

Are there malformations that might hurt my baby in utero or after birth or alter where and how I deliver?

Unbeknownst to most parents and many health care providers is the 2-3% incidence of congenital malformations in the general low risk populations (2 – 3 fetal malformations per 100 deliveries.)

- Congenital malformations represent the overwhelming majority of morbidity & mortality in the new born period.

- Congenital heart disease is the most common of these malformations.

- Abnormalities of the heart and great arteries are the most common congenital defects, accounting for approximately 20% of all stillbirths and 30% of neonatal death due to congenital defect.

*Office for National Statistics. Mortality statistics: childhood, infancy and perinatal, England and Wales. Series DH3, #35, Office for National Statistics 2002
*Hoffman. The Incidence of Congenital Heart Disease. Journal of American College of Cardiology 2002; 39: 1890-1900.
*Llurba. Ultrasound Obstetrics and Gynecology 2013; 42: 169-174

UNIVERSAL FETAL CARDIAC ULTRASOUND

The community standard that defines our imaging protocol is influenced by our proximity to tertiary care centers performing neonatal cardiac surgery. Antenatal detection of congenital heart disease with non- emergent delivery at such centers has proven to improve neonatal outcome.

Our protocols, therefore, must detect pathologies whose treatment will effect outcome and exclude those pathologies that would preclude delivery at a community hospital setting.

UNIVERSAL FETAL CARDIAC ULTRASOUND

Current imaging guidelines (American Institute of Ultrasound in Medicine) for cardiac assessment during the 18-22 week detailed anatomic exam (CPT 76811) and genetic/Level II obstetrical ultrasound only require four chamber cardiac view of the heart and suggest (but not necessitate) an "attempt" at outflow tract assessment "when technically feasible". Screening with four chambers views of the heart only detect 25-50% cardiac malformations and will miss the great majority of potentially cyanotic lesions such as tetralogy of Fallot, transposition, double outlet right ventricle, truncus arteriosus.

Journal of Ultrasound in Medicine 2005, p. 1752. Letter to Editor.

American Institute of Ultrasound in Medicine. AIUM Practice Guidelines for the Performance of an Antepartum Obstetrical Ultrasound Examination. Laurel: American Institute of Ultrasound in Medicine; 2003.
Available as: http://www.AIUM.org/publication/clinical/obstetrical.PDF

UNIVERSAL FETAL CARDIAC ULTRASOUND

- CHD is the most common and most serious of all structural malformations. 1/200 - 1/300 fetuses have heart malformations that necessitate delivery at a university hospital setting.

- The profound majority of fetuses with CHD are born to couples with no history nor risk factor.

Benacerraf. Accuracy of Fetal Echocardiography. Radiology 1987; 165: 847-849
Mitchell. Congenital Heart Disease in 56, 109 Births: Incidence of Natural History. Circulation 1971; 34:323-332.

UNIVERSAL FETAL CARDIAC ULTRASOUND

- CHD is the most frequently missed malformation at the time of "routine" detailed 18- week ultrasound.

- CHD is the number one cause for significant morbidity and mortality in the first year of life. 10% of newborns who die in the first year of life do so with an undiagnosed heart defect.

Yagel. Congenital Heart Defects: Natural Course and In Utero Development. Circulation 1997;96:550-555.

FAST FACTS ON FETAL CONGENITAL HEART DISEASE (CHD)

- CHD is the malformation most responsible for infant morbidity and mortality accounting for greater than one-third of infant deaths related to congenital malformation.

- Without a prenatal diagnosis, even severe forms of congenital heart disease commonly go undetected until after discharge to home leading to avoidable morbidity and mortality.

- 20-55% of infants with CHD are not diagnosed until after hospital discharge. Most obstructive left heart lesions (such as aortic coarctation) are not diagnosed at birth or at six weeks.

FAST FACTS ON FETAL CONGENITAL HEART DISEASE (CHD)

- Aortic coarctation is one of the three undiagnosed conditions (the others are hypoplastic left heart and interrupted arch) most likely to lead to death soon after discharge from hospital.
- During first year of life 25% of deaths due to CHD occur before the diagnosis of CHD.

UNIVERSAL FETAL CARDIAC ULTRASOUND

- 15% of fetuses with CHD have abnormal chromosomes so if the heart defect is missed you miss the opportunity to karyotype.

- 50% of Down syndrome fetuses have CHD, Screening for Down Syndrome is therefore incomplete without a dedicated study of the heart.

Nyberg. Diagnostic Imaging of Fetal Abnomalies, 2003. P. 453.

Ferentz, C. Perspectives in Pediatric Cardiology. Vol 4. Congenital Heart Disease: The Baltimore-Washington Infant Study, 1981-9. New York: Putura Publishing, 1993.

UNIVERSAL FETAL CARDIAC ULTRASOUND

Cardiac anomalies are the most overlooked lesions during prenatal ultrasound scanning and all the benefits of early prenatal diagnoses are withheld from the families if the cardiac diagnoses are missed.

Yagel. Congenital Heart Defects: Natural Course and In Utero Development. Circulation 1997;96:550-555.

UNIVERSAL FETAL CARDIAC ULTRASOUND

Fetal cardiac ultrasound is necessary to **complete and optimize** detection of malformations that alter obstetrical management, site of delivery, aneuploidy risk assessment, neonatal well being and minimize emergency transfer of sick newborns.

HVRA Outcome analysis - a ten year retrospective analysis of transfer rates of sick newborns

Outcome analysis - a ten year retrospective analysis of transfer rates of sick newborns with the ICD9 codes of congenital heart disease (CHD) delivered at Nyack Hospital demonstrates a transfer rate of 3-4 per 3,000 – significantly lower than the national average of 9-15 per 3,000 for similar sized hospital (biostatistics obtained from Center for Disease Control.)

UNIVERSAL FETAL CARDIAC ULTRASOUND

We report for a fetal cardiac exam accompanying a detailed 18 – 22 week anatomy scan.

" No signs of hypoplastic left heart, atrioventricular septal defect, tetrology of fallot, transposition nor aortic coarctation- the most common clinically significant patterns of CHD detectable on antenatal ultrasound. "

UNIVERSAL FETAL CARDIAC ULTRASOUND

The two accompanying articles from the December 2007 *Journal of Ultrasound In Medicine* substantiate HVRA's imaging philosophy that every pregnant woman should be offered a fetal cardiac ultrasound at the time of her detailed 18 – 22 week anatomy scan.

Improving Detection of Fetal Cardiac Anomalies

A fetal echo cardiogram for every fetus?

Bahtiyar and Copel. Pg 1639 - 1641. J. Ultrasound Med 26:2007.

Prenatal Detection of Congenital Heart Disease in Southern, Nevada. The need for universal fetal cardiac evaluation.

Acherman. Pg. 1715 - 1719. J. Ultrasound Med 26:2007.

UNIVERSAL FETAL CARDIAC ULTRASOUND

The existing guideline for "outflow tract imaging when possible" promulgated by CPT and AIUM guidelines has had no effect in improving the national detection rate of congenital heart disease 15-30%.

- 4 chamber heart view detects only 40 – 50% of potentially cyanotic patterns of CHD

*Prenatal Detection of Congenital Heart Disease in Southern Nevada
The Need for Universal Fetal Cardiac Evaluation
J. Ultrasound Med 26:1715-1719
*Barriers to Prenatal Detection of Congenital Heart Disease:
A Population based Study. Ultrasound Obstetrics Gynecology
2012. Vol. 40: 418-425

UNIVERSAL FETAL CARDIAC ULTRASOUND

It is not the mere performance of "outflow tract" imaging but the detailed knowledge of fetal cardiac pathology, its recognition, and the interpretative expertise that is necessary to substantially increase detection rate of CHD*.

It is these special skills that define 2D fetal cardiac 76825 assessments. HVRA's protocols and intervention identify CHD at rates concordant with the highest levels quoted in the literature (approximately 80%)

* Prenatal Detection of Congenital Heart Disease in Southern Nevada
The Need for Universal Fetal Cardiac Evaluation
J. Ultrasound Med 26:1715-1719

*Barriers to Prenatal Detection of Congenital Heart Disease:
A Population based Study. Ultrasound Obstetrics Gynecology
2012. Vol. 40: 418-425

2008 HVRA CENSUS FOR FETAL CARDIOVASCULAR MALFORMATIONS

- Total number of detailed 76811 exams – 2,718
- Total number of 2D fetal cardiac exams – 2,275
- Total number of cardiovascular malformations - 22, including isolated right aortic arch and isolated agenesis of ductus venosus both with normal heart - 6

MOST FREQUENTLY QUOTED INCIDENCE OF CLINICALLY SIGNIFICANT CONGENITAL HEART DISEASE AMONGST NEWBORNS – 1:200-1:300

HVRA'S 2008 DETECTION RATE

- All cardiovascular malformations – 22/2,718 = 2.4 cases per 300 newborns

- Not including isolated right aortic arch and isolated agenesis ductus venosus both with normal heart – 16/2,718 = 1.8 cases per 300 newborns.

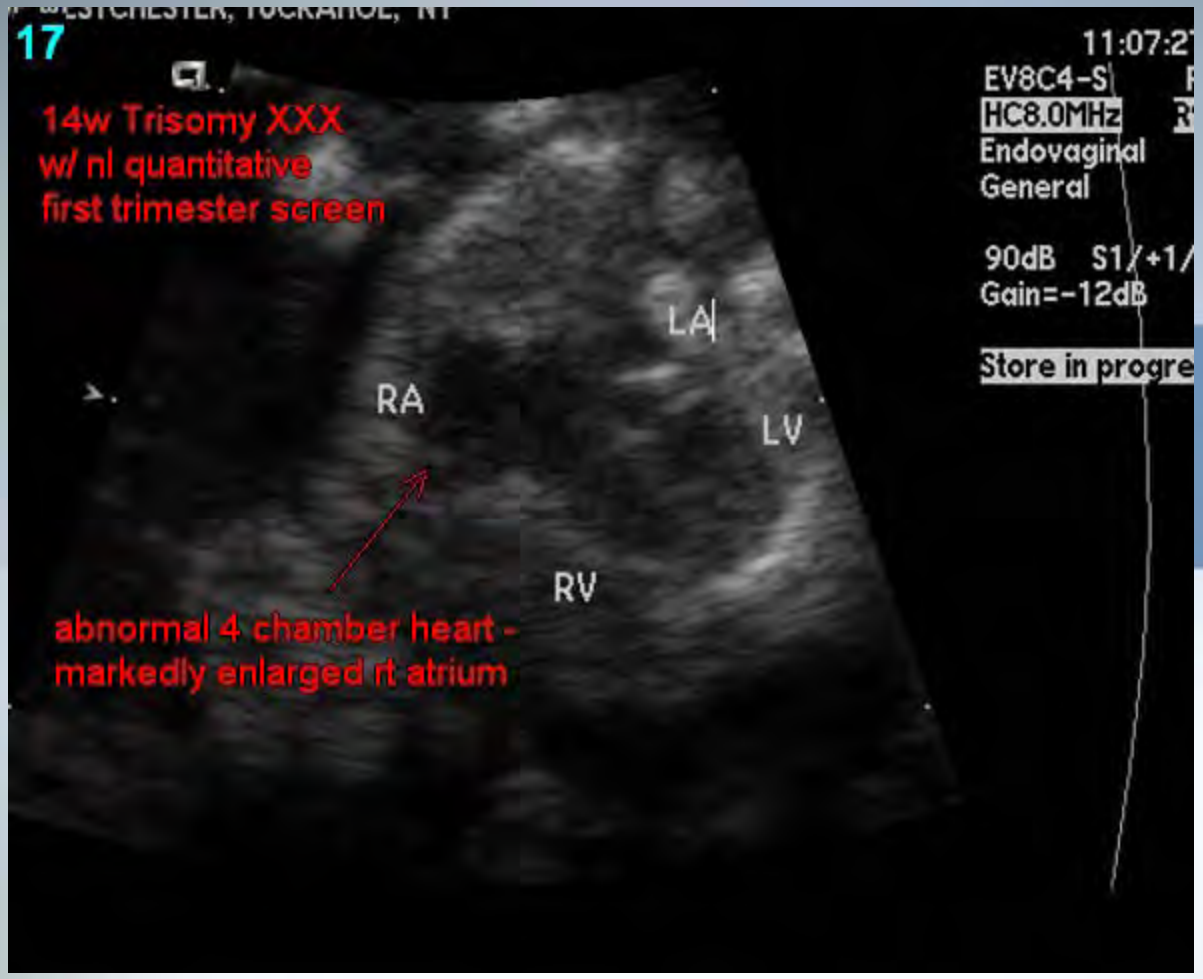
Cardiovascular Genetic/level II obstetrical ultrasound exam is an evidence-based pregnancy outcome verified program with a 90% Down syndrome detection rate which goes up to 98% after normal first or second trimester screening.

A 90% risk reduction can be applied to patients first trimester and/or second trimester screening results.

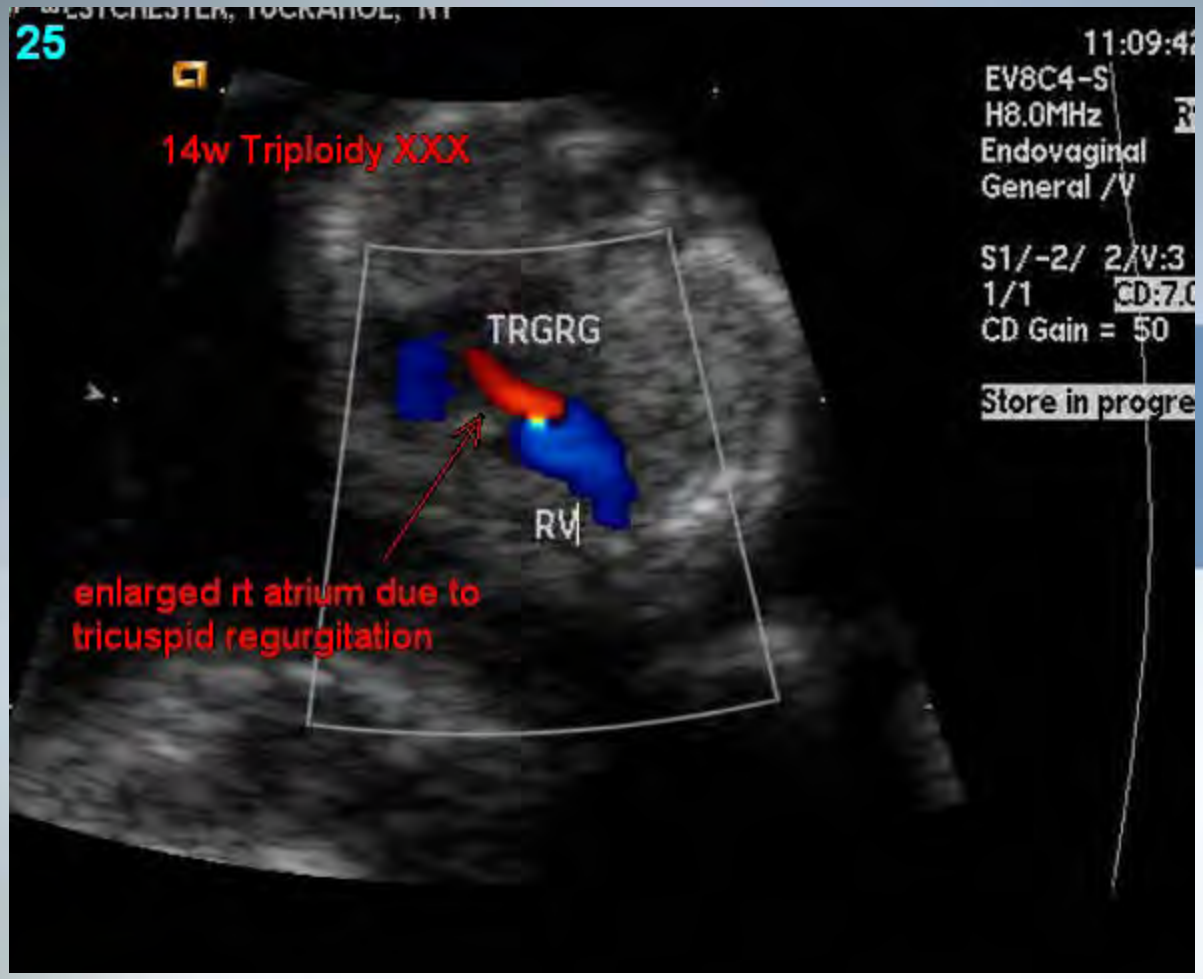
Example – For a patient whose screening results indicate an elevated 1:200 Down syndrome risk but is ambivalent or refuses amniocentesis, a negative cardiovascular genetic ultrasound will risk reduce to 1:1818 based on 90% risk reduction.

*Role of Second Trimester Genetic Sonography after Down Syndrome Screening. Obstetrics and Gynecology. Vol 114, #6. December 2009. p1189-1196. Aagaad-Tillery.
*Genetic Sonography: The Historical and Clinical role of Fetal Echocardiography. Ultrasound Obstetrics Gynecology 2010; 35:509-521. Devore
*Barriers to Prenatal Detection of Congenital Heart Disease: A Population Based Study. Ultrasound Obstetrics Gynecology 2012. Vol. 40: 418-425.

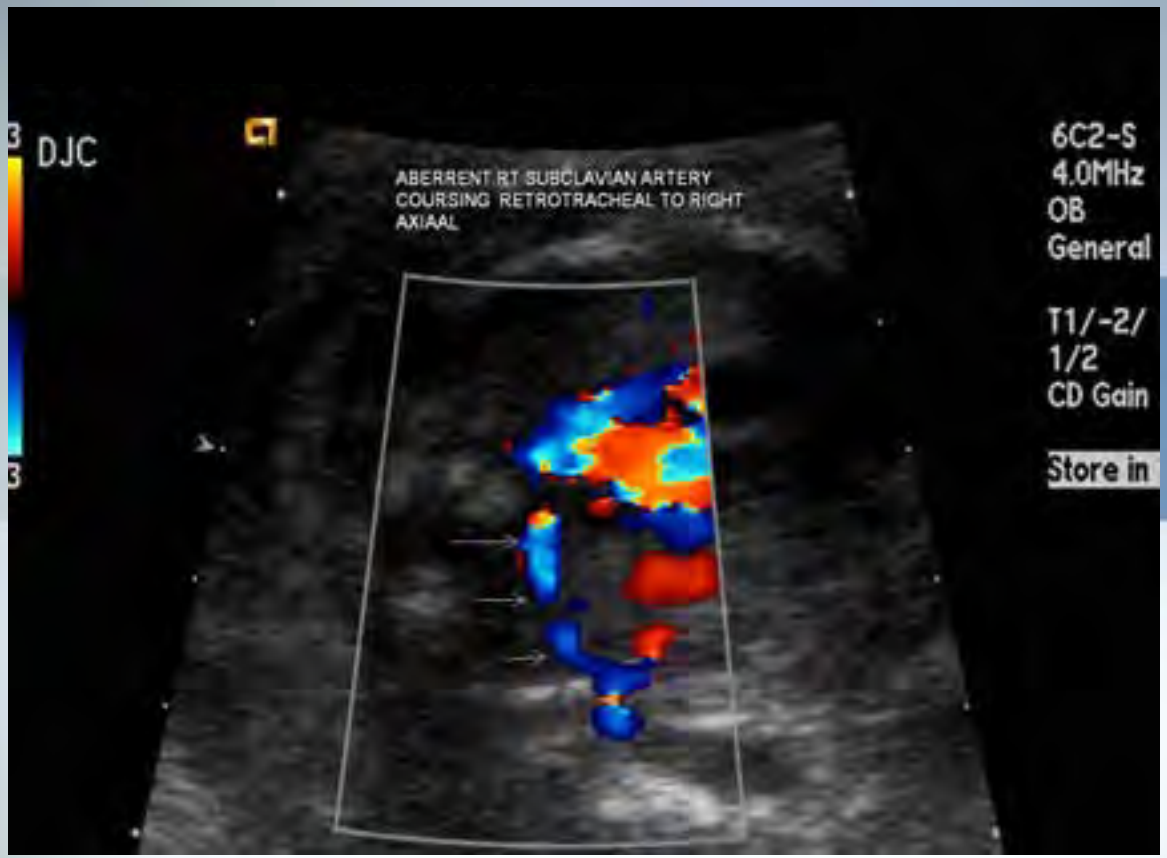
14-week triploidy XXX – enlarged right atrium



14-week triploidy XXX – tricuspid regurgitation

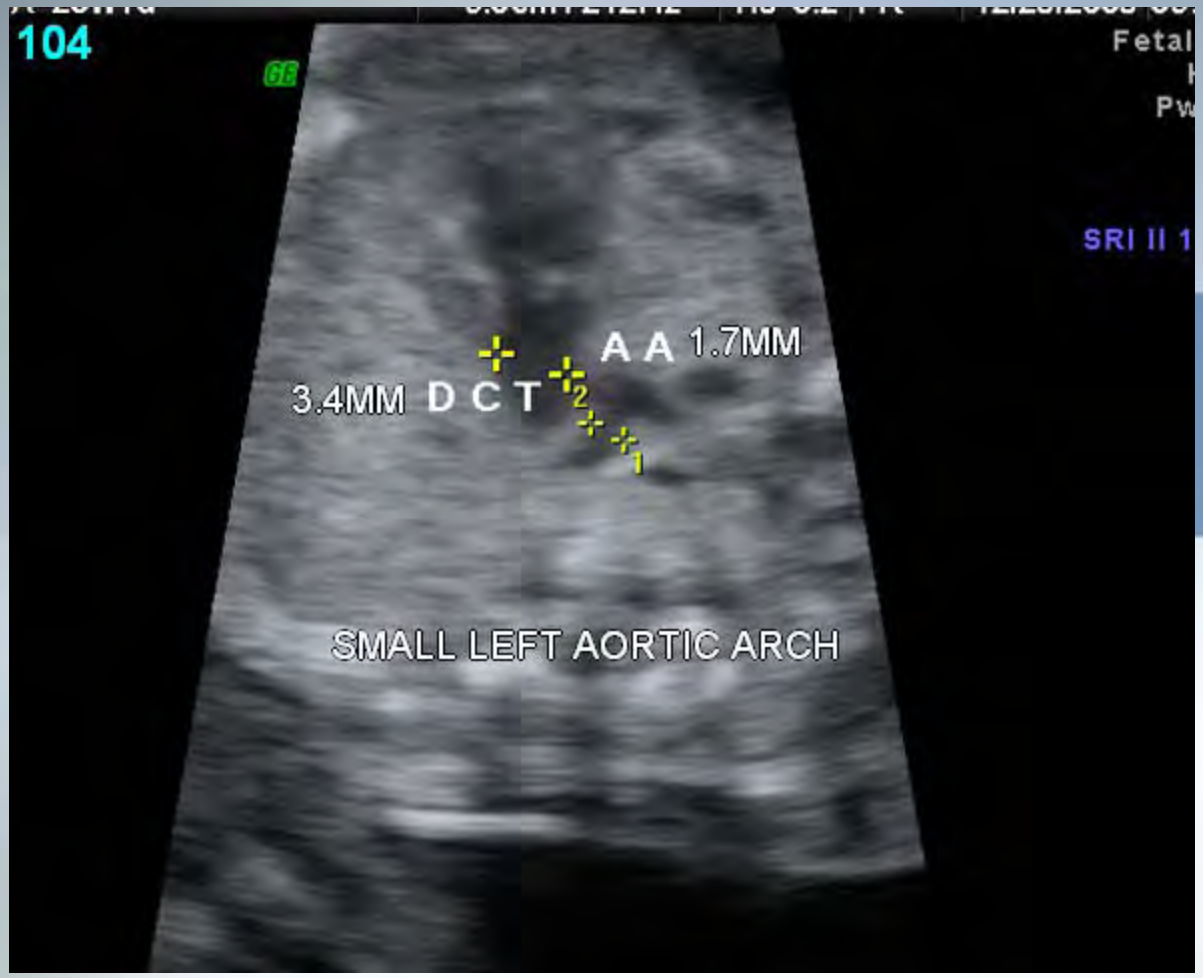


Aberrant right subclavian artery – Down syndrome marker. Likelihood ratio of 20

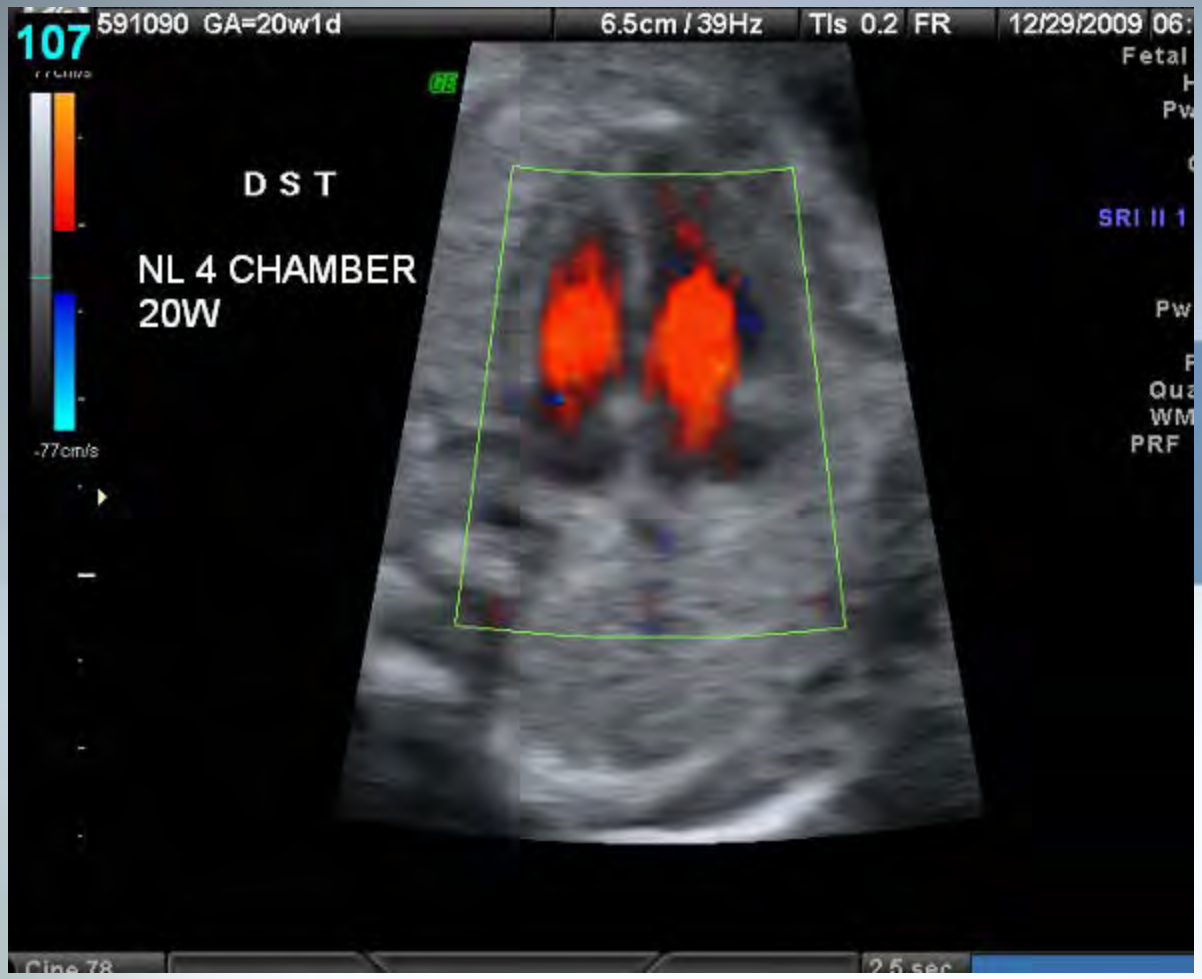


Aortic coarctation – syndromic marker.

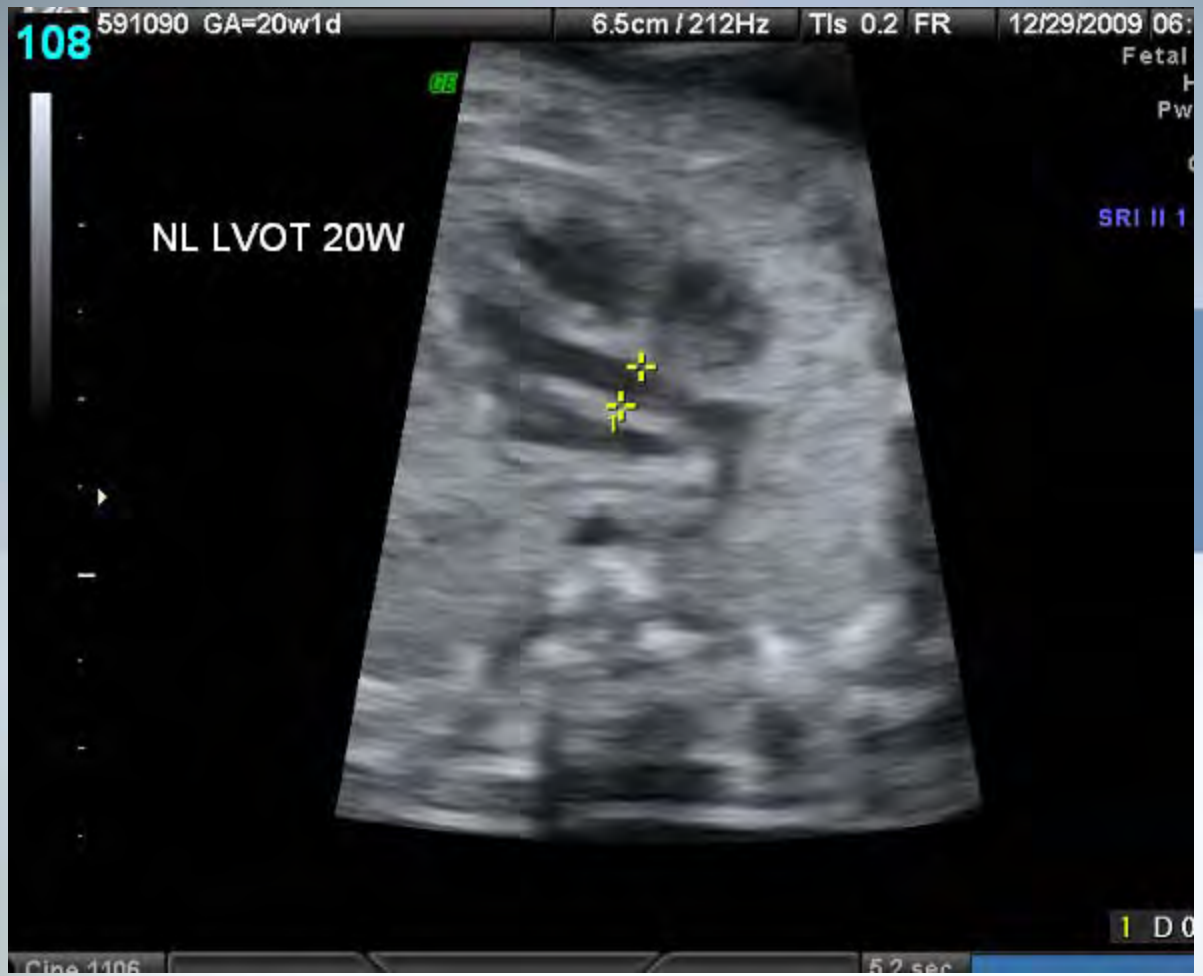
Deliver at tertiary care center



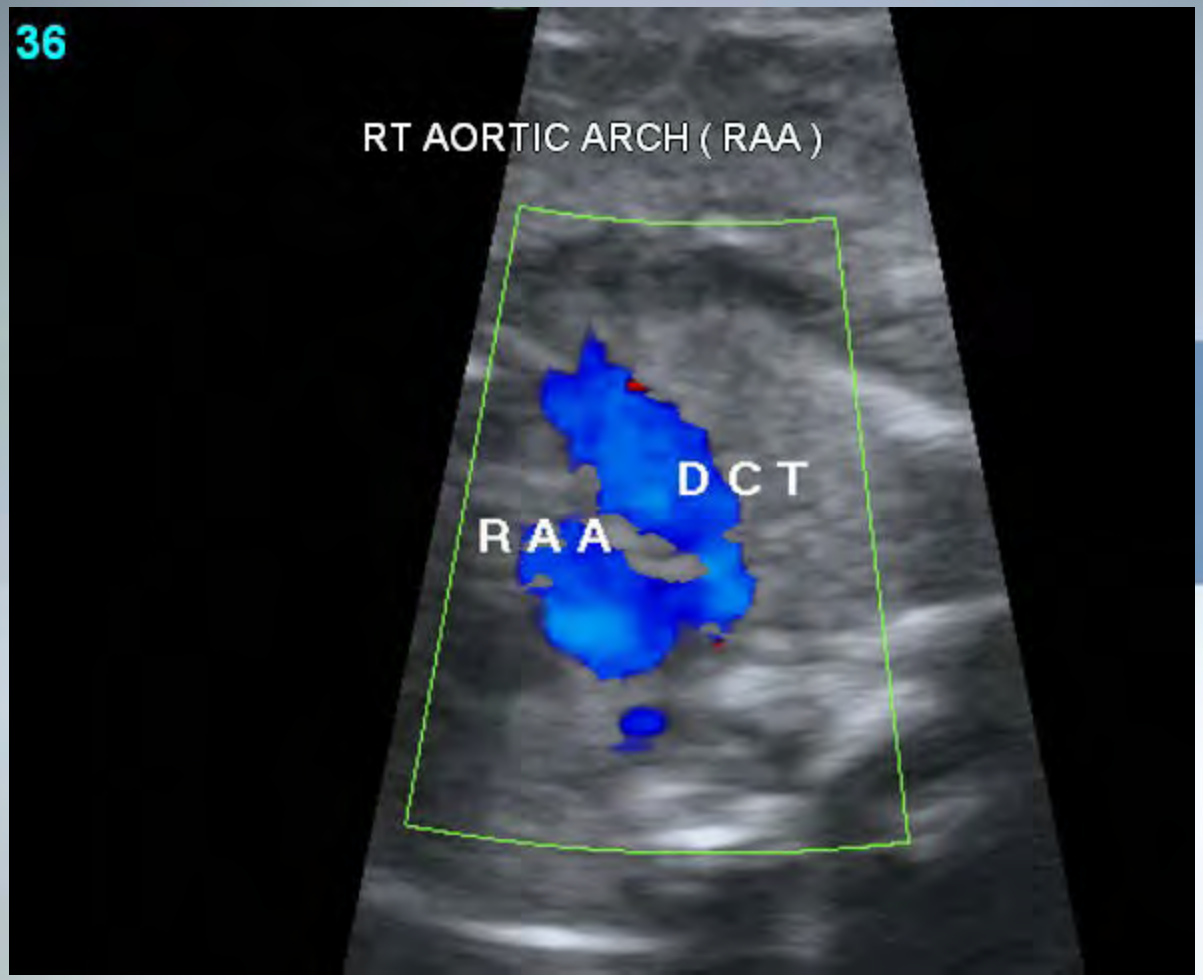
Normal four chamber heart in fetus with aortic coarctation



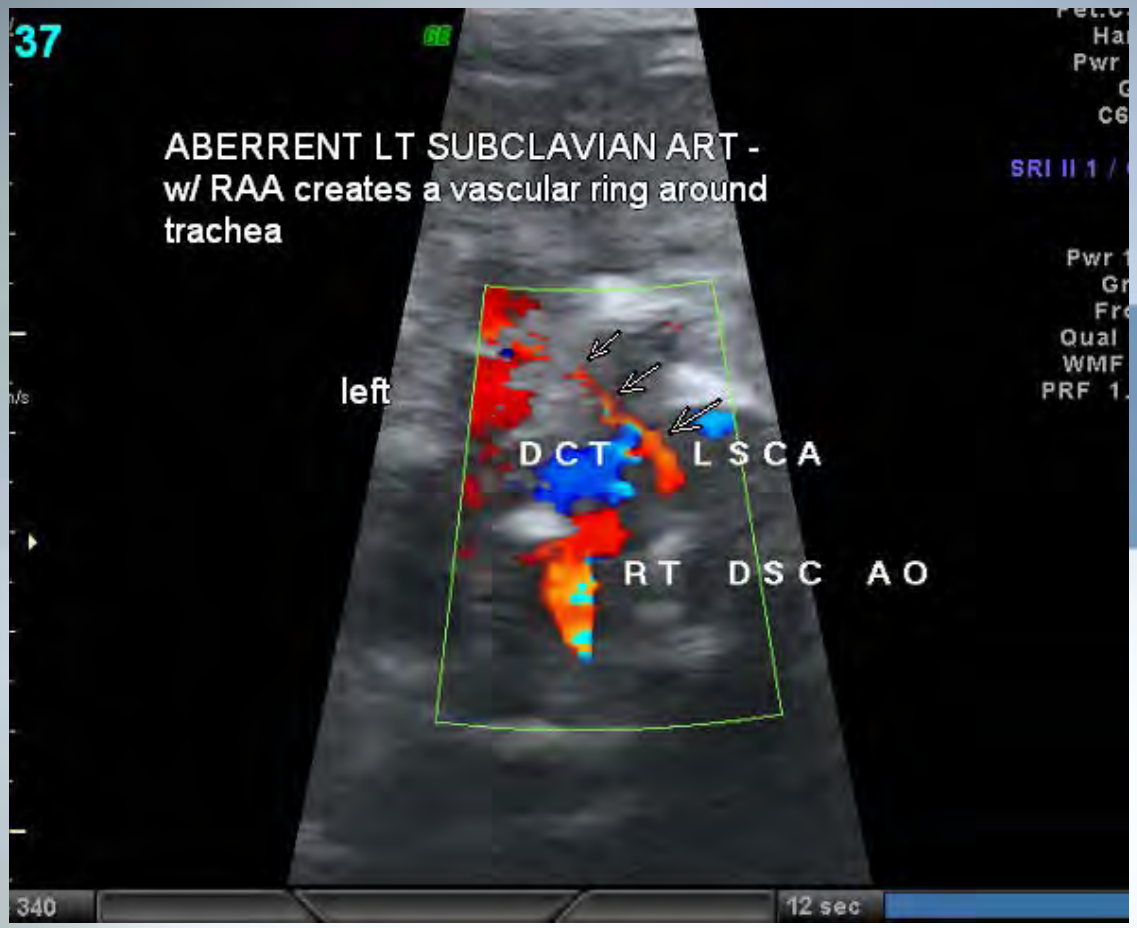
Normal left ventricular outflow tract in fetus with aortic coarctation



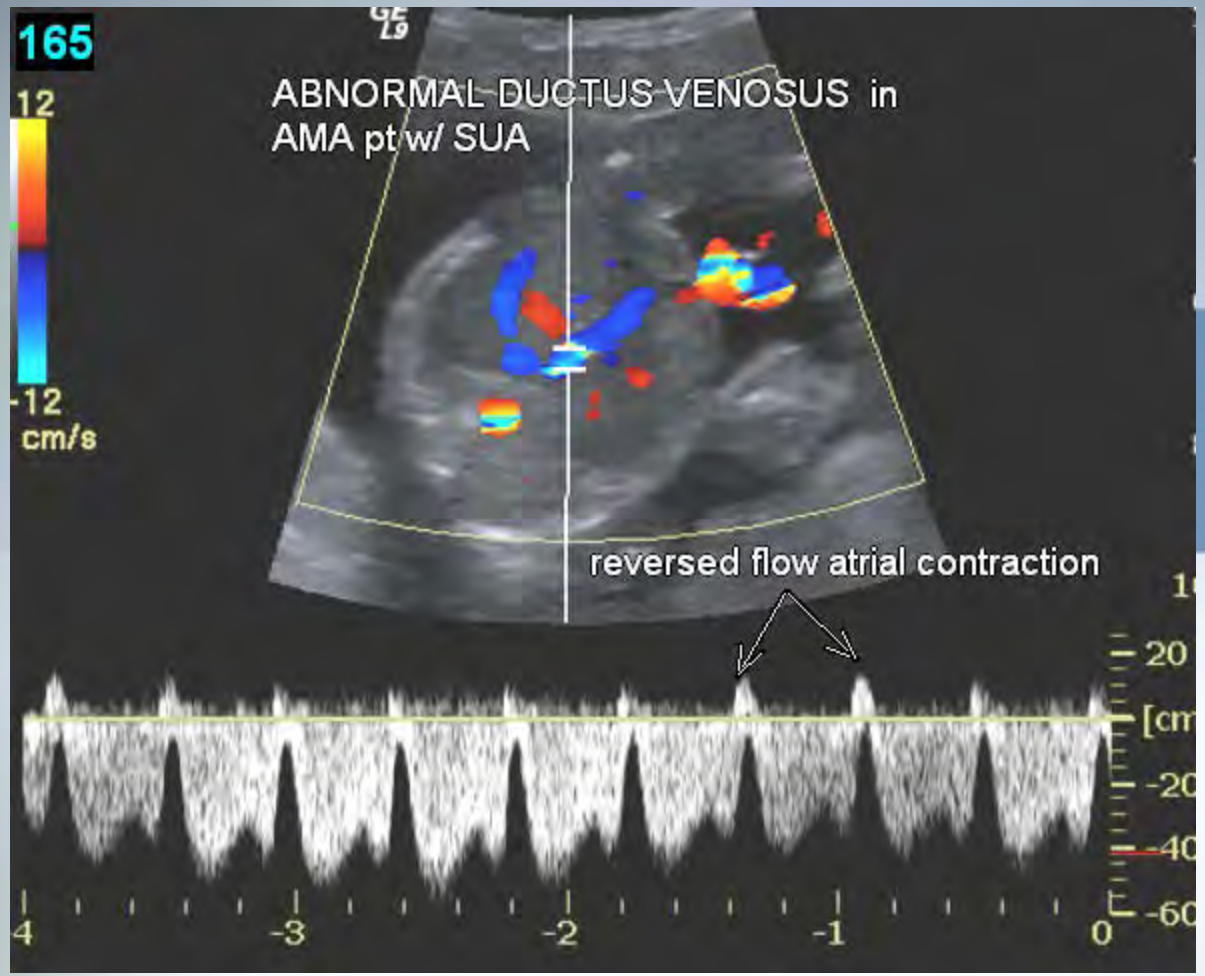
Right aortic arch (RAA) – syndromic marker



RAA with aberrant left subclavian artery – vascular ring, deliver at tertiary care center



Unscreened AMA with single umbilical artery. A-V fistula. Fetal death two weeks later



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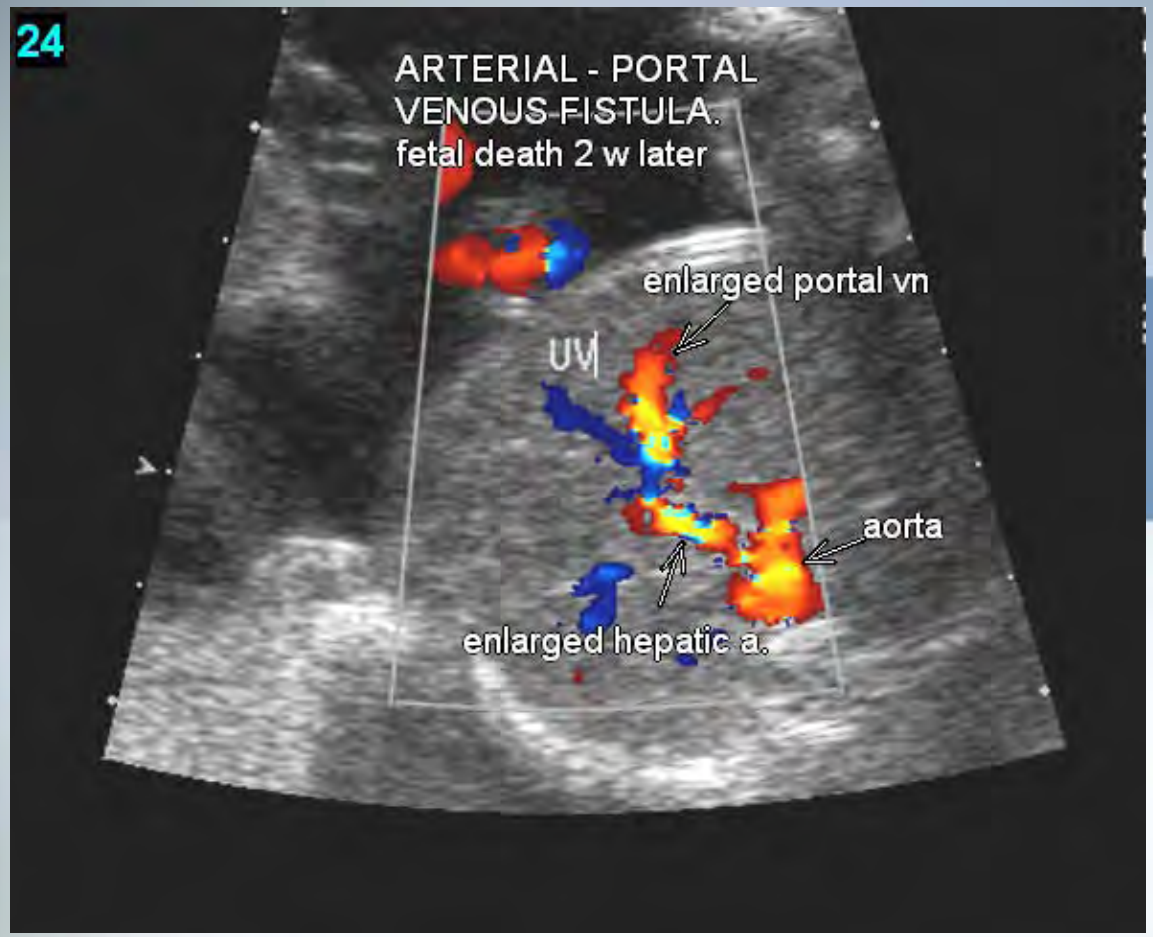
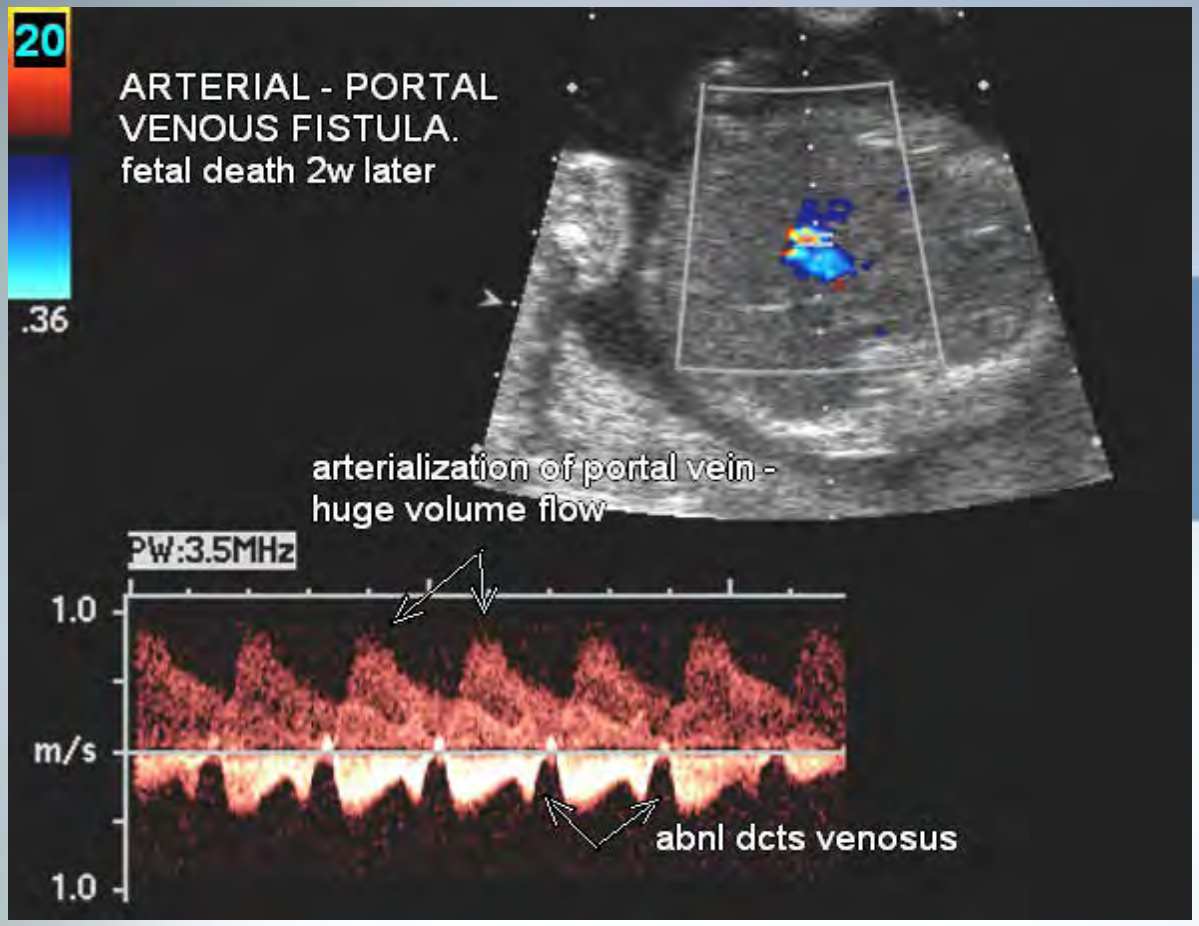
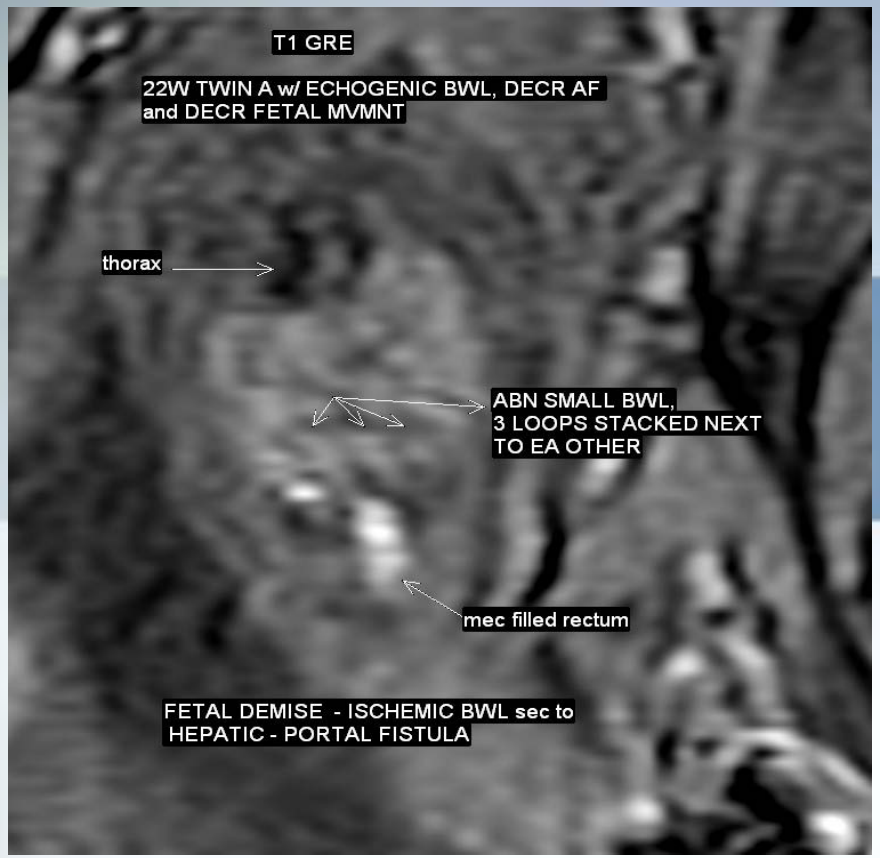


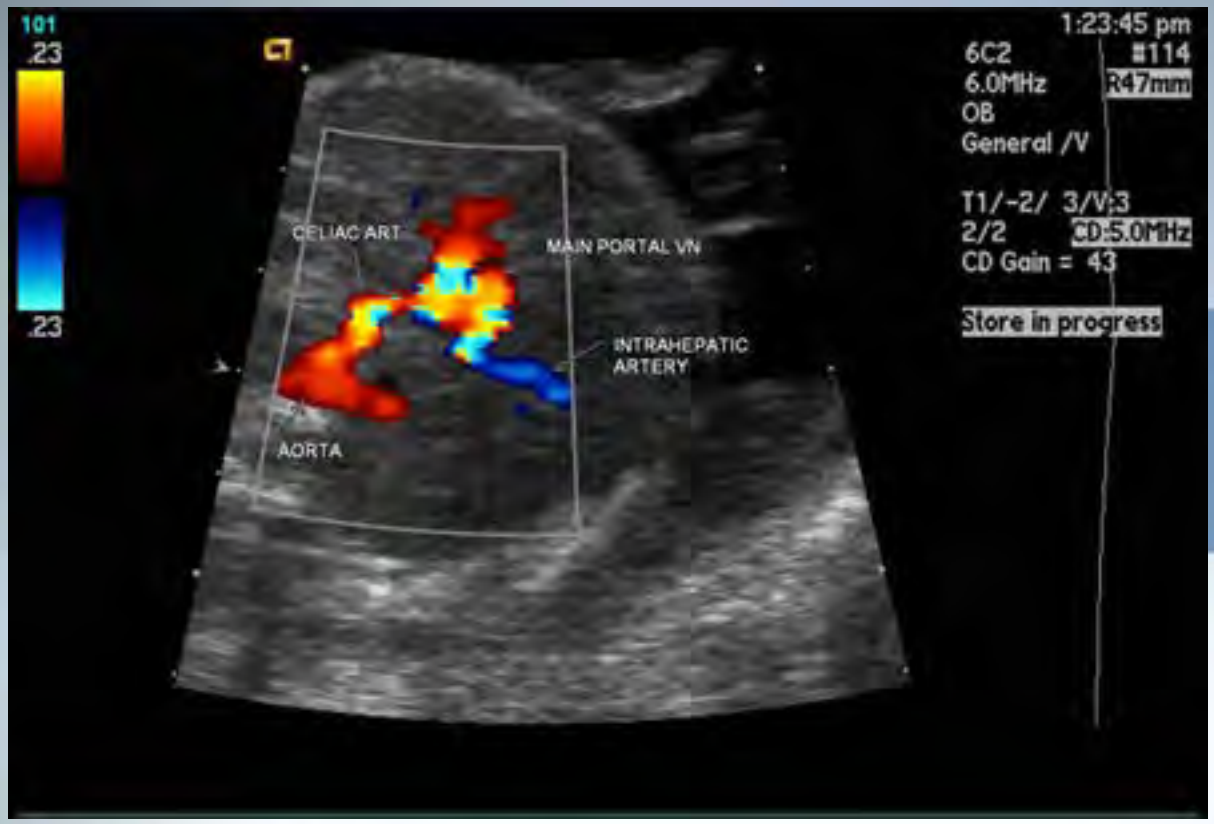
Unscreened AMA with single umbilical artery. A-V fistula. Fetal death two weeks later



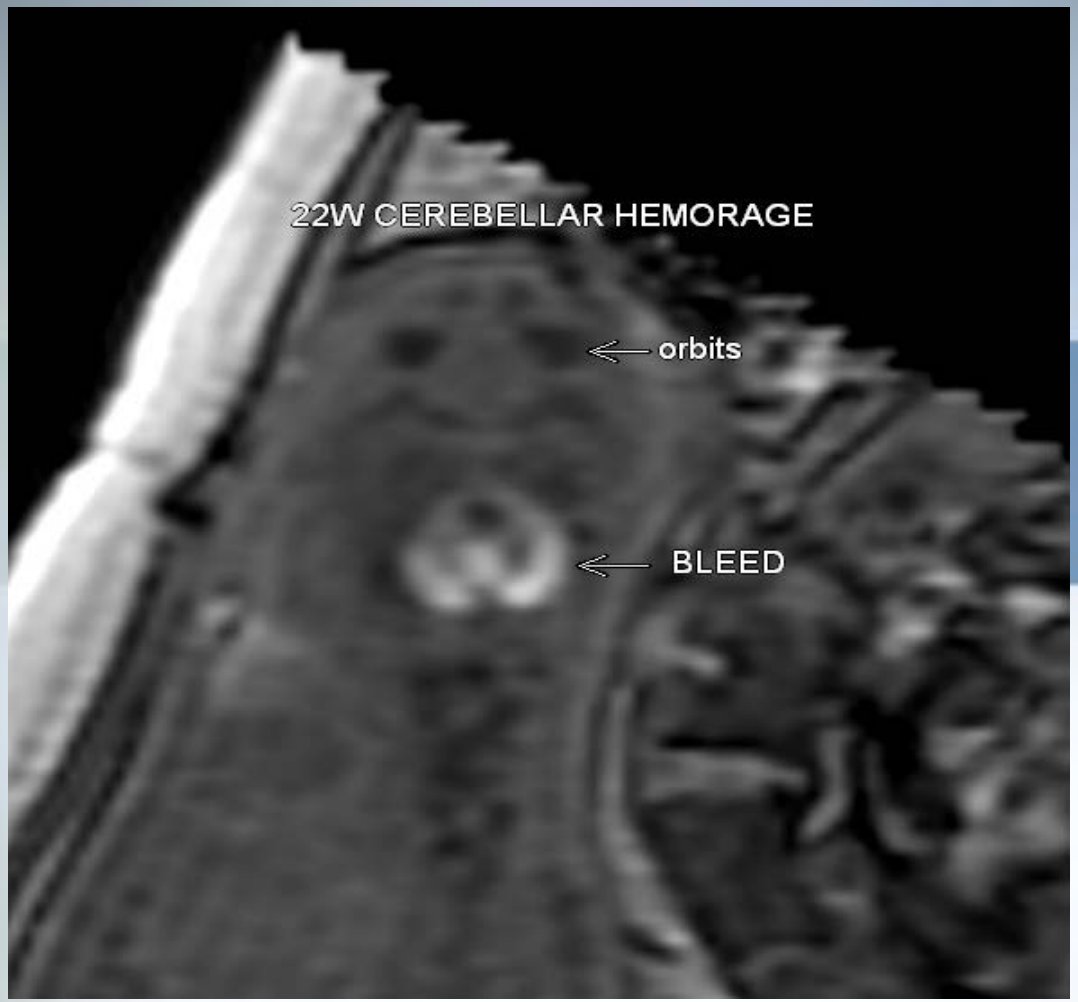
Ischemic bowel secondary to A-V fistula. Fetal death.



Fistula – celiac artery to portal vein. Fetal death two weeks later.



Fistula – aorta to IVC. Severe IUGR. Cerebellar bleed.



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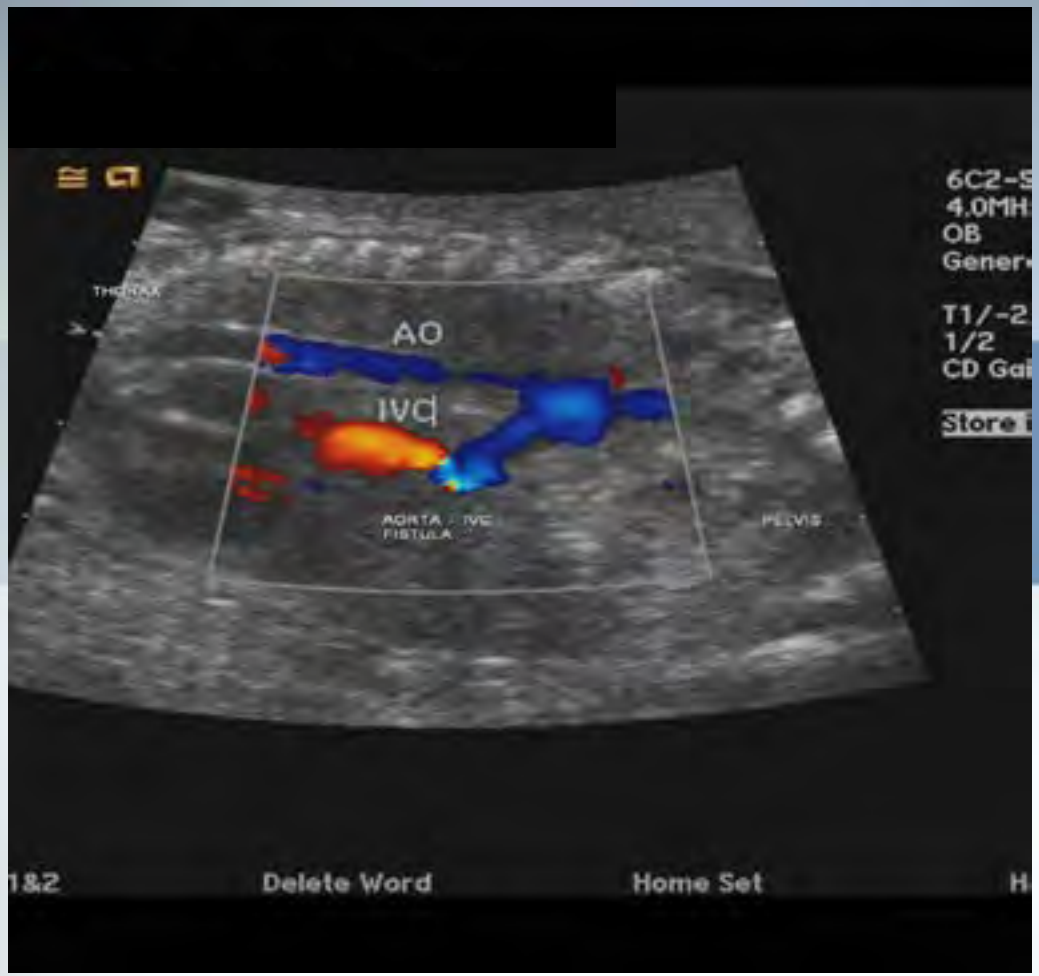


Obstetrical MR

What Every Obstetrician Should Know:

Fetal Malformations. Placental Dysfunction

Daniel J. Cohen, MD
Hudson Valley Radiology Associates

Clinical and ultrasound indications for obstetrical MR
MR's unique problem solving attributes in comparison to ultrasound
Role of collaborative multidisciplinary care by prenatal and postnatal specialists

FETAL MR INTRODUCTION

The goal of our fetal MR is to optimize patient counseling and fetal/neonatal management by providing unique, clinically relevant contributions to fetal anatomic assessment above and beyond that which can be offered by high quality transabdominal/transvaginal ultrasound.

SPECIFICITY OF TERTIARY LEVEL FETAL IMAGING

- In general practice, MR and ultrasound are typically conducted by two different types of specialists – MR performed by radiologists who usually do not have a background in fetal medicine and who do not perform hands –on fetal ultrasound on a regular basis –

- Ultrasound often performed by physicians who do not have special skills in neuroanatomy and neuroimaging (Pilu, 1992-93)

SPECIFICITY CAN BE ENHANCED BY THE SAME PHYSICIAN PERFORMING BOTH MR EXAM AND TERTIARY LEVEL HANDS-ON ULTRASOUND IF NECESSARY.

Laurent Guibaud. *Prenatal Diagnosis* 2009, Vol. 29, pp 420-433.

MOST COMMON INDICATION FOR FETAL MR – INTRACRANIAL ABNORMALITIES ON ULTRASOUND

- Mild ventriculomegaly
- Nonvisualization or abnormal appearing cavum septum pellucidum.

"ISOLATED" MILD INTRACRANIAL VENTRICULOMEGALY

- Definition – lateral ventricles 10-12 mm, normal karyotype, no extracranial ultrasound abnormalities including normal fetal cardiac.
- MR will identify additional intracranial abnormalities that are clinically significant in approximately 10% of cases.

PATIENT COUNSELING FOR ISOLATED MILD VENTRICULOMEGALY

Definition of isolated mild ventriculomegaly:
10 -12 mm; versus 10-14 mm.

“Isolated” implies normal karyotype including microarray, normal fetal cardiac, no other malformations, non-progressive on follow up ultrasound.

PATIENT COUNSELING FOR MILD ISOLATED VENTRICULOMEGALY

When truly isolated, an optimistic prognosis can be provided. Outcome studies vary in their length of followup and in the type of neurodevelopmental and cognitive testing employed.

Less than 10% incidence of usually mild neurodevelopmental delays and/or learning challenges that are usually treated with early intervention.

PATIENT COUNSELING FOR ISOLATED MILD VENTRICULOMEGALY

Some authors feel better neurodevelopmental outcome in male than female fetuses

Better outcome in unilateral than bilateral mild isolated ventriculomegaly.

HISTORY: 22-week gestation with outside ultrasound demonstrating mild ventriculomegaly.

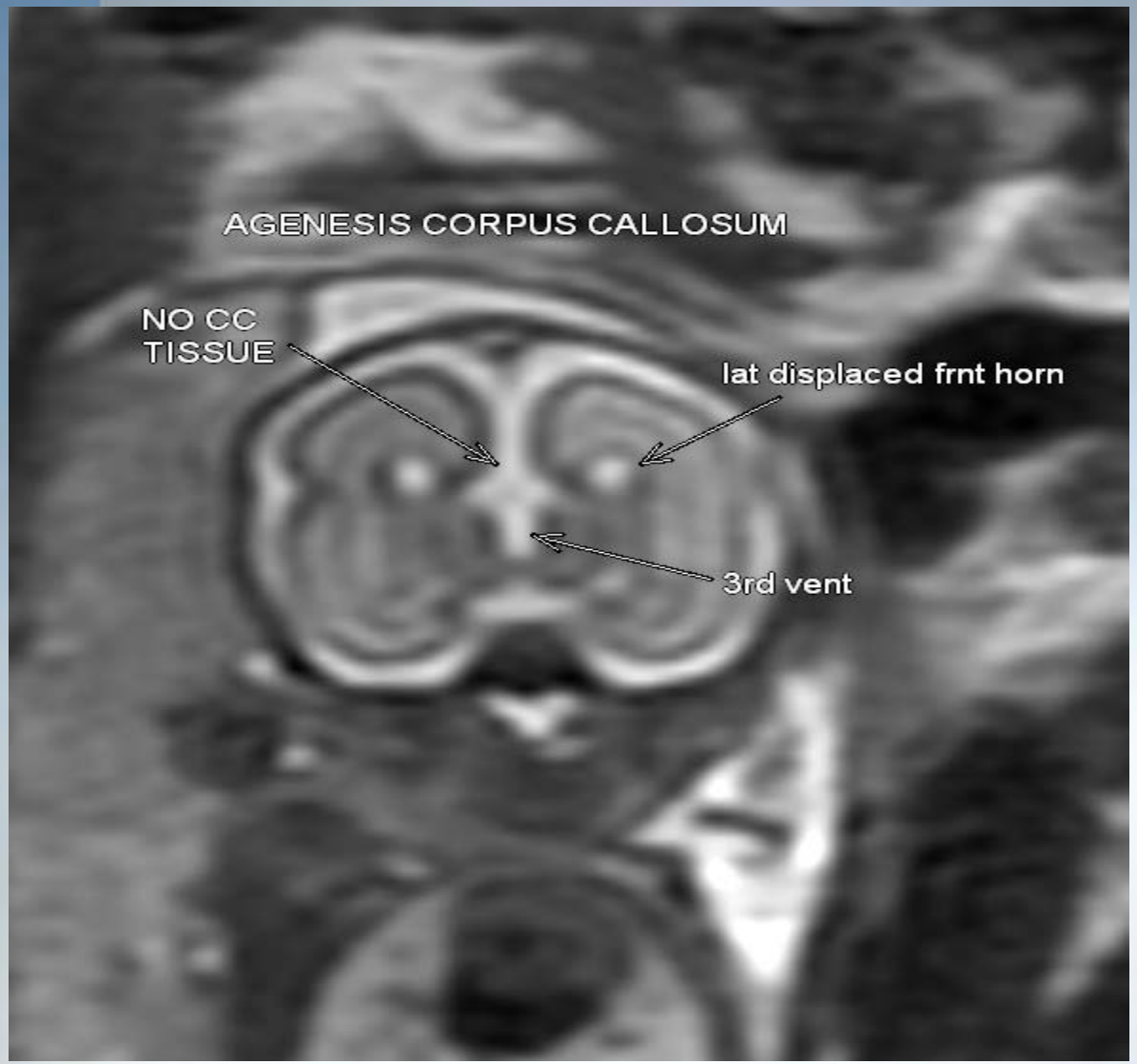
MR FINDINGS:

Complete agenesis of the corpus callosum and a schizencephalic defect – a malformation of cortical development (MCD).

TEACHING POINT:

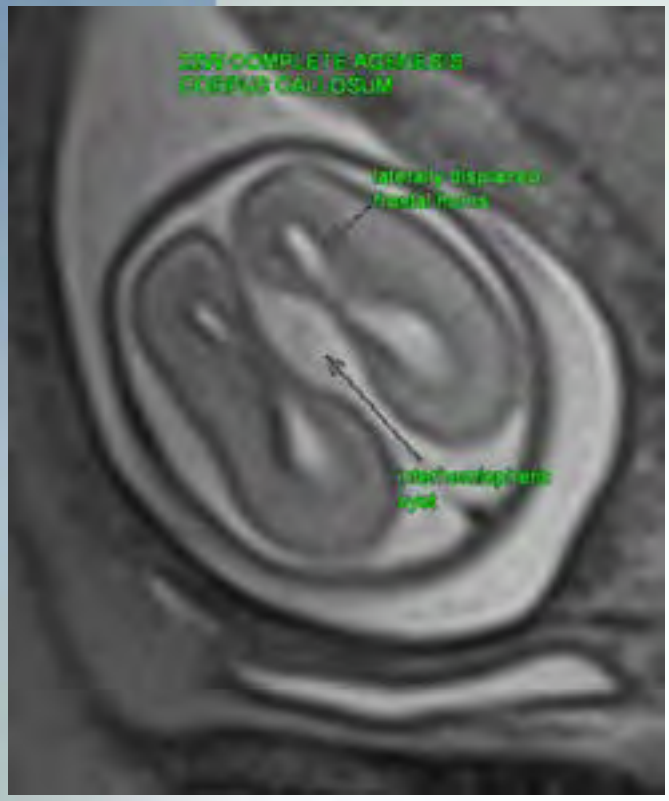
MR uniquely images MCD, conferring additional poor prognosis.

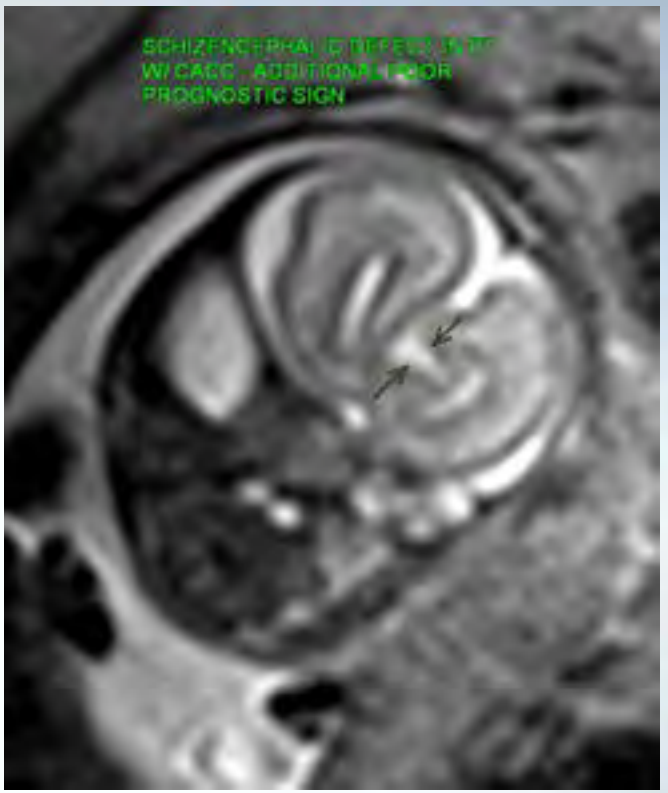
No Normal CSP - 2nd Most Common Indication



MR demonstrates pathology unable to be seen on ultrasound.

Agenesis of corpus callosum with schizencephalic defect – additional poor prognostic sign





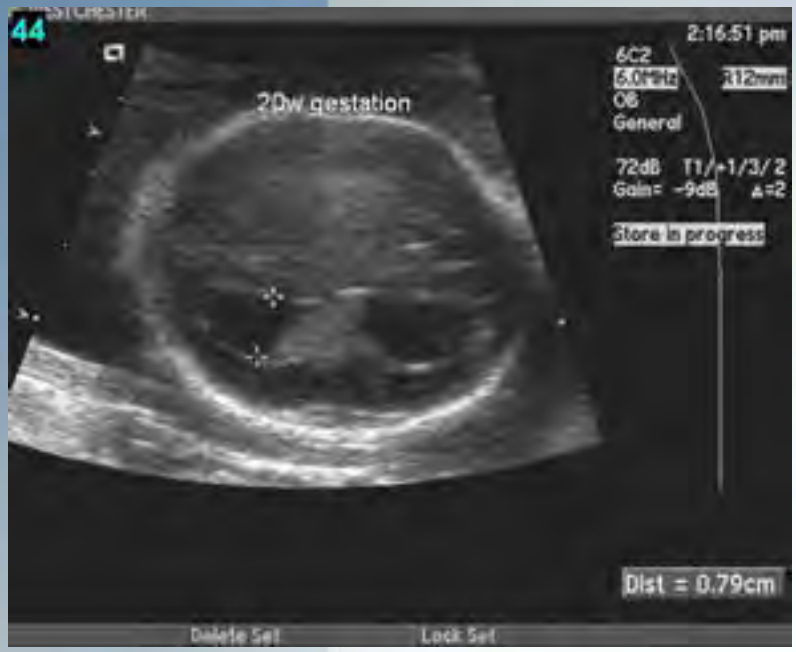
Intracranial Bleed Not Suspected on MFM Ultrasound

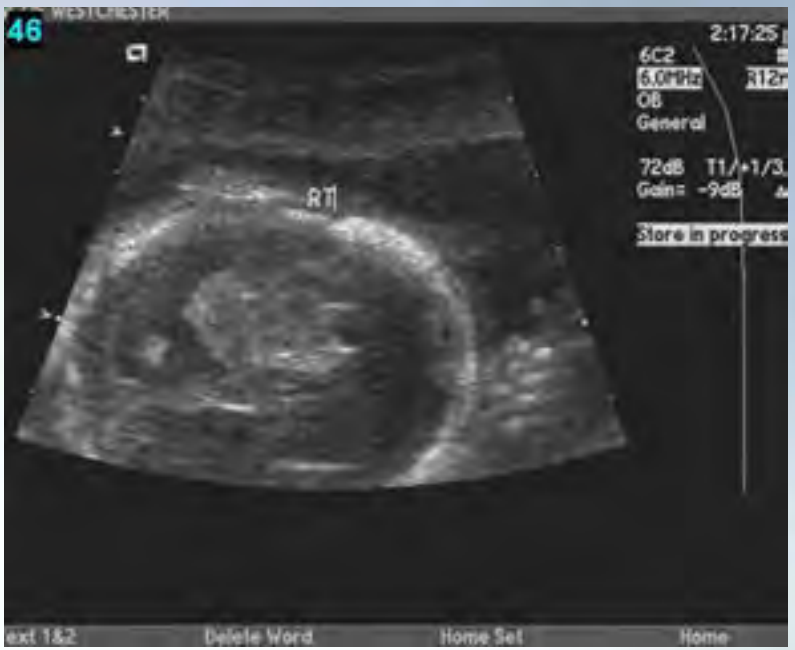
HISTORY: 20-week gestation with outside ultrasound studies describing prominent ventricles and single umbilical artery.

MR FINDINGS: Grade III germinal matrix bleed with diffusion pulse sequence demonstrating cerebral cytoxic edema.

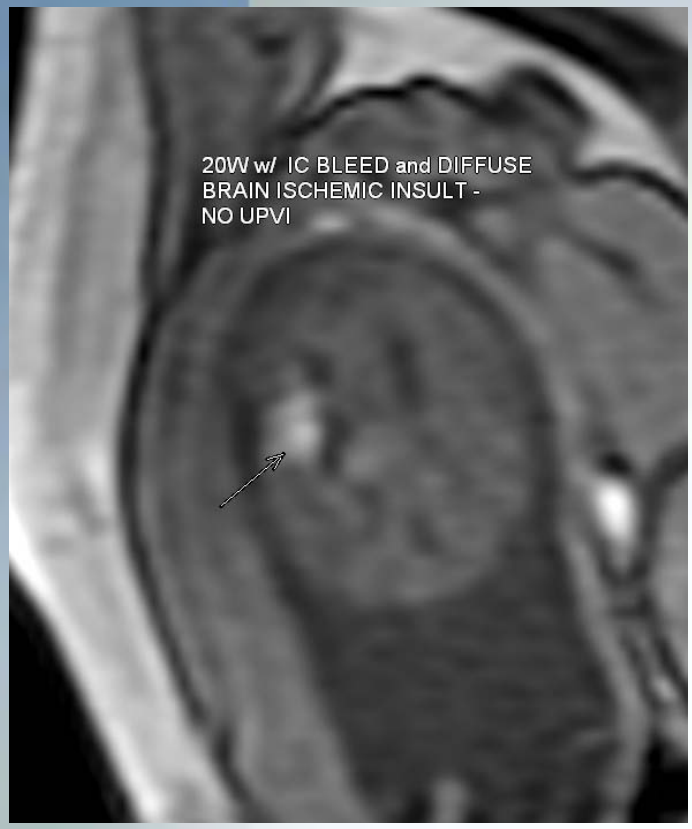
OUTCOME: One-month follow up ultrasound demonstrated no calvarial growth consistent with microcephaly -profoundly poor prognosis.

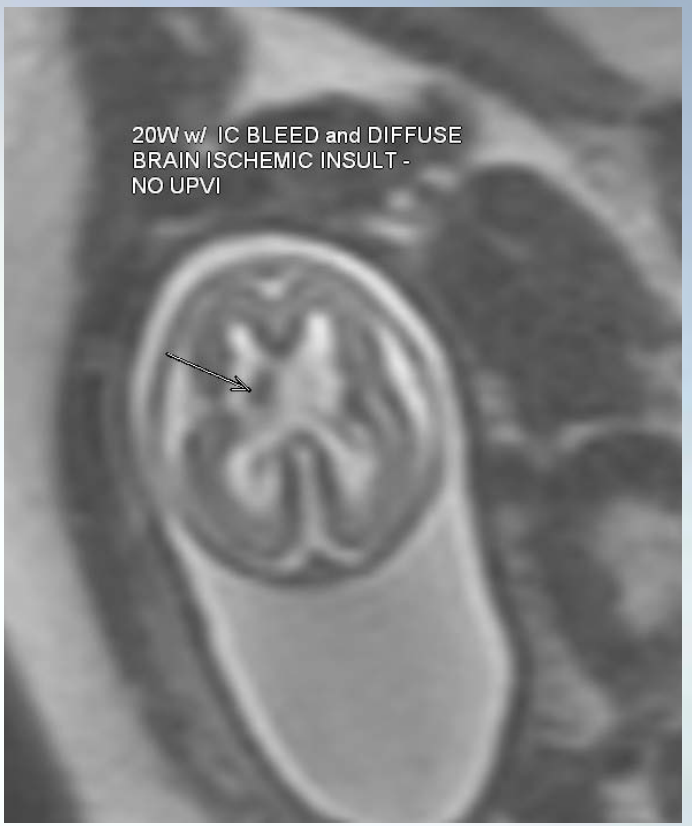
Intracranial Bleed Not Seen on MFM Ultrasound





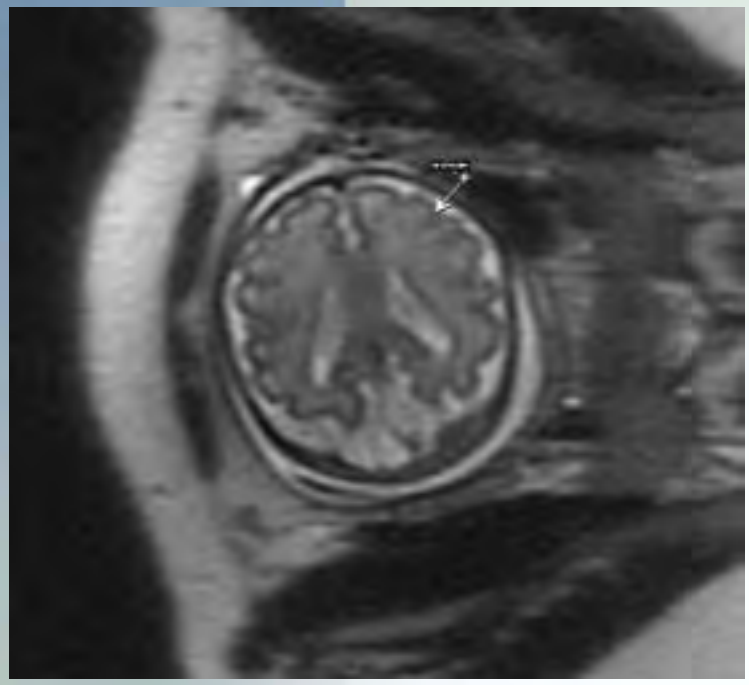
Intracranial Bleed Not Suspected on MFM Ultrasound



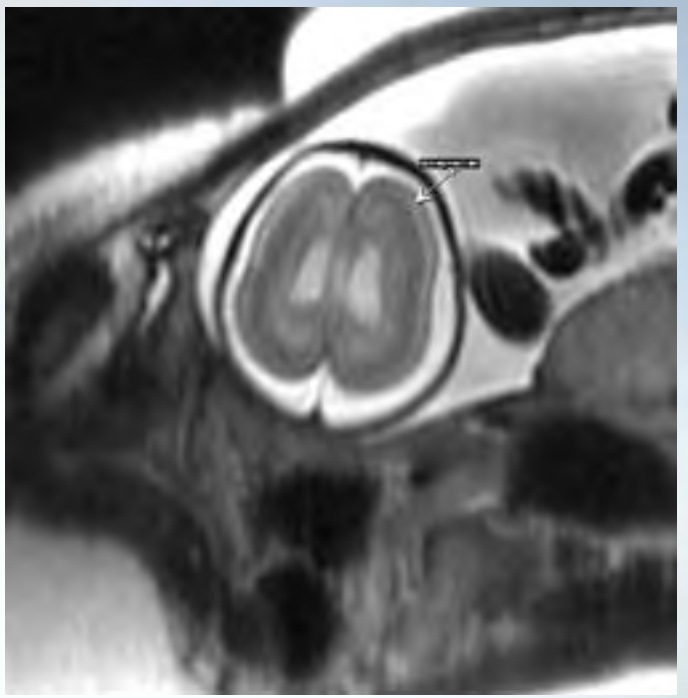


MFM ultrasound demonstrated 3rd trimester mild ventriculomegaly

MR dg = Lissencephaly
32 Weeks



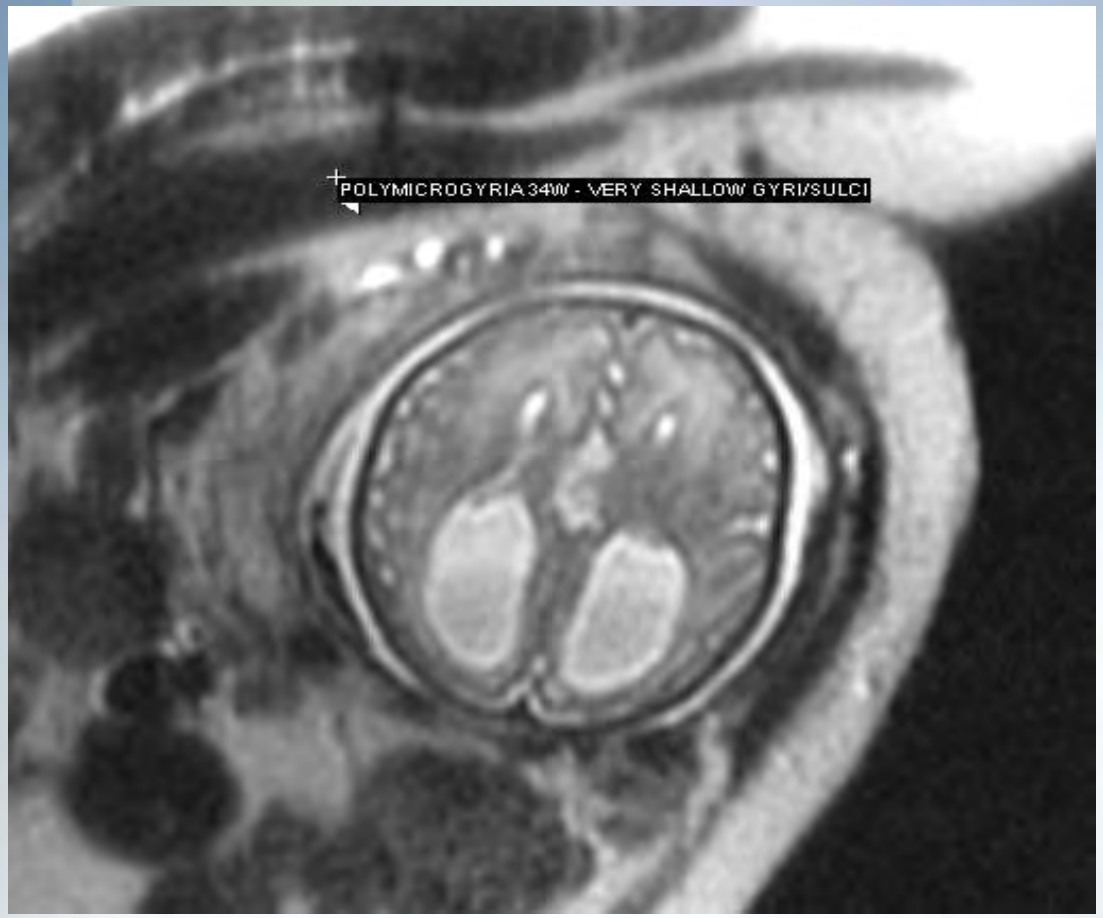
Normal gyral development



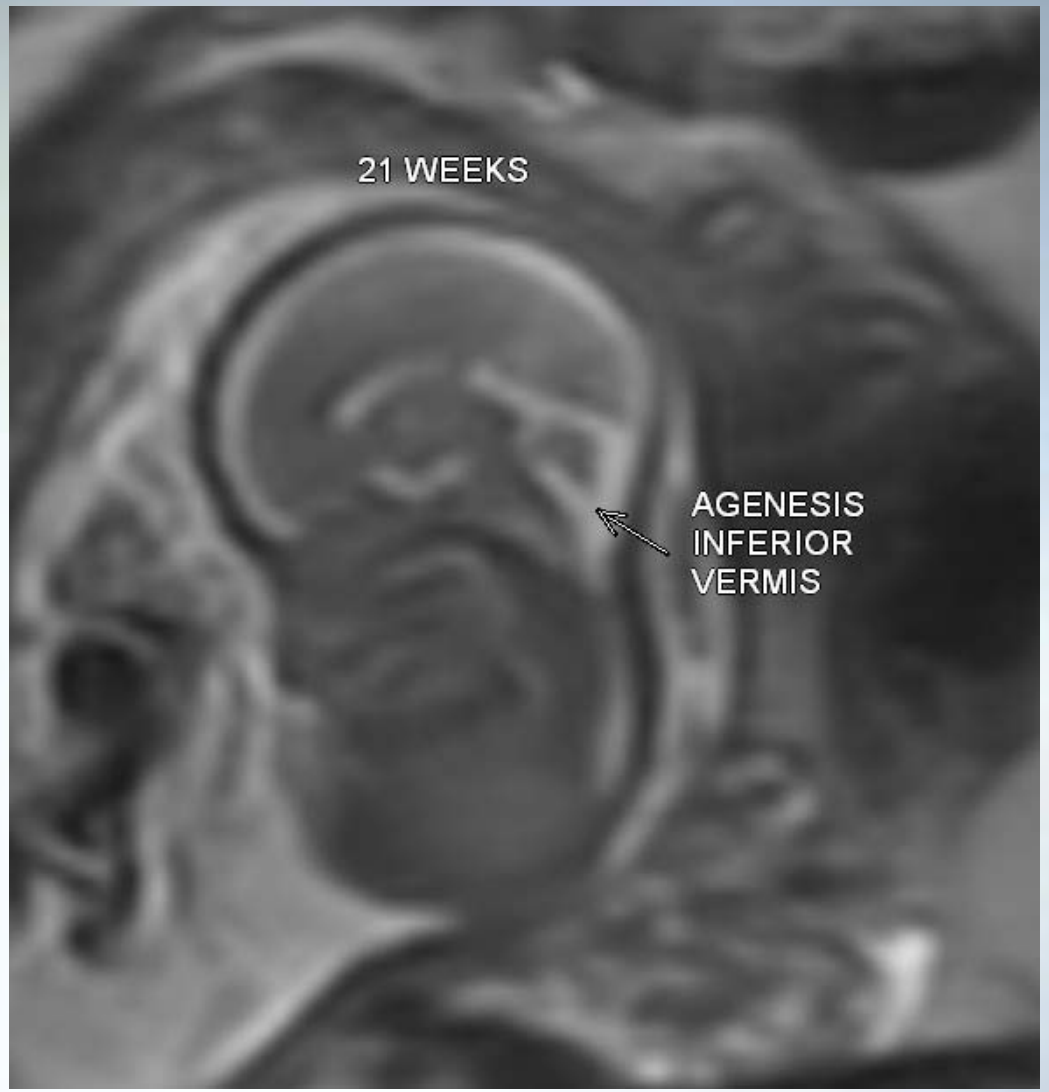
Complete agyria

MR demonstrates pathology rarely seen on ultrasound

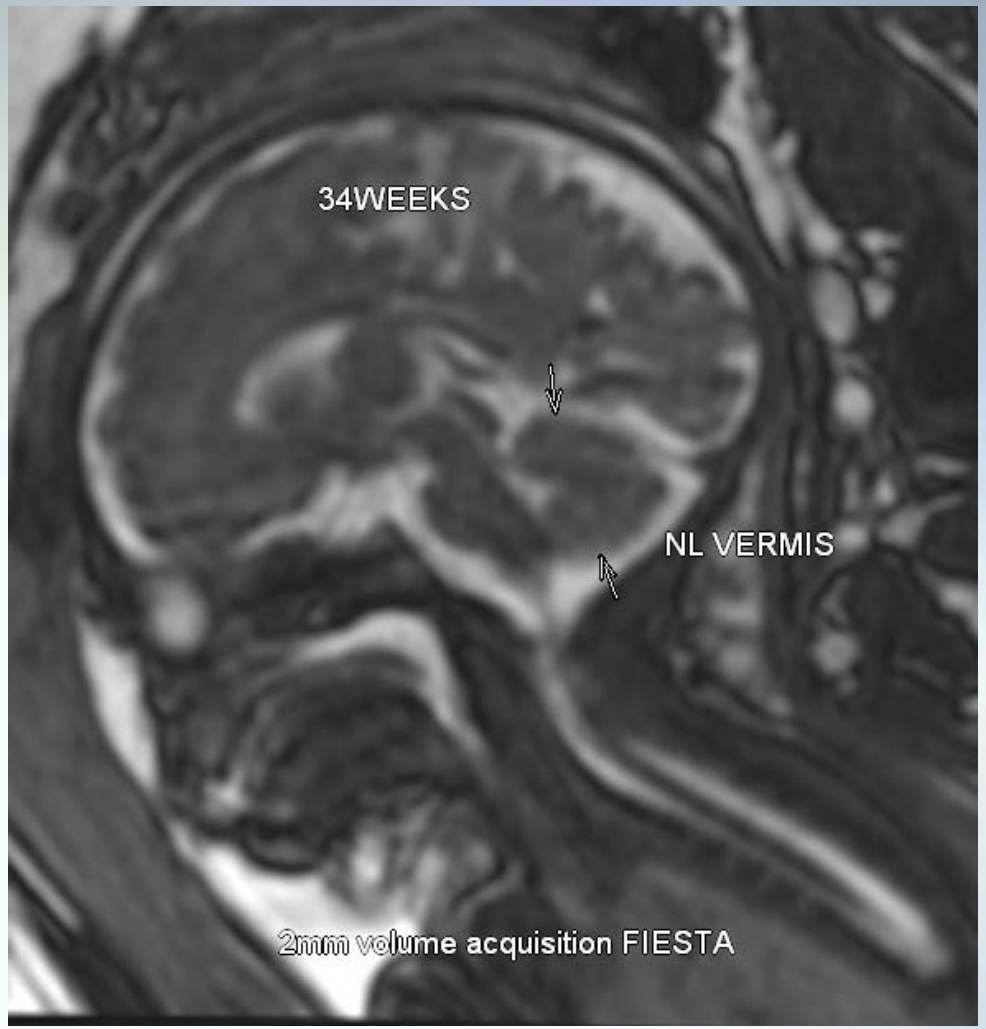
On US - 3rd TM onset of mild ventriculomegaly
MR dg = polymicrogyria



US dg – enlarged Cisterna Magna, R/O DWM
MR dg = NL Variant Agenesis Inferior Vermis



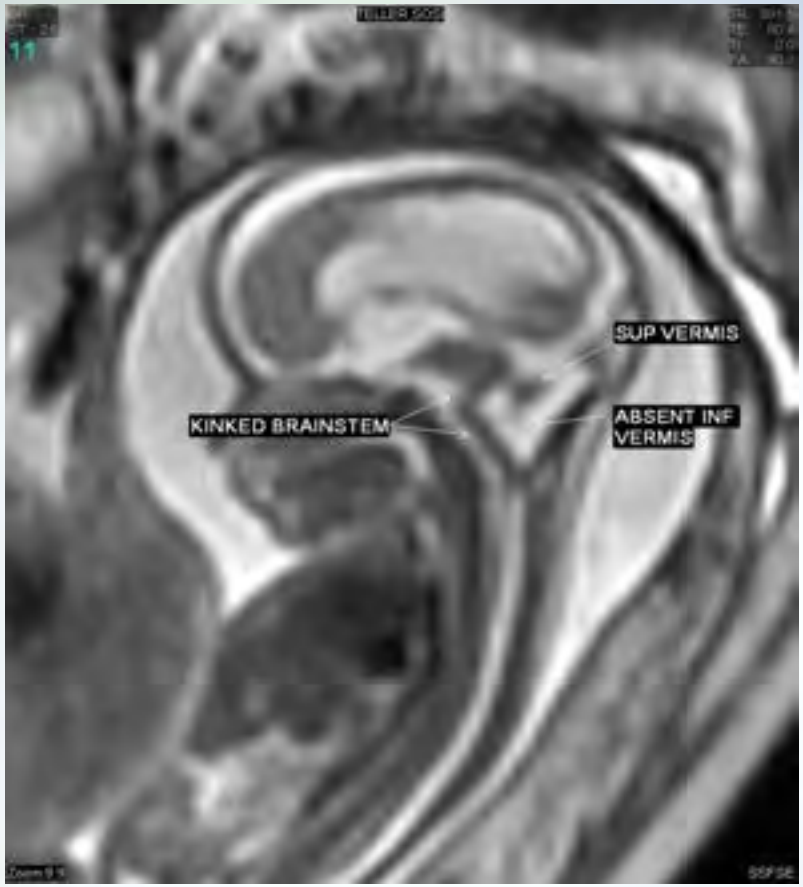
NL Variant Agenesis Inferior Vermis



MR demonstrates pathology – kinked brainstem- unable to be seen on Ultrasound

US dg - 20 w Hydrocephalus

kinked brainstem reflects late first trimester embryologic defect. Profoundly poor prognosis even if successfully shunted.



History:

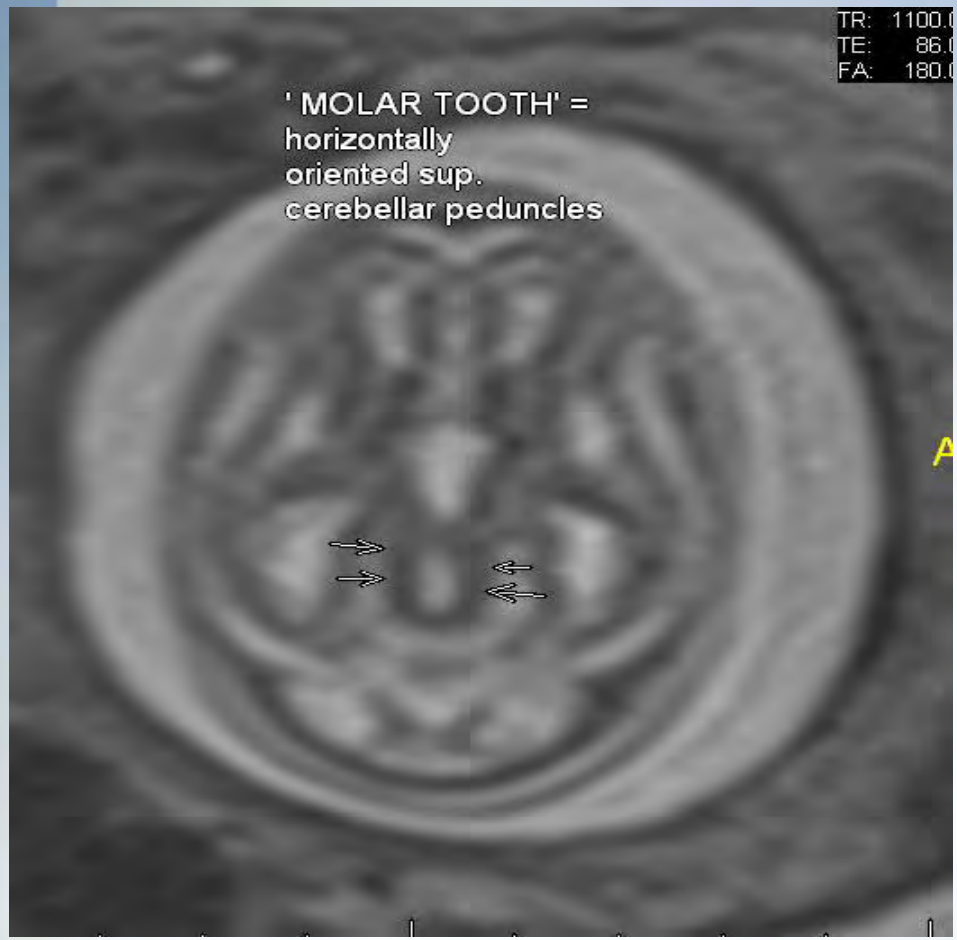
21 w GA whose MFM outside of ultrasound study demonstrated BPD and HC 12-14 days smaller than expected (2nd standard deviation) with non normalization of CSP, SUA, micrognathia and abnormal posturing of the lower extremities.

MR Findings:

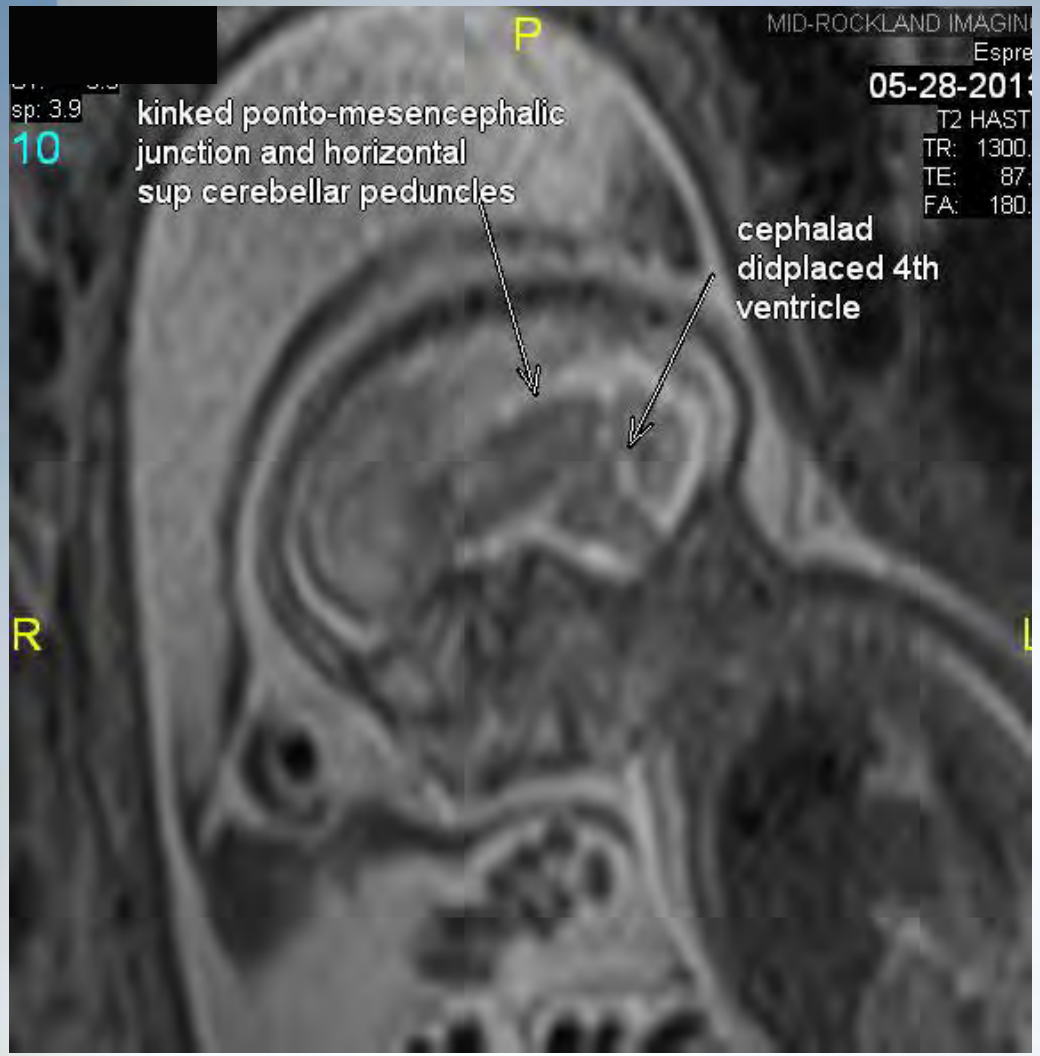
- Agenesis of the corpus callosum.
- Horizontal, thickened orientation to the superior cerebellar peduncles creating the molar tooth sign
- Cephalad displacement of the fourth ventricle with moderate brainstem kinking.
- Direct occipitofrontal brain diameter 3.5 standard deviations smaller than expected consistent with microcephaly.

MR demonstrates pathology unable to be seen on Ultrasound

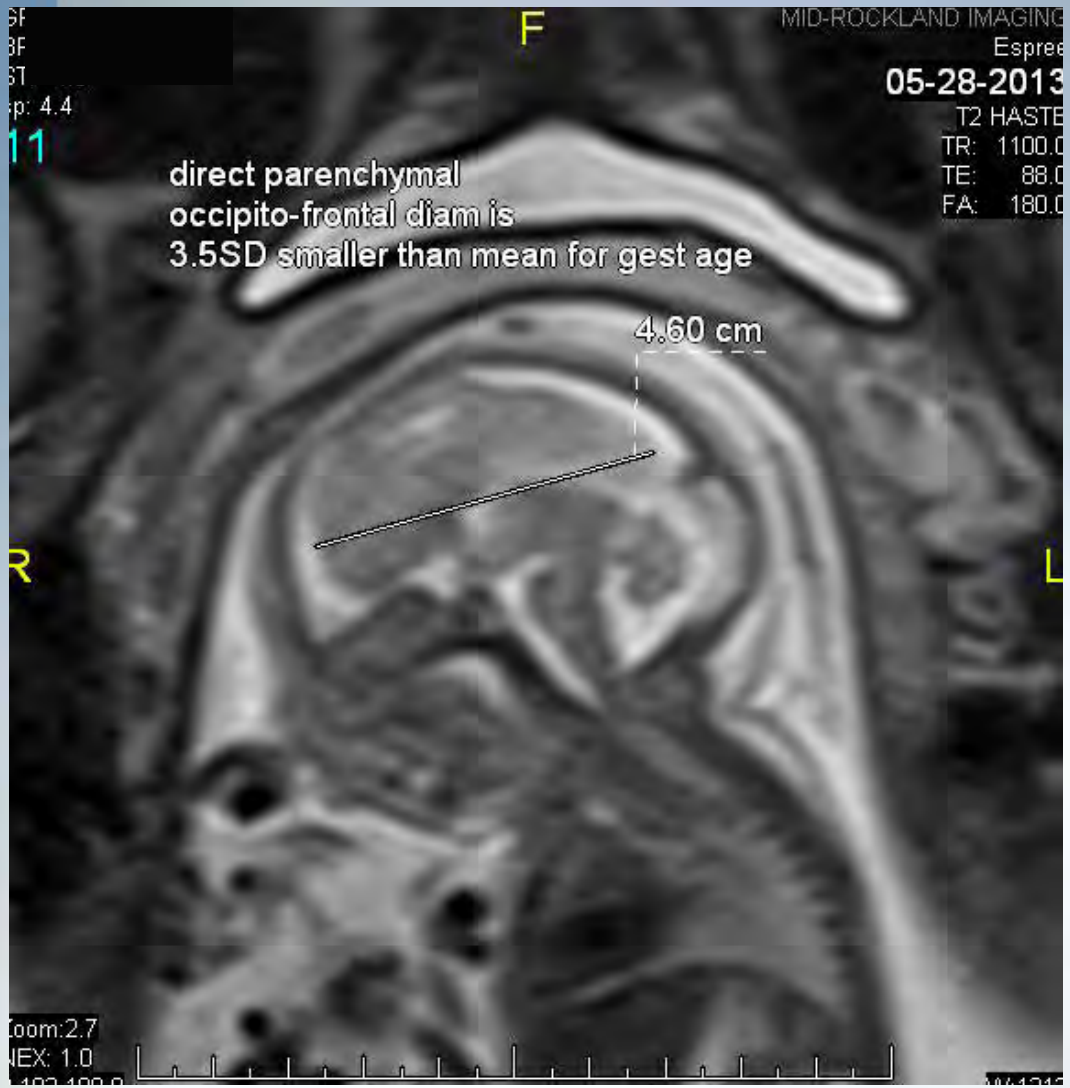
"Molar tooth"sign: ponto-mesencephalic (hindbrain-midbrain) dysmorphology



Ponto-mesencephalic dysmorphology
Not seen on ultrasound



MR images direct brain microcephaly before boney ultrasound measurements of BPD + HC



Clinical follow up: Patient terminated pregnancy. Whole -exome sequencing negative.

Teaching Point: MR's ability to diagnose multiple intracranial poor prognostic signs of types not able to be imaged on ultrasound.

Consider whole- exome sequencing in cases with multiple malformations when metaphase cytogenetics and microarray are negative.

* Clinical Whole –exome Sequencing for the Diagnosis of Mendelian Disorders. NEJM Oct 2, 2013. Yang -

Abstract Conclusion: Whole –exome sequencing identified the underlying genetic defect in 25% of consecutive patients referred for evaluation of a possible genetic condition.

HISTORY: 23-week gestation with outside ultrasound identifying cerebellar hypoplasia. Normal karyotype. Isolated cerebellar hypoplasia is worrisome but not specific for bad outcome.

FINDINGS: MR identifies not only cerebellar but also significant pontine hypoplasia. Direct cerebral parenchymal biometry is at the fifth percentile.

MR IMAGING DIAGNOSIS:

Hypoplasia not only of the posterior fossa anatomy but also of the cerebral hemispheres -greater specificity for poor prognosis.

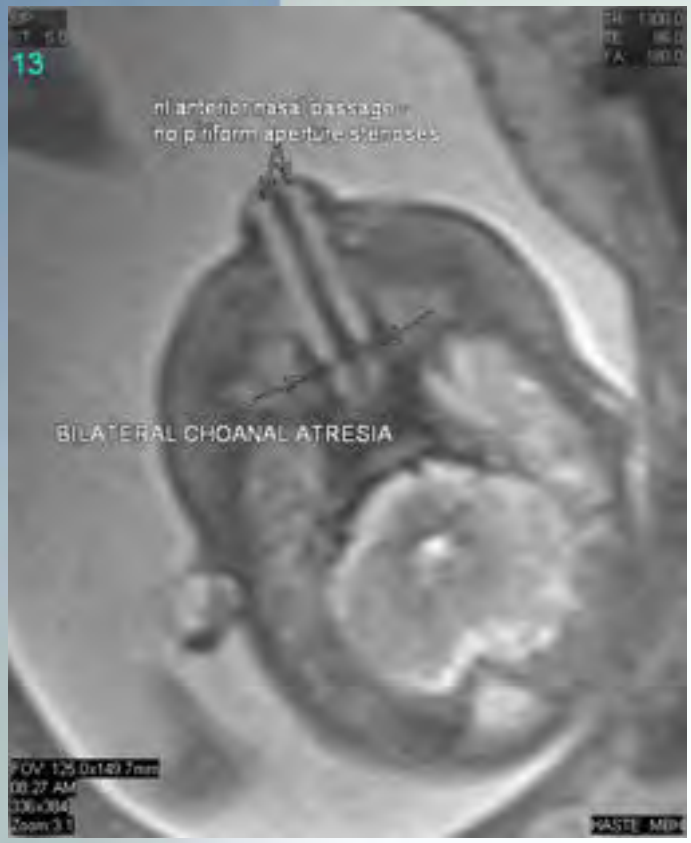
TEACHING POINTS:

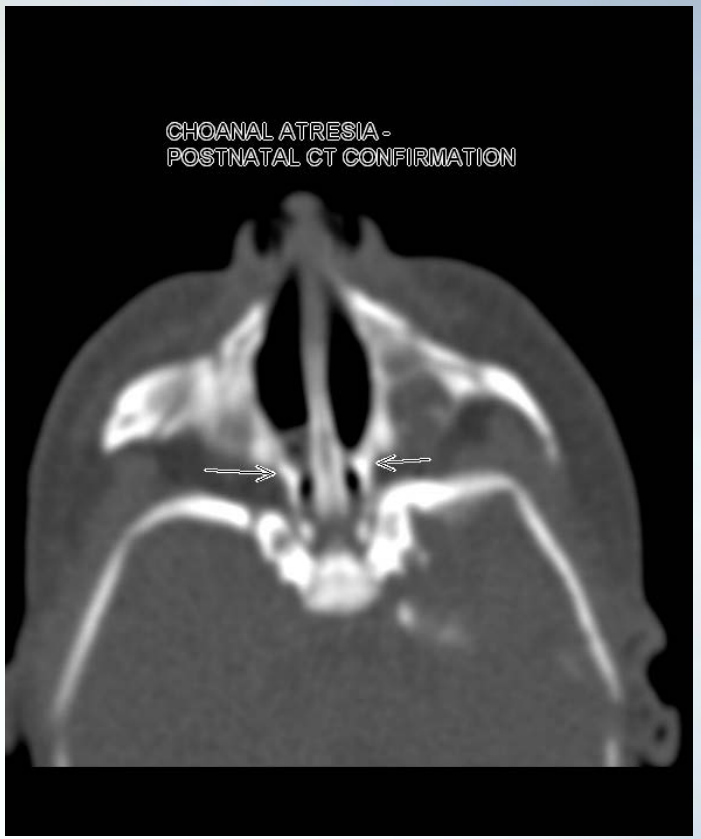
MR can provide a direct brain parenchymal measurement in the assessment of impending microcephaly prior to diminution in measurements of the bony calvarium.

MR demonstrates pathology unable to be seen on ultrasound
29w ga referred for mild ventriculomegaly

nasal passage obstruction requiring neonatal surgery

Choanal atresia. CHARGE syndrome





MR demonstrates pathology unable to be seen on ultrasound

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Nasal passage obstruction requiring neonatal surgery

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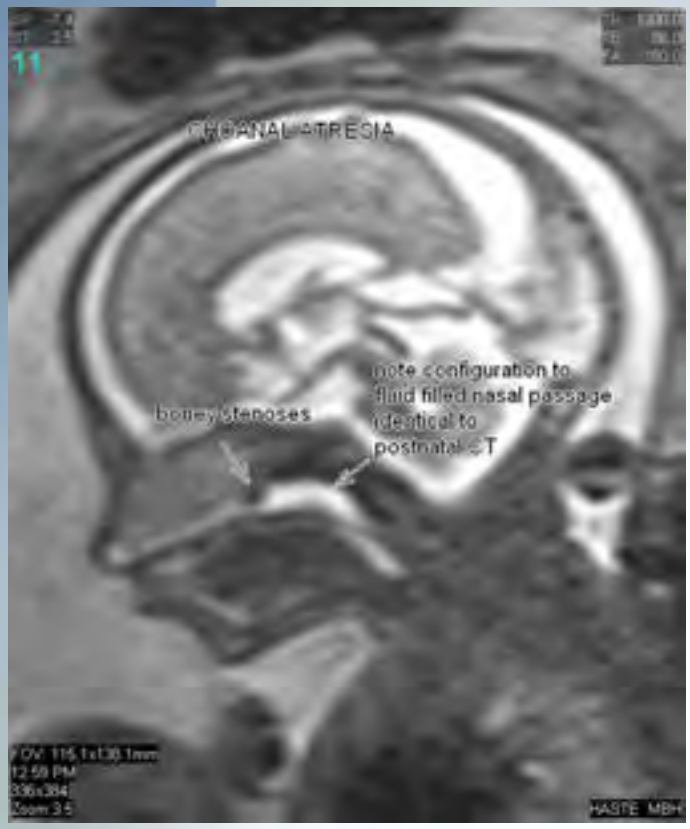
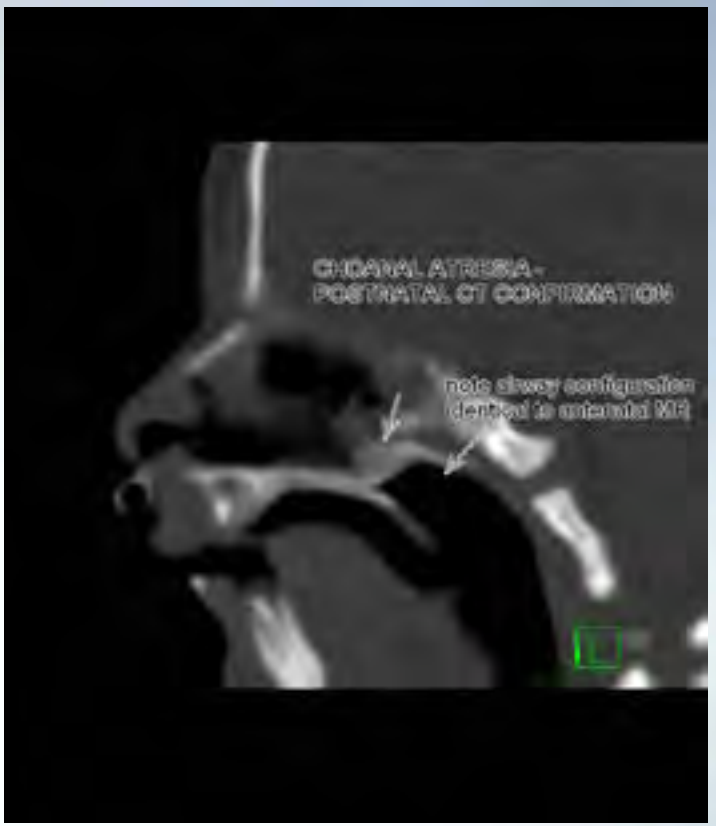




30 W Twin B referred for mild ventriculomegaly and non normalization of corpus callosum.

Final Diagnosis – Nasal passage obstruction due to *pyriform aperture stenosis and basal frontal encephalocele with dermoid* requiring intubation and tracheostomy.

HISTORY: IVF pregnancy with normal karyotype.

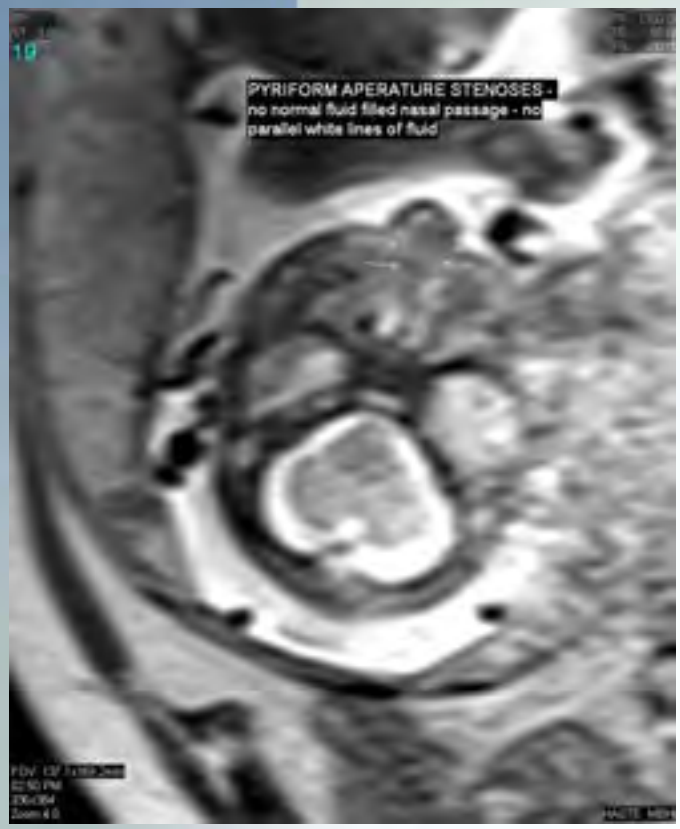
Fetal MR confirmed complete agenesis of the corpus callosum and demonstrated no other identifiable intracranial abnormalities.

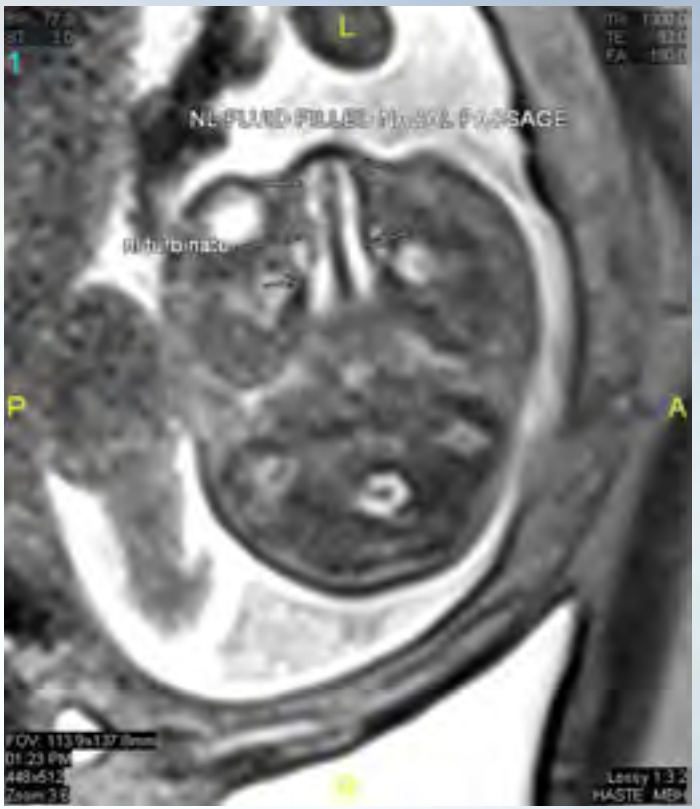
The twin with agenesis of the corpus callosum (ACC) had difficulty breathing and feeding, Requiring intubation and then tracheostomy. Postnatal MR and CT for the evaluation of upper airway issues identified two sources of nasal passage obstruction – pyriform aperture stenosis (PAS) and a small 6 x 4 mm anterior nasal encephalocele with an accompanying fatty dermoid component. Clinical exam and imaging demonstrated a large single midline maxillary incisor – a frequent accompaniment to PAS.

Pyriform aperture stenosis (PAS) creates an obstruction to the anterior aspect of the nasal passage (in comparison to choanal atresia which is obstruction to the posterior aspect of the nasal passage).

MR demonstrates pathology unable to be seen on ultrasound

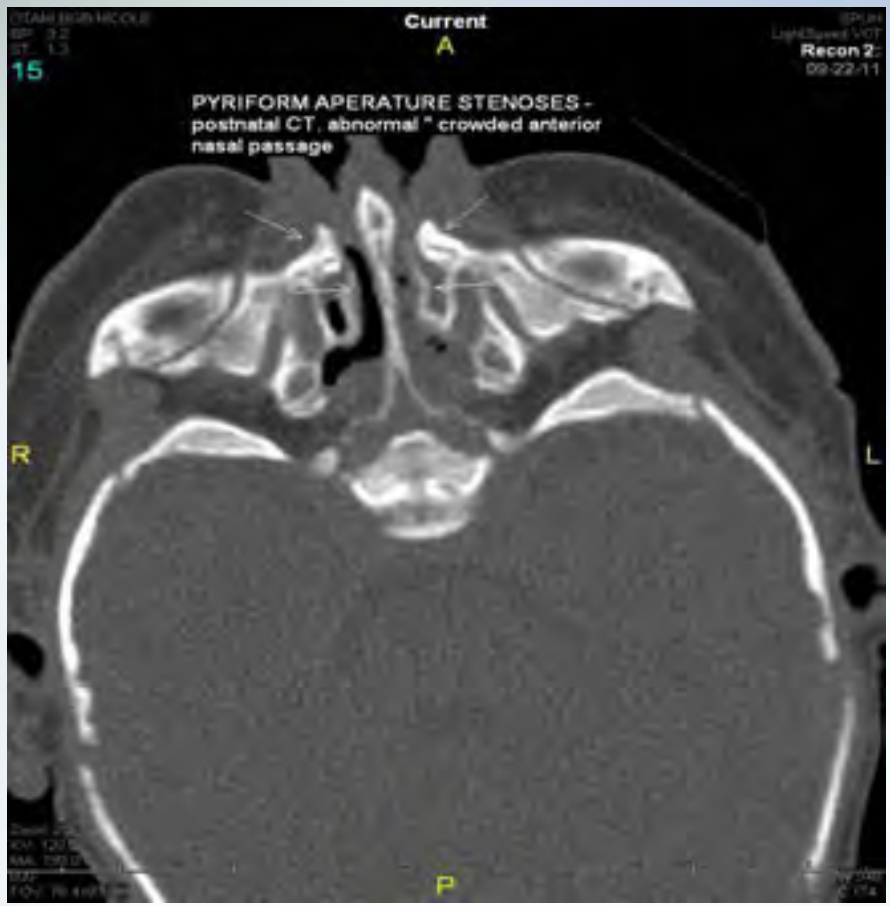
Pyriform aperture stenoses.
nasal passage obstruction requiring intubation and tracheostomy.





MR demonstrates pathology unable to be seen on ultrasound

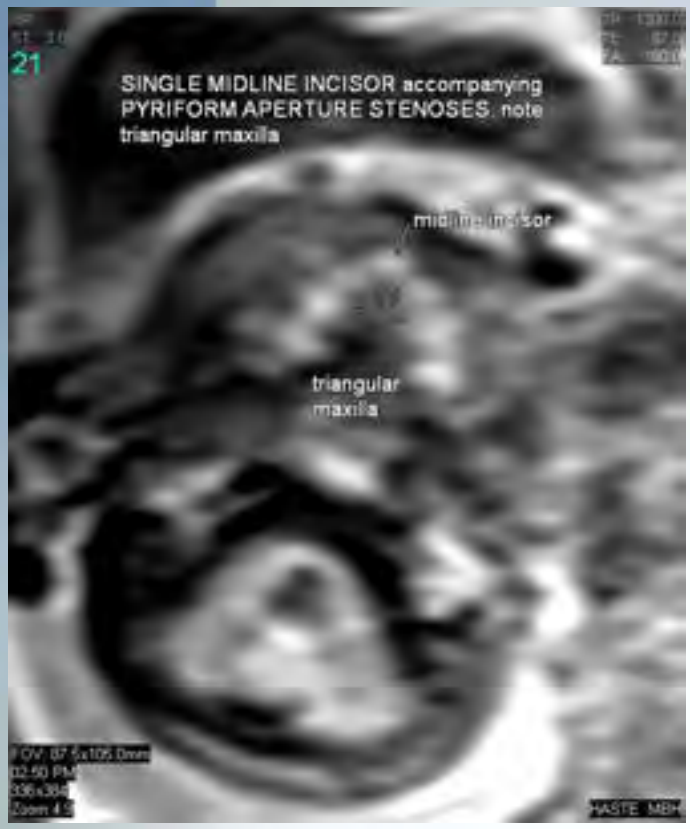
Pyriform aperture stenoses.
nasal passage obstruction requiring intubation and tracheostomy.

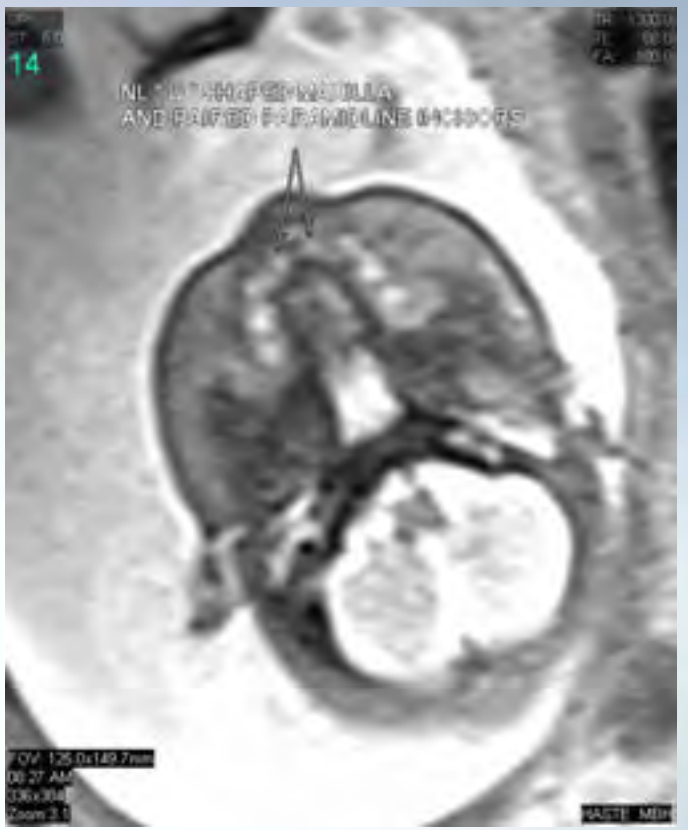


MR demonstrates pathology unable to be seen on ultrasound

Pyriform aperture stenoses with midline maxillary incisor

nasal passage obstruction requiring intubation and tracheostomy.

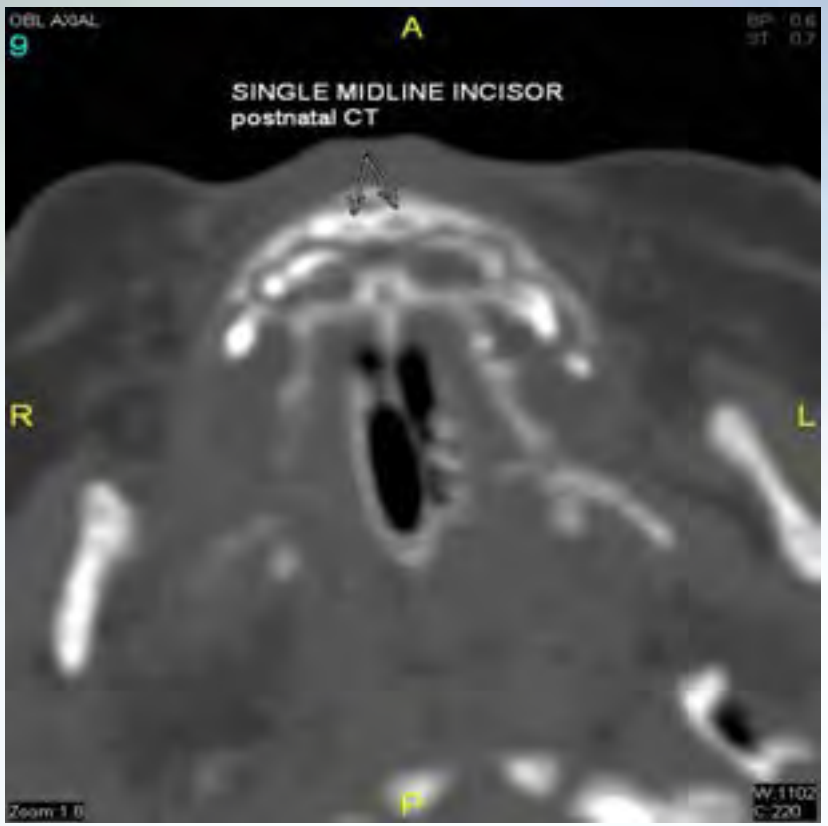




MR demonstrates pathology unable to be seen on ultrasound

Pyriform aperture stenoses with midline maxillary incisor

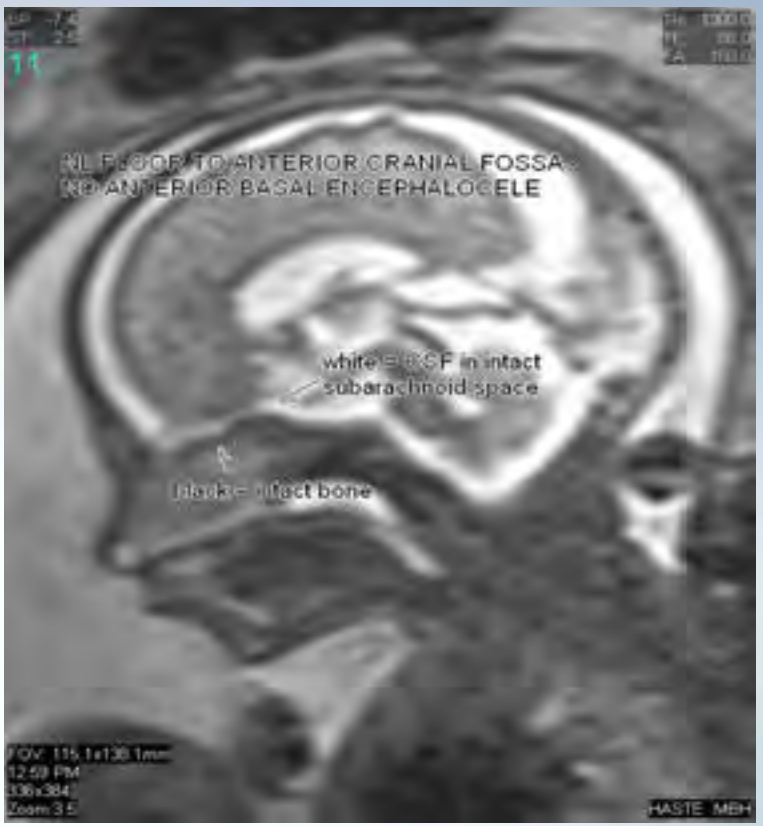
nasal passage obstruction requiring intubation and tracheostomy.



MR demonstrates pathology unable to be seen on ultrasound

Basal encephalocele with dermoid

nasal passage obstruction requiring intubation and tracheostomy.



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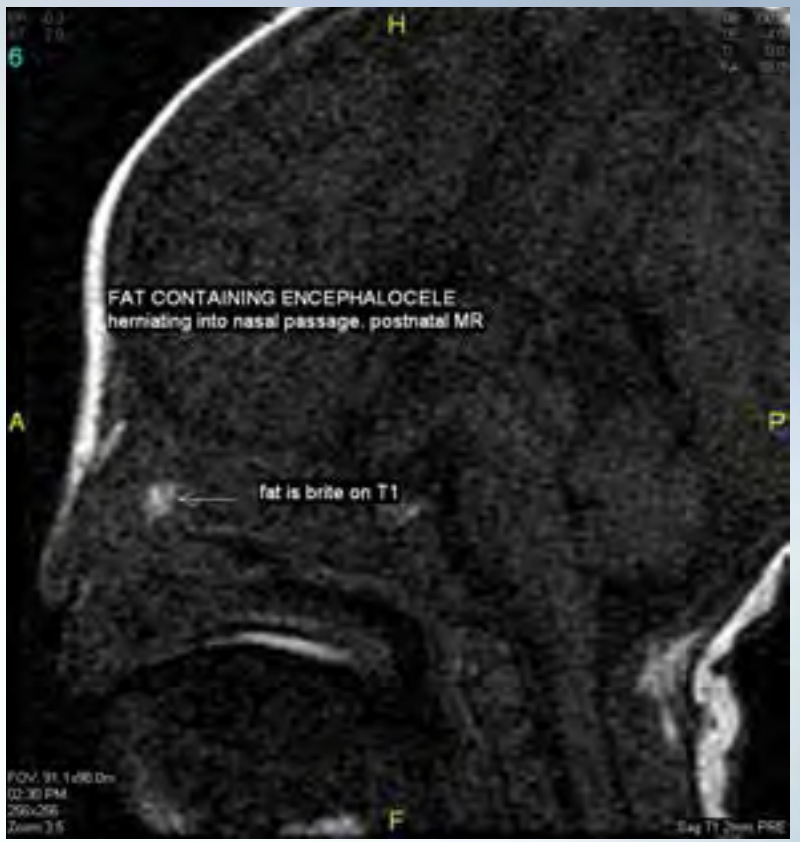
MR demonstrates pathology unable to be seen on ultrasound

Basal encephalocele with dermoid

nasal passage obstruction requiring intubation and tracheostomy.



Bilateral atresia of external auditory canal with middle ear dysplasia – trisomy 18

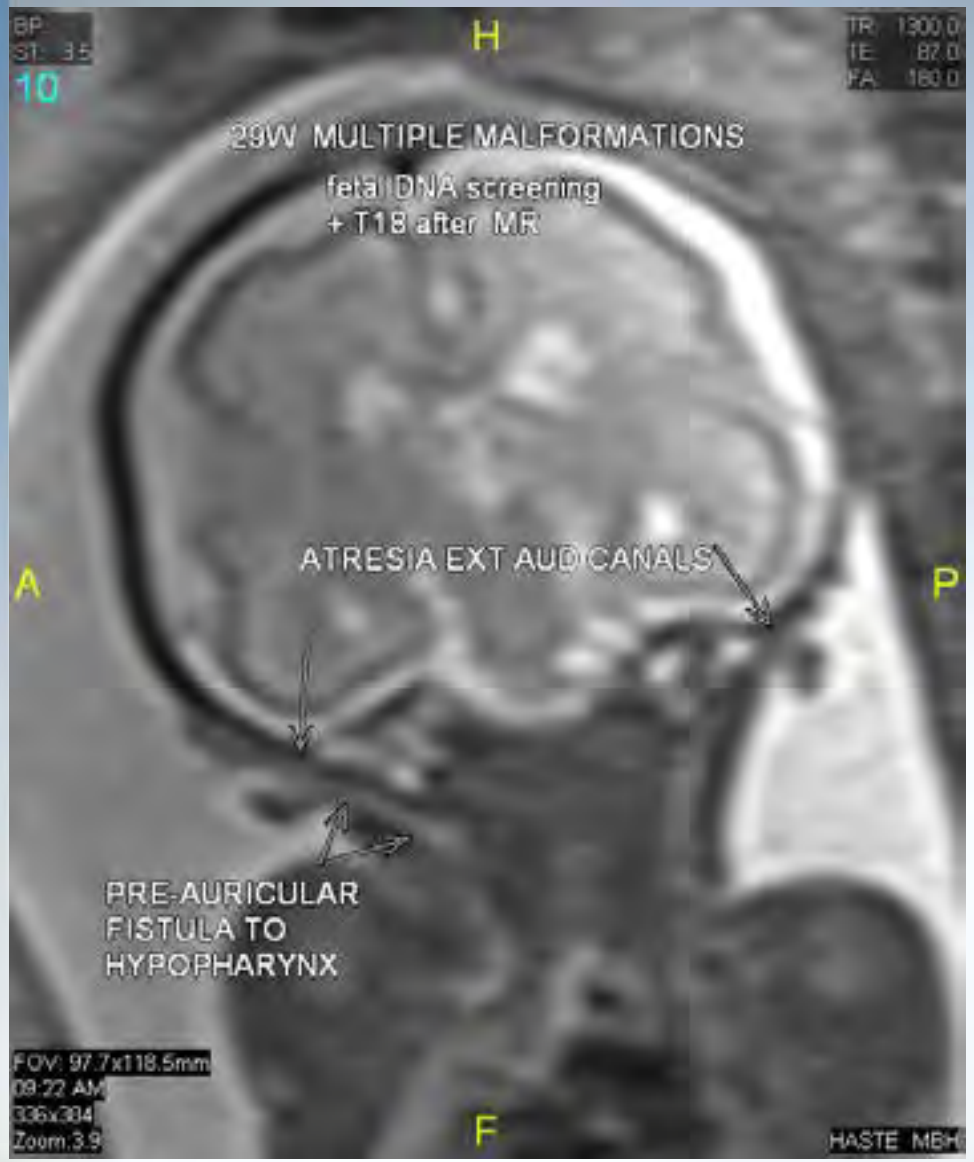
History: 29w late registrant whose outside MFM ultrasound demonstrated multiple malformations including enlarged cisterna magna, mild ventriculomegaly, abnormal hand and feet posturing and polyhydramnios. *At the time of MR, fetal DNA screening results were not known.*

MR demonstrated:

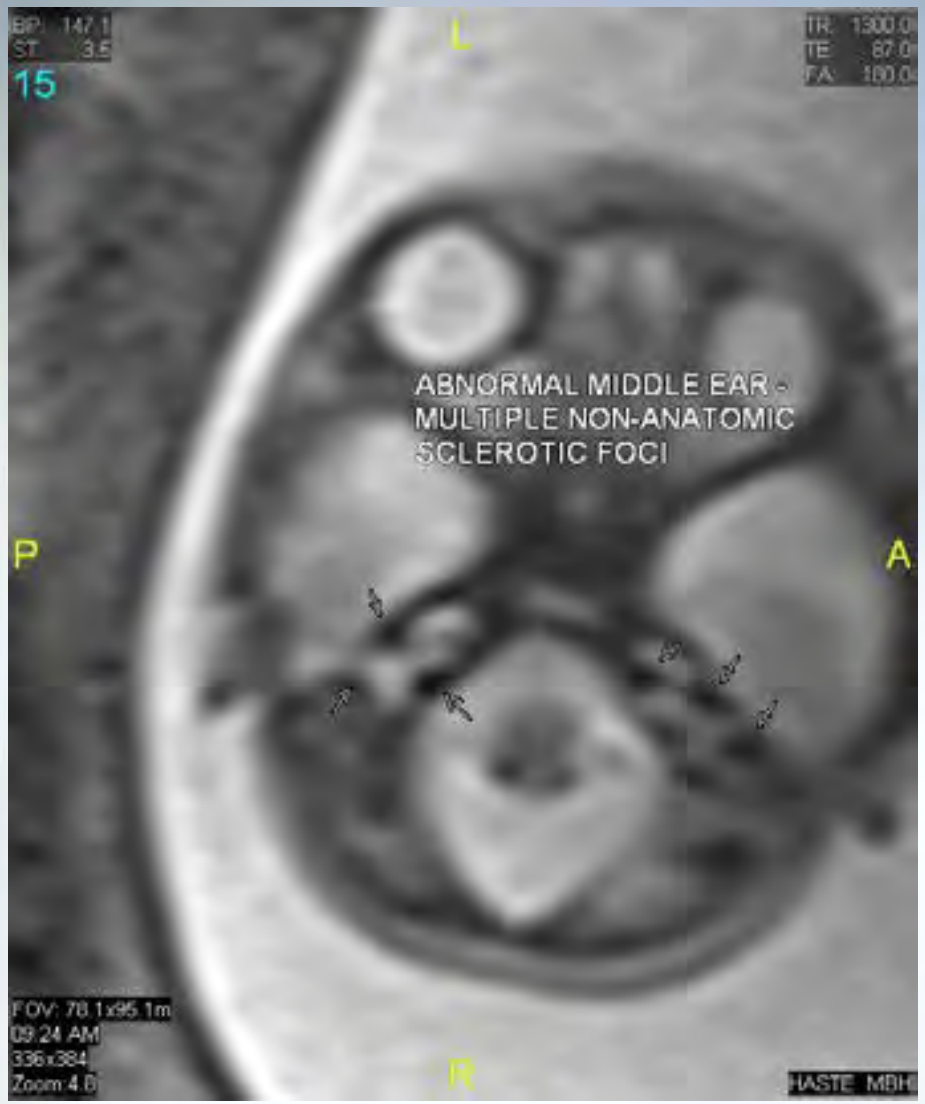
- Cerebellar and pontine hypoplasia with intact vermis. No DWM.
- Bilateral atresia of external auditory canals with bilateral preauricular fistula to hypopharynx.
- Bilateral abnormal mesotympanum with multiple non anatomic sclerotic foci.

One week after MR, fetal DNA in maternal serum screening was positive for trisomy 18.

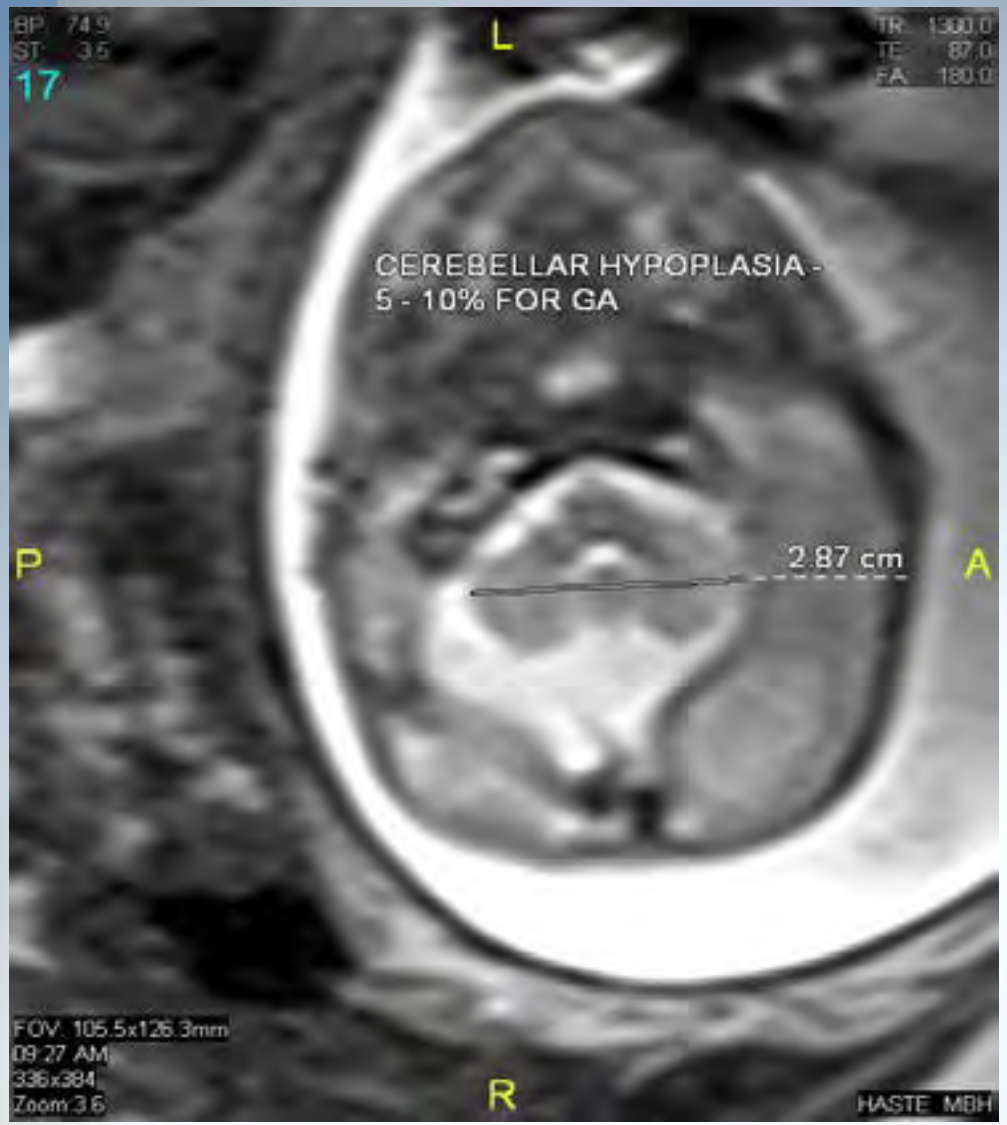
MR demonstrates malformations of the auditory anatomy unable to be seen on ultrasound



MR demonstrates auditory malformations unable to be seen on ultrasound



Enlarged Cisterna Magna on Ultrasound – MR changes dg. from DWM to cerebellar and pontine hypoplasia



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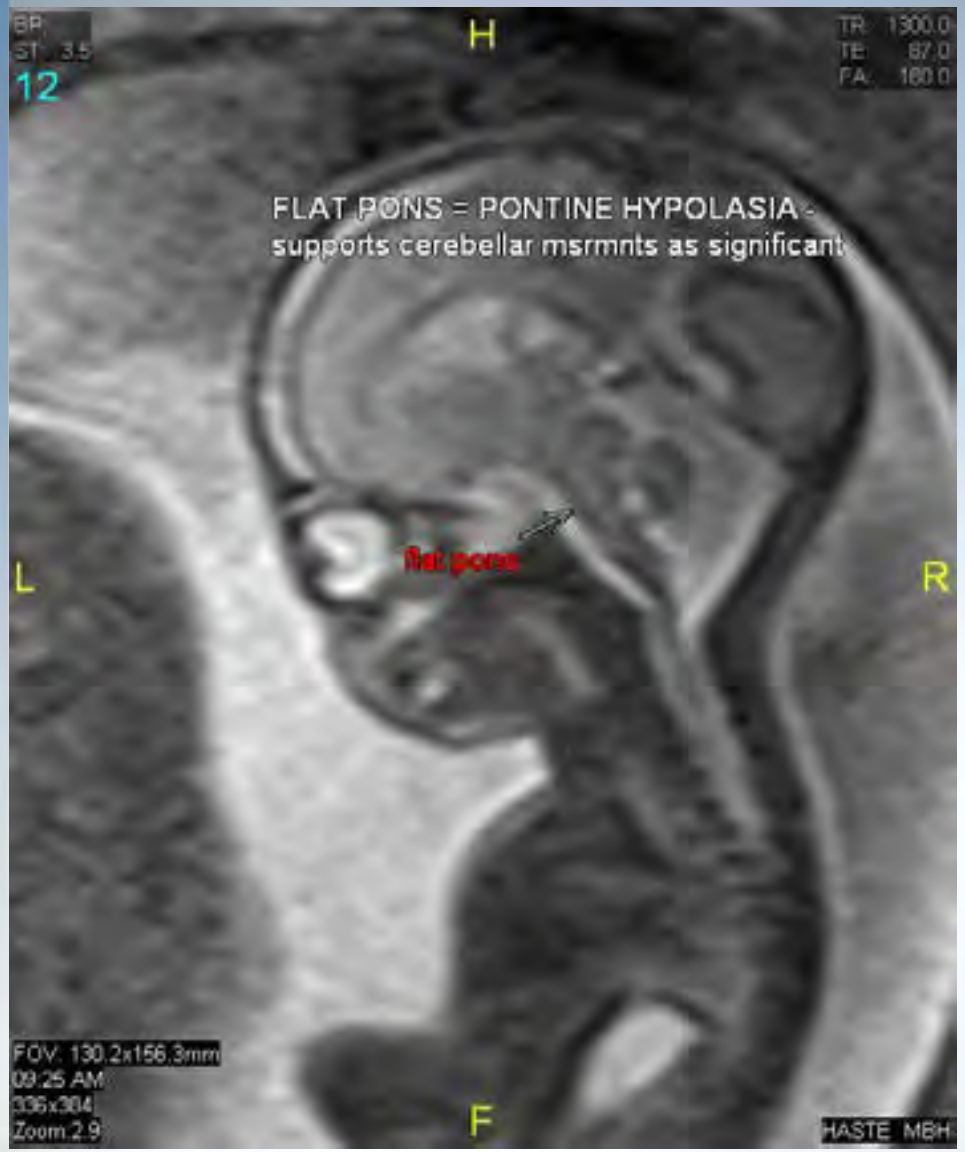
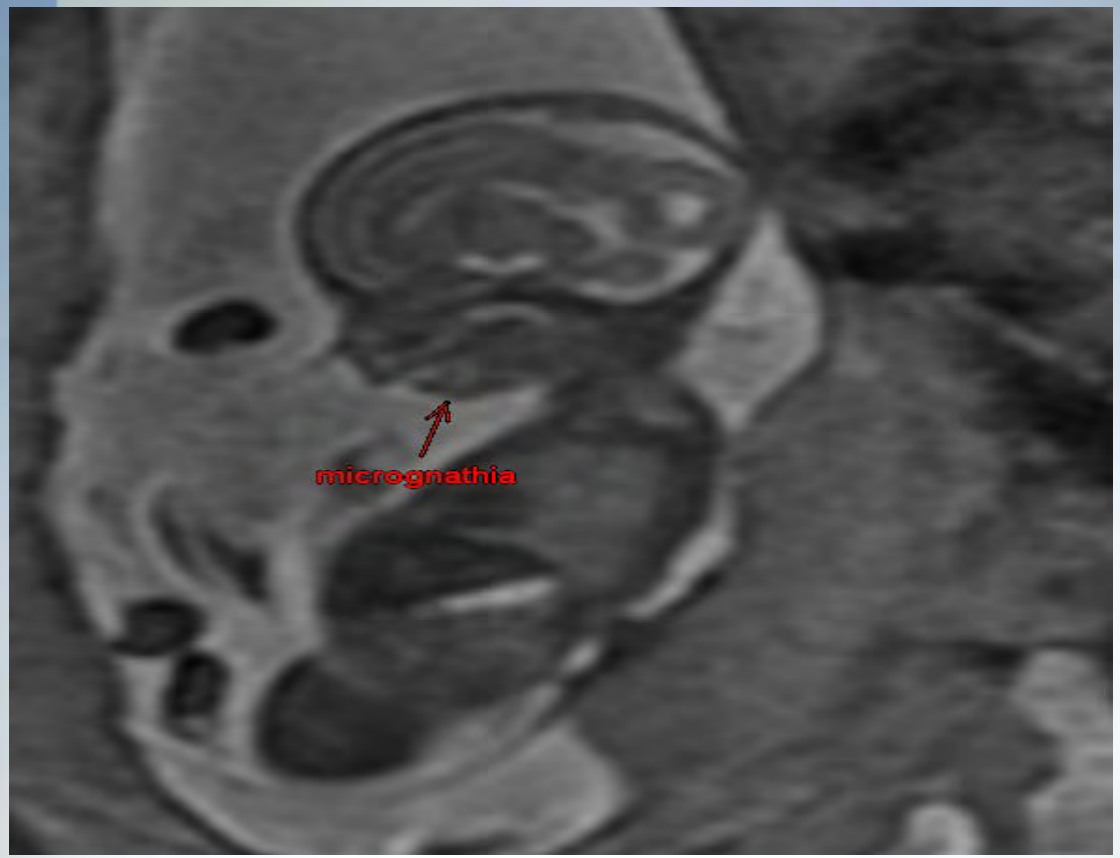


GOLDENHAR SYNDROME
MFM Ultrasound Identified an Arachnoid Cyst. MR Identifies Additional Pathologies as Clues to Syndromic Condition



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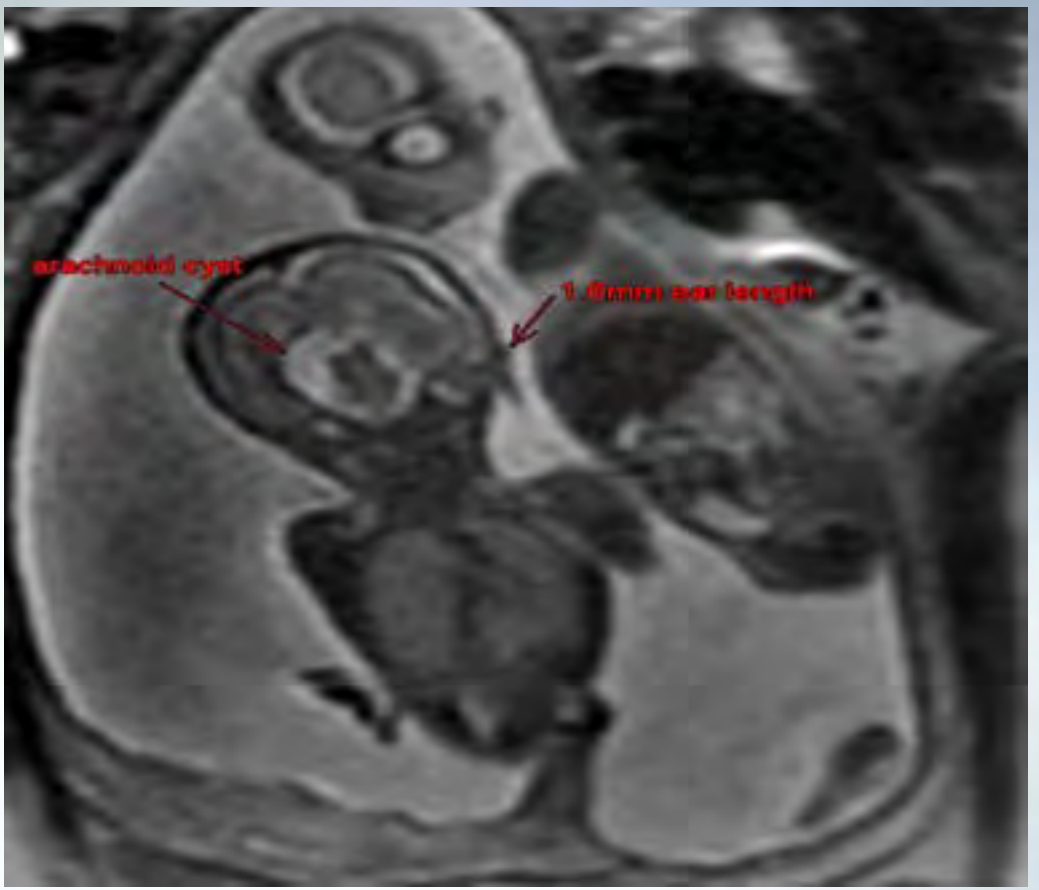
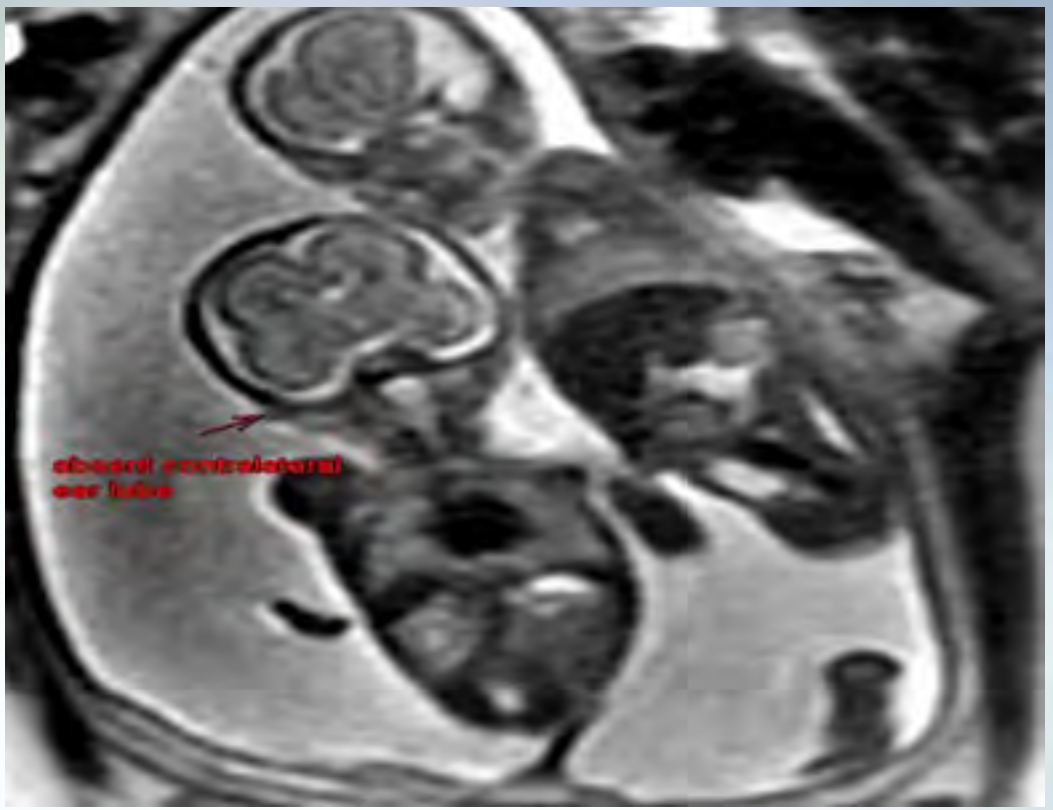


GOLDENHAR SYNDROME
Absent ear

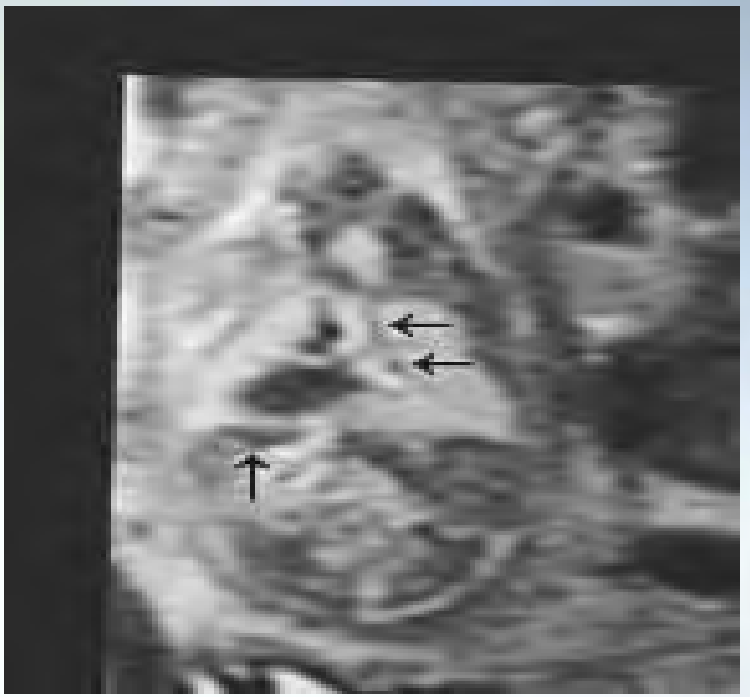


MR changes diagnoses from guarded optimism to lethal

MFM Ultrasound Diagnosed Bilateral Congenital Cystic Adenomatoid Malformation (CCAM)

MR dg = Laryngeal Atresia

Dilated trachea and major bronchi, bilateral enlarged lungs with flattened diaphragm consistent with upper airway obstruction most commonly laryngeal atresia.

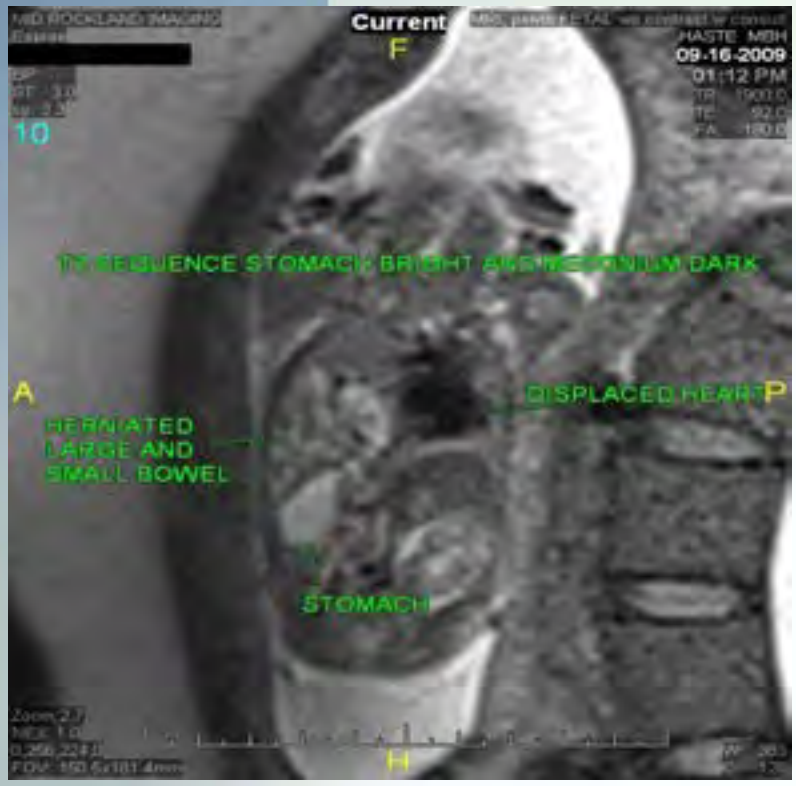


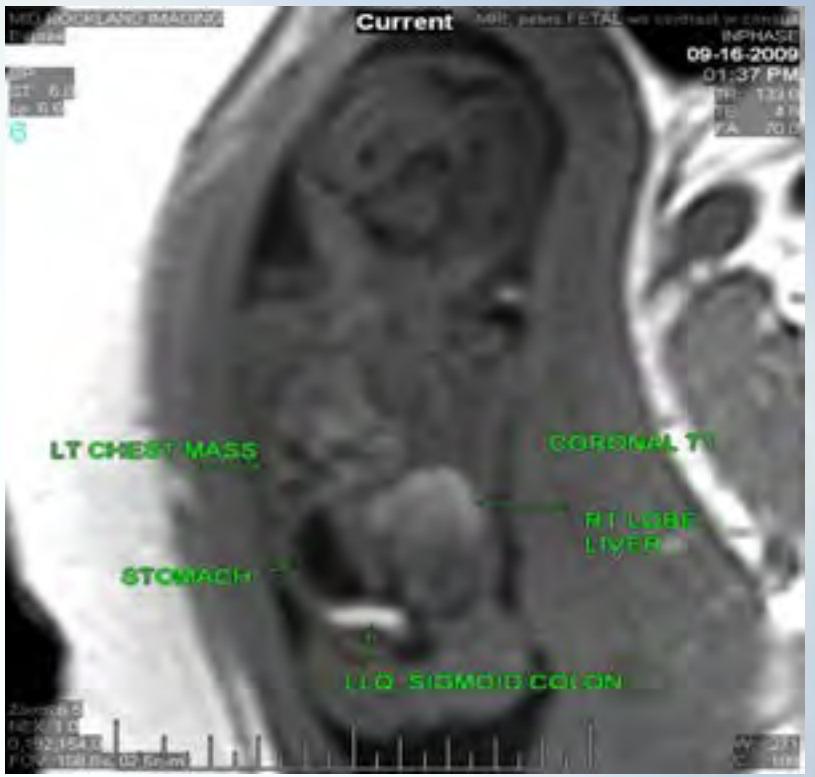
- **Teaching Point:** MR is the modality of choice for evaluating fetal chest masses. In this case MR was able to distinguish laryngeal atresia from bilateral congenital cystic adenomatoid malformations.

MR changes Ultrasound Diagnosis

HISTORY: 21-week gestation with outside ultrasound studies demonstrating rightward cardiac displacement and echogenic left thoracic mass – CCAM vs BPS

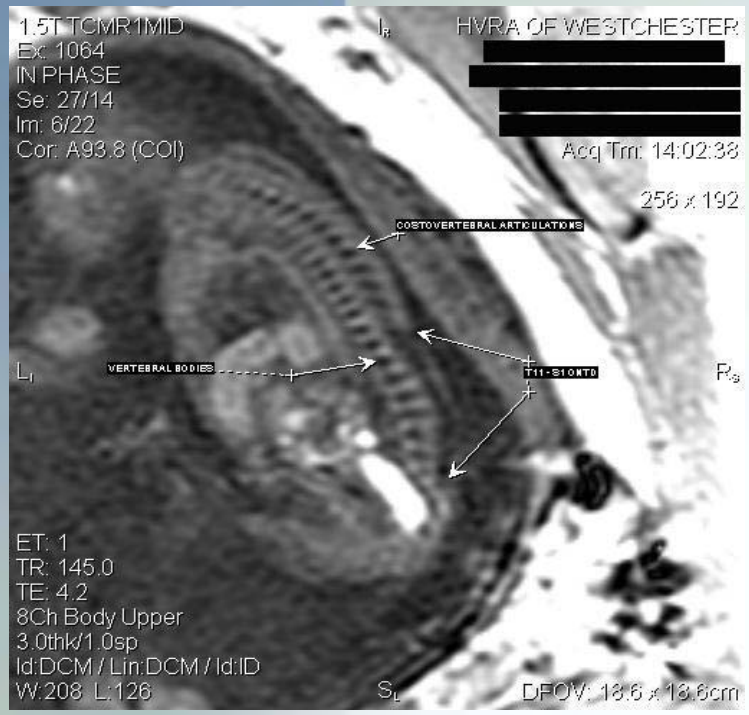
DIAGNOSIS: Left congenital diaphragmatic hernia. Intra- abdominal stomach.

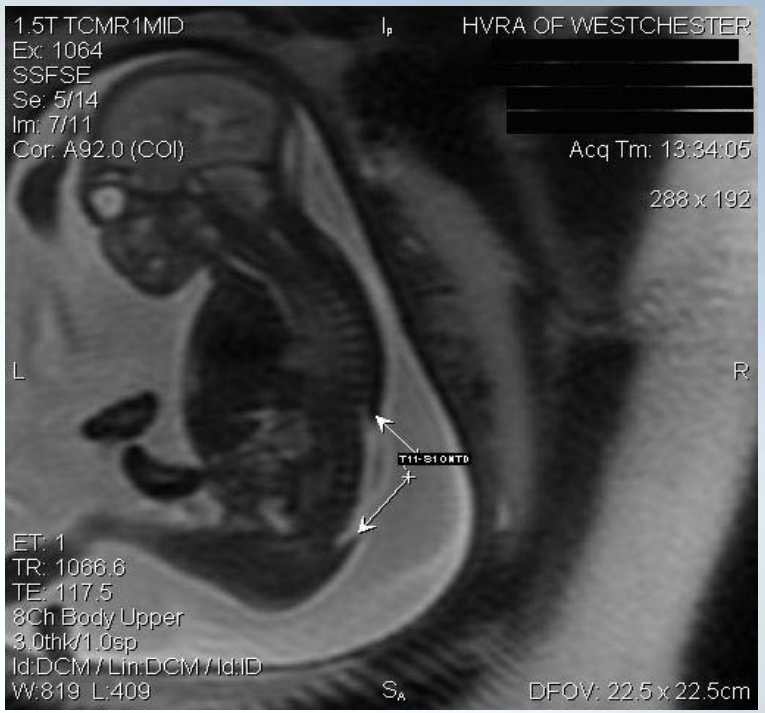




T12 – SI ONTD with Anal Atresia

Ultrasound Uncertain As to Length of ONTD. MR Diagnoses Additional Pathology Not Seen on Ultrasound

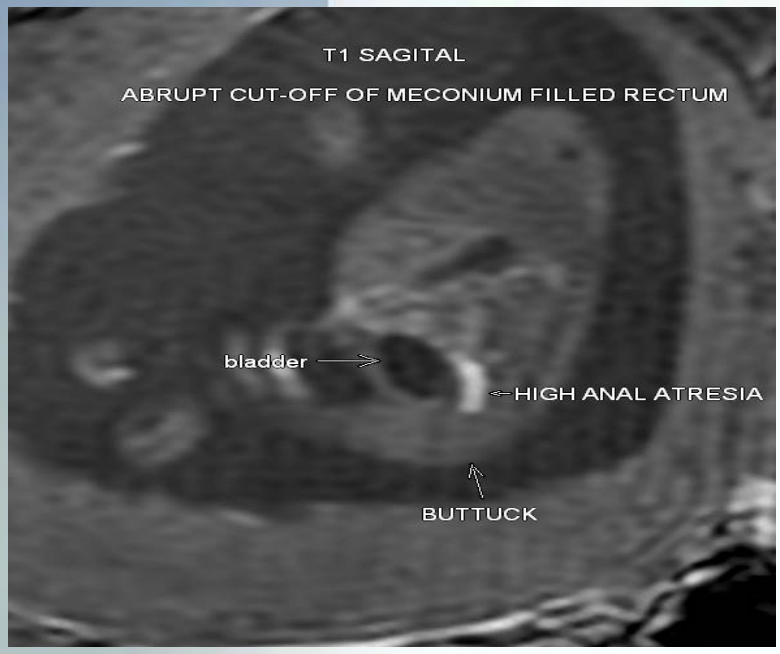


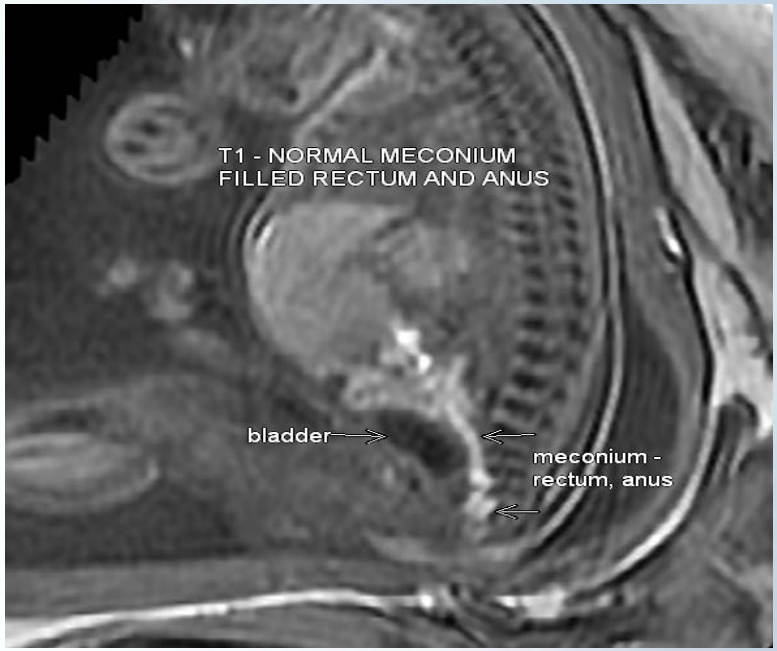


MR identifies significant pathology unable to be seen on ultrasound
Anal atresia in patient with closed sacral NTD

HISTORY: 20-week gestation with outside ultrasound studies suggesting a sacral spinal defect. Our ultrasound studies confirmed a skin-covered sacral spina bifida "occulta" with male appearing phallus with shawl-type scrotum.

Unexpected MR Findings: High anal atresia





Anal atresia with recto-urethral fistula

History: 21w GA whose outside ultrasound studies identified dilated intestine- large versus small bowel uncertain, etiology uncertain.

MR findings: T2 imaging demonstrated a dilated rectosigmoid that did not extend caudal to the anal region consistent with anal atresia. A fistulous tract was identified extending to the urethral region.

Ultrasound and MR evidence of fistulous admixture of urine and meconium.

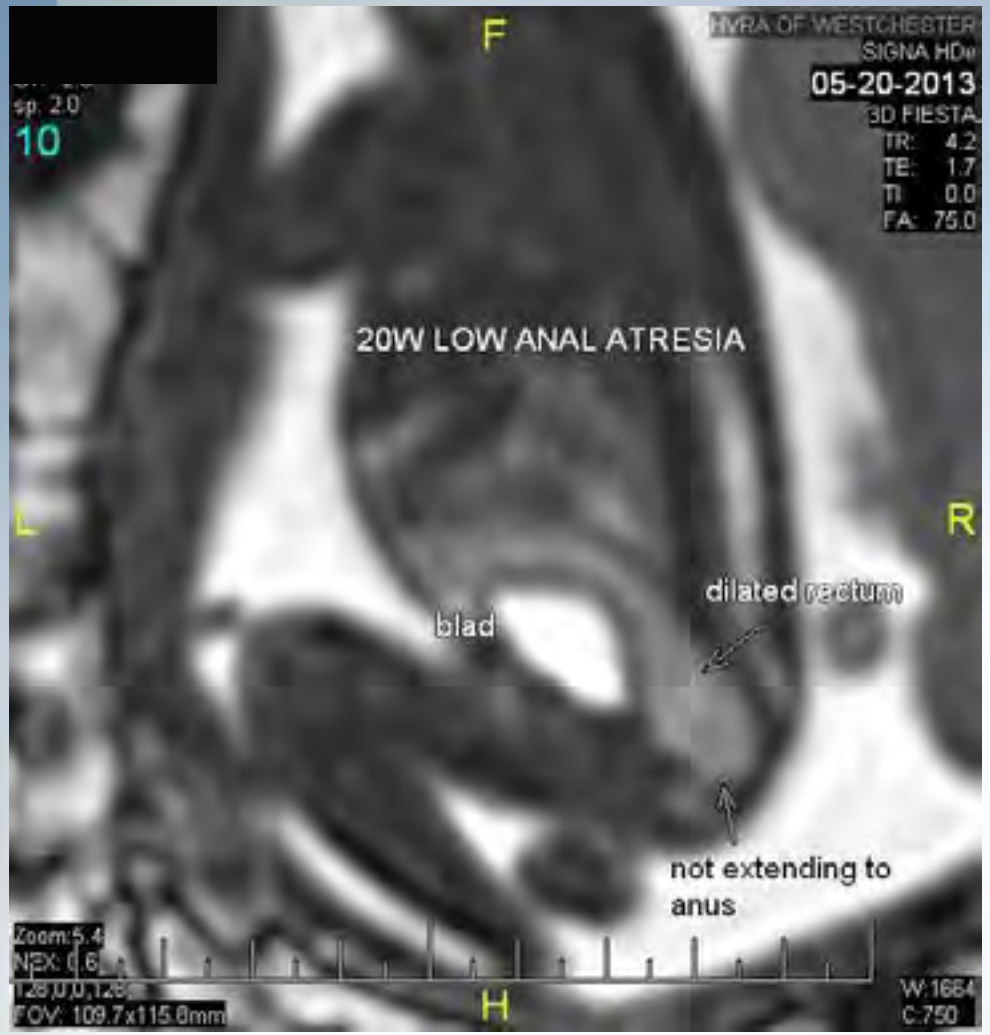
- Absence of T1 meconium hypersignal intensity
- Presence of an enterolith (calcification) within the stool of the dilated colon

Clinical outcome: Patient was born full term with anal atresia. Urine analysis confirmed fecal content consistent with fistula. VCU pending.

Patient treated with diverting colostomy pending definitive surgery when older.

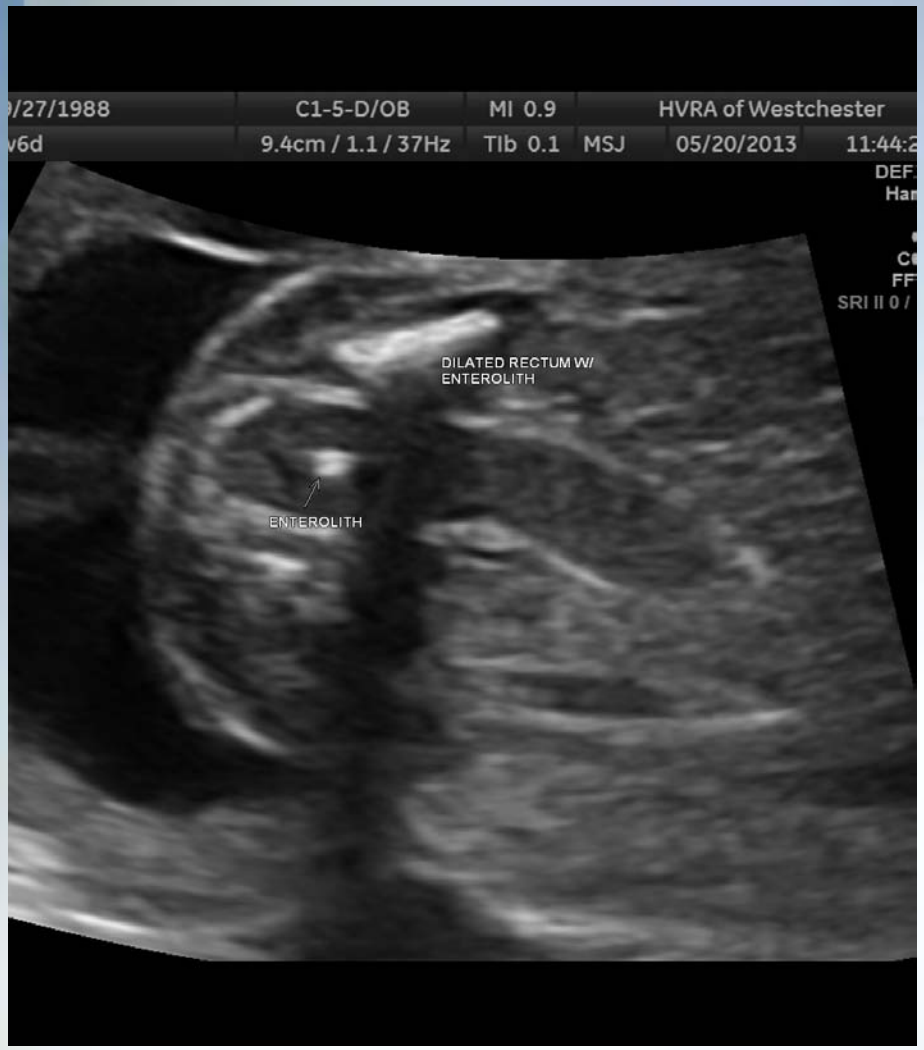
Teaching point: MR is complementary and often superior to ultrasound in diagnosing GI and GU pelvic pathology.

MR establishes a Dg rarely made on ultrasound
Anal atresia with recto-urethral fistula



MR establishes a Dg rarely made on ultrasound
Anal atresia with recto-urethral fistula

MR establishes a Dg rarely made on ultrasound
Anal atresia with recto-urethral fistula



Esophageal atresia with TE fistula

History: 32w GA whose outside ultrasound demonstrated a small stomach. Patient was screened positive for T21 but declined karyotyping.

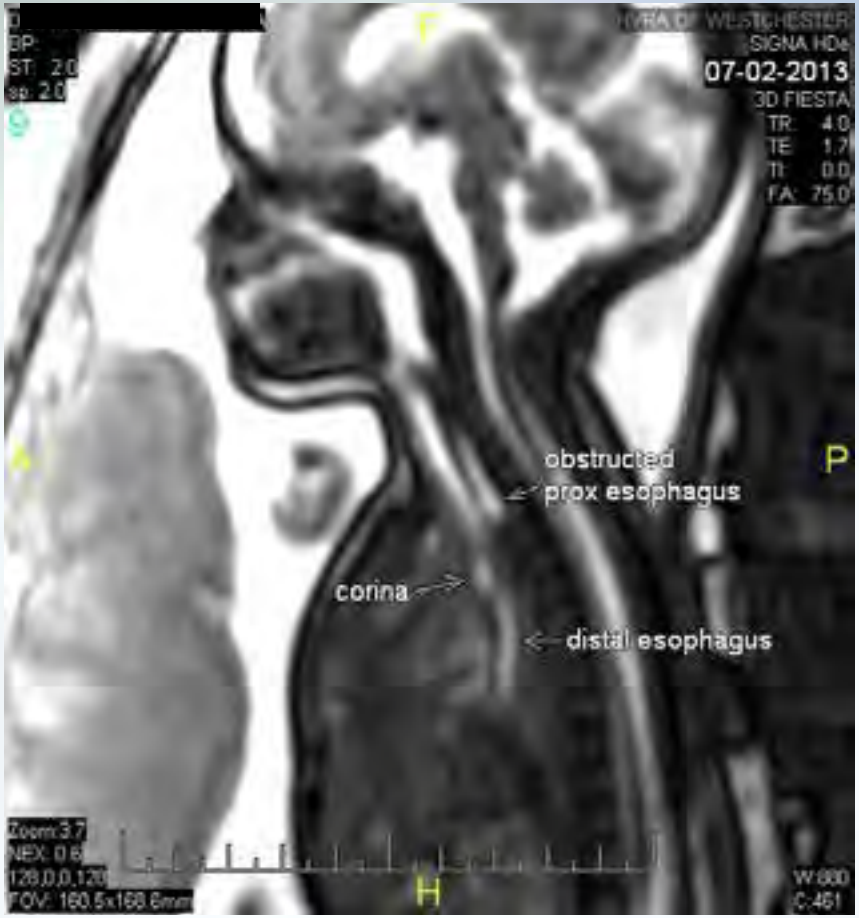
MR demonstrates transient dilatation of the proximal esophagus, the “pouch” sign and visualizes the TE fistula between the carina and the distal esophageal segment.

Teaching point: Consider MR in the evaluation of otherwise unexplained polyhydramnios and in the evaluation of malformations for which esophageal atresia may coexist in syndromic association –

- VACTERL
- CHARGE

MR establishes a Dg rarely made on ultrasound

Esophageal atresia with TE fistula



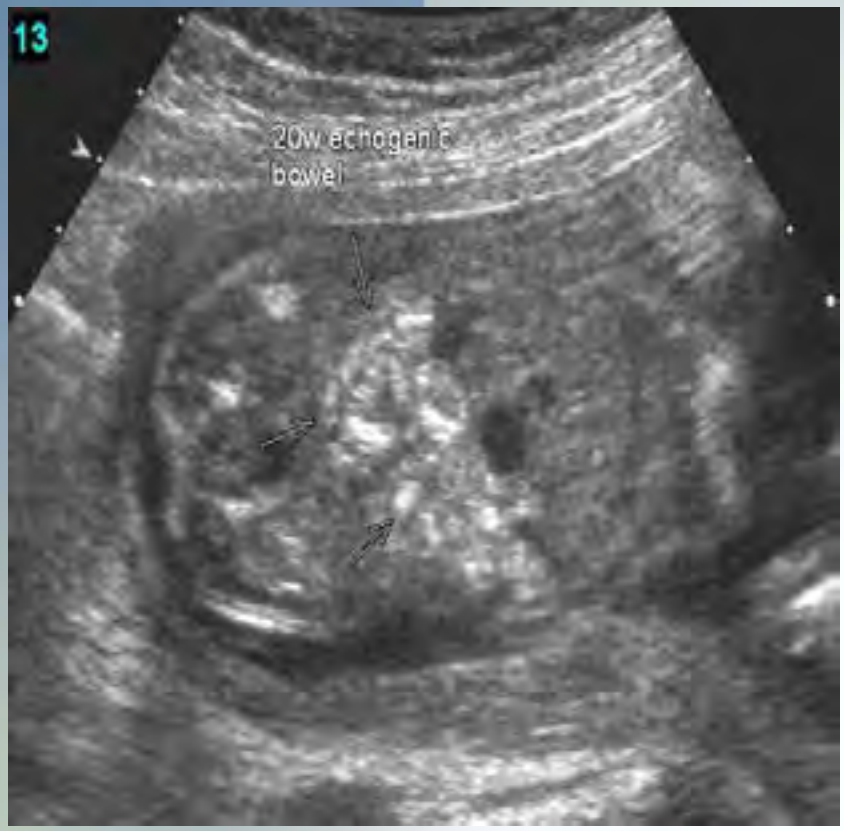
Fetal bowel dilation - small vs. large bowel on Ultrasound, uncertain

History: 21w GA with detailed ultrasound demonstrating a single 8 mm dilated loop of bowel and large amount of non dilated echogenic bowel. Aneuploidy, infectious and cystic fibrosis screening studies were negative.

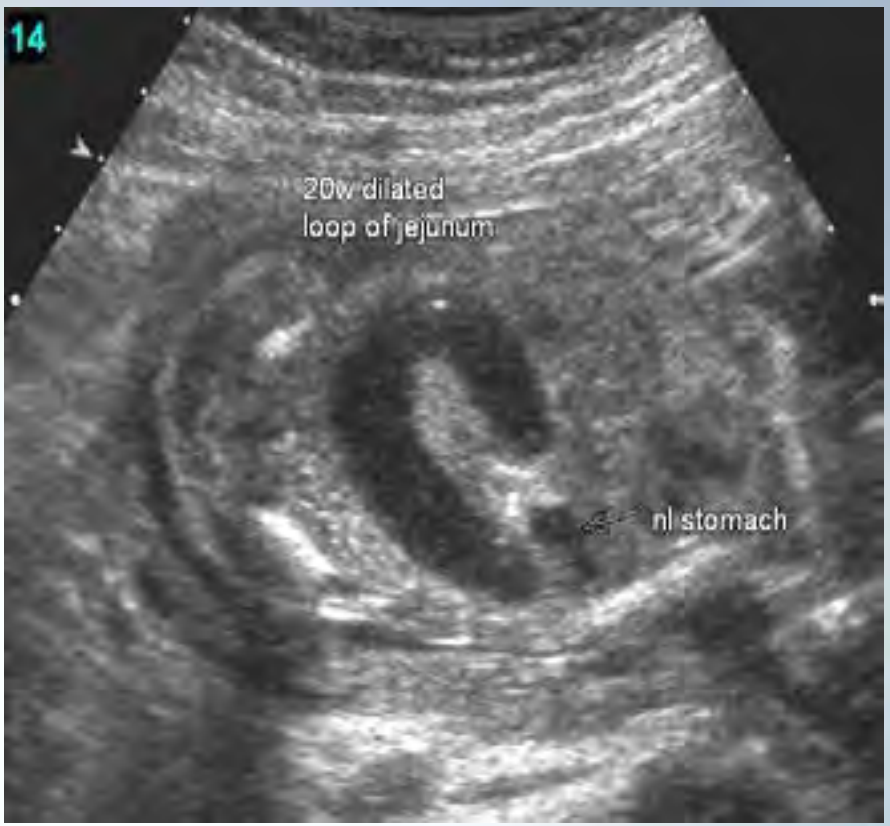
MR demonstrated dilated loop to be of small bowel origin with small, but intact meconium-filled rectum being traced to the anal region (anorectal atresia excluded).

Followup US at tertiary care center demonstrated unchanged small bowel dilatation until 32 weeks, at which time a dilated stomach developed.

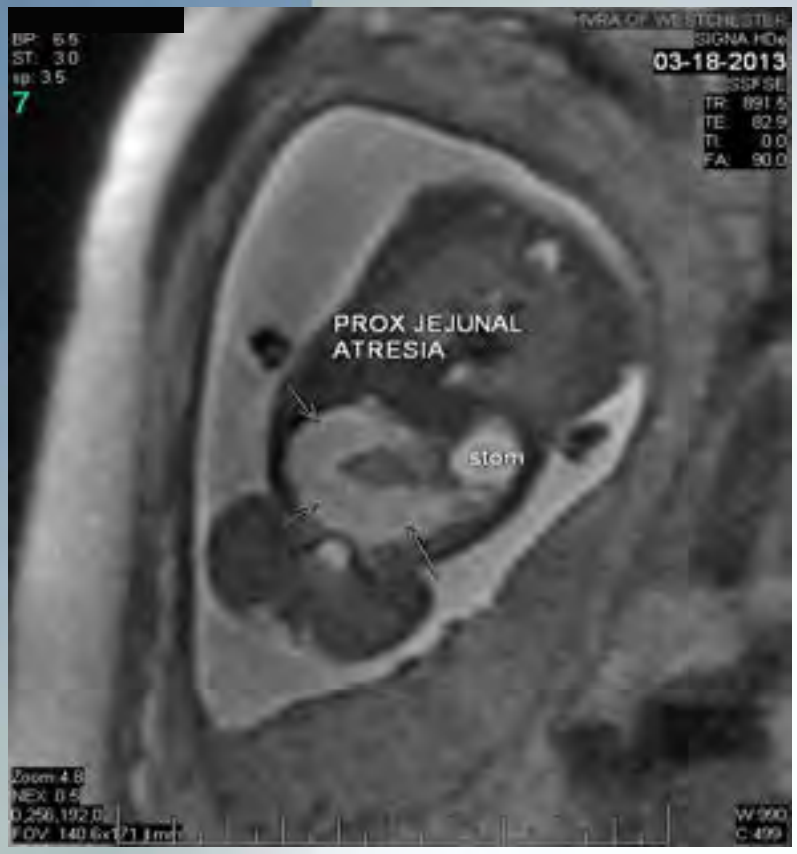
Large vol. of echogenic bowel and dilated jejunum suggests meconium ileus.

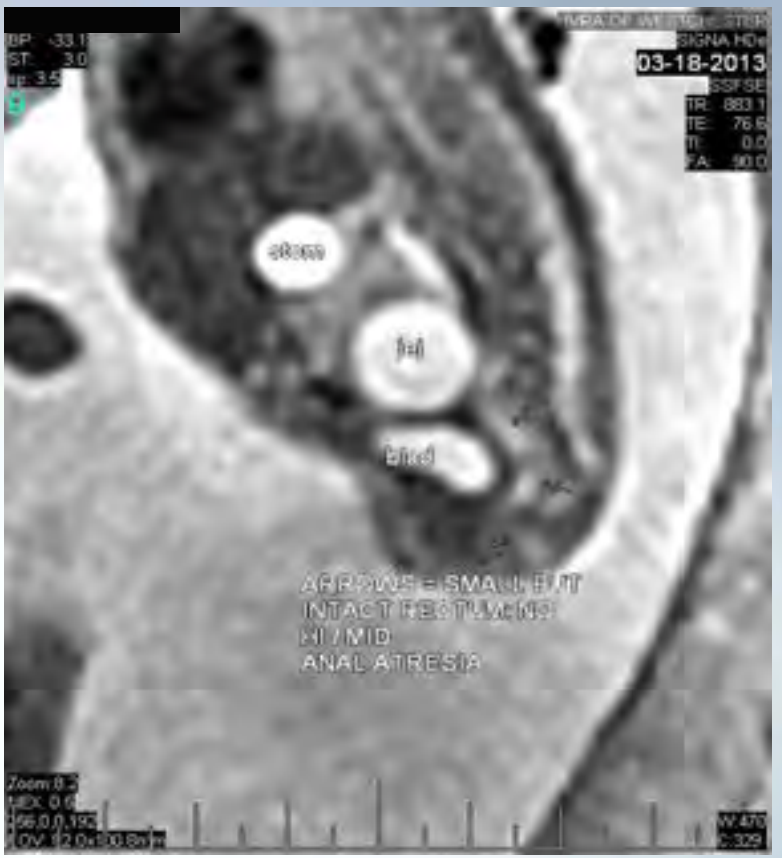


Ultrasound demo - Fetal bowel dilatation small vs. large bowel?



Fetal bowel dilatation - MR assigns organ of origin and answers the question- “is there anal atresia”?





At surgery – gastric antral atresia with 7 sites of small bowel and colonic atresias.

Clinical workup consistent with combined immunodeficiency syndrome.

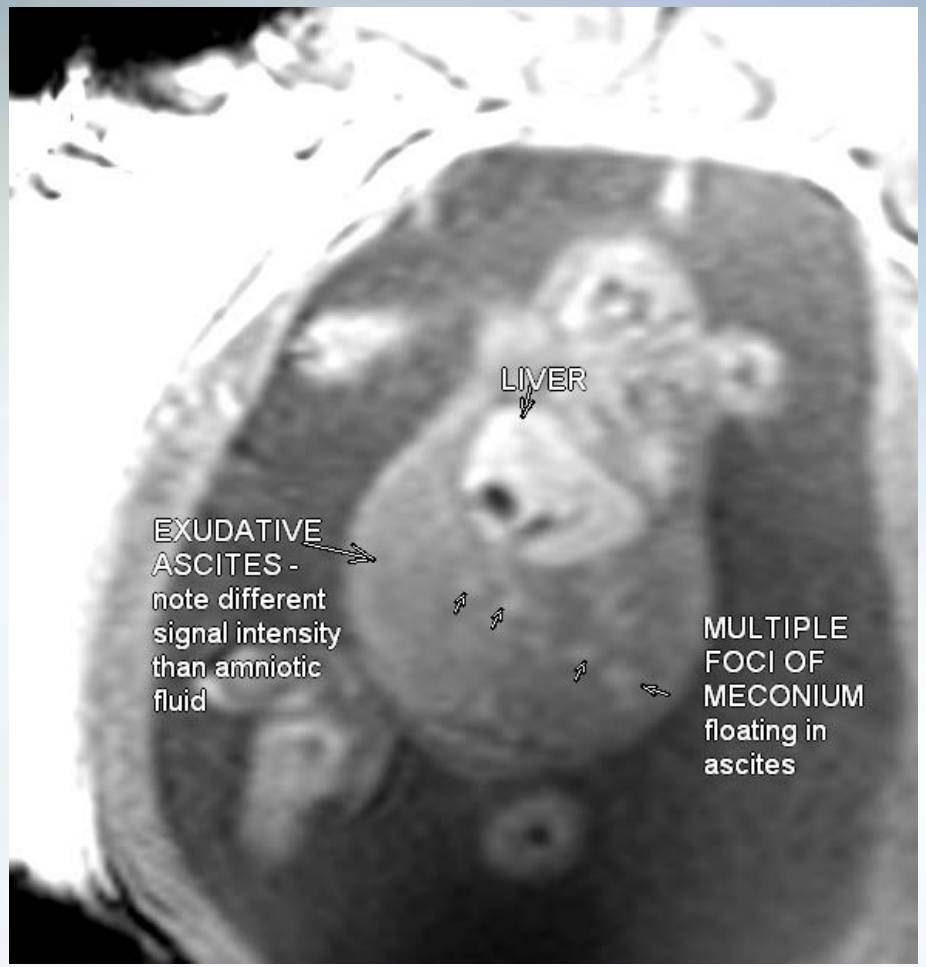
Two months after surgery, the neonatal GI tract remains nonfunctioning. Poor prognosis. ? bone marrow transplant: ? intestinal transplant.

Teaching point:

1.When ultrasound identifies dilated bowel, MR is the modality of choice to establish small versus large bowel organ of origin and generate differential diagnosis.

2. If imaging demonstrates multiple sites of bowel dilatation, immunodeficiency syndromes should be considered in the differential diagnosis.

26 w US demo. ascites of unknown etiology. MR diagnosis – meconium peritonitis



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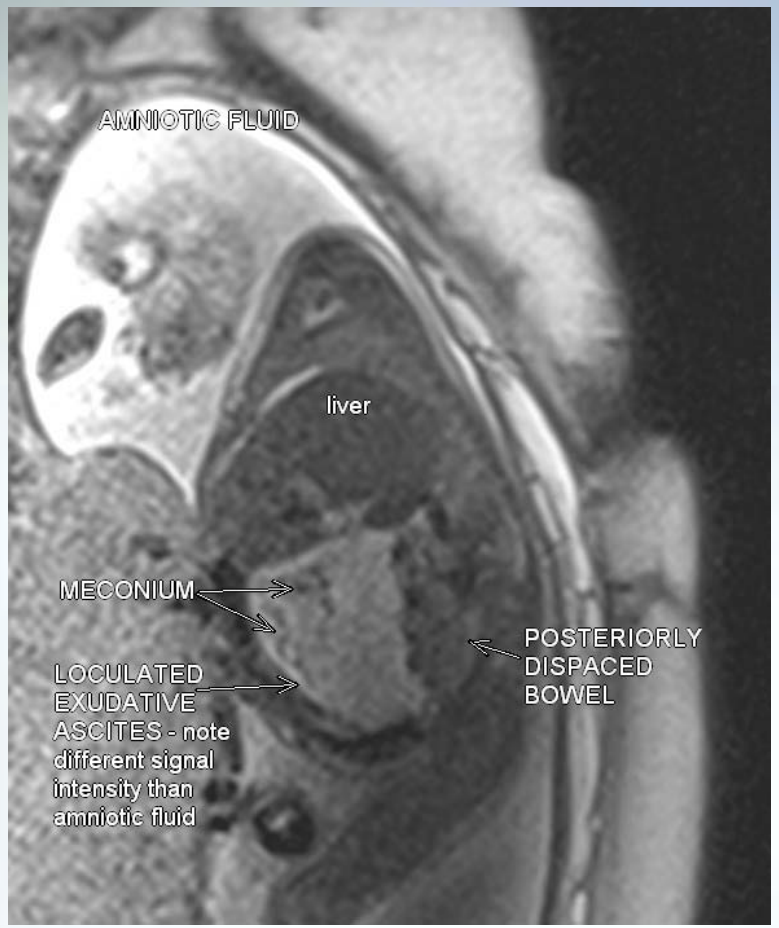


MR establishes a Dg unable to be made on US – hemosiderosis

Anhydramnios and fetal hydrops of unknown etiology.

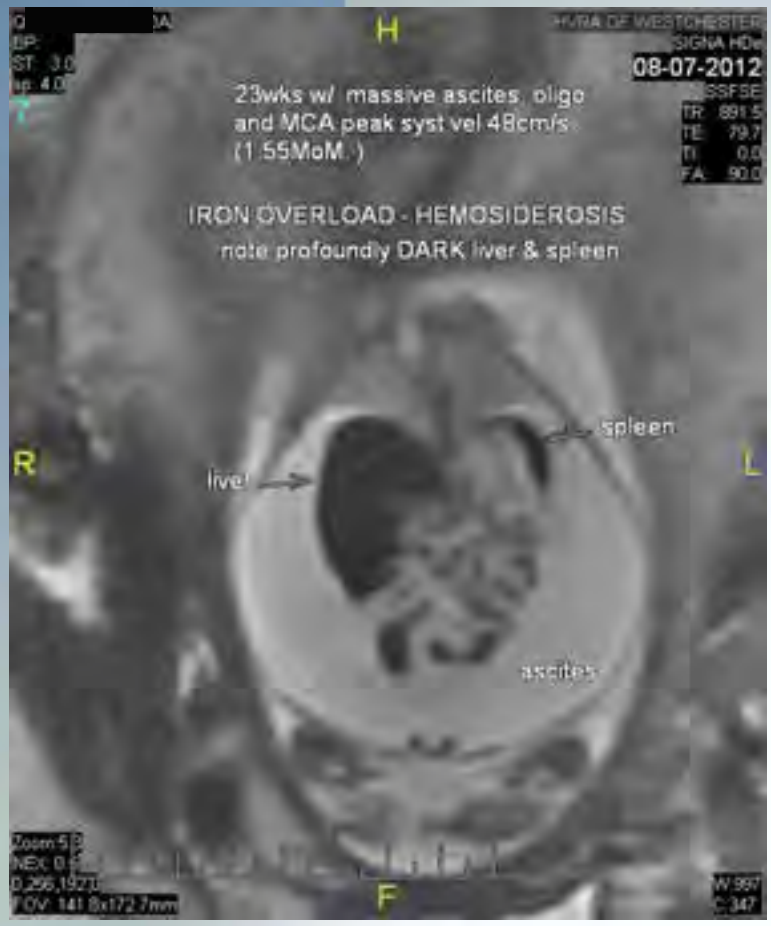
History : 23w GA with MFM outside studies demonstrating anhydramnios, nonnormalized kidneys, ascites and hydrops of unknown etiology. *At the time of MR study results of infectious workup were not known.* Negative maternal workup for immune incompatibility. Normal fetal echocardiography during MR appointment. Middle cerebral peak systolic velocity 48 cm/second (1.55 MoM) cw severe anemia.

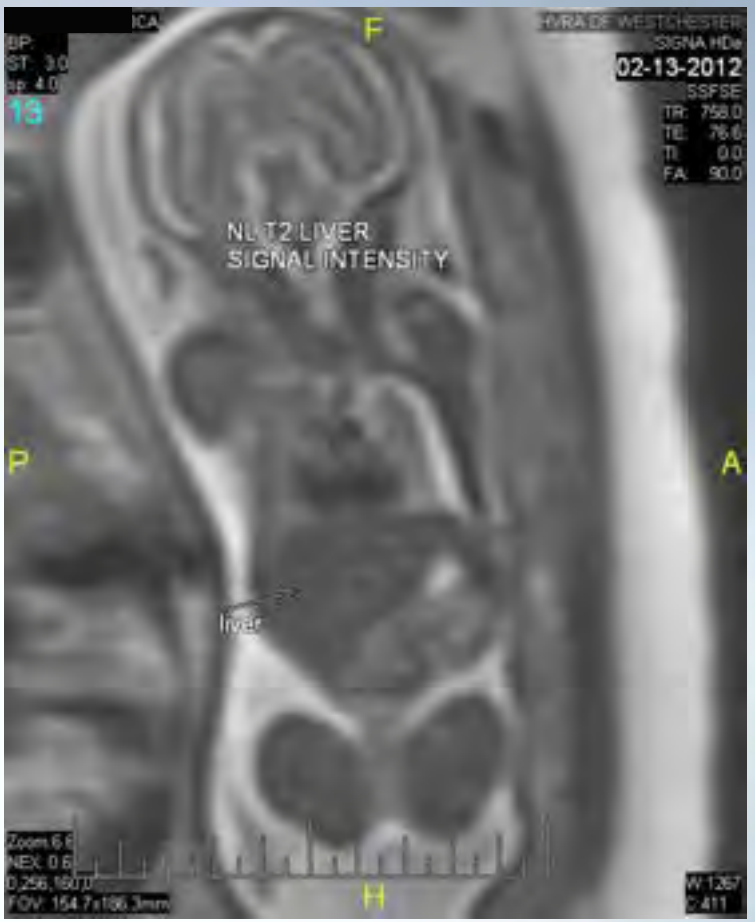
MR demonstrated normal kidneys, featureless transudative-appearing ascites and marked hepatic hyposignal intensity.

Differential diagnosis for marked decreased fetal liver signal intensity statistically favors iron overload (hemosiderosis) due to either various etiologies of fetal anemia (hemolysis) or congenital alloimmune hepatitis (fetal/neonatal hemochromatosis).

MR establishes a dg unable to be made on ultrasound – hemosiderosis

Anhydramnios and fetal hydrops of unknown etiology.





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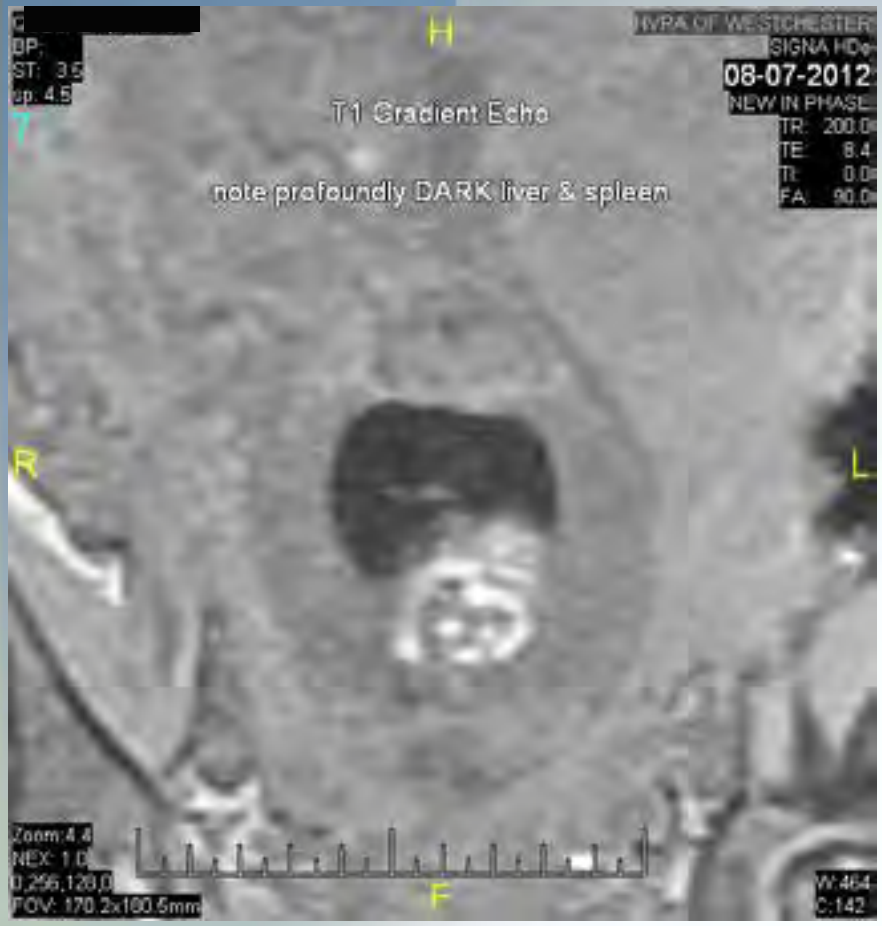
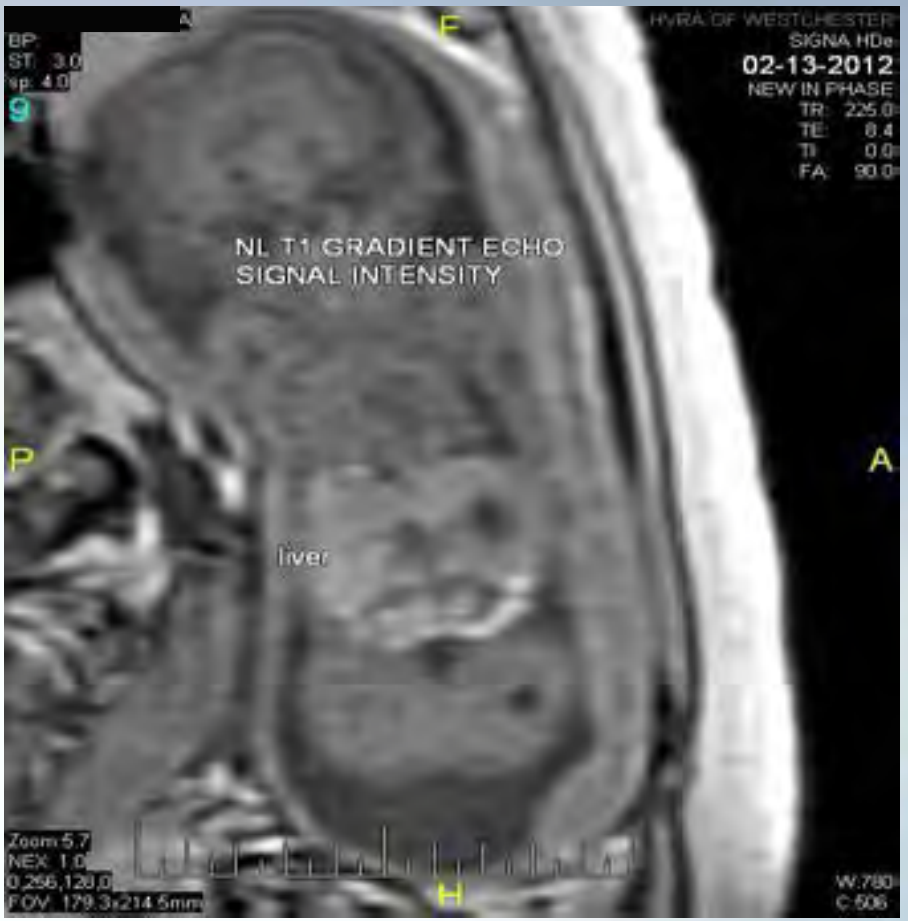
MR establishes a dg unable to be made on ultrasound – hemosiderosis

Anhydramnios and fetal hydrops of unknown etiology.





MR establishes a Dg unable to be made on US – hemosiderosis

Anhydramnios and fetal hydrops of unknown etiology.

Clinical outcome:

IUFD after fetal transfusion.
After demise lab workup returned + CMV.

Teaching point:

1. With oligohydramnios/anhydramios, MR is superior to ultrasound in evaluation of fetal anatomy.

2. In the context of ascites/hydrops of unknown etiology, MR's ability to diagnose hepatic iron overload is unique. *(Congenital hemochromatosis is the most common cause for neonatal liver failure and has a high rate of recurrence in future pregnancies.)*

MR establishes severe placental pathology unable to be seen on ultrasound

HISTORY: 31-week gestation whose outside ultrasound demonstrated an enlarged solid right kidney suspicious for mesoblastic nephroma. The fetus had recently exhibited ascites that had resolved. Past OB history of 29-week intrauterine fetal demise.

FINDINGS: MR demonstrates abnormal appearing kidneys of heterogeneous solid echotexture and multiple triangular hemorrhagic placental infarcts.

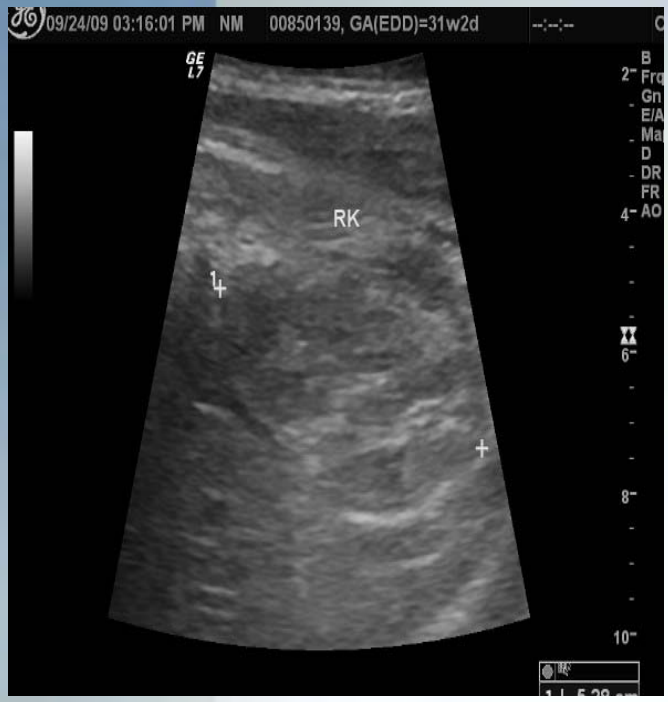
Still birth 2 weeks after MR exam.

AUTOPSY DIAGNOSIS:

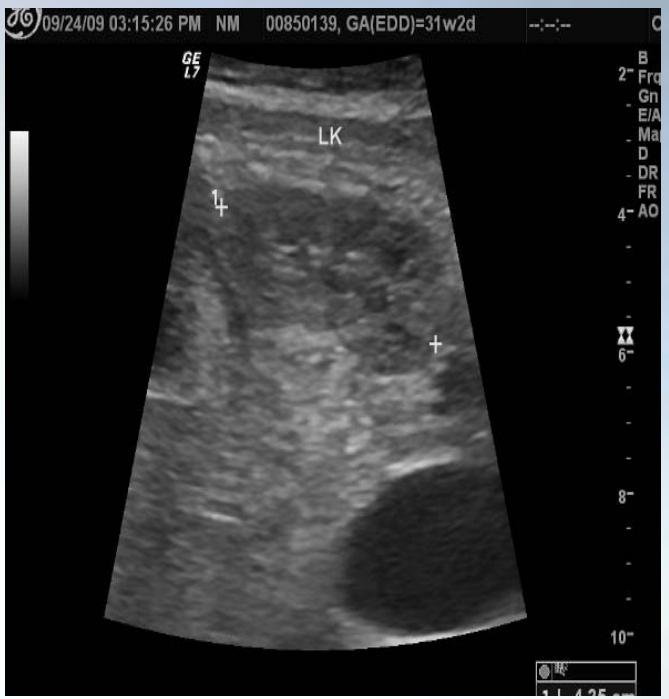
Hemorrhagic vasculopathy of placenta and fetus.

MR changes diagnosis from neoplasm to vasculopathy.

Hemorragic vasculopathy of fetus and placenta
US: enlarged, heterogenous R kidney.



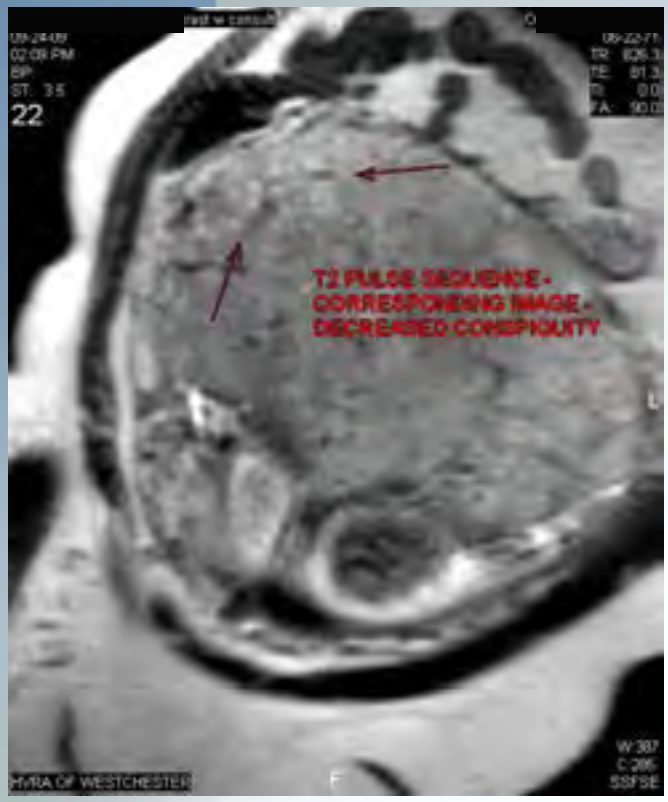
abnormal right kidney

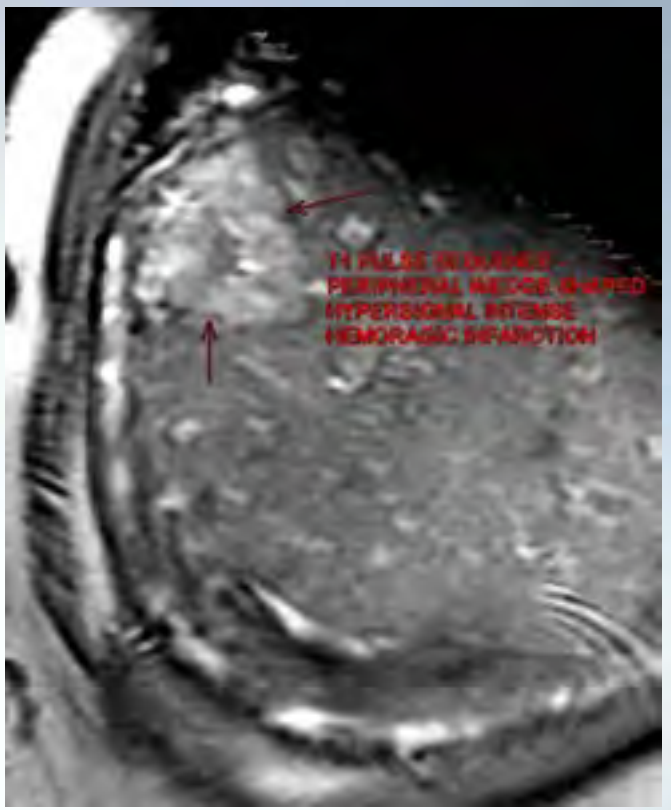


normal left kidney

MR changes diagnosis from neoplasm to vasculopathy.

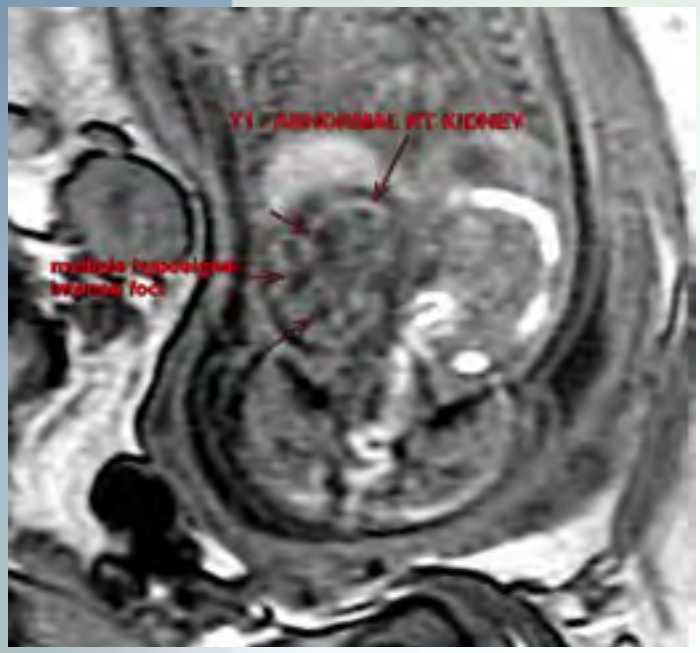
Hemorrhagic vasculopathy of fetus and placenta

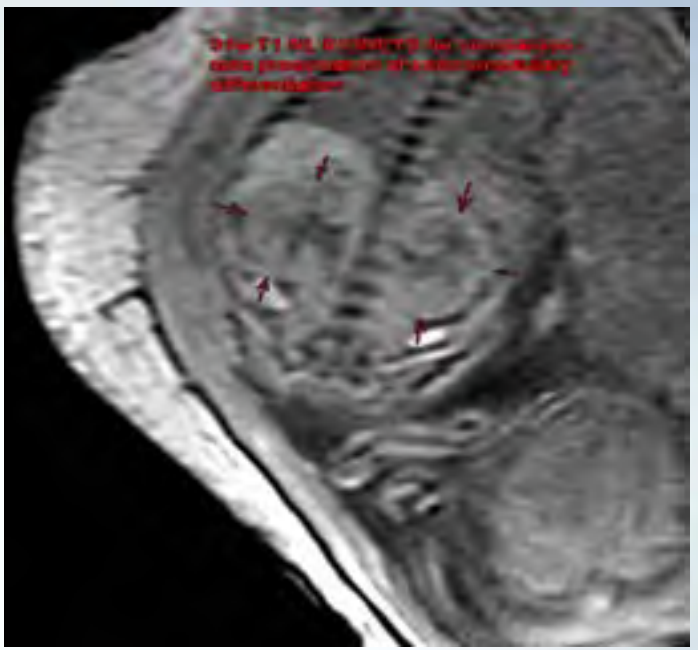




MR changes diagnosis from neoplasm to vasculopathy. Hemorrhagic vasculopathy of fetus and placenta

R. kidney demonstrates multiple foci of dark T1 signal – blood by products vs fibrous tissue.





Gestational age matched normal for comparison

MR establishes severe placental pathology unable to be seen on ultrasound

Abnormal placenta – IUFD 26w - impaired maternal placental perfusion

History: 20w GA referred for unexplained elevated maternal serum AFP (7.2 MoM) with outside MFM ultrasound biometry two weeks smaller than expected.
Normal amniocentesis including microarray.
Negative maternal workup for thrombophilia, SLE and anticardiolipin antibodies.

MR results: Small placenta demonstrating homogeneous diffuse decreased (dark) signal intensity.
Echoplanar diffusion imaging demonstrated decreased apparent diffuse coefficient (ADC).

Uterine artery Doppler demonstrated bilateral early diagnostic notching with markedly elevated resistive indices for gestational age.
Umbilical arterial resistive index is 0.92 at the 4th standard deviation higher than expected for gestational age.

Abnormal placenta – IUFD 26w - impaired maternal placental perfusion

Clinical outcome: IUFD at 26w.

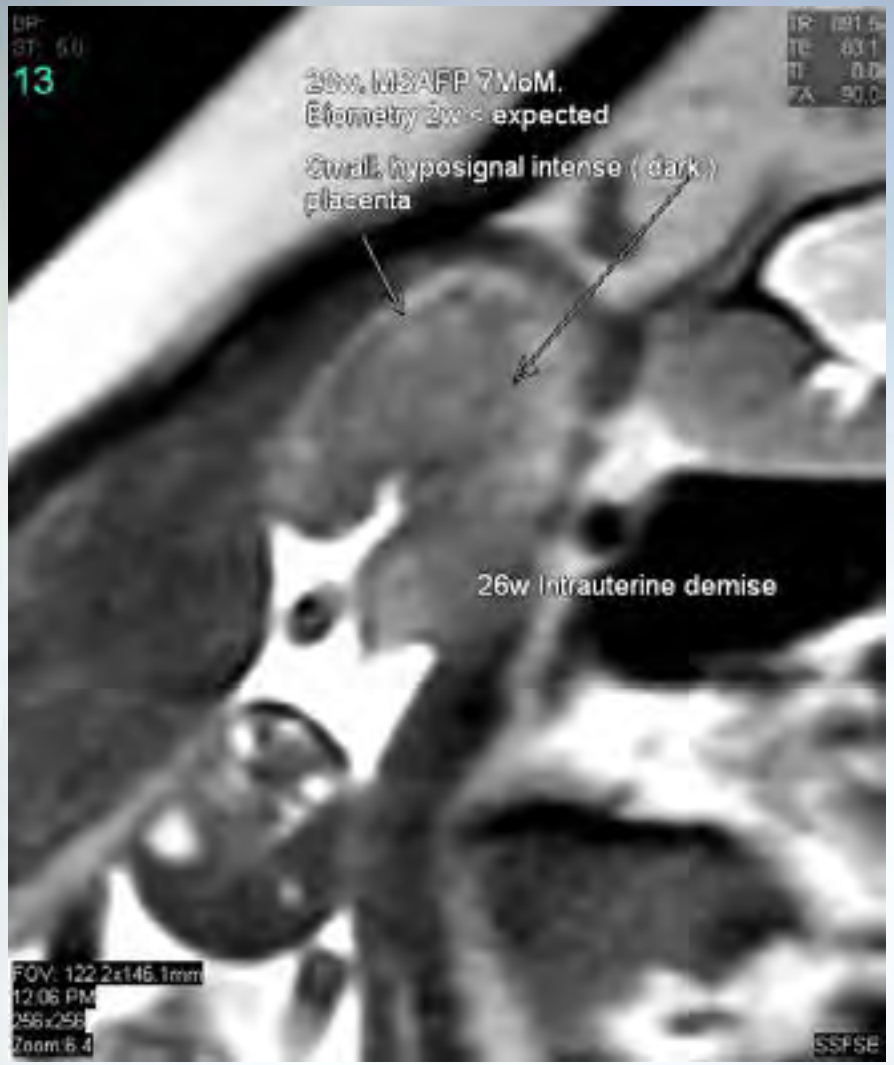
Placental pathology:

- 95 g placenta, less than the 10th percentile for gestational age.
- Diffuse hypoxic-ischemic villous changes.
- Focal villous stromal mineralization.
- Luminal septation within fetal stem blood vessel secondary to intraluminal clot.
- Increased numbers of circulating nucleated red blood cells in fetal circulation consistent with chronic fetal hypoxia secondary to placental vascular lesions.

Teaching point: Diffusion weighted placental imaging may identify abnormalities that corroborate with placental insufficiency. ADC values reflect restricted water diffusion in the extracellular extravascular space suggesting restricted oxygen diffusion.

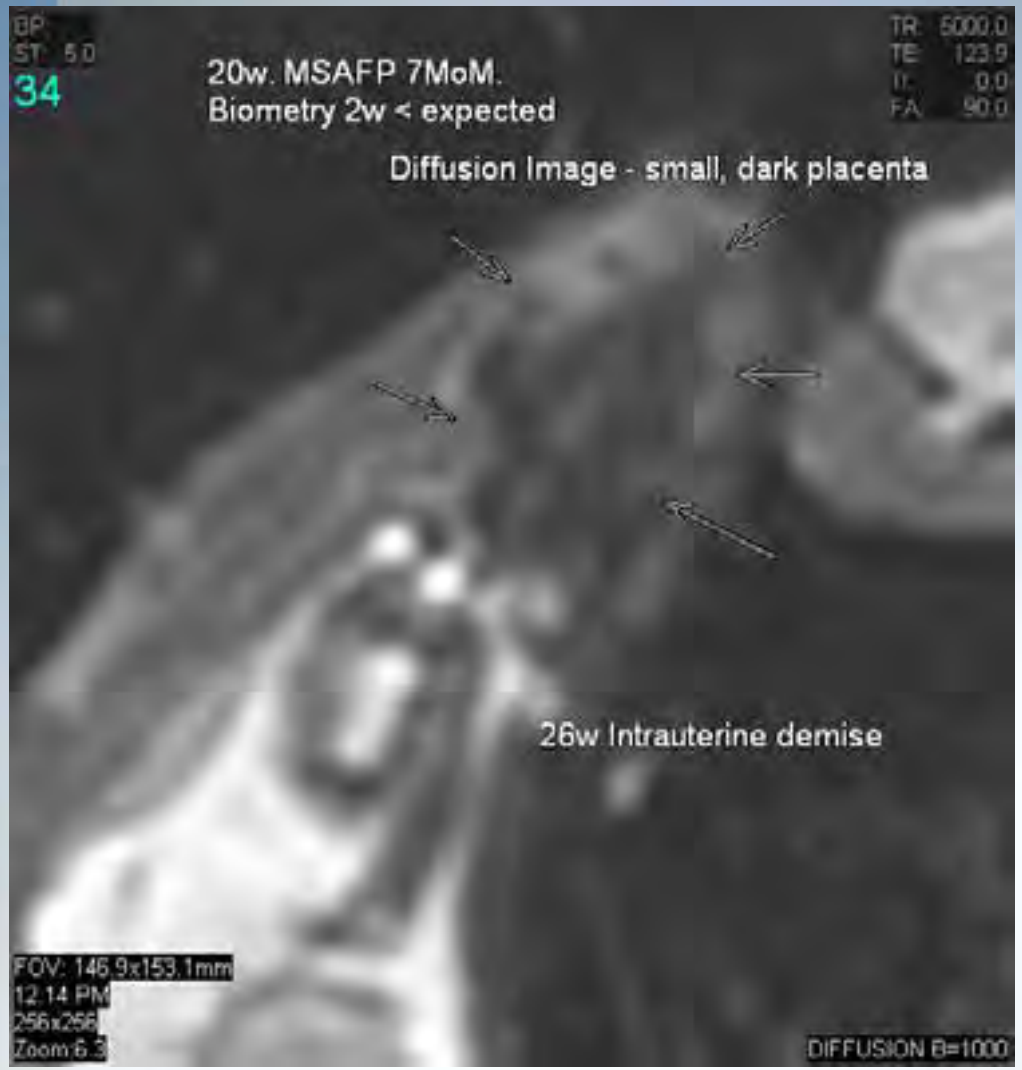
*Diffusion weighted MR imaging of the placenta in fetuses with placental insufficiency. Radiology Vol. 257:December 2010.

MR establishes severe placental pathology unable to be seen on ultrasound
Abnormal placenta – IUFD 26w - impaired maternal placental perfusion



MR establishes severe placental pathology unable to be seen on ultrasound

Abnormal placenta – IUFD 26w - impaired maternal placental perfusion



Abnormal placenta – IUFD 26w - massive perivillous fibrin deposition

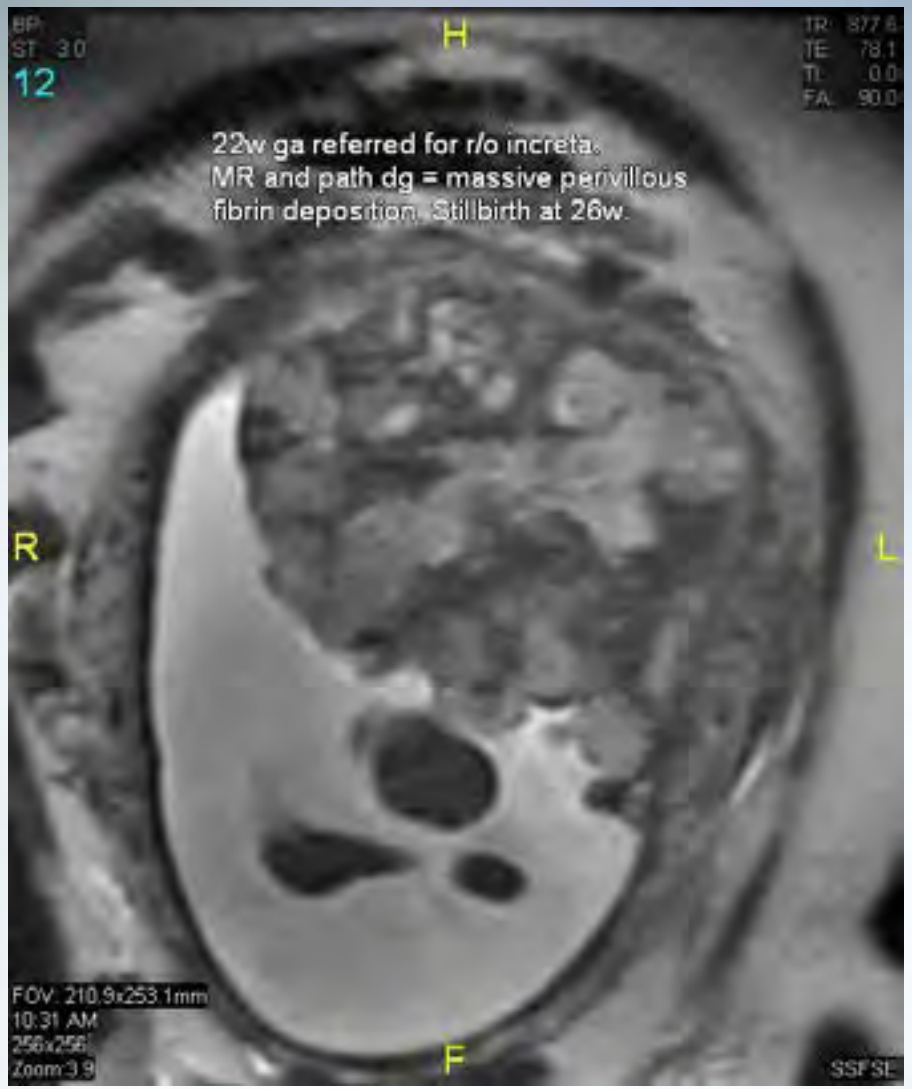
History : 22w GA referred for suspected placenta increta. Patient has unexplained elevated maternal serum AFP.

MR: Profoundly heterogeneous placenta with large curvilinear bands of hyposignal intense tissue. The distribution has an appearance similar in configuration to the convolutions of the cerebral cortex.

Clinical follow up: IUFD at 26w.

Teaching point: MR of the placenta can identify severe placental pathologies associated with early onset IUGR and marked morbidity/mortality such as hemorrhagic vasculopathy and massive perivillous fibrin deposition

MR establishes severe placental pathology unable to be seen on ultrasound
Abnormal placenta – IUFD 26w - massive perivillous fibrin deposition



Abnormal placenta – IUFD 26w - massive perivillous fibrin deposition

Figure 2. a) Basal (maternal) surface of placenta showing abnormal pale tan-grey cerebriform appearance that contrasts with the normal lobulated and beefy red-purple appearance (inset). b) Cut sections of the placental disc showing diffuse deposition of waxy pale grey fibrinoid material.

MR is underutilized in the detection of severe placental pathologies in the context of euploid early onset growth restriction – hemorrhagic placental infarction, massive perivillous fibrin deposition, severe maternal-placental perfusion defects

These pathologies are associated with extremely high risk of stillbirth, preterm delivery and abruption.

Most frequent CNS indications for fetal MR

- Ventriculomegaly
- Nonnormalization of cavum septum pellucidum
- Enlarged cisterna magna

MR is underutilized in the detection of malformations in the head and neck for which ultrasound is weak

- Orbits - coloboma; microphthalmia
- Nasal passage stenosis – choanal atresia, piriform aperture stenosis
- Ear – atresia external auditory canal, abnormal mesotympanum, preauricular fistula
- Pharyngeal airway obstruction

MR is underutilized in the evaluation of the following processes when otherwise unexplained –

- Polyhydramnios; oligohydramnios
- Bowel dilatation
- Hydrops