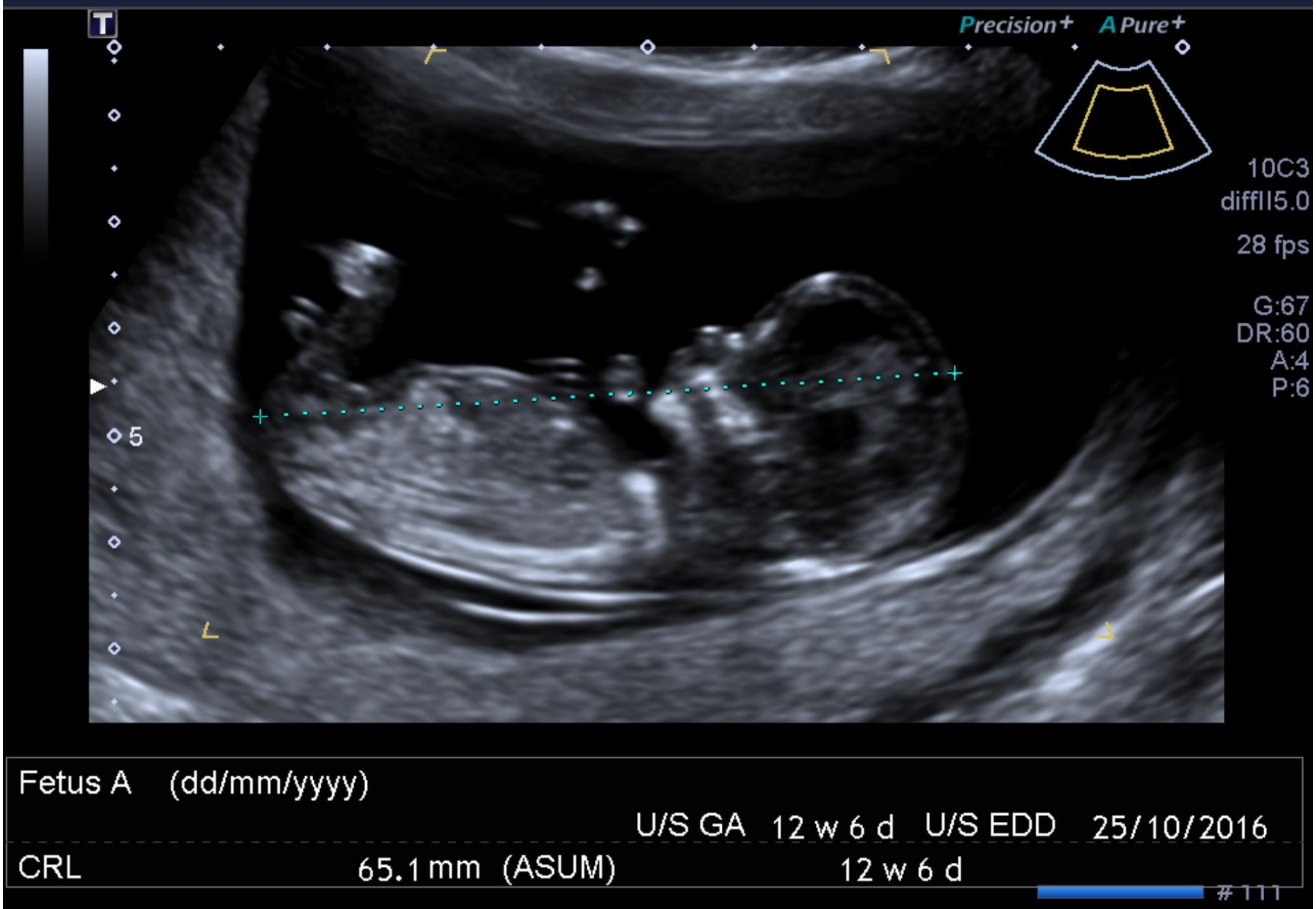
First trimester ultrasound screening: detection of fetal structural anomalies

- Screening for trisomy 21 and other chromosomal defect
- Early anatomic evaluation
- CRL (must be 45 to 84 mm, gestation age 11 weeks 3 days to 13 weeks 6 days)
- NT,FHR,NB,anencephaly,holoprosencephaly,ductus venosus flow
- gastroschisis omphalocele,megacystis

CRL

- Fetus fill the majority of the image
- Fetal spine midsagittal
- Profile,spine,and rump are visible
- Spine is in line with the head
- Fluid is visible between fetal chest and chin
- Angle of insonation perpendicular to fetus
- Fetus horizontal on image
- Calipers are placed on the outer border of the skin at crown and rump


Precision+
A Pure+
10C3
diffII5.0
28 fps
G:67
DR:60
A:4
P:6
5
Fetus A (dd/mm/yyyy)
U/S GA 12 w 6 d U/S EDD 25/10/2016
CRL 65.1 mm (ASUM) 12 w 6 d
111

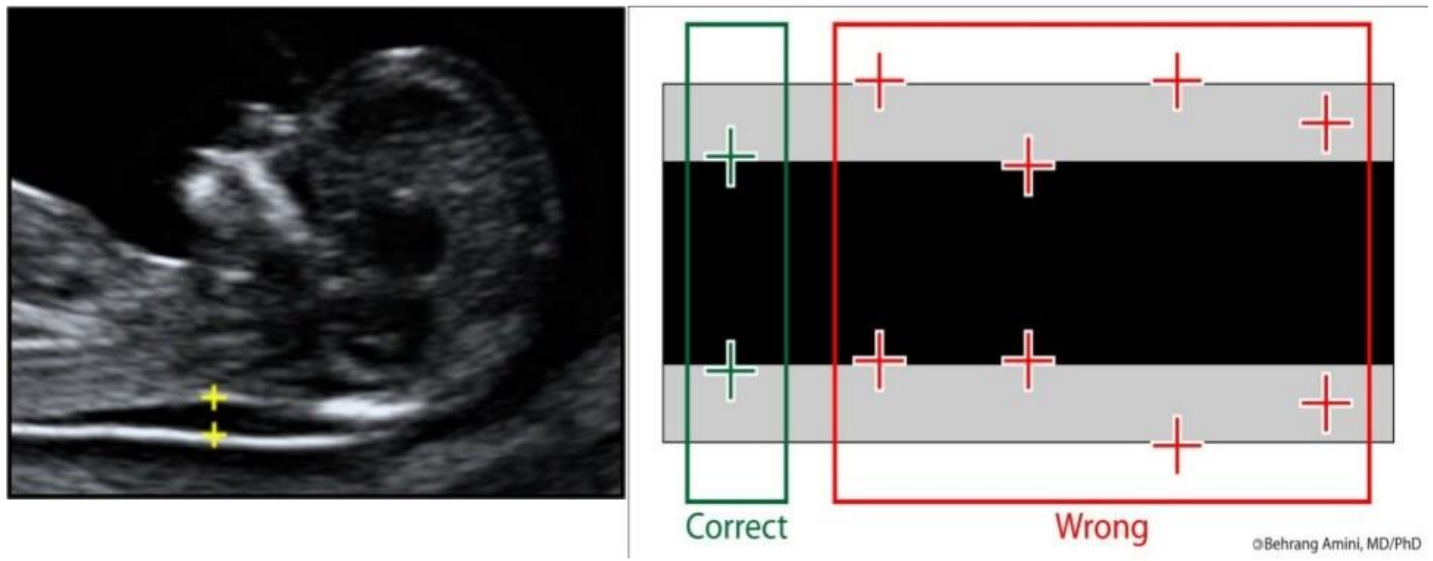
- The fetal CRL must be between 45 and 84 mm although variations exist among laboratories that perform the biochemical component. The accuracy of the NT measurement and the CRL is critical because the NT measurement is converted into multiples of the median (MoM) based on the CRL. An adequate sonographic image to measure the CRL requires the fetus to occupy a majority of the image space and be in a neutral position. The longest straight line between the fetal crown and rump is measured at least three times and the average of three good measurements is used.

NT

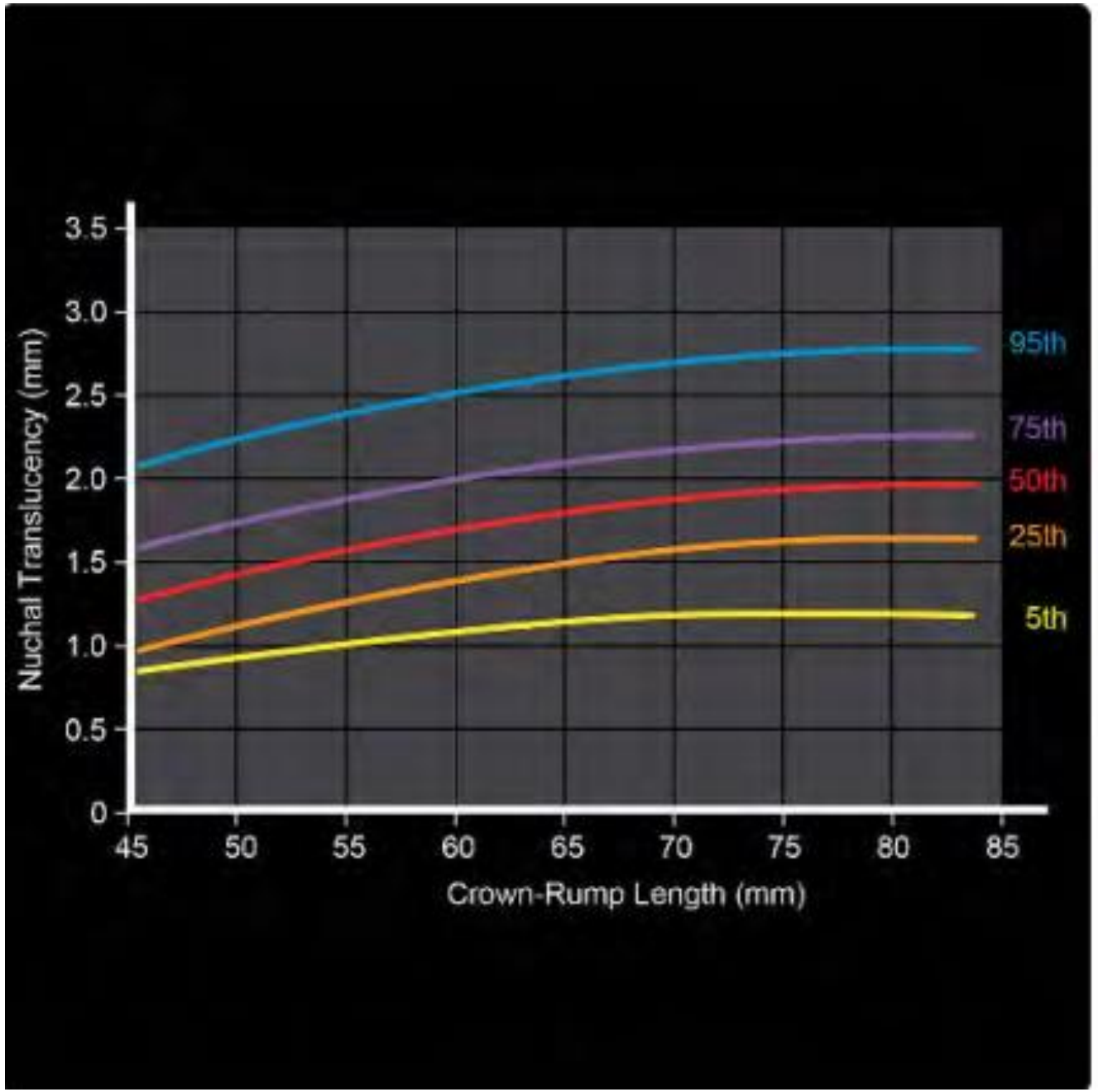
- Angle of insonation perpendicular to NT space
  Fetus horizontal on image
  TIPS for optimal imaging:
  1. Optimize your focal zone
  2. Reduce your dynamic range
  3. Reduce the gain
  4. Review harmonics. Possible edge enhancement optimized with harmonics off
  5. Avoid post freeze zoom
  6. Narrow your sector

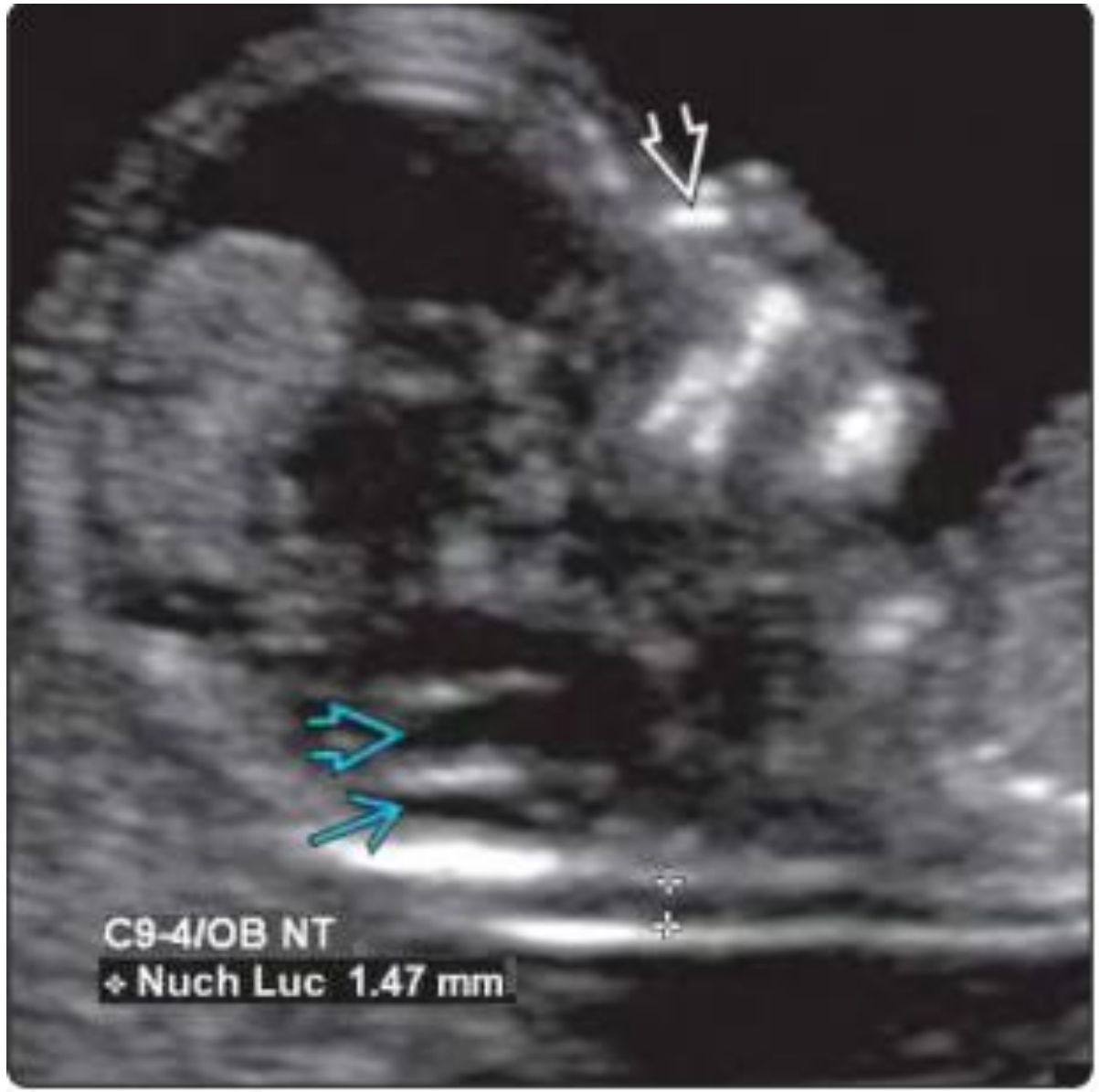
- Fetal spine midsagittal in thoracic and cervical region
- Tip of nose in profile
- Third and fourth ventricles in brain demonstrated
- Head, neck, and upper thorax Fill image
- Fetus occupies more than 50% of image space
- Head in line with the spine
- Pocket of fluid should be visible between chin and neck
- Fetus is seen away from uterine wall and separate from the amnion
- Calipers (cursors) must be + (plus/positive)
- Crossbar of caliper is on the NT line at the inner border adjacent to the lucency
- Measurement is perpendicular to long axis of fetus
- Measurement is taken at the WIDEST part of the lucency (If nuchal cord, measure above and below the cord and average)

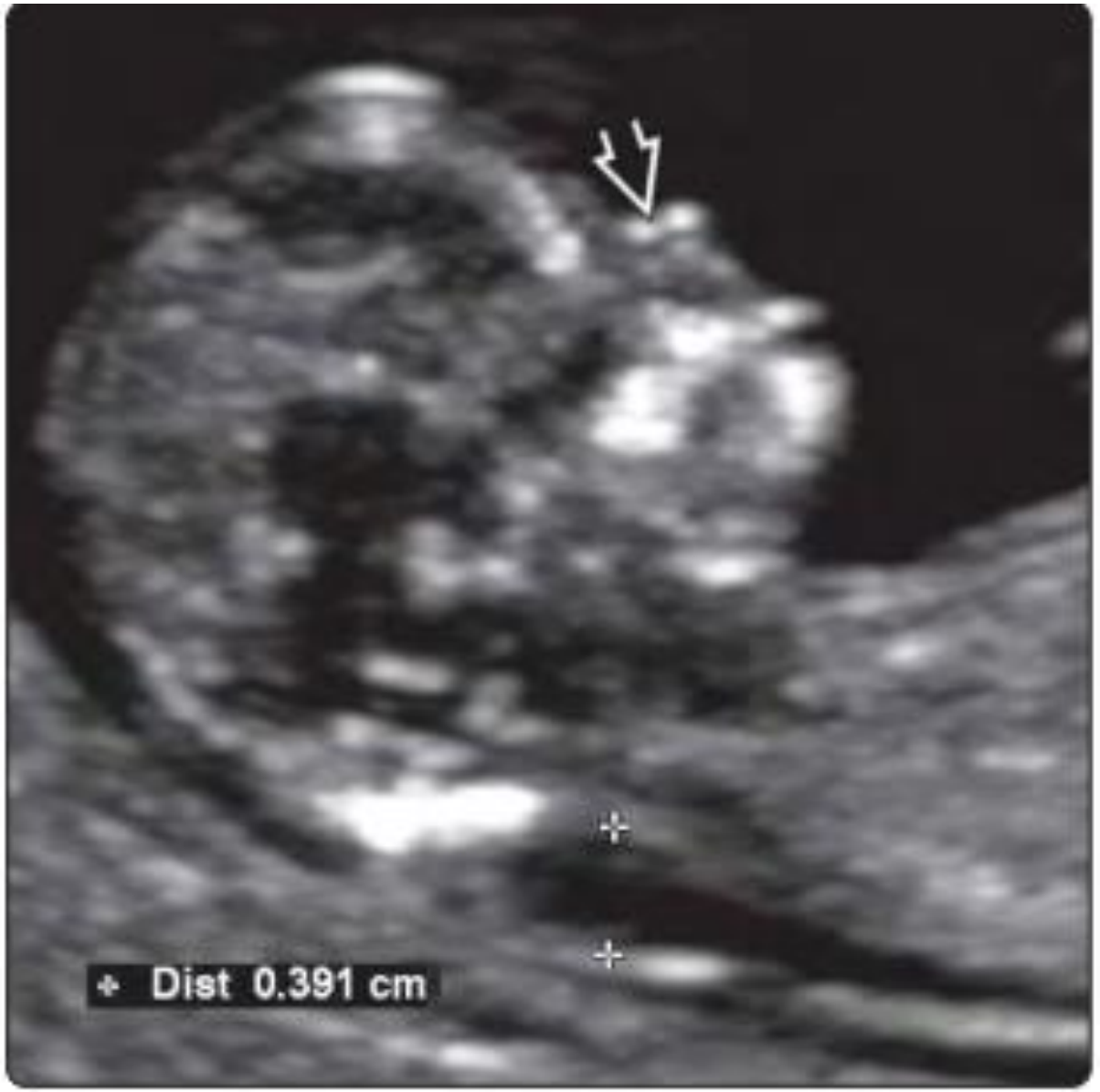
Callipers



- The crossbar of the calliper should be hardly visible as it merges with the white line of the border and not in the nuchal fluid.


Nuchal Translucency (mm)
3.5
3.0
2.5
2.0
1.5
1.0
0.5
0
45
50
55
60
65
70
75
80
85
Crown-Rump Length (mm)
95th
75th
50th
25th
5th

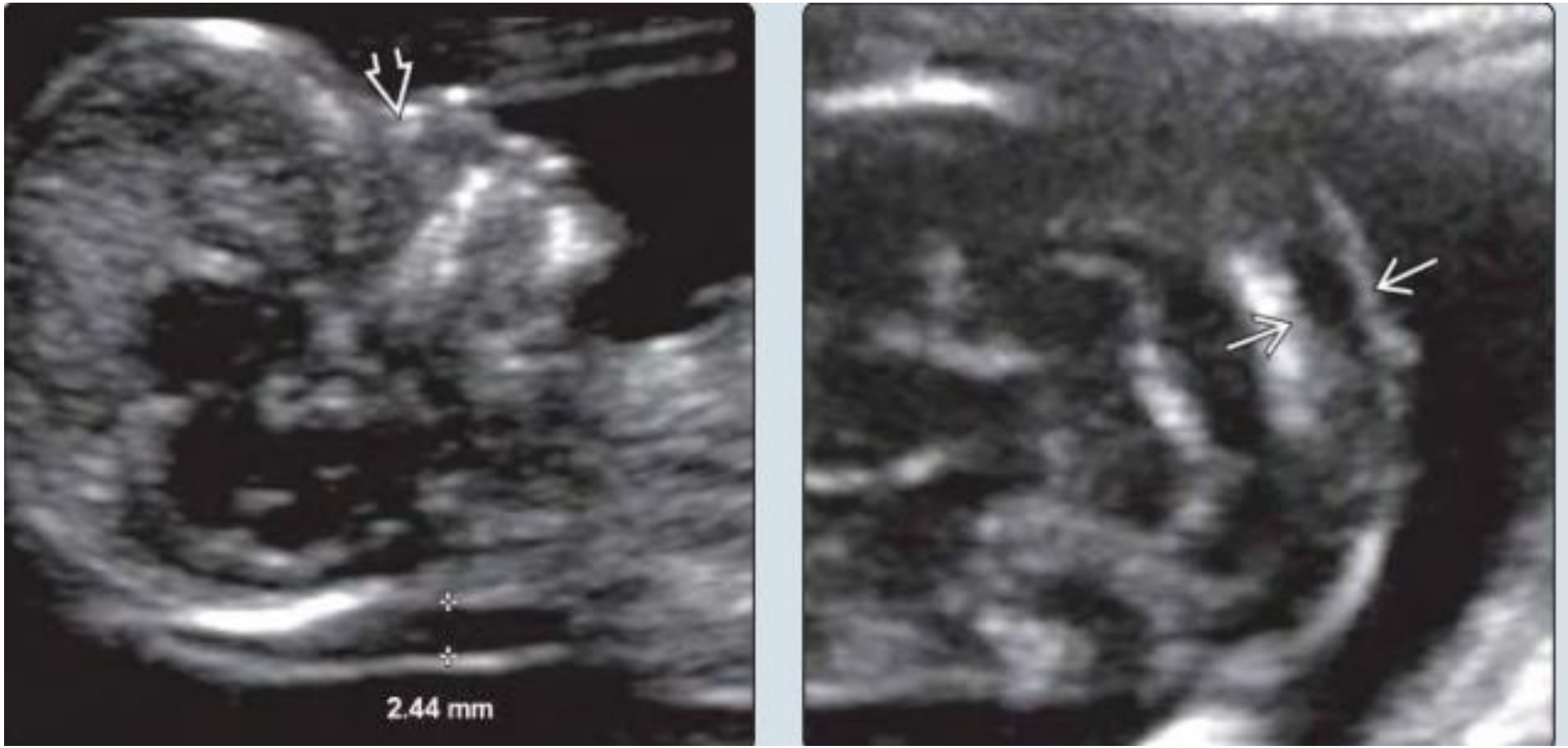

C9-4/OB NT
Nuch Luc 1.47 mm

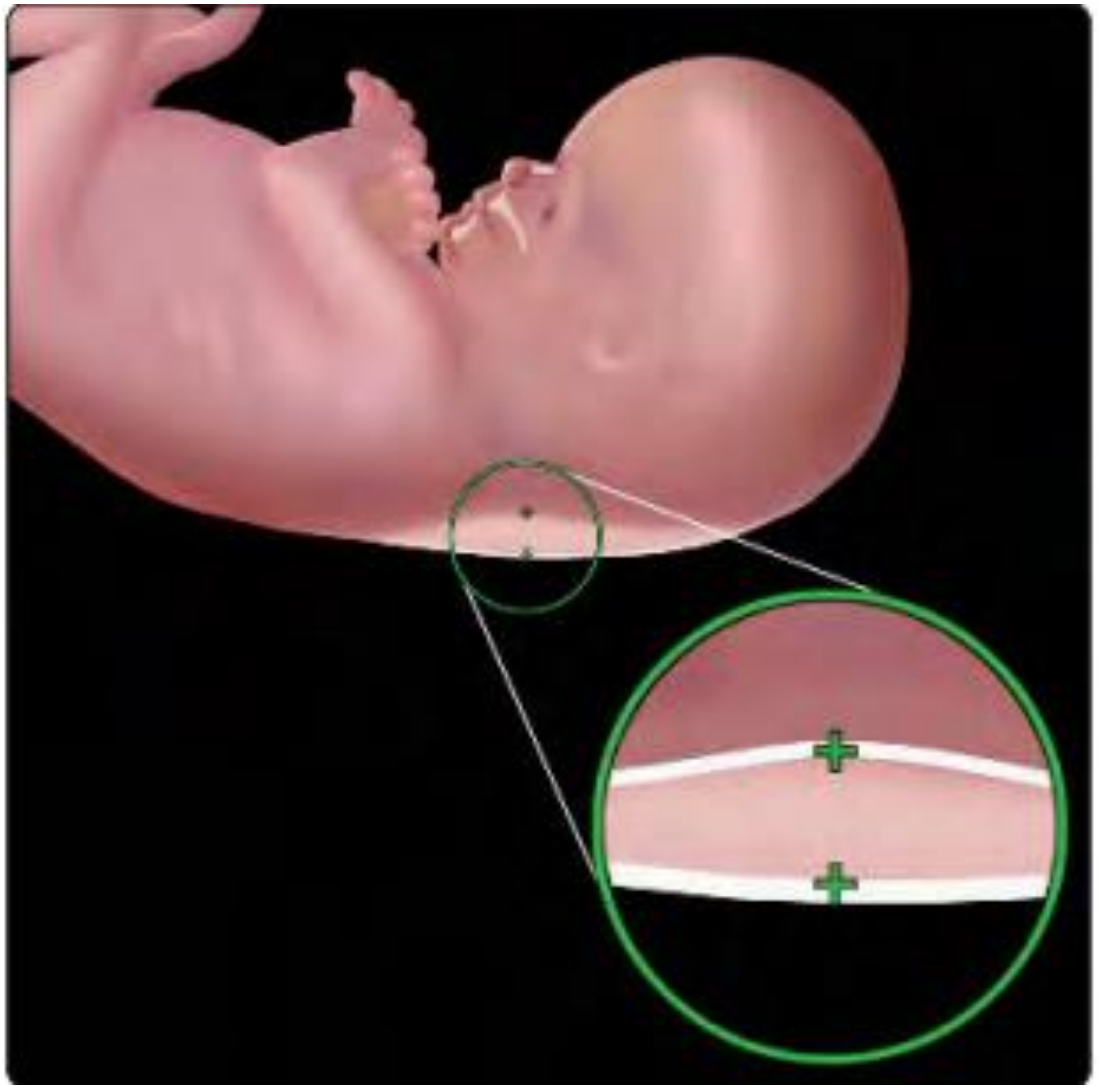

Dist 0.391 cm

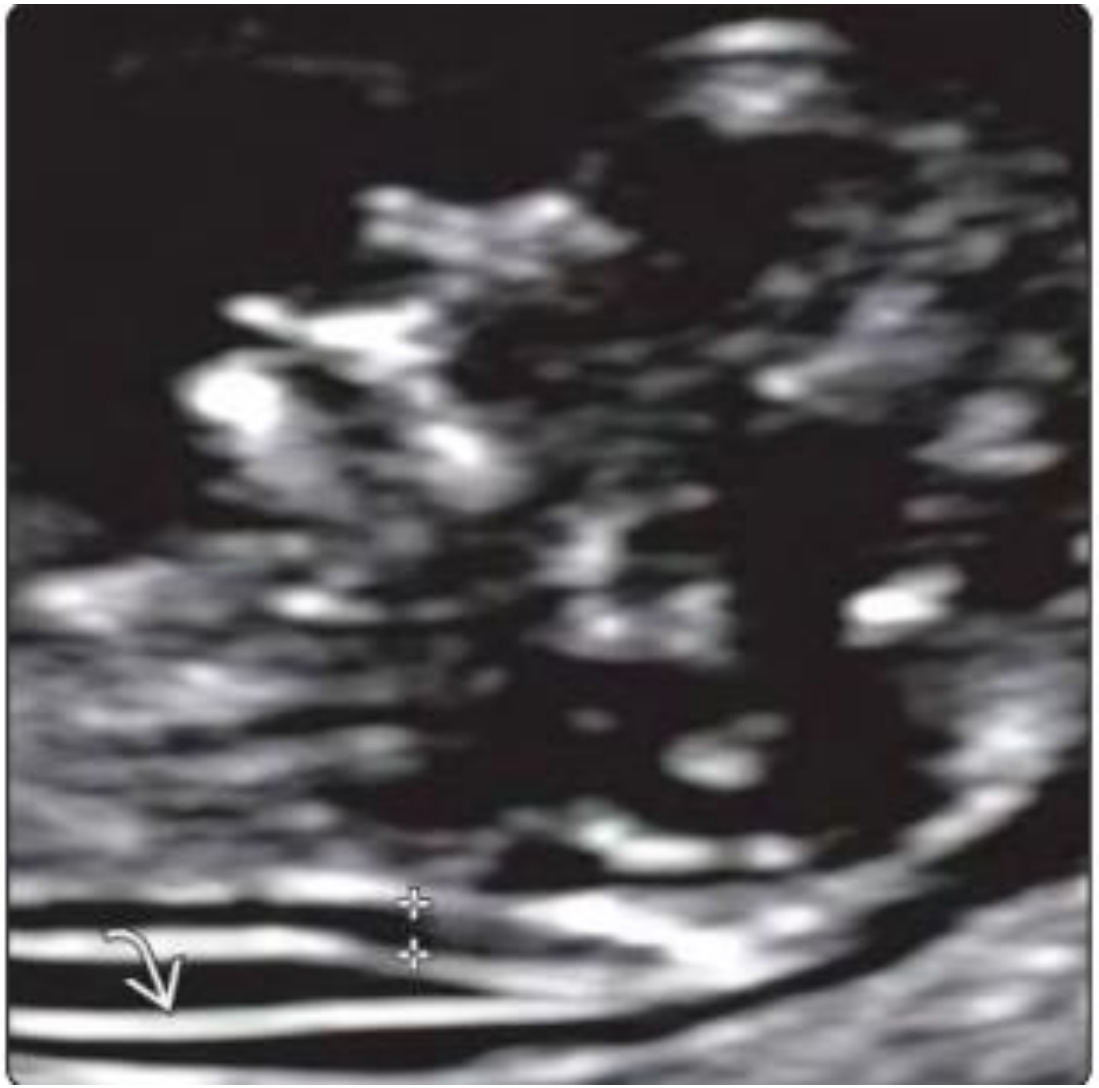
Trisomy 21

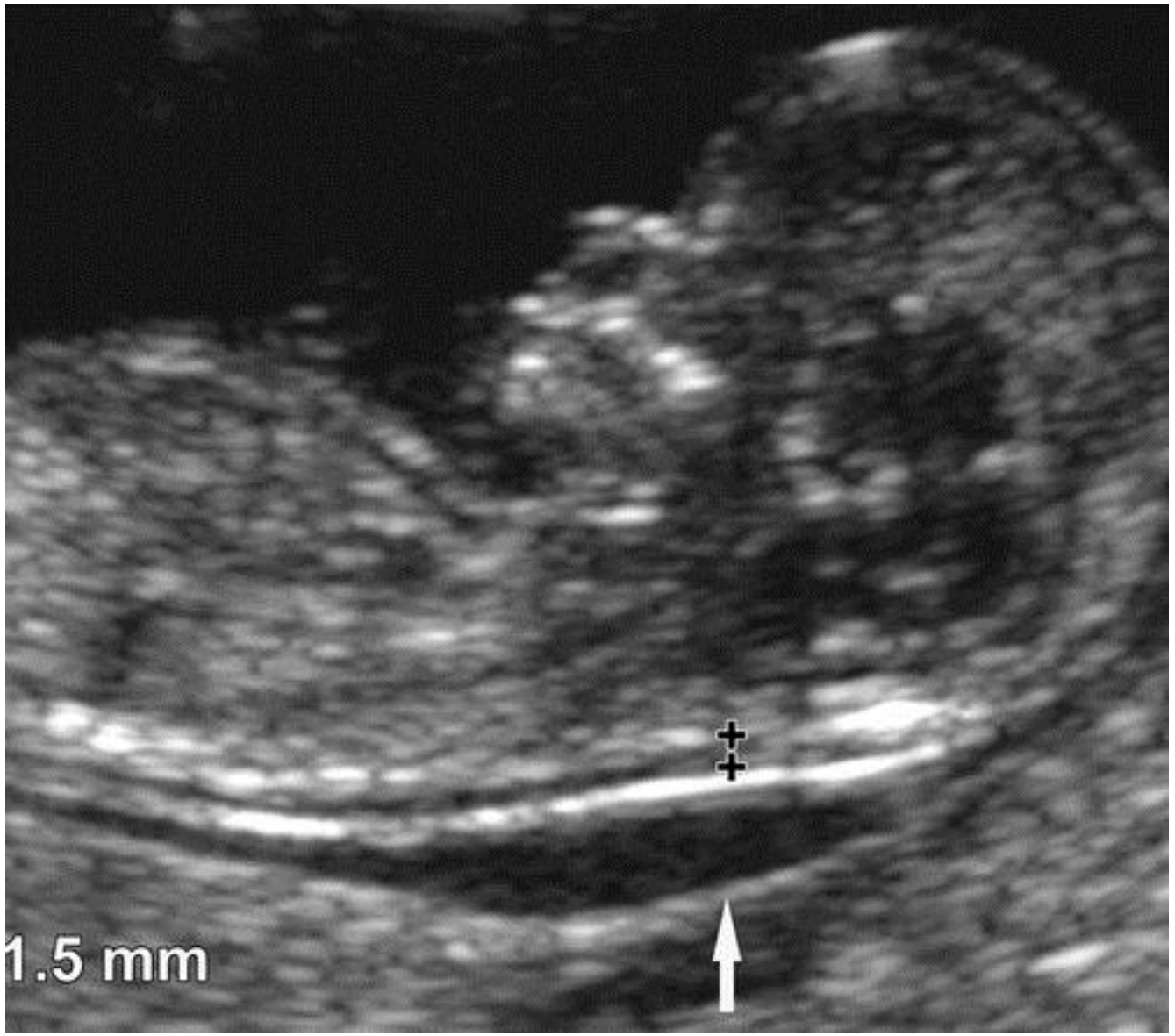
1st-trimester findings (11- to 14-week scan)
- ↑ NT: ↑ fluid behind neck on midsagittal view
- Absent nasal bone
- Abnormal ductus venosus and tricuspid flow
- Other anomalies may be seen

- For the NT, magnified view of fetal profile is obtained; neck is not overly flexed or extended, & calipers are placed so that only fluid is measured
  • Nasal bone (NB) is deemed present or absent on a midsagittal profile view; ultrasound beam is perpendicular to nose so that tip of nose, nasal bone, & frontal bone are all seen separately
- Additional anatomy survey can be performed at this time, a& up to 2/3 major anomalies are detectable at time of NT screening; anatomic survey includes visualization of normal falx with "butterfly choroid," facial profile, heart, cord insertion site, stomach, bladder, & extremities; in addition, number of umbilical cord vessels & placental cord insertion site can be seen well


2.44 mm

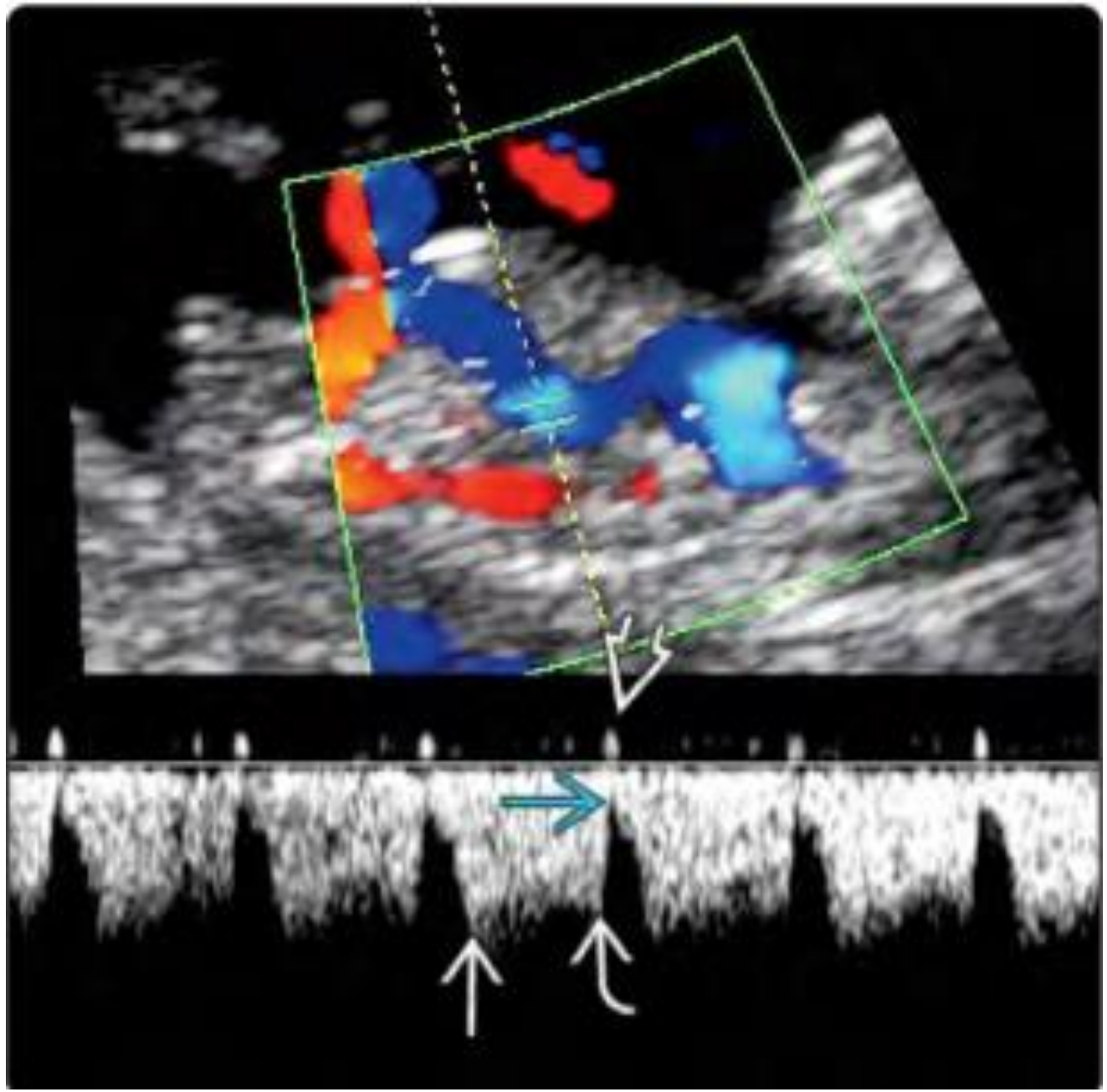


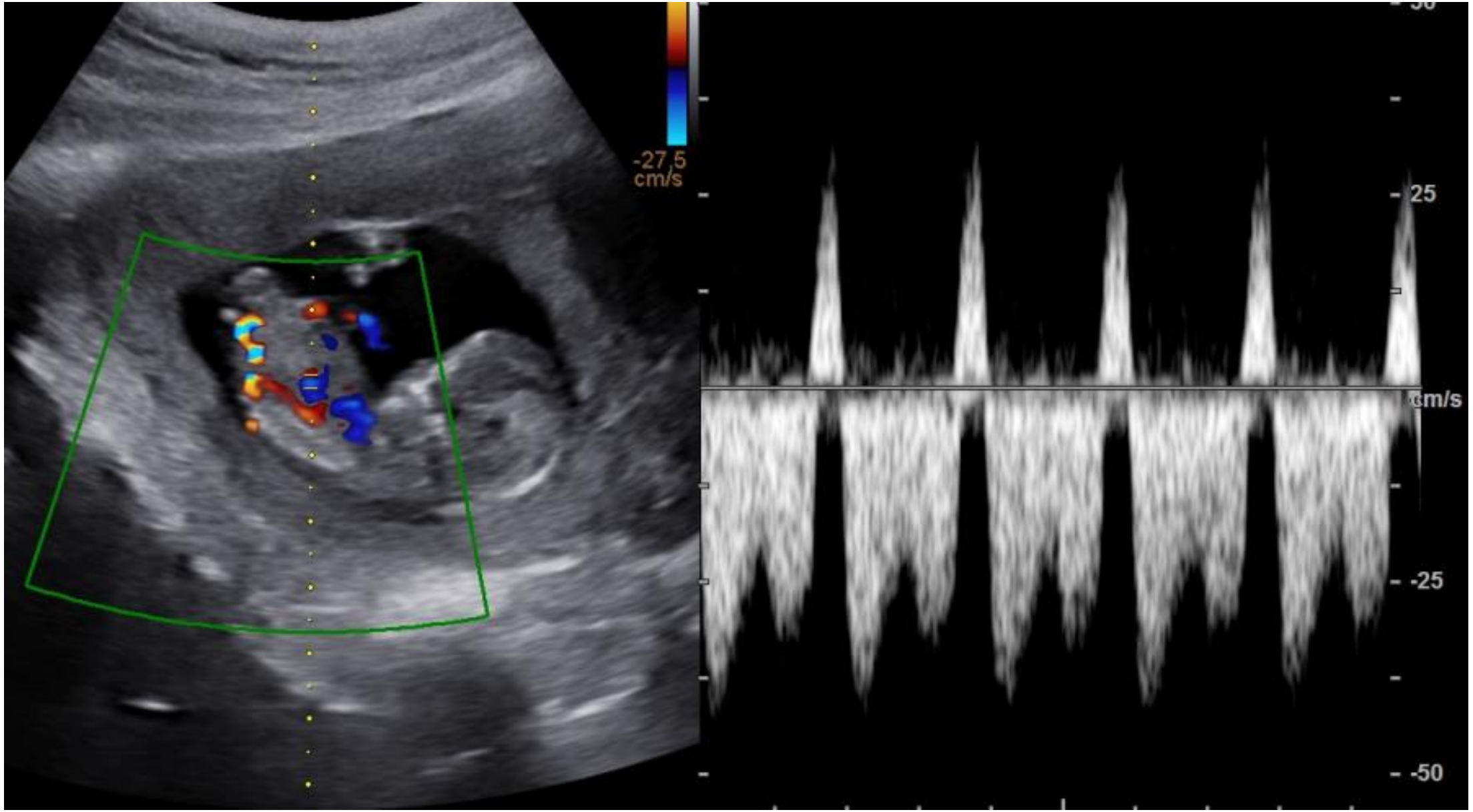



1.5 mm

Ductus Venosus

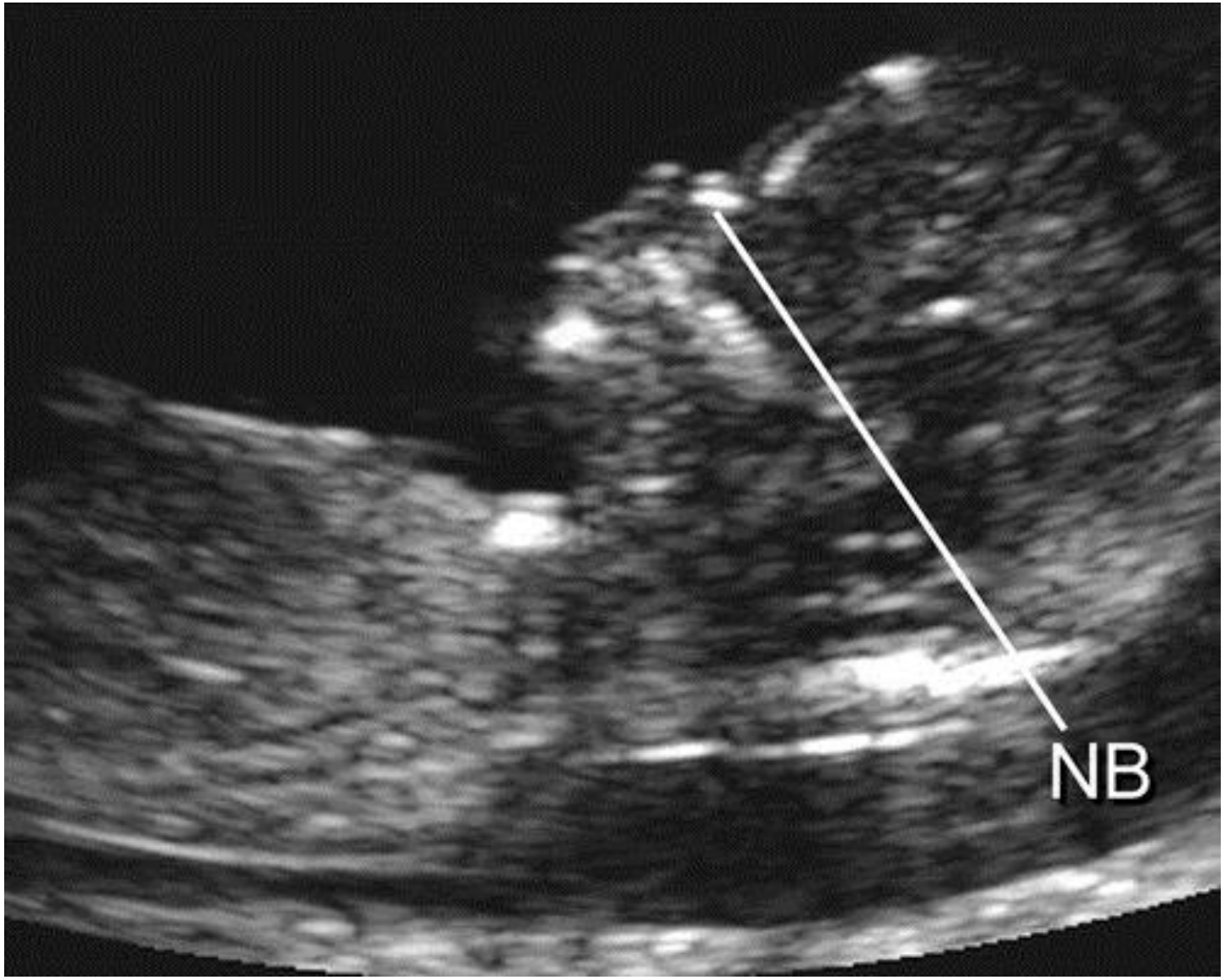
- Ductus venosus is small vessel with turbulent flow seen on sagittal view of lower chest & upper abdomen; normal flow is consistently toward heart, & retrograde flow is considered abnormal.

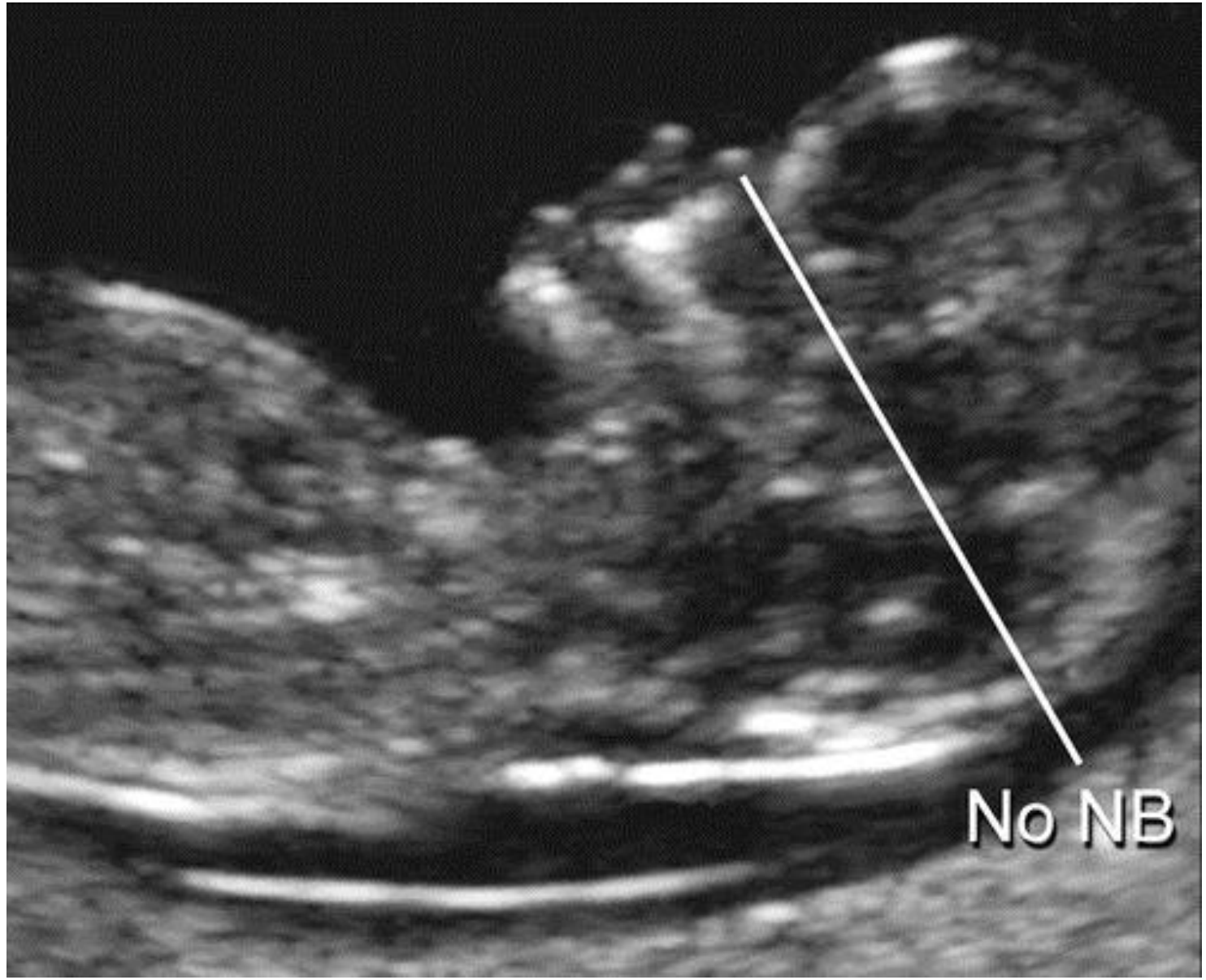


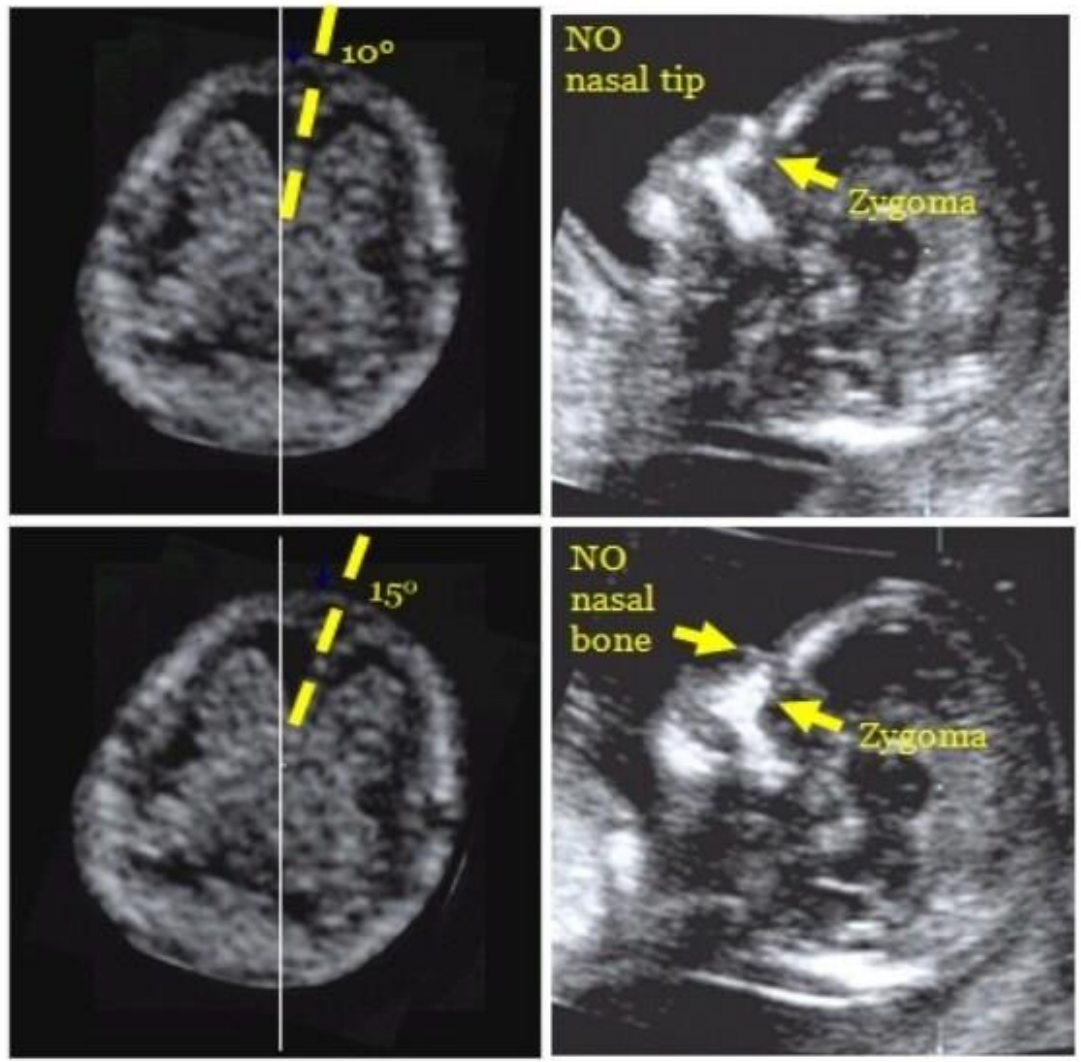

-27,5
cm/s
25
cm/s
-25
-50

NB

- Margins of fetal anatomy clear without ambiguity in nasal anatomy
- Fetus in midsagittal plane
- Fetal spine midsagittal in thoracic and cervical region
- Tip of nose clearly seen in proile and the skin edge over the nasal bridge is identiied
  Care must be taken to demonstrate the skin edge separately from the NB so that an equal (=) sign is apparent
- Third and fourth ventricle are identified in the brain
- Fetus occupies majority of image Head, neck, and upper thorax fill image
- Fetus occupies more than 50% of width and length of image
  Angle of insonation 45 degrees to fetal profile, perpendicular to NB
  Echogenicity of NB is comparable to other bony structures and similar or brighter than the overlying skin

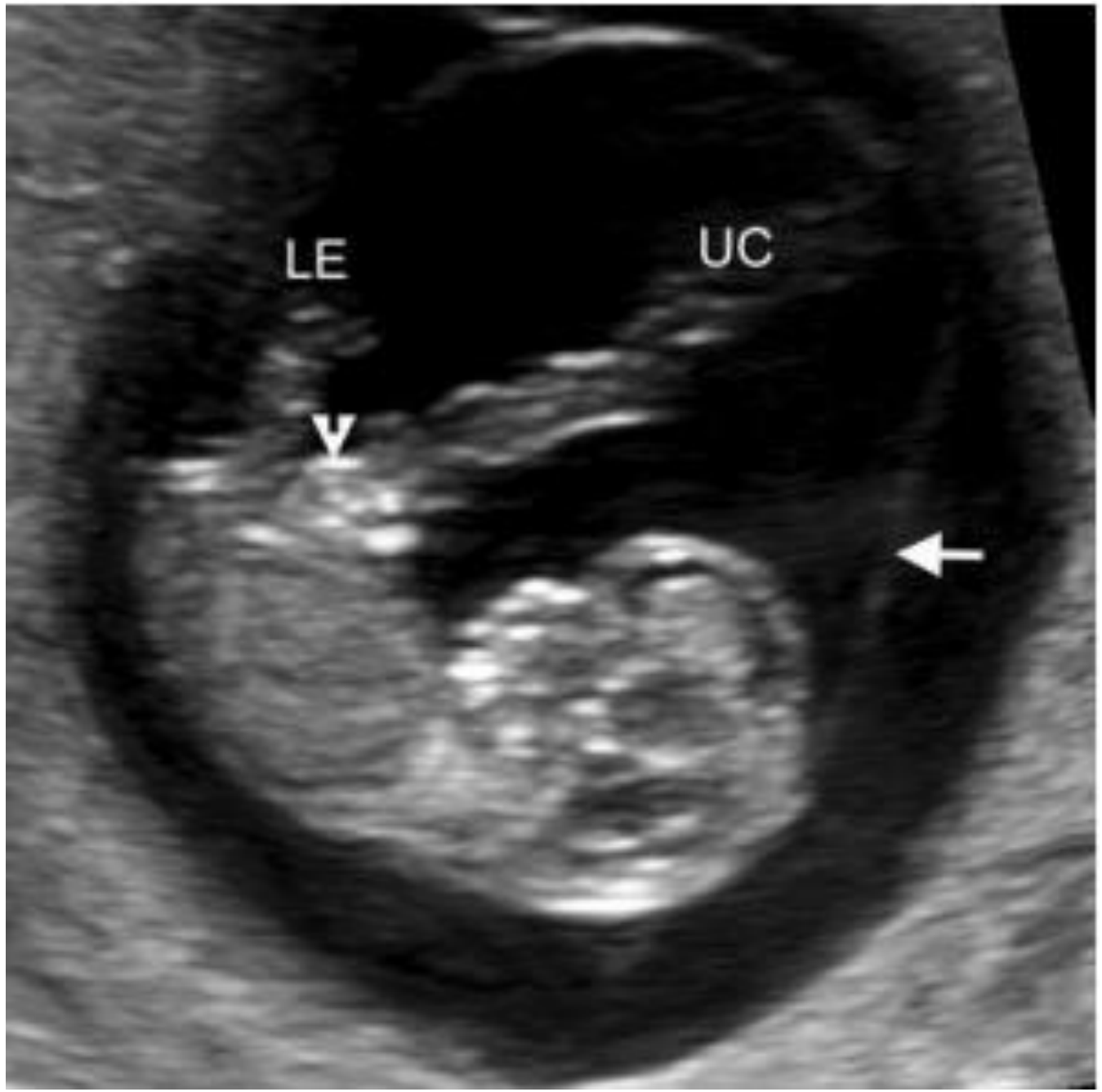

NB

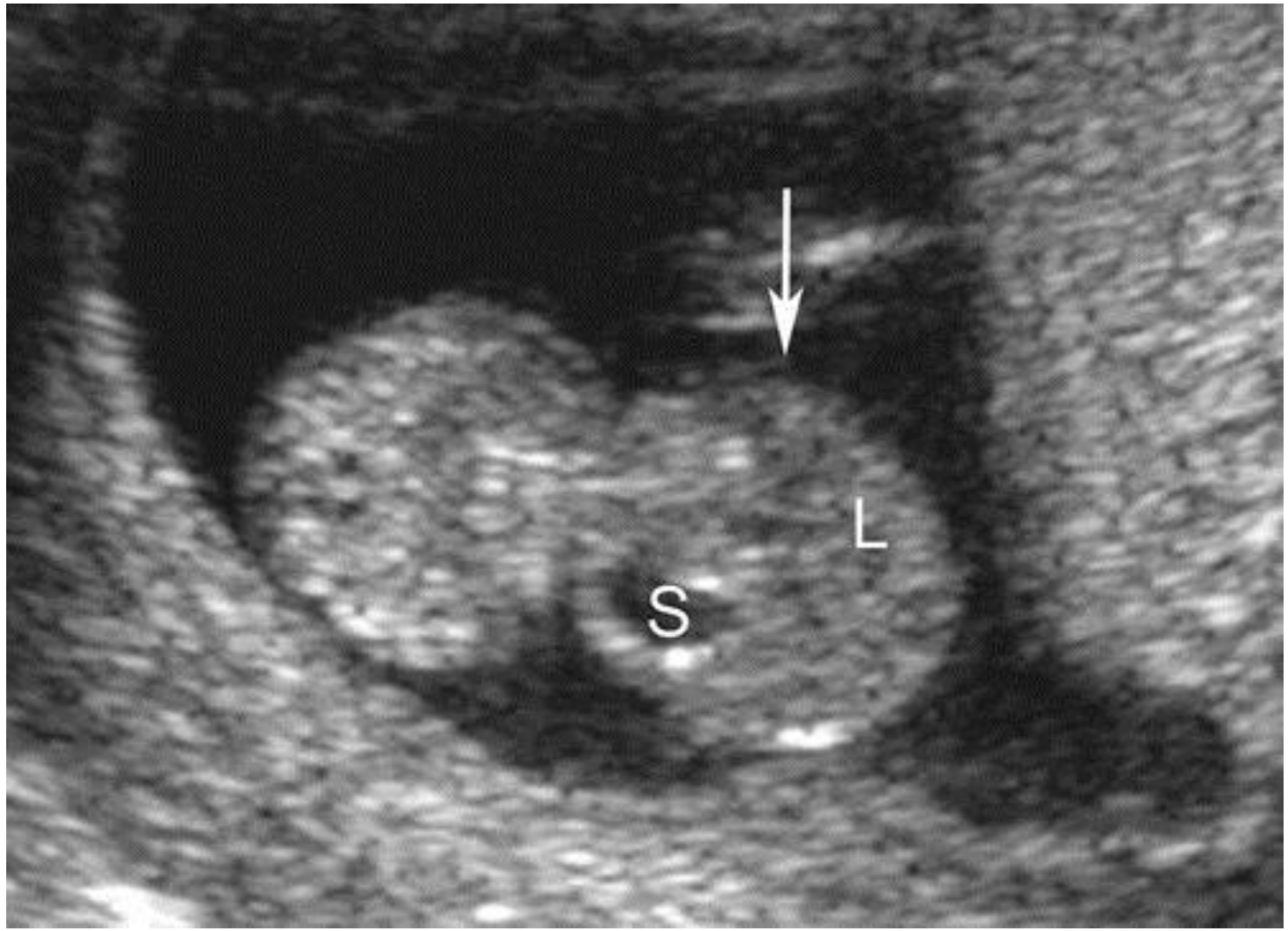

No NB


10°
NO
nasal tip
Zygoma
15°
NO
nasal
bone
Zygoma

Omphalocele

- Omphalocele refers to herniation of the intestine and other abdominal organs into the base of the umbilical cord through an enlarged umbilical ring, with the umbilical cord inserting at the apex of the herniated sac. The herniated content is covered by amnion and peritoneum .
- In the first trimester, **physiologic midgut herniation** can be mistaken for an omphalocele. this normal finding is limited to herniation of bowel loops and should resolve by 12 weeks. It should never include the liver, and the herniation should not be more than 1 cm into the cord.
- The presence of even small herniation into the base of the umbilical cord beyond 12 weeks is therefore diagnostic of omphalocele


LE
UC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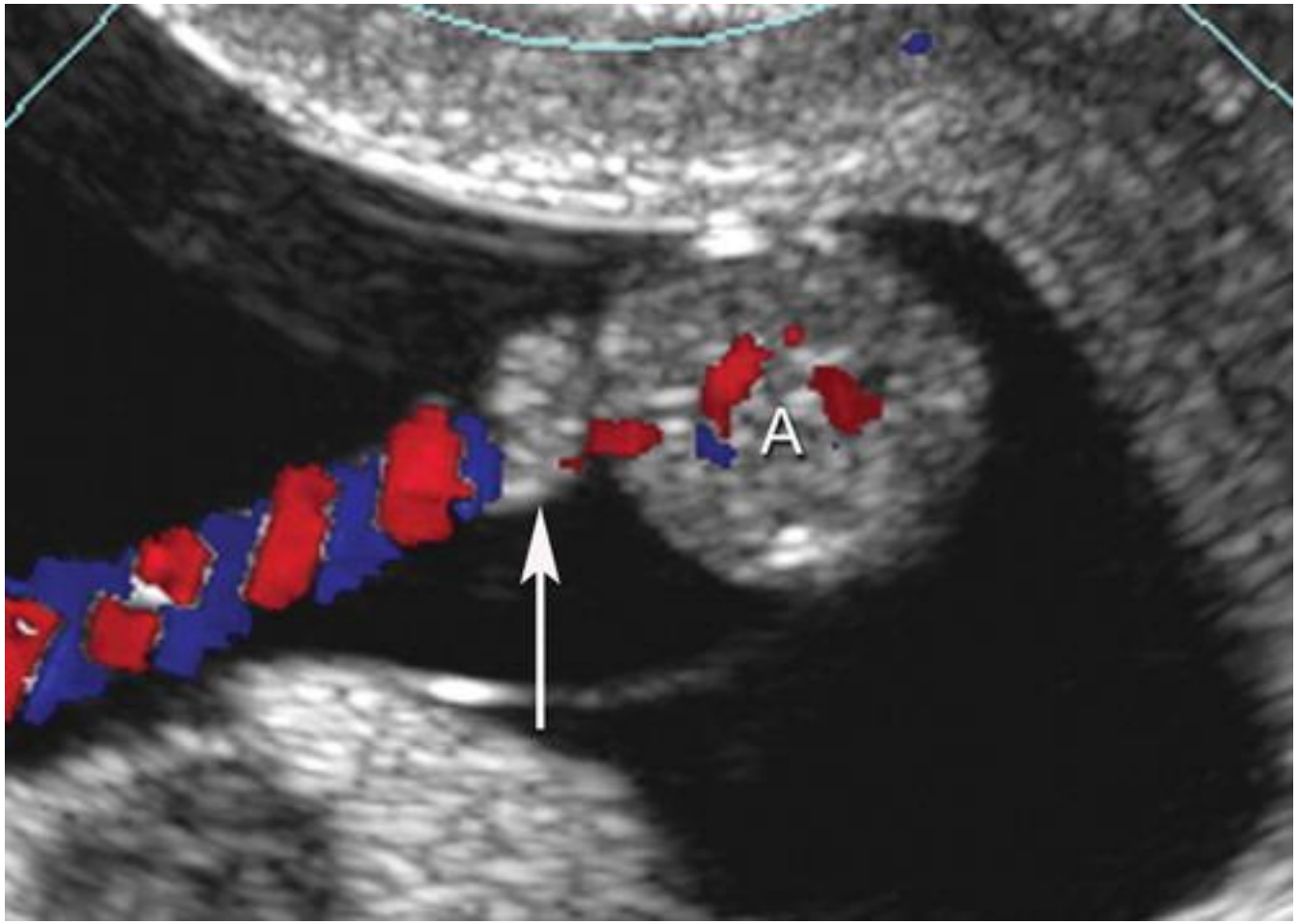
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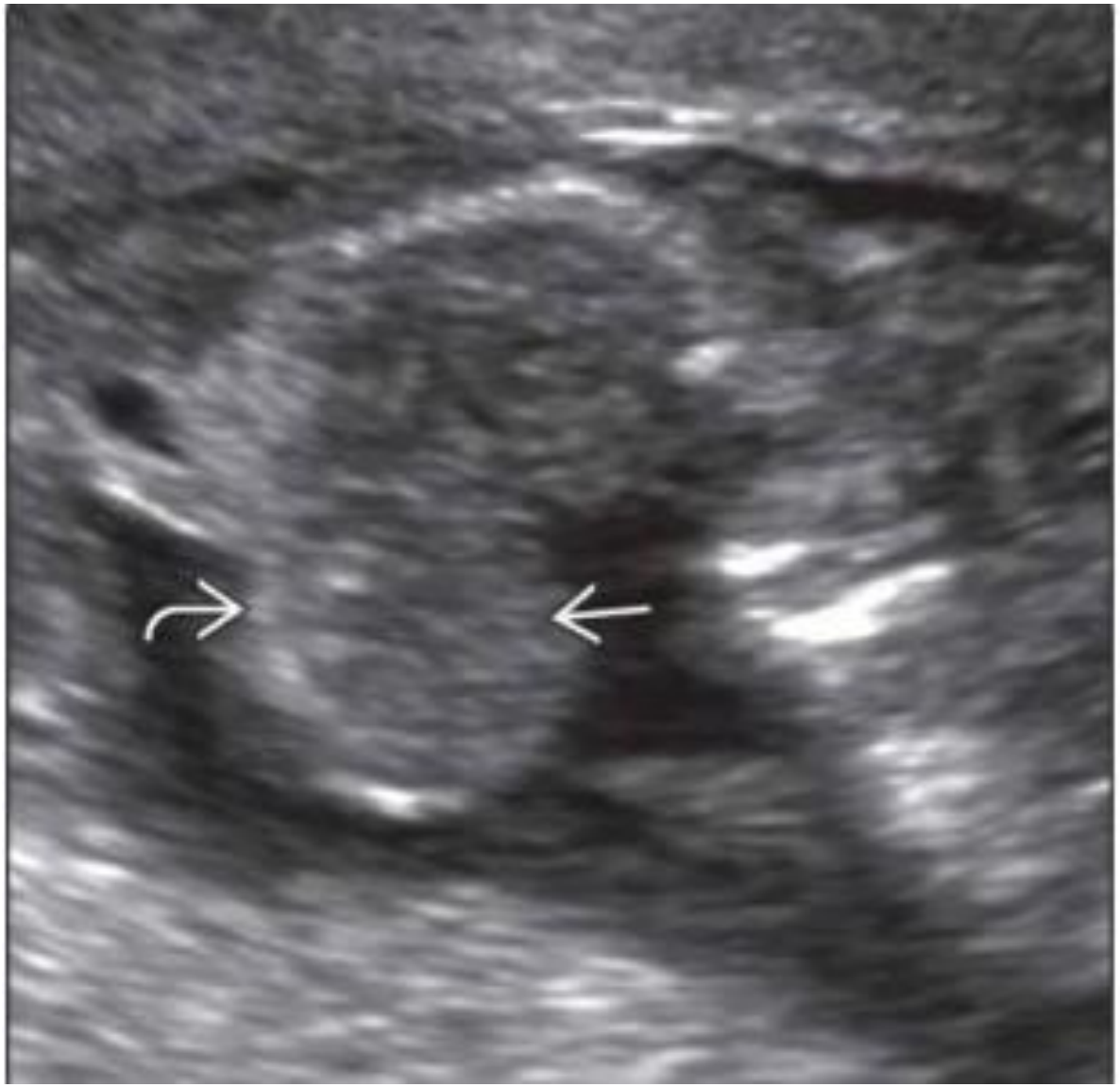
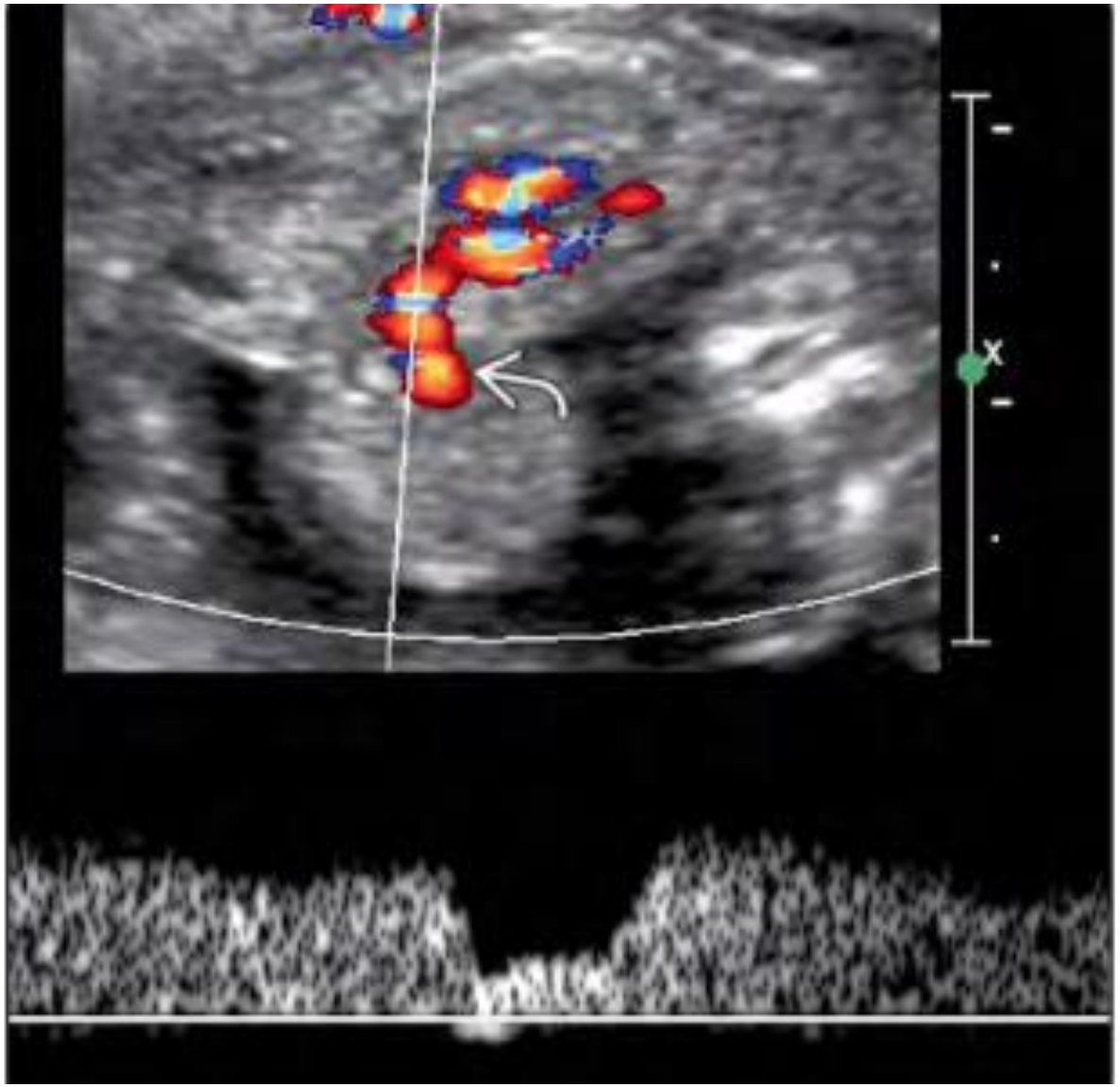
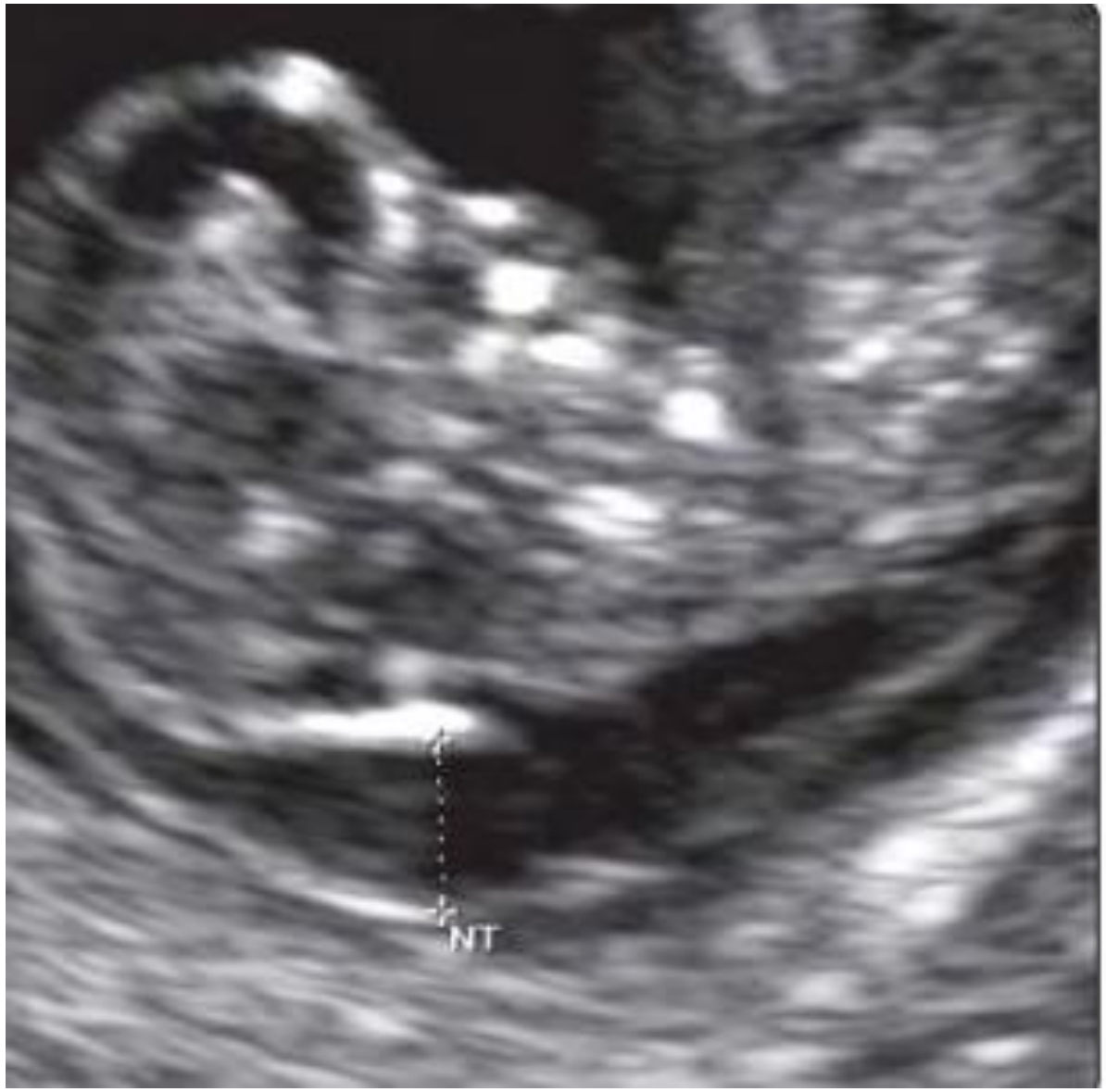
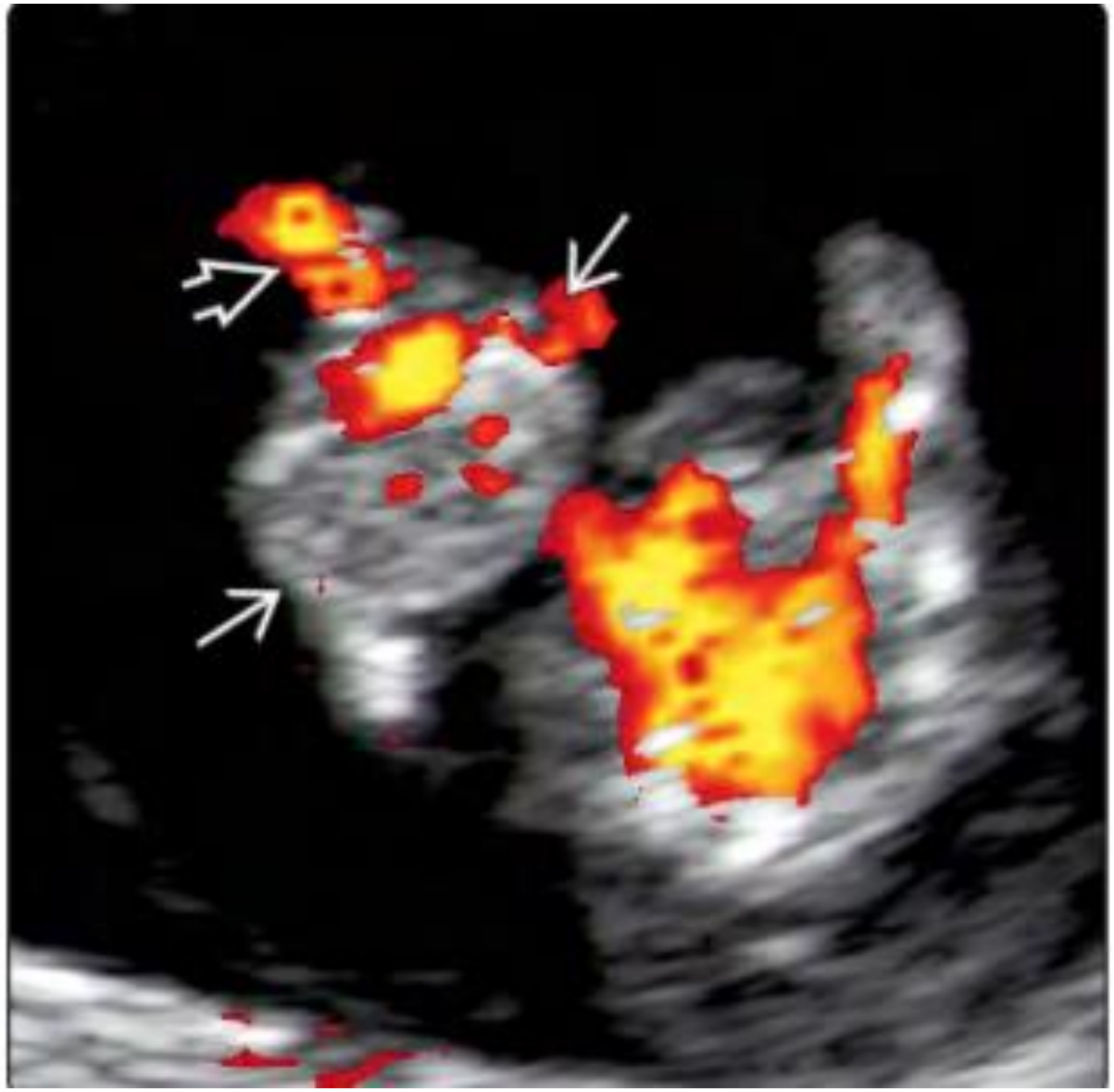
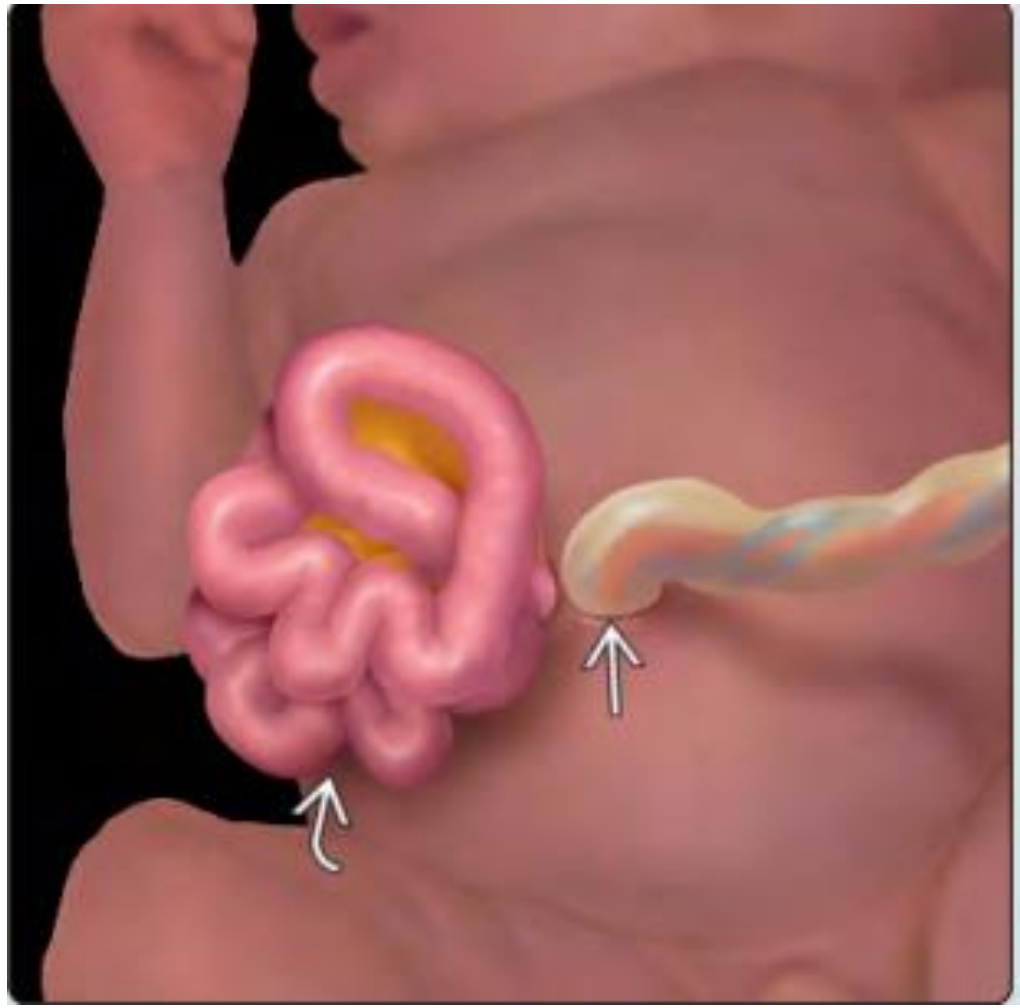


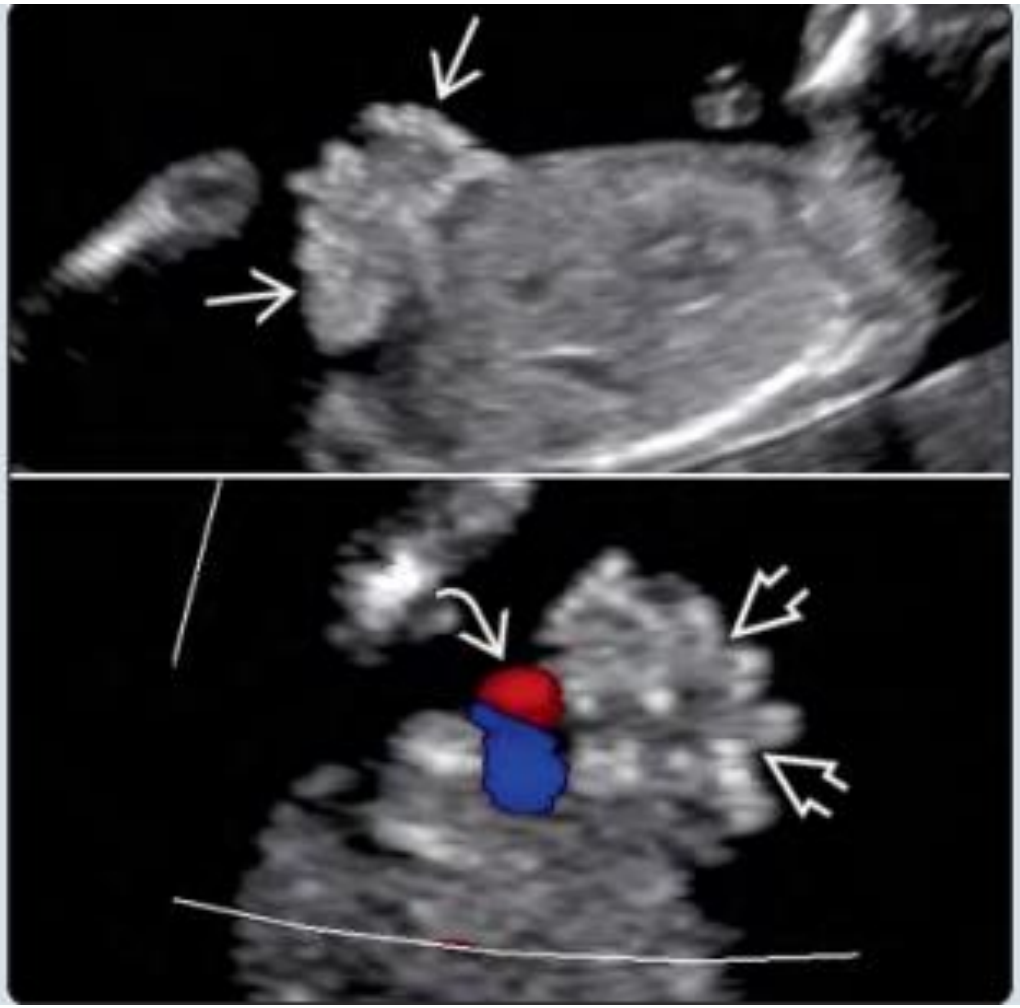


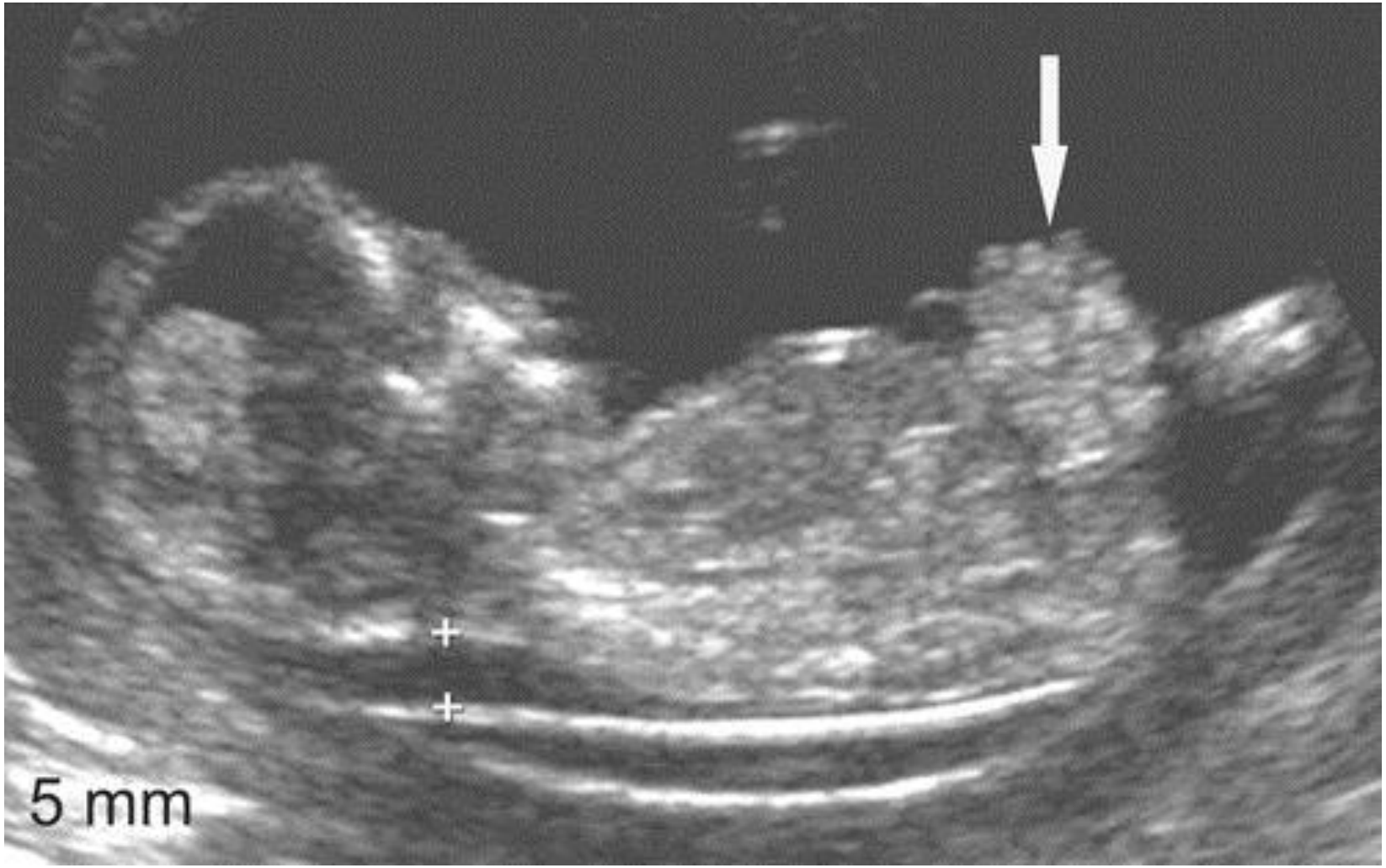

NT

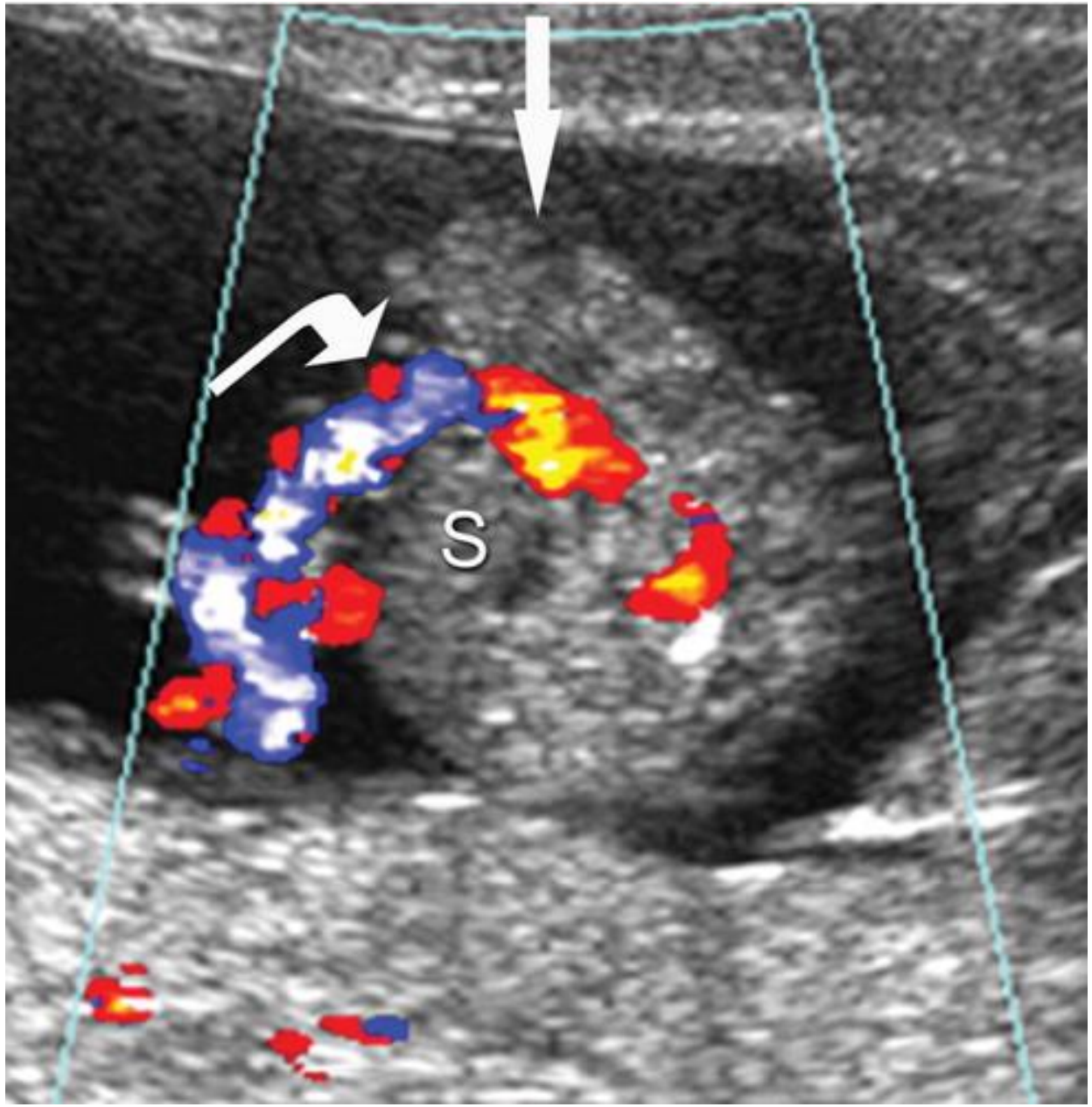


- **Gastroschisis** is a relatively small (<4 cm in most cases), full thickness paraumbilical defect of the abdominal wall, most often located to the right of the umbilicus. Free-floating loops of bowel in the amniotic fluid are the key finding on ultrasound






5 mm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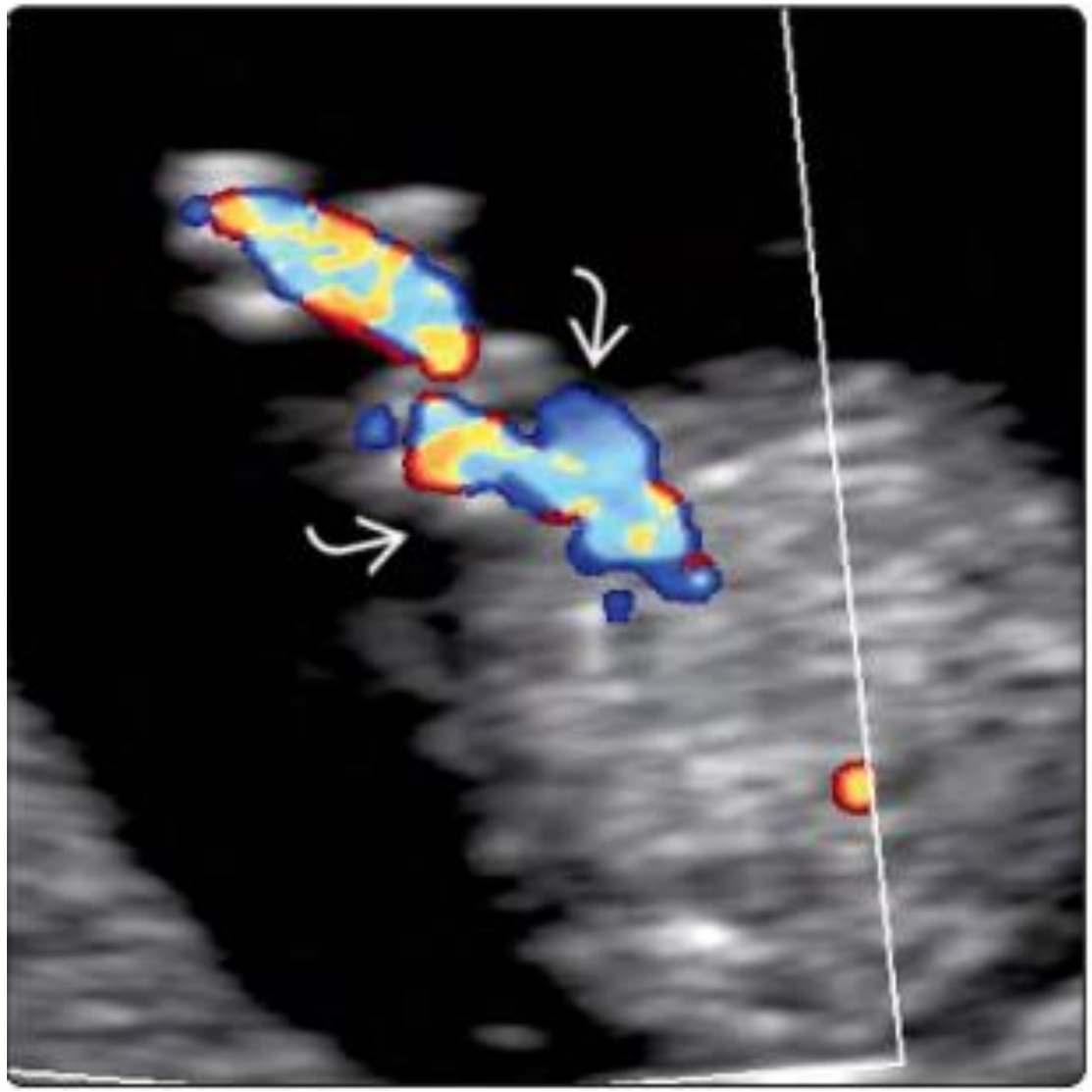
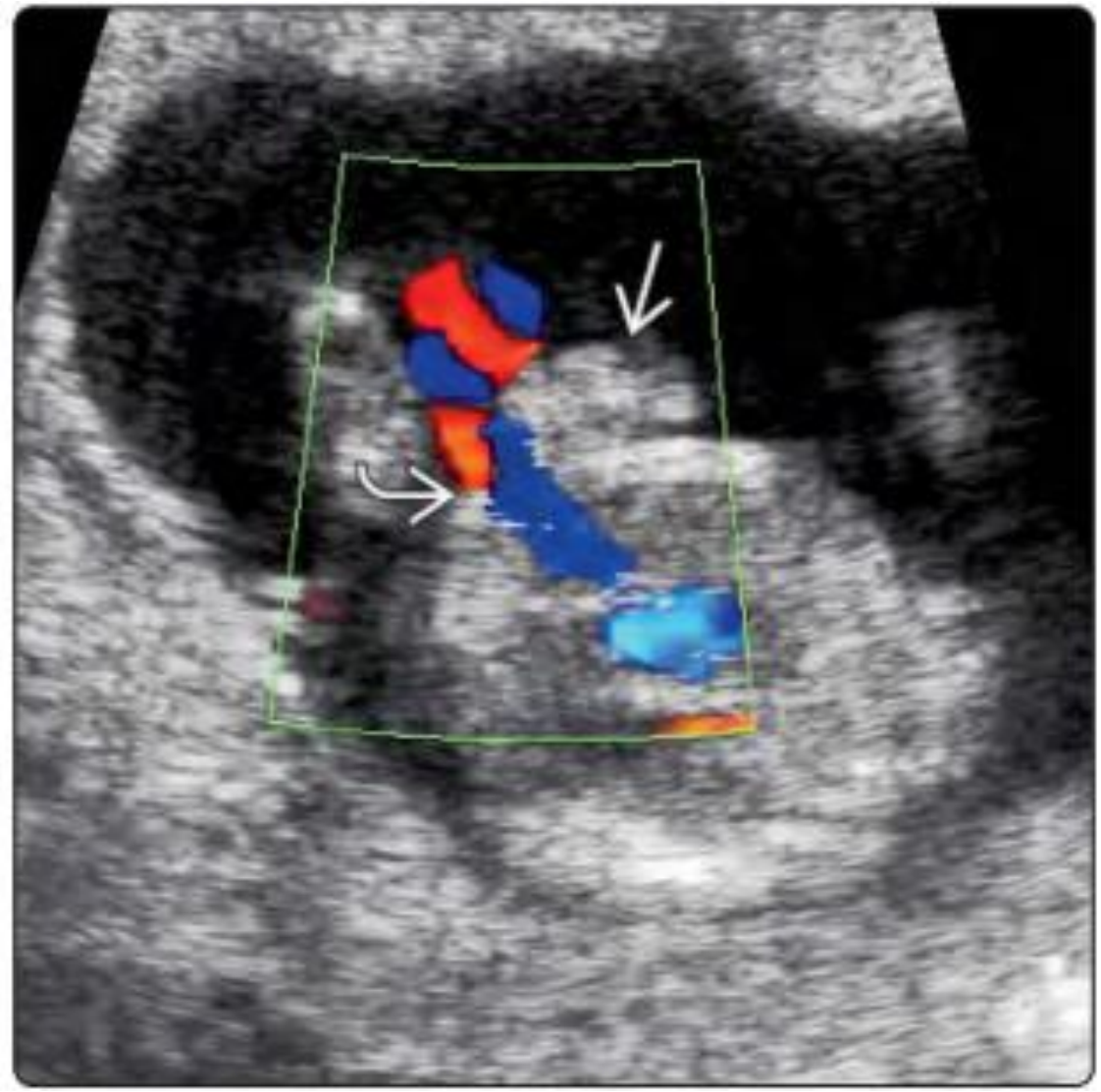
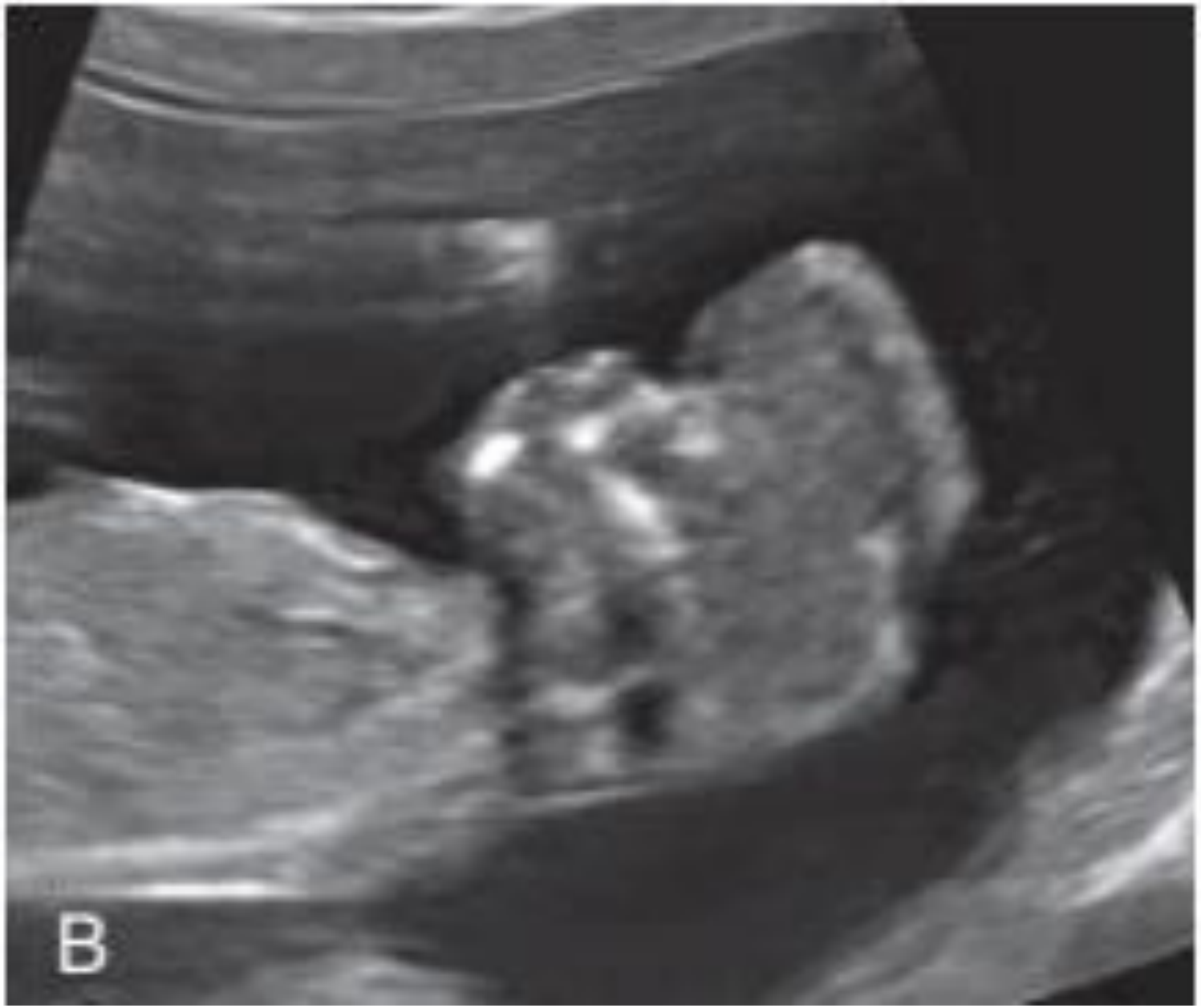




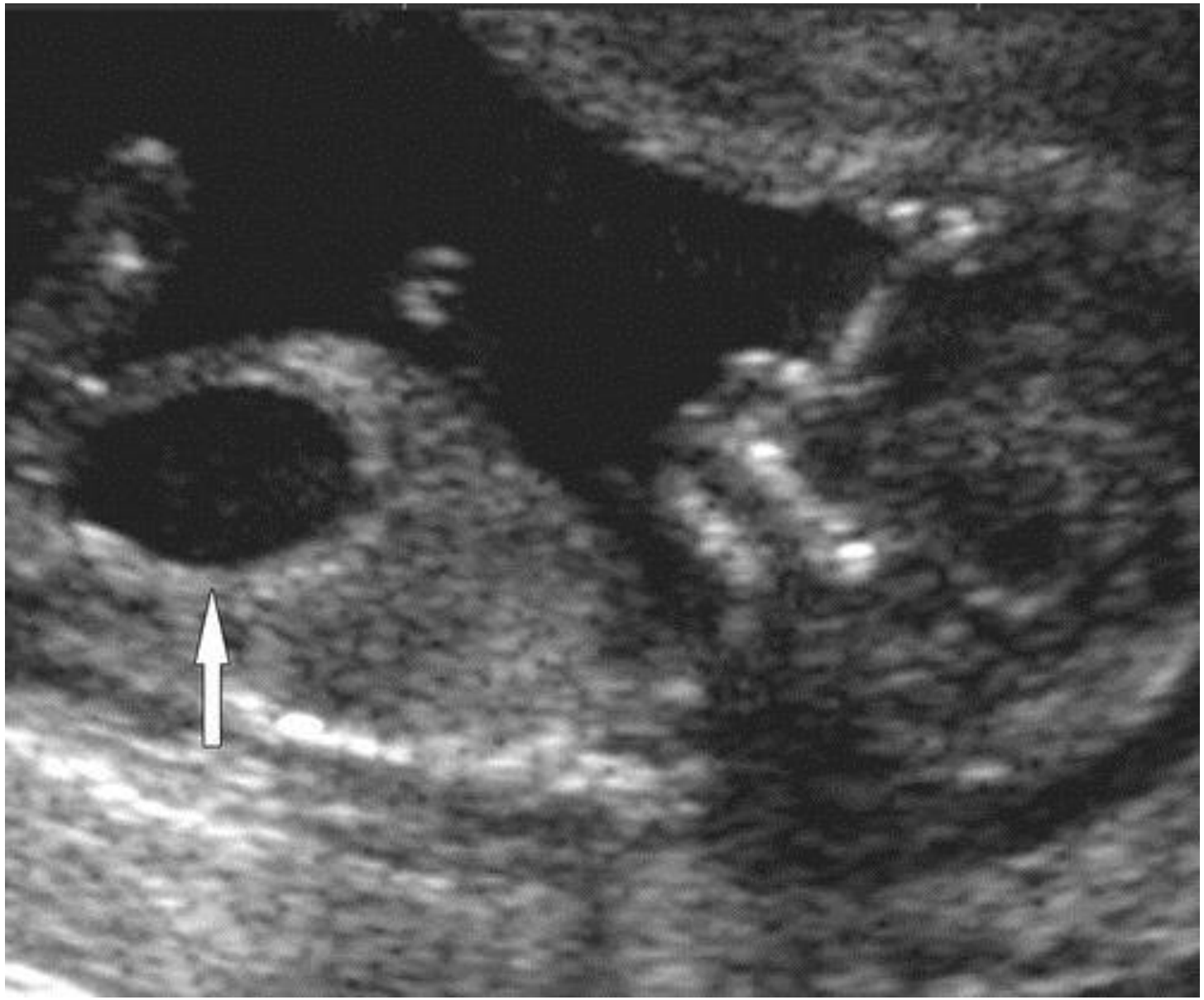
Anencephaly

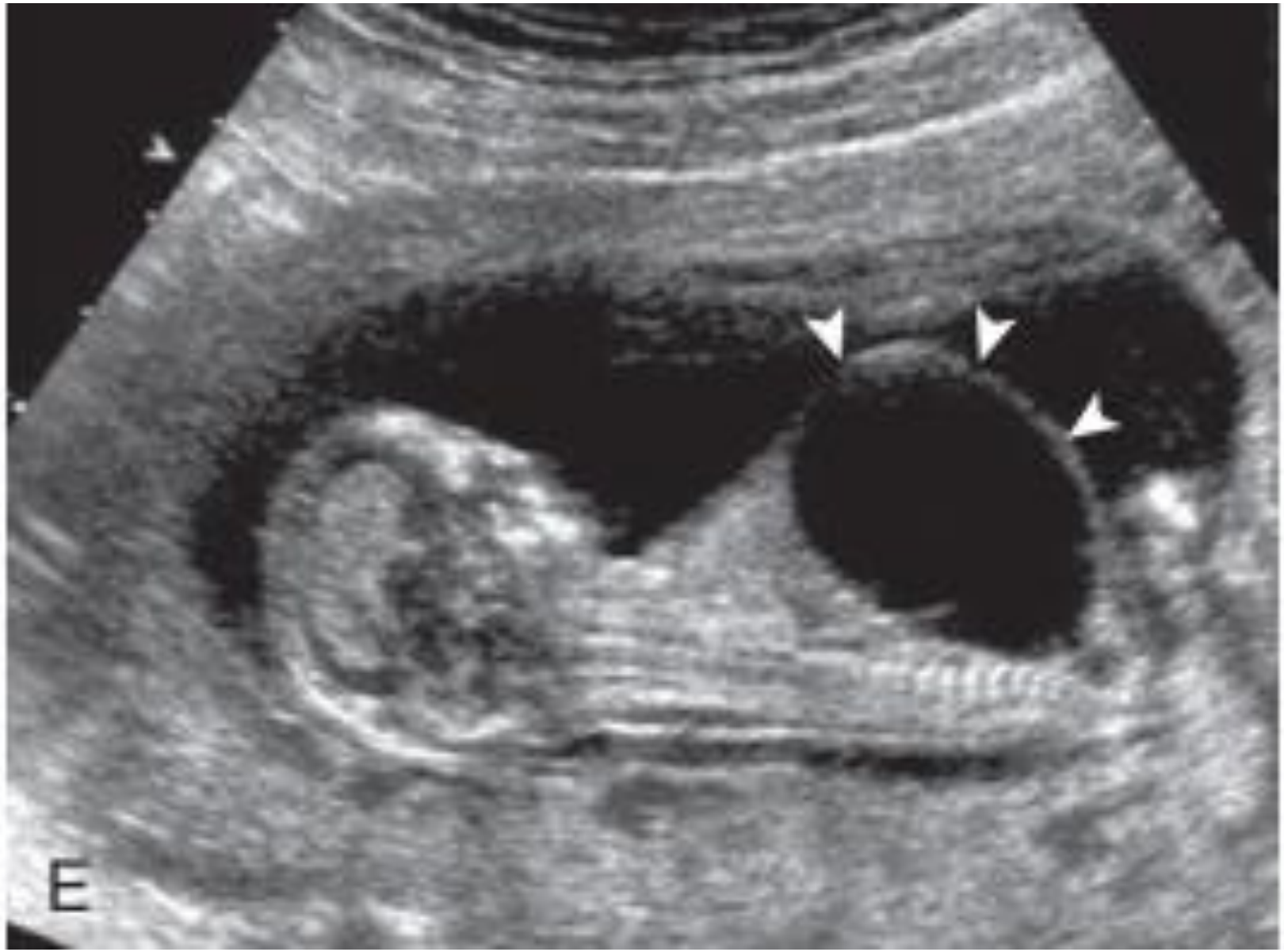
- **Anencephaly** follows failure of closure of the rostral neuropore leading to failed cranial vault development and unprotected exposed brain tissue **(exencephaly),** the subsequent destruction of which leaves a flattened, amorphous vascular-neural mass (area cerebrovasculosa) characteristic of anencephaly.
- Chromosomal abnormality is seen in 2%, and additional abnormalities are common.
- Detection of anencephaly before 14 weeks can be difficult because a relatively normal-appearing brain structure can be present, but the diagnosis has been suggested as early as 10 12 weeks.The diagnosis can be missed unless the examiner speciically looks for ossified cranial bones. Ultrasonically visible ossification of frontal bones may not be apparent until 10 weeks, and anencephaly should not be diagnosed before this gestational age. Commonly, however, the cerebral mass is deformed, often looking like “Mickey Mouse ears.”
- The outcome of anencephaly is invariably fatal, and pregnancy termination is offered at any gestational age


B

Fetal megacystis

- **Fetal megacystis** refers to the presence of an unusually large urinary bladder in a fetus.
- bladder diameter >7 mm in the first trimester.
- on a first trimester scan (10-14 weeks) if the longitudinal bladder diameter is 7-15 mm, the risk of chromosomal defects is estimated at ~25% ·if the bladder diameter is >15 mm, the risk of chromosomal defects is estimated at ~10%.
- oligohydramnios
- megacystis microcolon intestinal hypoperistalsis (MMIH) syndrome (Berdon syndrome)
- megacystis megaureter syndrome,prune belly syndrome
- If the fetus is chromosomally normal and there is megacystis on the 1st trimester scan:there is a spontaneous resolution of the megacystis in about 90% of cases when the 1st trimester longitudinal bladder diameter is between 7-15 mm

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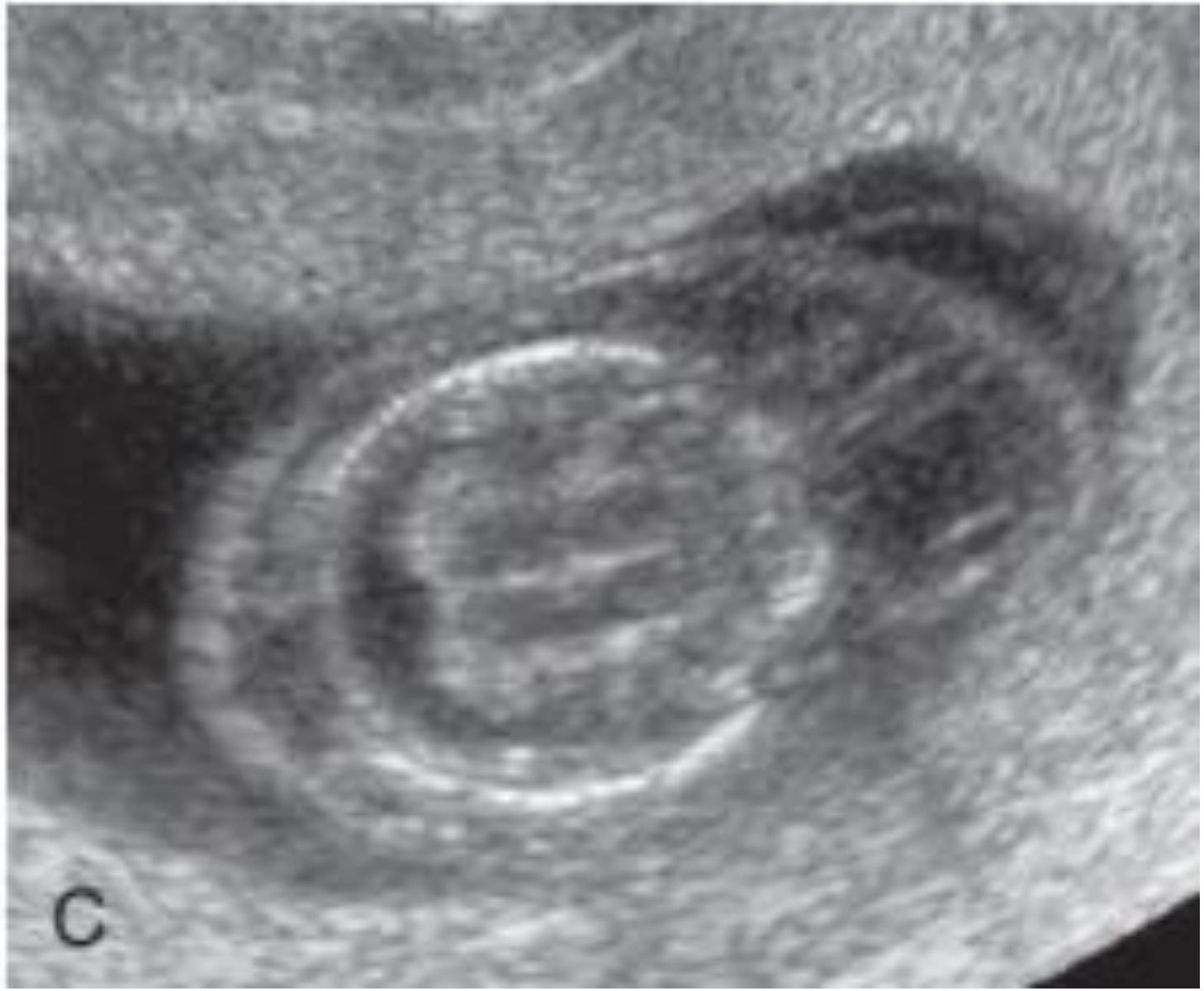
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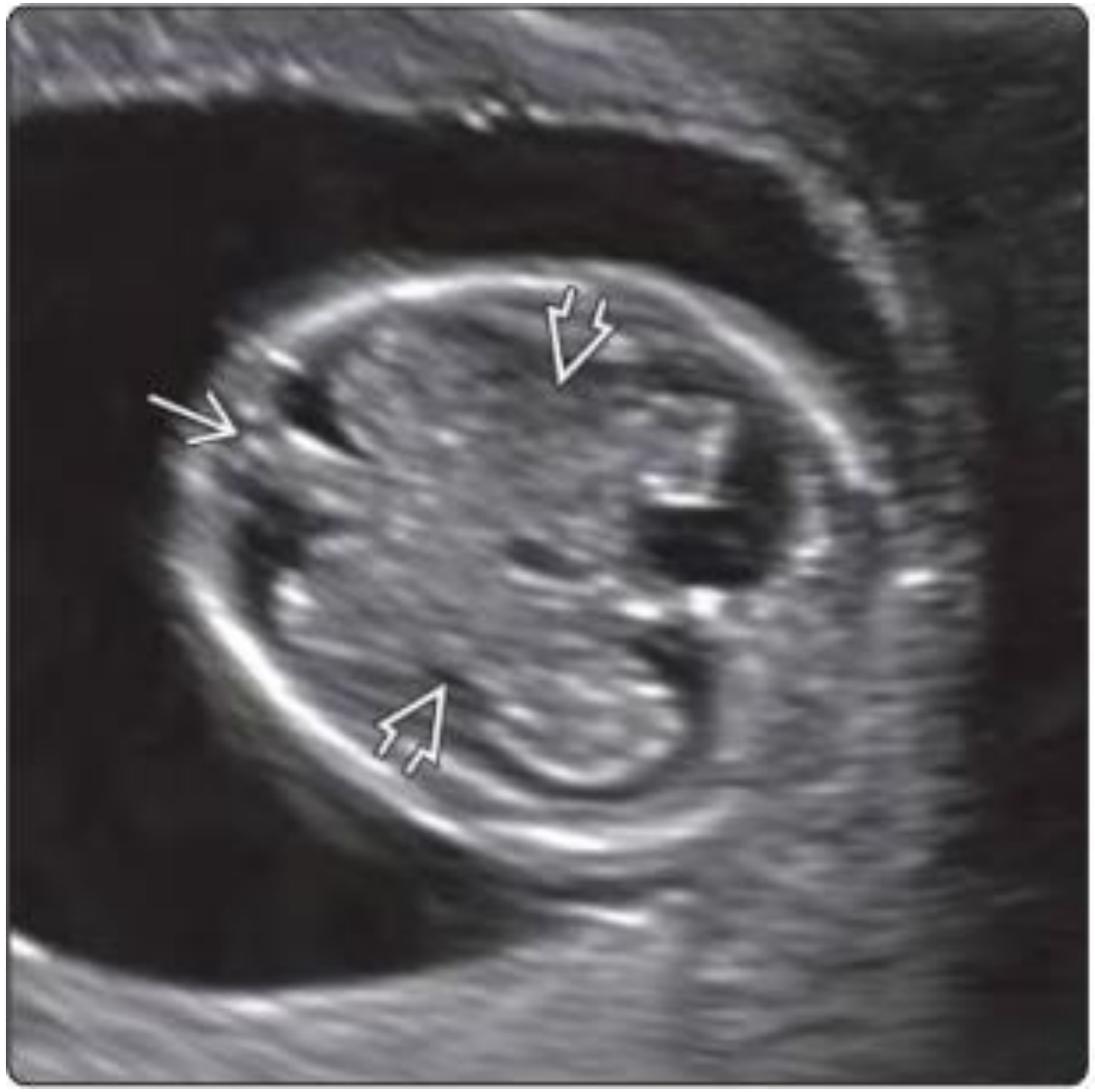

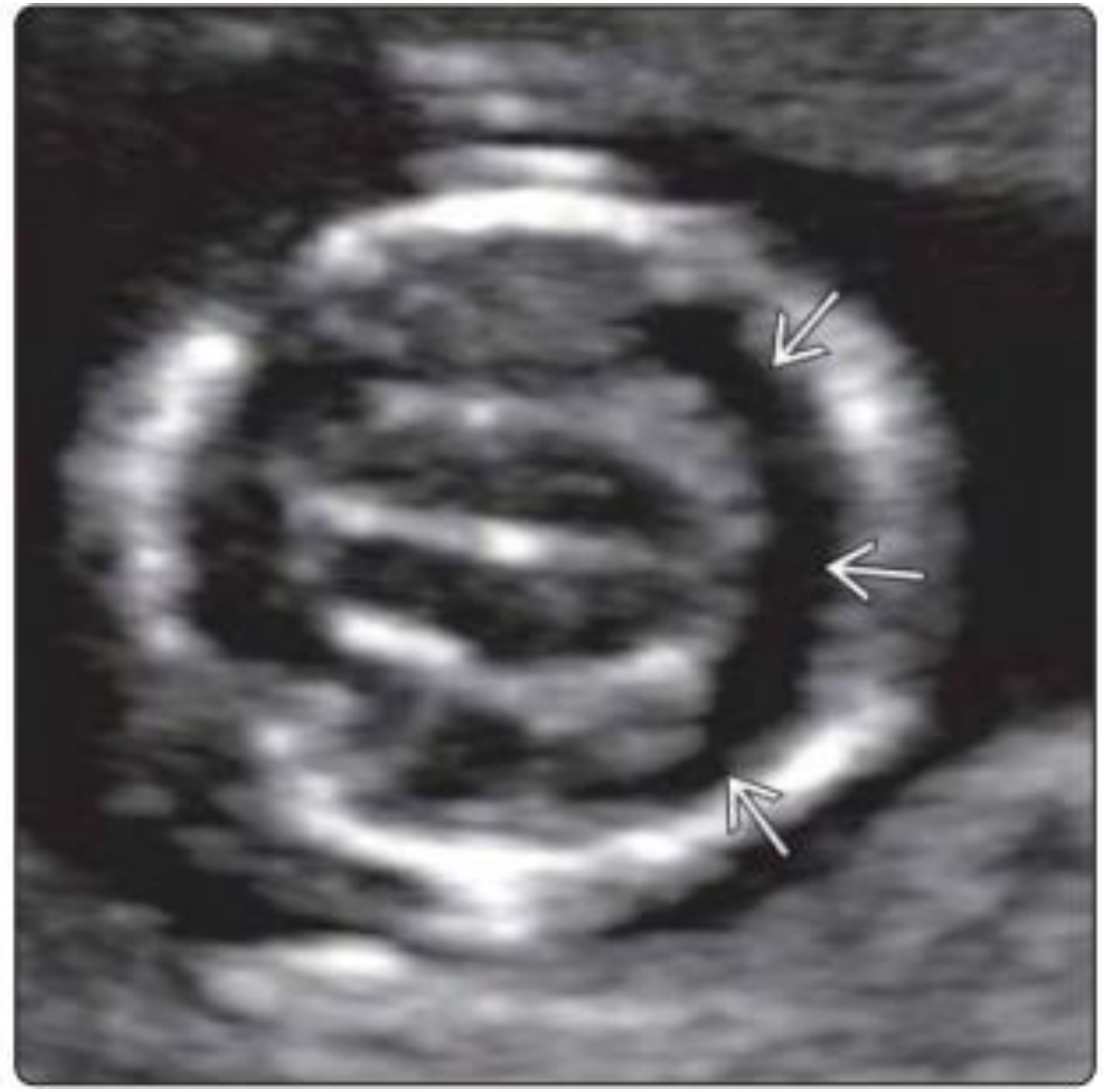

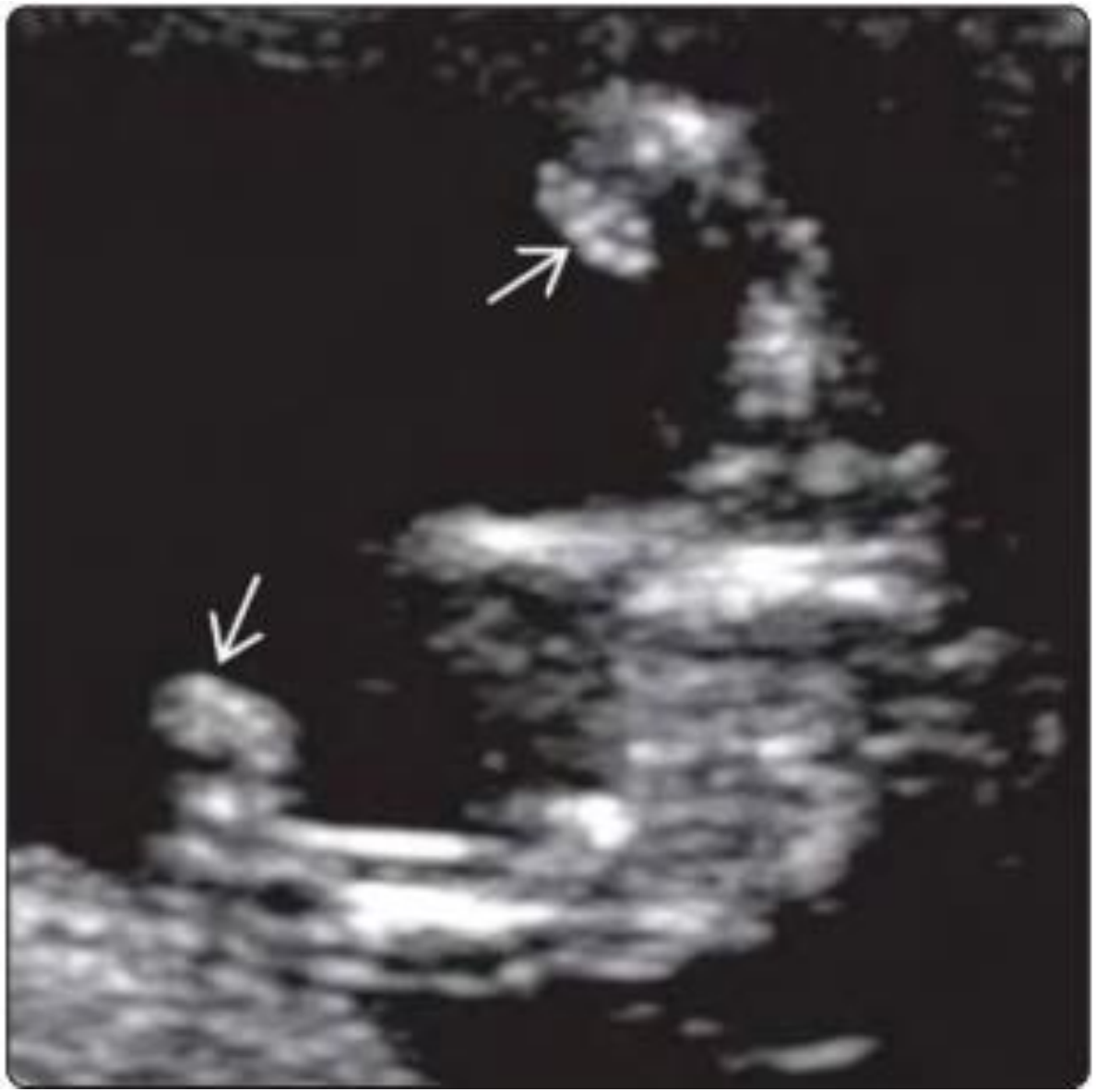

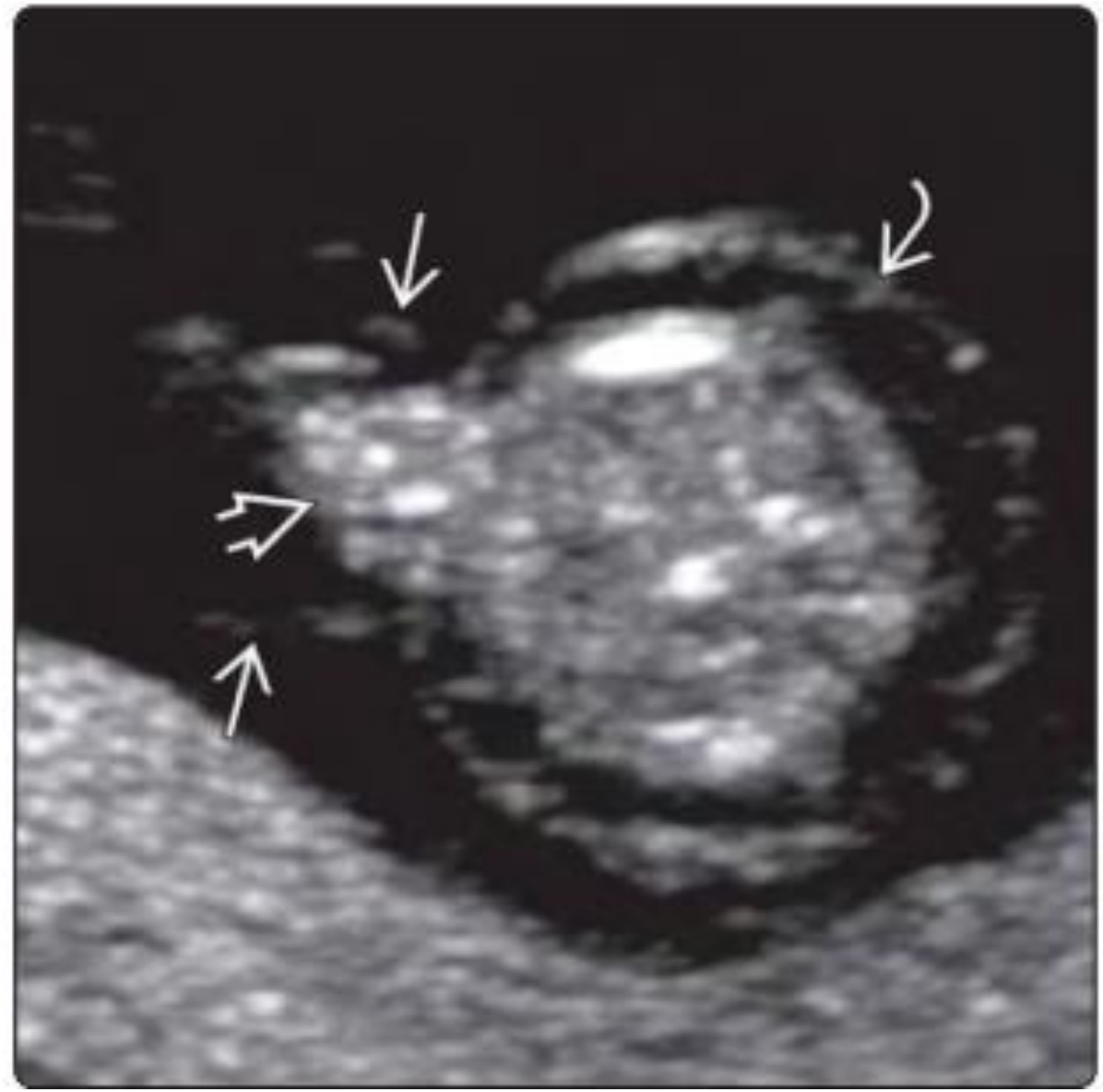

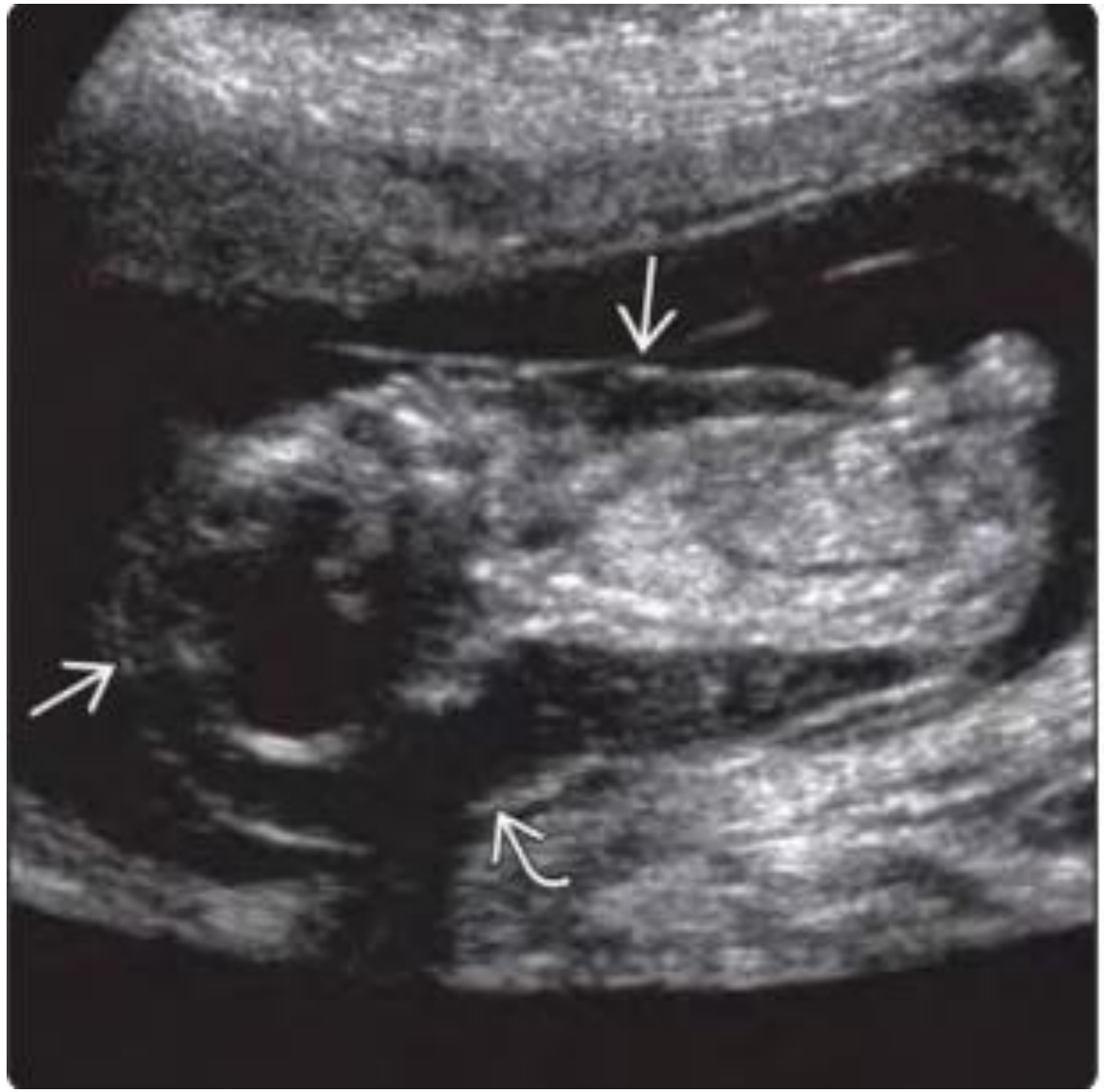

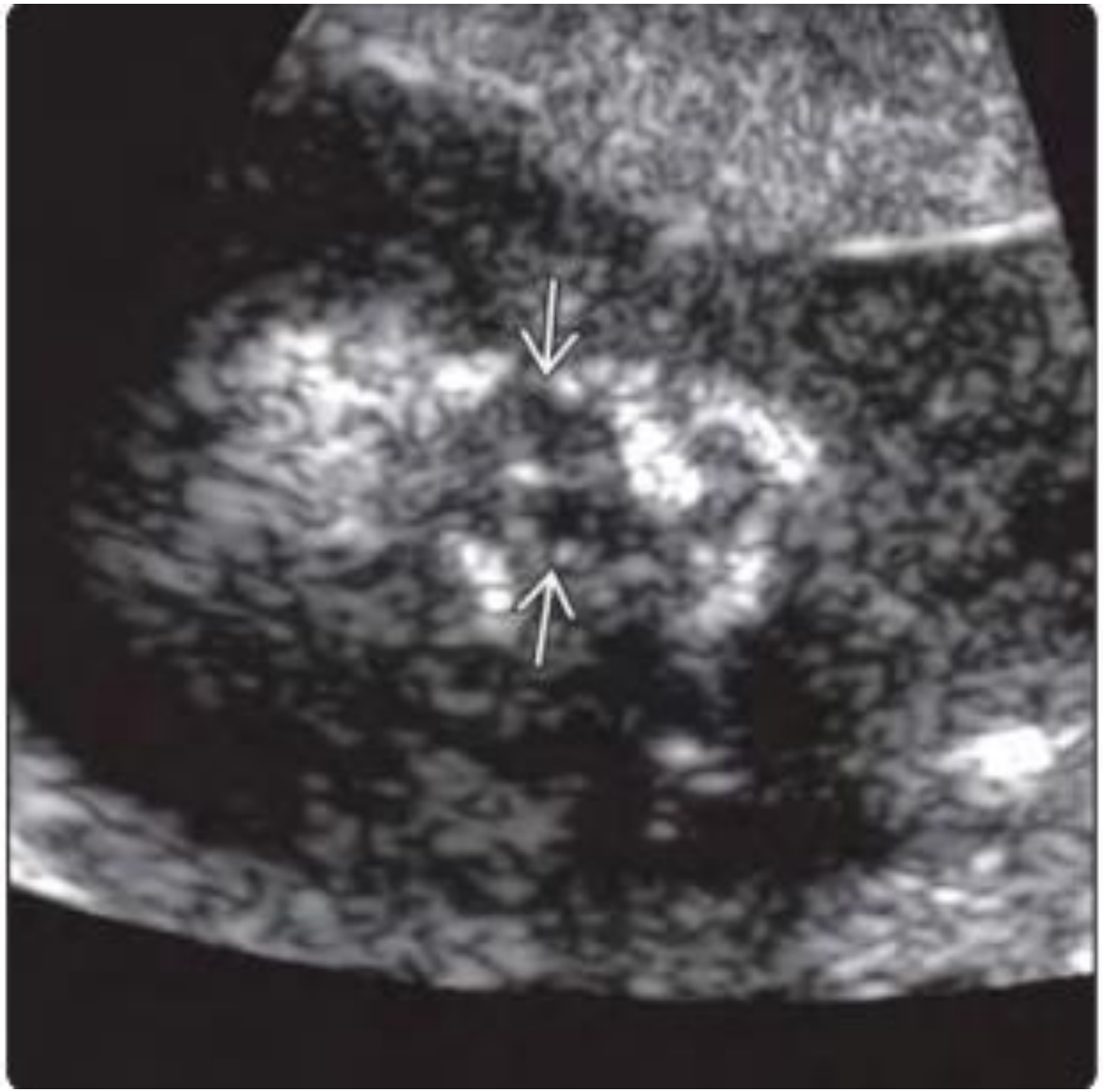

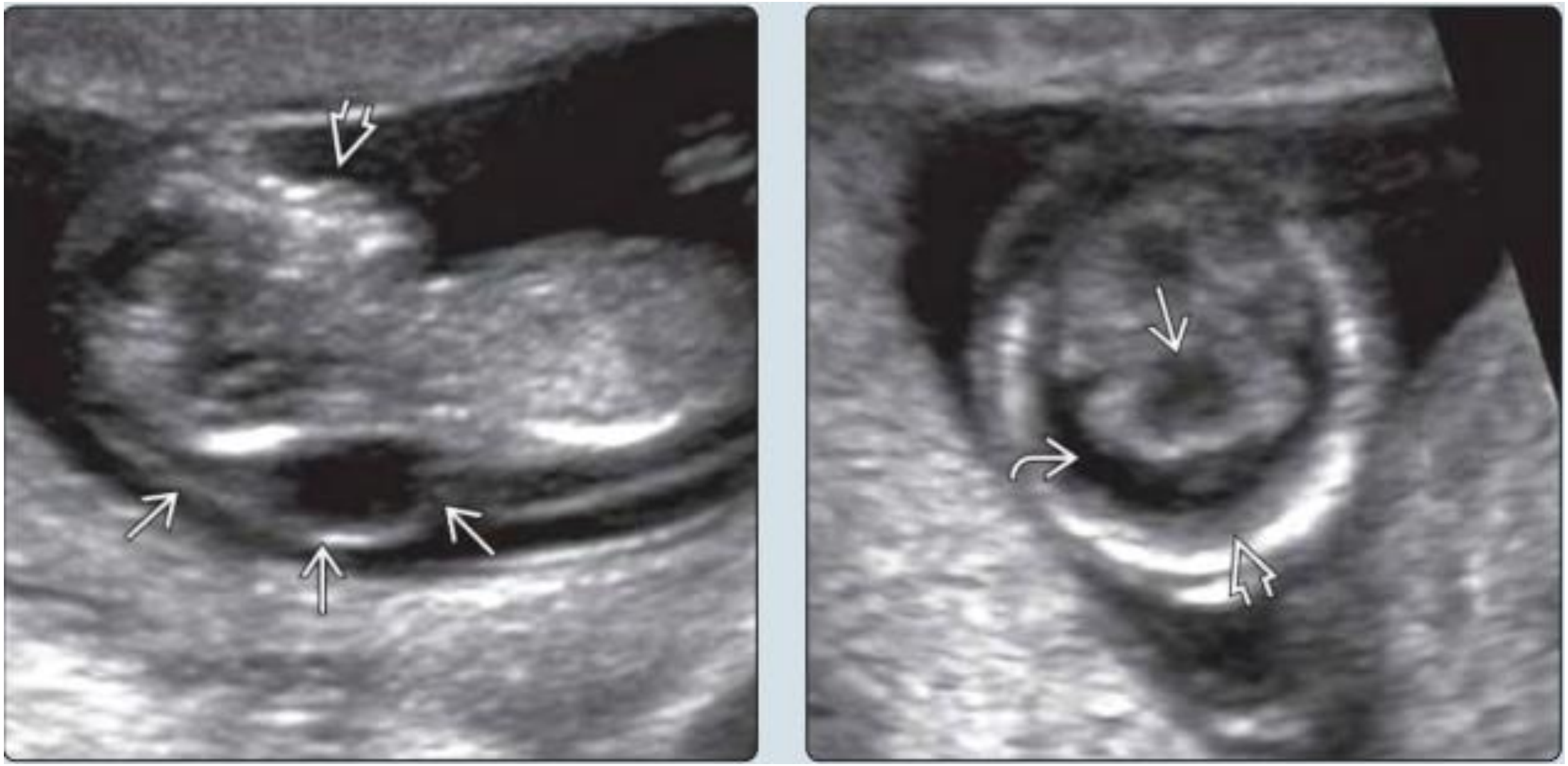

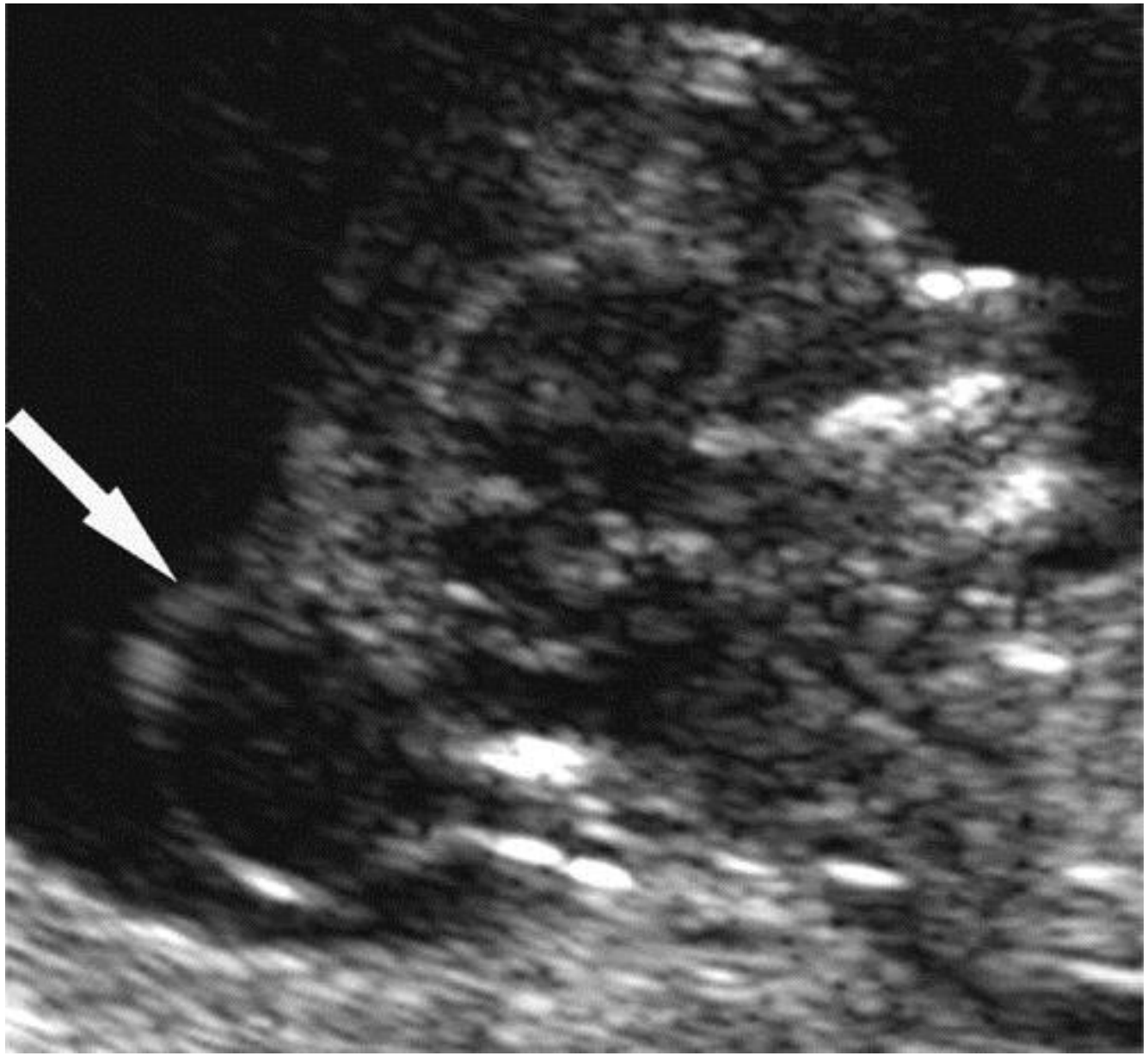

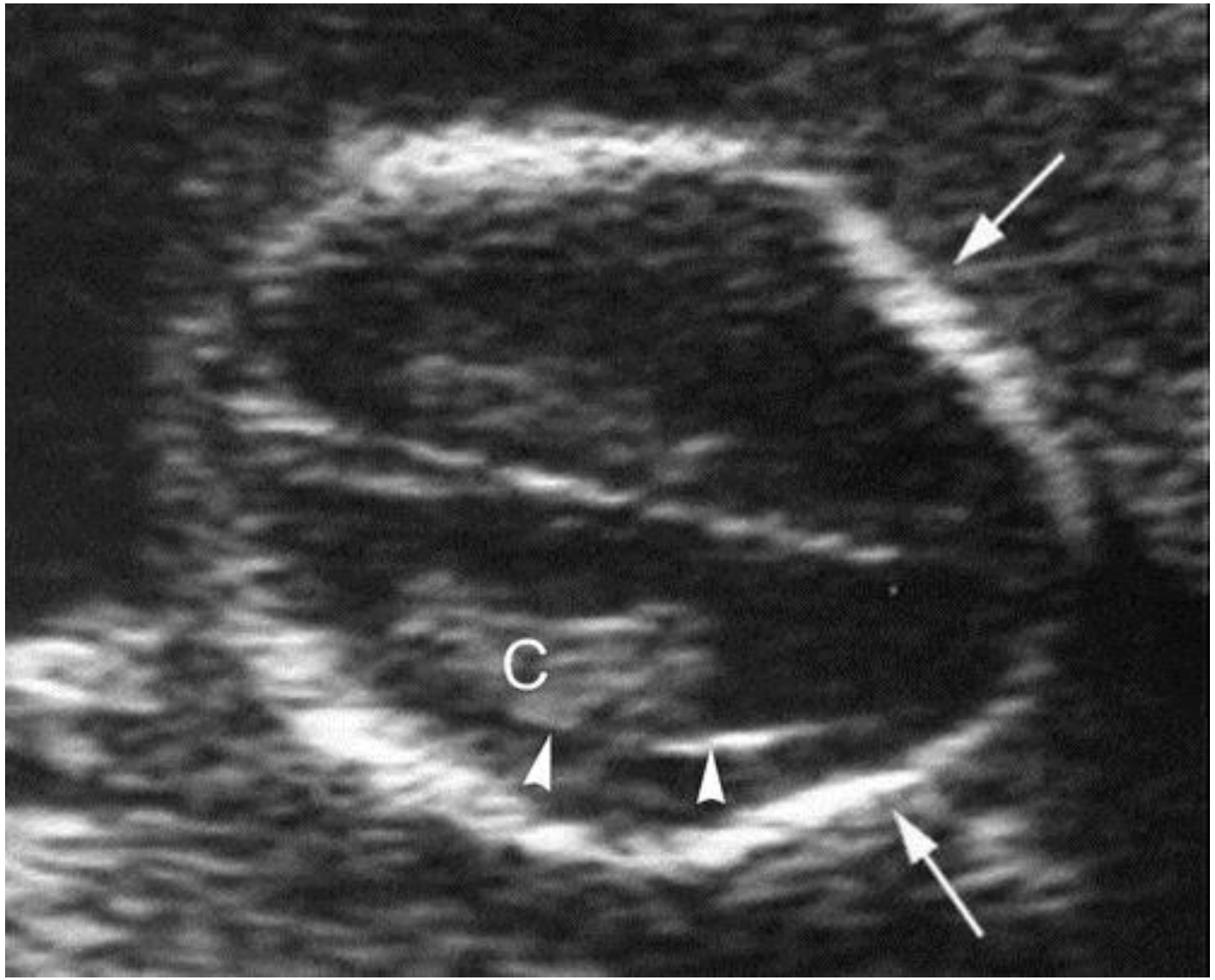
C