Prenatal Screening and Diagnosis

(Based on ACOG screening recommendations, 2007; ACOG Committee Opinions Recommendations, 2012)

Pretest counseling/assess patient risk for fetal aneuploidy

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)

Serum marker screening

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 microarray +/- chromosomes (or just microarray)

OTHER HIGH RISK (NIPT may not be appropriate)
- 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

Amniocentesis or CVS

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

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

Low risk pending 2nd specimen

Genetic counseling with NIPT or amniocentesis/CVS

Low risk for DS or T18

SECOND TRIMESTER:

Integrated Screen, Specimen #2

Sequential Screen, Specimen #2

Quad Screen

High risk

Low risk

EDD is correct within 10 days

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

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

Genetic counseling with NIPT or amniocentesis

High risk

Low risk

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

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