Prenatal Screening and Diagnosis for Chromosomal Abnormalities and Neural Tube Defects
(Based on ACOG Screening Recommendations, 2016; ACOG Committee Opinion Recommendations, 2015)

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Screening Recommendations
- All women, regardless of age, should have the option of invasive testing
- Maternal age of 35 years alone should not be used as a cutoff to determine who is offered screening versus who is offered invasive testing; however, maternal age does play a role in determining a priori risk for certain fetal abnormalities
- This algorithm provides a guideline; women may choose screening options alternate to what is recommended by their risk category after appropriate pretest counseling

Pretest counseling/assess patient risk for fetal aneuploidy

LOW RISK
- Patient <35 years old at delivery
- Normal fetal ultrasound or no ultrasound
- No previous fetus/baby with aneuploidy
- Neither parent is a known carrier of a chromosome rearrangement (translocation or inversion)

FIRST TRIMESTER:
Integrated Screen, Specimen #1
Sequential Screen, Specimen #1

SECOND TRIMESTER:
Integrated Screen, Specimen #2
Sequential Screen, Specimen #2
Quad Screen

High risk
Low risk

If nuchal translucency >3.5mm and aneuploidy screens are negative, offer patient genetic counseling with cfDNA or amniocentesis/CVS; targeted US or fetal echo, or both

High risk
Low risk

If cfDNA is positive, offer genetic counseling and amniocentesis/CVS to confirm
If cfDNA is negative in the presence of US anomalies, offer genetic counseling and amniocentesis/CVS for microarray +/- chromosomes (or just microarray)
If cfDNA is negative in the absence of US anomalies, offer 2nd trimester AFP (only) screen for ONTD

If EDD changes by ≥10 days based on US (Quad and Serum Integrated only) – recalculate

If EDD is correct within 10 days
Level II US to confirm dating and presence of twins and/or fetal/placental abnormalities

If BNP is positive, offer genetic counseling and tandem mass spectrometry

Genetic counseling with cfDNA or amniocentesis
EDD is correct within 10 days

Level II US to confirm dating and presence of twins and/or fetal/placental abnormalities

Low risk
High risk
Low risk

Low risk for T18 or T21
Low risk for T18, T21, and ONTD; no further testing recommended

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