AMSER Case of the Month March 2021

AS is a 33 y/o female G6P0032 who presented to OB clinic at 19 weeks GA to establish care.

Alexander Tran, MS3
University of Utah School of Medicine

Anne M. Kennedy, MB, BCH, BAO
University of Utah School of Medicine

HEALTH UNIVERSITY OF UTAH

AMSER

Patient Presentation

- Pregnancy history:
  - 3 spontaneous abortions, no D&C necessary for any
  - G6P0032 (unknown term vs. preterm births)

- Pregnancy was dated by LMP without first trimester ultrasound
  - Ultrasound at presentation (19 weeks gestation) showed
    - Size < dates
    - Low amniotic fluid volume

US at presentation

- S<D
  - No formal early scans to confirm gestational dates
- Subjective low fluid

What is the differential?

- Incorrect dating of gestation
- Ruptured membranes
- Early onset growth restriction
  - Abnormal fetus
  - Abnormal placenta
  - Congenital infection (TORCH)
  - Aneuploidy

What is the differential?

- Incorrect dating of gestation
- Ruptured membranes → Ruled out clinically
- Growth restriction
  - Abnormal fetus
  - Abnormal placenta
  - Congenital infection (TORCH) → Negative for routine OB infection screen (*Chlamydia trachomatis*, *Neisseria gonorrhea, Hepatitis B, Hepatitis C, HIV, Syphilis, Rubella).*
  - Aneuploidy

Serum screening was abnormal

Positive screen for Down syndrome
Positive screen for open spina bifida
Negative screen for Trisomy 18

	MoM (multiples of the median)	Value
AFP	2.90	94.8 ng/mL
UE3	0.30	0.32 ng/mL
HCG	8.34	212848 mIU/mL
DIA	12.09	1655.28 pg/mL

What Imaging Should We Order?

Applicable ACR Appropriateness Criteria

Variant 2: **Growth disturbance. High risk for fetal growth restriction. Initial evaluation.**

Procedure	Appropriateness Category	Relative Radiation Level
US pregnant uterus transabdominal	Usually Appropriate	O
US duplex Doppler velocimetry fetal umbilical artery	Usually Appropriate	O
US pregnant uterus biophysical profile	Usually Appropriate	O
US duplex Doppler velocimetry ductus venosus	May Be Appropriate	O
US duplex Doppler velocimetry maternal uterine artery	May Be Appropriate	O
US duplex Doppler velocimetry fetal middle cerebral artery	Usually Not Appropriate	O

Variant 2: **Second and third trimester screening for fetal anomaly. High-risk pregnancy. Initial imaging.**

Procedure	Appropriateness Category	Relative Radiation Level
US pregnant uterus transabdominal detailed scan	Usually Appropriate	O
US echocardiography fetal	May Be Appropriate	O
MRI fetal without IV contrast	May Be Appropriate (Disagreement)	O
US pregnant uterus transabdominal anatomy scan	May Be Appropriate (Disagreement)	O
MRI fetal without and with IV contrast	Usually Not Appropriate	O

Applicable ACR Appropriateness Criteria

Variant 2: **Second and third trimester screening for fetal anomaly. High-risk pregnancy. Initial imaging.**

Procedure	Appropriateness Category	Relative Radiation Level
US pregnant uterus transabdominal detailed scan	Usually Appropriate	O
US echocardiography fetal	May Be Appropriate	O
MRI fetal without IV contrast	May Be Appropriate (Disagreement)	O
US pregnant uterus transabdominal anatomy scan	May Be Appropriate (Disagreement)	O
MRI fetal without and with IV contrast	Usually Not Appropriate	O

A detailed scan was ordered given abnormal serum screen results and concern for early onset growth restriction. When growth restriction was confirmed, umbilical artery Doppler was added to assist with determination of the cause.

Variant 2: **Growth disturbance. High risk for fetal growth restriction. Initial evaluation.**

Procedure	Appropriateness Category	Relative Radiation Level
US pregnant uterus transabdominal	Usually Appropriate	O
US duplex Doppler velocimetry fetal umbilical artery	Usually Appropriate	O

Detailed Ultrasound Findings (unlabeled)

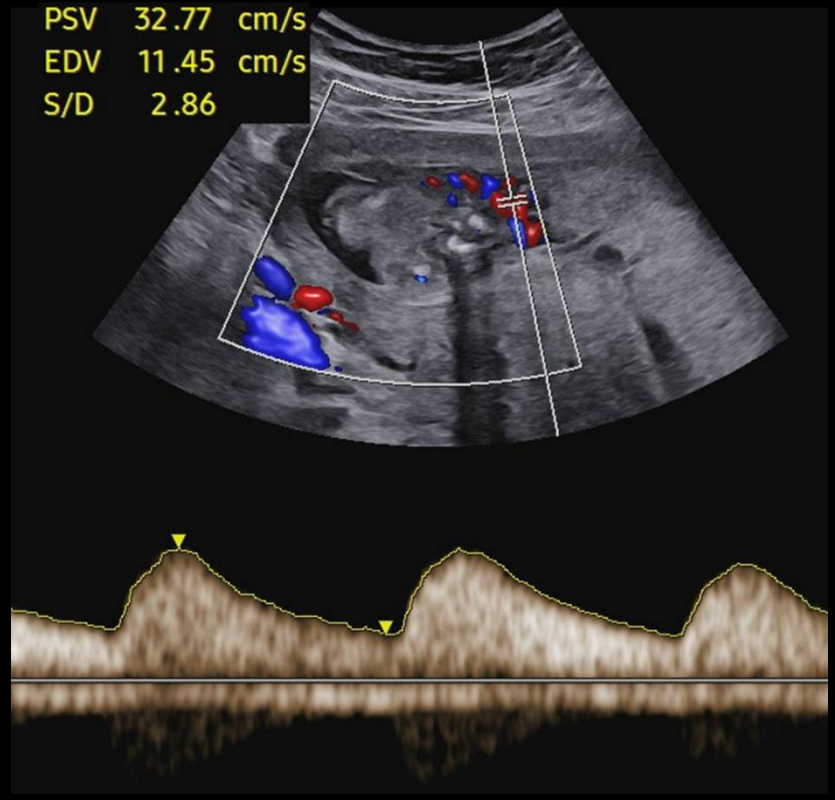
A.

B.

Detailed Ultrasound and Doppler Findings (unlabeled)

C.

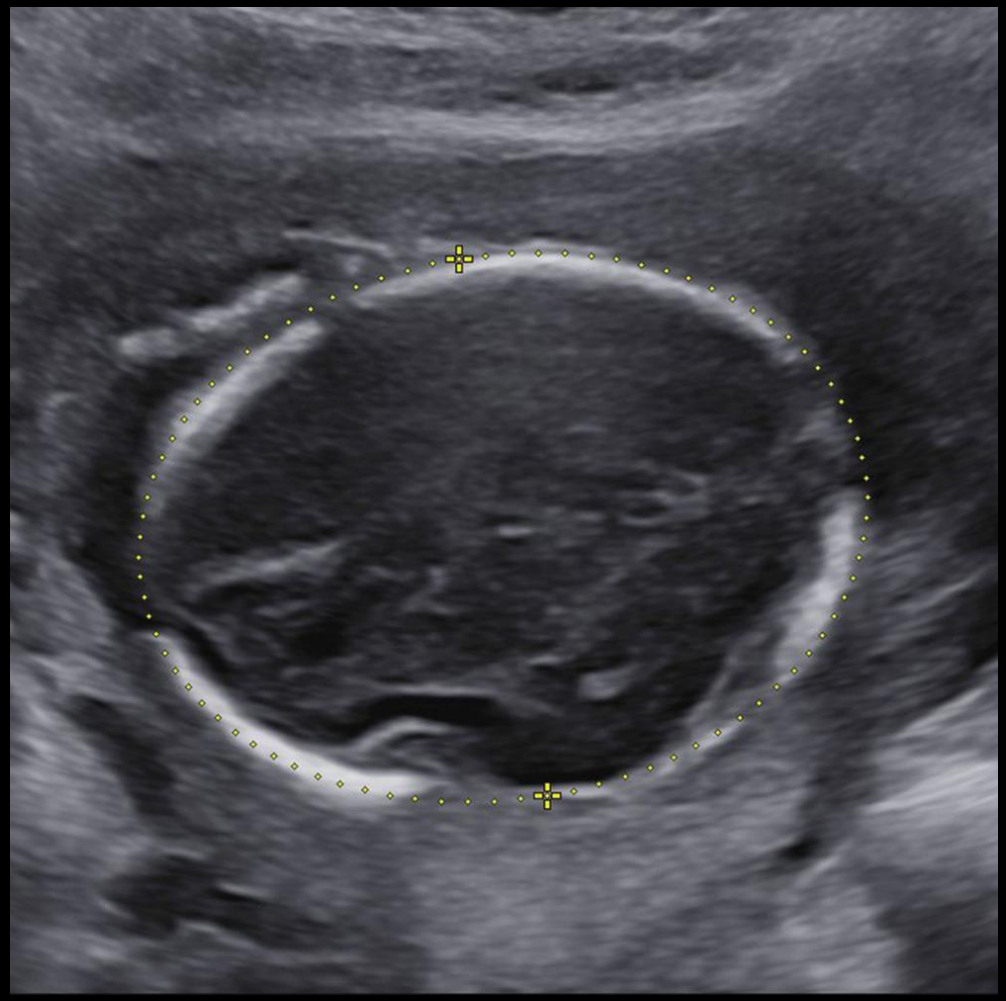
D.

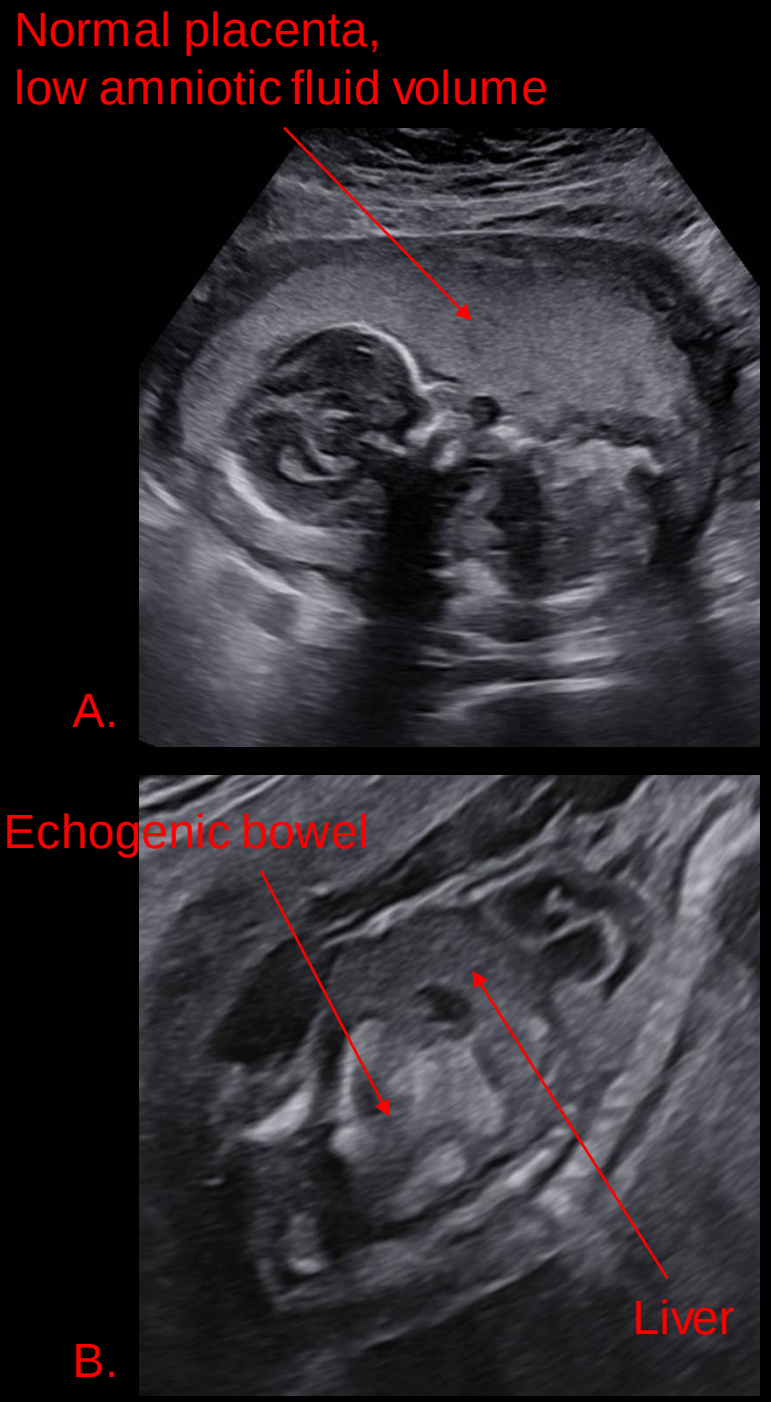


Detailed Ultrasound Findings (unlabeled)

E.

F.





Findings (labeled)

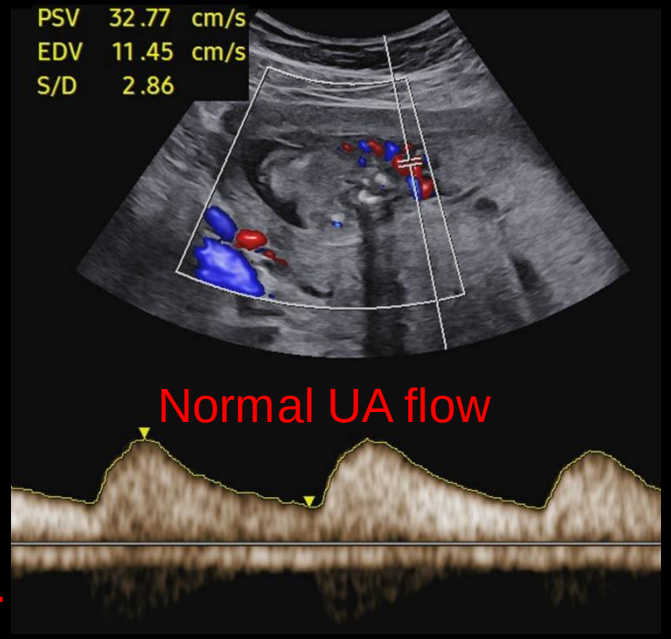
A) This image shows normal placental echotexture, which suggests that this is not a primary placenta problem. The placenta is neither cystic nor calcified . This image also shows low fluid; there is no fluid visible around the fetus which is very abnormal in a 21-week pregnancy. Incorrect gestational dating would not explain low fluid.

B) This image shows a small chest and echogenic fetal bowel (as bright as bone in the adjacent spine). Echogenic bowel is nonspecific but can be seen in aneuploidy, infection, cystic fibrosis or with chronic ischemia. There were no visible structural malformations, but the scan was incomplete due to limited acoustic access in the setting of oligohydramnios.

Findings (labeled)

Bladder contains urine

C.



D.

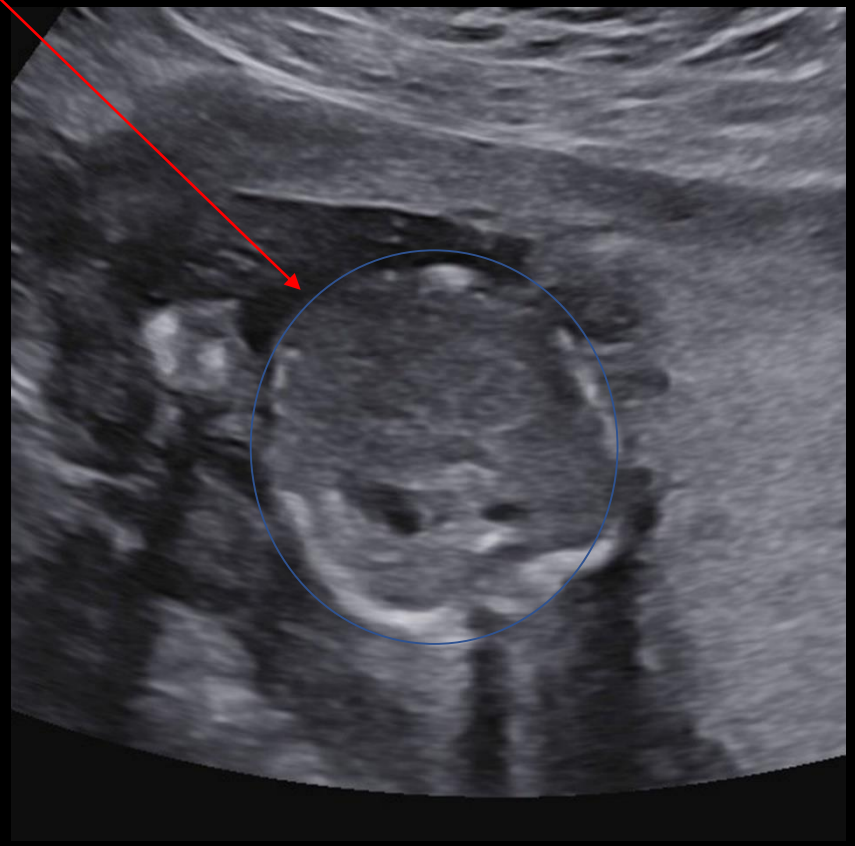
These images are used to narrow the differential diagnosis.

Image C reveals urine in the bladder of the fetus and other views showed normal kidneys therefore oligohydramnios is not due to a renal anomaly.

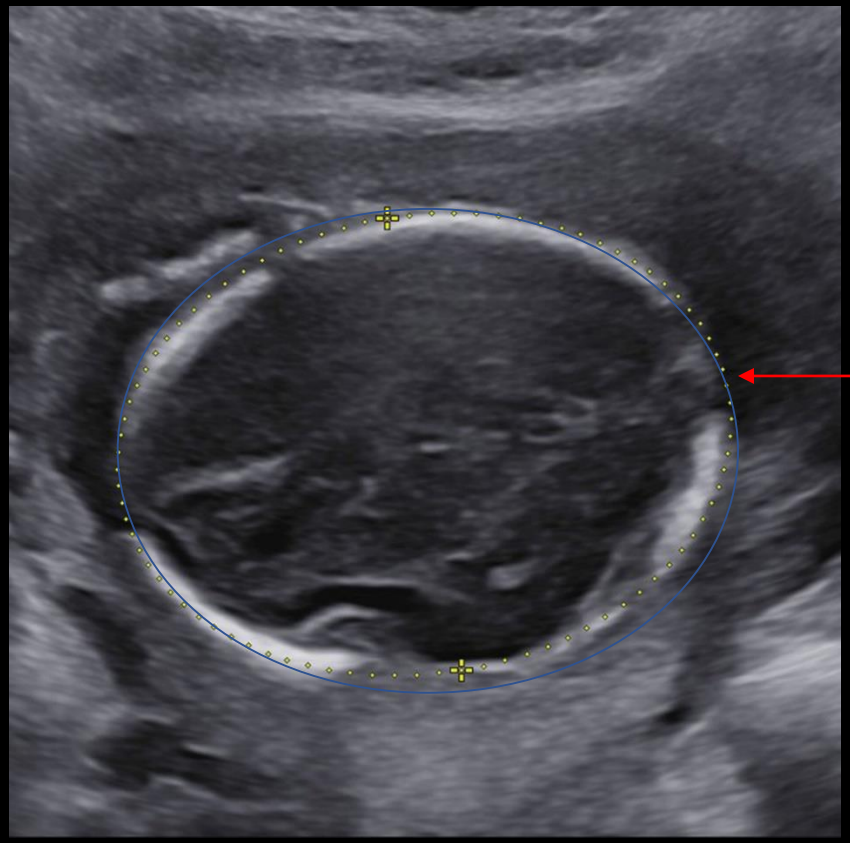
Image D shows a normal Doppler waveform in the umbilical artery (UA). If the cause of growth restriction and low fluid was placental insufficiency we would expect abnormal UA Doppler with decreased diastolic flow or even absent/reversed end diastolic flow.

AMSER

Findings (labeled)



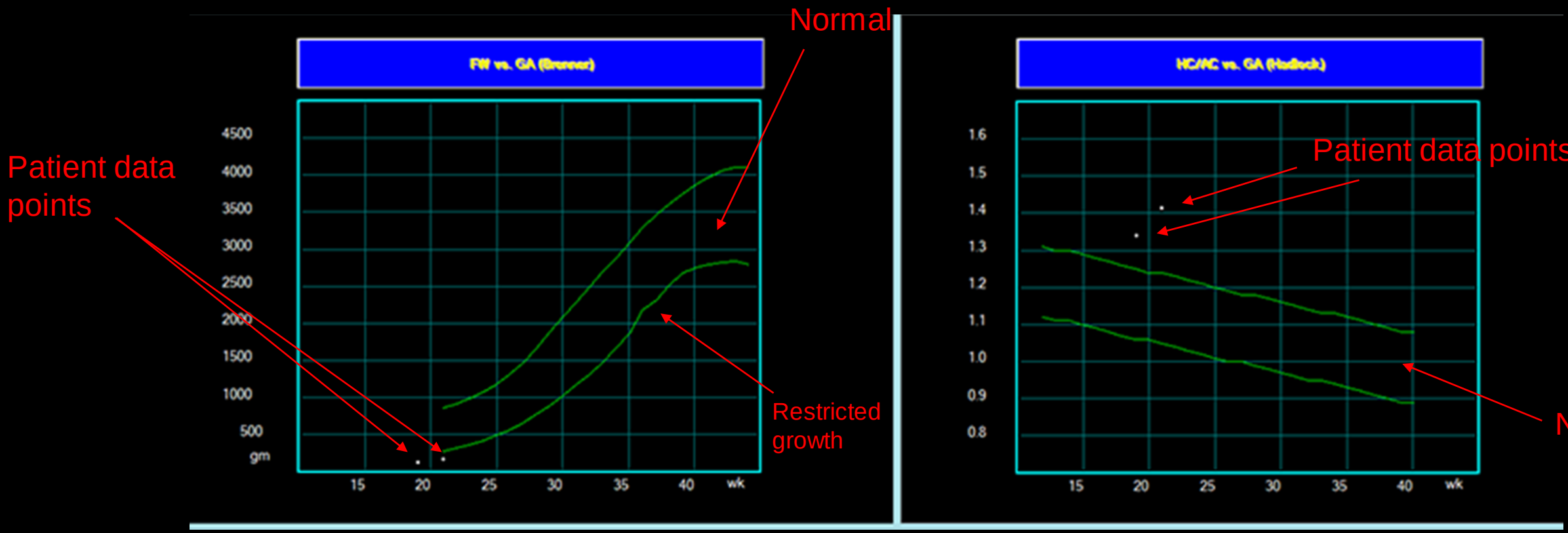
E.



F.

Image E shows an ultrasound image of the fetal abdomen and Image F shows an image of the fetal head at the second US visit when the fetus was confirmed to have growth restriction. The fetal head circumference measured 15 cm and the fetal abdominal circumference measured 10 cm. ***When head size is significantly greater than abdominal circumference, in the setting of severe, early-onset growth restriction it is concerning for digynic triploidy.***

Growth Restriction



The graphics show the fetal data points at 19 and 21 weeks plotted on graphs of the 10th to 90th percentiles for gestational age.

-The graph on the left shows fetal weight on the y-axis against gestational age on the x-axis. -

-The graph on the right shows head circumference/abdominal circumference on the y-axis against gestational age on the x-axis.

This fetus is well below normal for fetal weight and displays an elevated head circumference to abdominal circumference ratio compared to normally developing fetuses.

Final Diagnosis made by Amniocentesis:

Triploidy

Two types of triploidy

Diandric triploidy (more common)

- Extra set of chromosomes is paternal (69 XYY)
- Cystic placenta
- Pathogenesis
  - Caused either by dispermy (more common) or fertilization with diploid sperm
    - Results in partial mole with hydropic placenta, symmetric fetal growth retriction

Digynic triploidy

- Extra set of chromosomes is maternal (69 XXY)
- “Head sparing” with severe asymmetric fetal growth restriction
- Pathogenesis
  - Caused by fertilization of diploid ovum
    - Results in extreme placental hypoplasia characterized by marked hypermature and fibrotic villi

Take home points

- **Triploidy is a lethal diagnosis**
- **Think about triploidy when**
  - **Severe early onset fetal growth restriction**
  - **Low fluid associated with early growth restriction**
  - **Head size > abdominal circumference suggest digynic**

Acknowledgements

- Dr. Anne Kennedy, MB, BCH, BAO
- University of Utah Department of Radiology
- University of Utah School of Medicine

References:

1) Witters, G., Robays, J.V., Willekes, C., Coumans, A., Peeters, H., Gyselaers, W., Fryns, J.P. (2011). Trisomy 13, 18, 21, Triploidy and Turner syndrome: the 5T's. Look at the hands. *Facts, Views and Vision. Issues in Obstetrics, Gynaecology and Reproductive Health in ObGyn* (3), 15-21.

2) Kang, H. and Rosenwaks, Z. (2008). Triploidy – the breakdown of monogamy between sperm and egg. *The International Journal of Developmental Biology* (52), 449-454.

3) Benn, P., Cuckle, H., Pergament (2013). Non-invasive prenatal testing for aneuploidy: current status and future prospects. *Ultrasound in Obstetrics and Gynecology* (42), 15-33.