Prenatal Screening and Diagnosis for Chromosomal Abnormalities and Neural Tube Defects

ARUP Consult

(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)

HIGH RISK
At least one of the following:
- Patient ≥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 (cfDNA may not be appropriate)
- Either parent is a known carrier of a chromosome rearrangement (translocation or inversion)
- Abnormal ultrasound not consistent with trisomy 13, 18, 21, or Turner syndrome

Serum marker screening

cfDNA

Amniocentesis or CVS

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

Patient presents in 1st trimester

Patient presents in 2nd trimester

FIRST TRIMESTER:

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

Integrated Screen, Specimen #1

Sequential Screen, Specimen #1

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

High risk

Low risk

Low risk pending 2nd specimen

Genetic counseling with cfDNA or amniocentesis/CVS

Low risk for T18 or T21

SECOND TRIMESTER:

Integrated Screen, Specimen #2

Sequential Screen, Specimen #2

Quad Screen

High risk

Low risk

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

EDD is correct within 10 days

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

Genetic counseling with cfDNA or amniocentesis

High risk

Low risk

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

AFP – alpha fetoprotein, CRL – crown rump length, CVS – chorionic villus sampling, DIA – dimeric inhibin A, DR – detection rate, EDD – estimated delivery date, hCG – human chorionic gonadotropin, cfDNA – cell-free DNA, NT – nuchal translucency, ONTD – open neural tube defect, PAPP-A – pregnancy-associated placental protein A, SPR – screen positive rate, T18 – trisomy 18, T21 – trisomy 21 (Down syndrome), uE3 – unconjugated estriol