Prenatal Screening and Diagnosis: Key Clues to Antepartum Surveillance

IMAGE OF THE MONTH

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CASE PRESENTATION

A 25-year-old, gravida 6, para 1041, white female presented to the clinic for prenatal care. Her period was 2 weeks late and she was experiencing breast tenderness. Her urine pregnancy test was positive. She weighed 200 lbs, her menses was "regular," and she had never used any form of contraception. Past medical history was remarkable for obesity. She denied taking any prescription or over-the-counter medications or supplements. She reported no history of tobacco, alcohol, and/or illicit drug use. She and her boyfriend had been "together" for 1 year. Both family histories were unremarkable and negative for birth defects, mental disability, or chromosomal abnormalities. Her obstetric history was remarkable for a spontaneous abortion at 8 weeks gestation, 3 elective abortions, and a healthy infant (7 lbs, 4 oz) delivered post-dates (42 weeks) via primary, low-transverse cesarean section. The patient verbalized excitement about her pregnancy.

PRENATAL SCREENING

Antepartum surveillance is a process of ongoing risk identification and assessment critical to the well-being of the mother and fetus.

First Trimester: Laboratory Tests

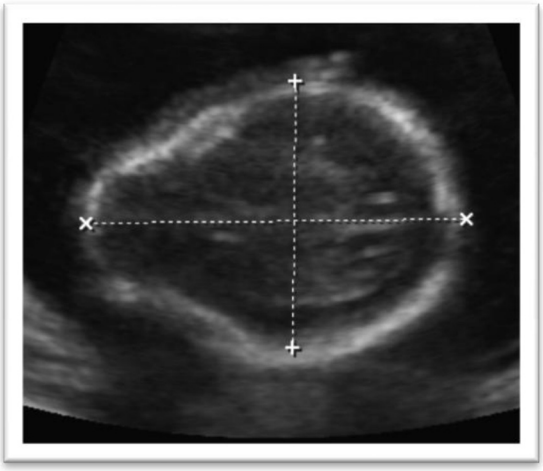
The patient presented for her prenatal visit at 8 weeks gestation (estimated due date [EDD] was based on her reported last menstrual period). Vital signs were normal and fetal heart rate was 150 beats/min. As expected, there was no fetal movement. Routine prenatal laboratory tests[1] were drawn and remarkable for A-negative blood type.

Second Trimester: Imaging Studies and Genetic Screening

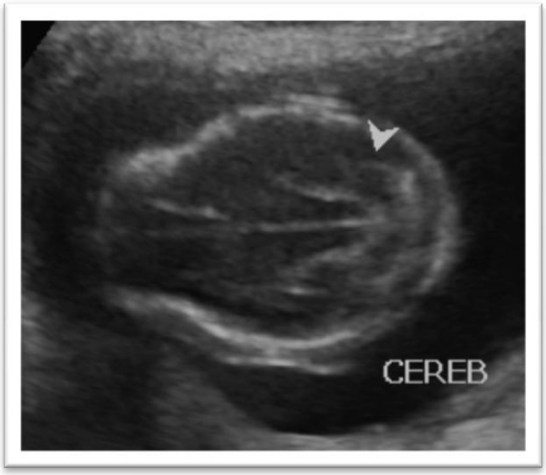
During the 12 6/7-week visit, the patient's vital signs were normal (fetal heart rate 150 beats/min), and she reported quickening. An ultrasound examination was performed to confirm EDD and to obtain fetal nuchal translucency (1.9 mm/normal).[2] EDD was confirmed by fetal measurements and last menstrual period. She returned 2 days later for a Sequential Integrated Screen, Part 1 blood test. Laboratory results stated "Increased risk for Down syndrome." This 2-part blood test screens for trisomy 18, 21, and open neural tube defects (ONTDs). Part 1 is performed during the first trimester between 10 and 13 weeks and includes a nuchal translucency measurement. Part 2 is performed during the second trimester between 14 and 21 weeks. The results from parts 1 and 2 are combined into a single screening test result.[3]

The patient returned next at 17 1/7 weeks. Vital signs were normal, fundal height 17 cm, fetal heart rate 140 beats/min, and active fetal movement. Her Sequential Integrated Screen, Part 2 was drawn 2 days earlier. The results, indicating "Screen positive for Down syndrome," were discussed. She agreed with the recommendation to see the maternal fetal medicine (MFM) specialist. Her appointment was scheduled for 2 weeks. In the interim, the MFM specialist requested screening with a noninvasive prenatal test (NIPT). NIPTs

Level II Ultrasound. US view of fetal biparietal diameter (BPD); Lemon Sign.



Level II Ultrasound. US view of fetal head circumference; posterior fossa; Banana Sign.



involve blood screening that analyzes cell-free DNA for risk of aneuploidy (eg, trisomy 18 and 21). NIPTs do not provide information about risk for anomalies such as ONTDs or ventral wall defects. Although not diagnostic, NIPTs are highly accurate and remain the recommended, first-line genetic screening method in pregnant women.[4] The patient returned for her Harmony Prenatal Test™ (NIPT).

PRENATAL DIAGNOSIS OF GENETIC DISORDERS

Invasive Genetic Testing and Imaging Studies

At 19 1/7 weeks, the patient presented for her initial MFM appointment. Her NIPT was pending; average time from blood draw to results is 10 days. The MFM specialist conducted a level II fetal anatomy ultrasound. Findings included:

> "…single living intrauterine fetus estimated fetal weight 165 grams (25th percentile) corresponding to very small for gestational age with lagging clinical dates by 3 weeks. Fetal anatomy limited by small fetal size; following anatomical abnormalities: scalloping of frontal cranial bones; lower back there appears to be cystic structure—concerning for neural tube defect; fetal hands appear clenched throughout study; views of heart very limited but on real time imaging seems that only left ventricle functioning (noted by color Doppler)—high suspicion congenital heart defect; …findings make likelihood for genetic syndrome—vs. aneuploidy quite high…"

A Lemon Sign was noted when measuring biparietal diameter and a Banana Sign when

Level II Ultrasound. Side-by-side US views of the fetal lower sping; abnormal cystic structure.