Prenatal Screening and Diagnosis
(Based on ACOG screening recommendations, 2007; ACOG Committee Opinions Recommendations, 2012)

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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

LOW RISK
- Pt <35 years old at delivery
- Normal fetal ultrasound or no ultrasound
- No previous fetus/baby with aneuploidy
- Neither pt nor partner are known carriers of a chromosome rearrangement (translocation or inversion)

HIGHER RISK
- At least one of the following:
  - Pt ≥35 years old at delivery (advanced maternal age)
  - An increased risk for trisomy 13, 18 or 21 by traditional maternal serum aneuploidy screen
  - An abnormal fetal ultrasound with findings consistent with trisomy 13, 18, 21 or Turner syndrome
  - Previous fetus/baby with chromosome aneuploidy

OTHER HIGH RISK
- Either Pt or partner are known carriers of a chromosome rearrangement (translocation or inversion)
- Abnormal ultrasound not consistent with trisomy 13, 18, 21 or Turner syndrome

Pretest counseling/assess patient risk for fetal aneuploidy

Serum marker screening

NIPT

amniocentesis or CVS

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

Patient presents in 1st trimester

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

Integrated Screen, Specimen #1

Sequential Screen, Specimen #1

Low risk pending 2nd specimen

First Trimester Screen Note: Regardless of screen results, 2nd trimester AFP (Only) should be offered

low risk

high risk

Low risk for DS or T18

Genetic counseling with NIPT or amniocentesis

SECOND TRIMESTER:

Integrated Screen, Specimen #2

Sequential Screen, Specimen #2

Quad Screen

low risk

high risk

low risk

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

Genetic counseling with NIPT or amniocentesis

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

EDD is correct within 10 days

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


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