ARUP Consult

Prenatal Screening and Diagnosis

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

Click here for topics associated with this algorithm

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

- 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)

HIGH 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 (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

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 microarray +/- chromosomes (or just microarray)

If NIPT is negative in the absence of US anomalies, offer 2nd trimester AFP (only) screen for NTD

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 NIPT 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 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

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 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

AFP – Alpha Fetoprotein, CRL – Crown Rump Length, CVS – Chorionic Villus Sampling, DIA – Dimeric Inhibin A, DR – Detection Rate, DS – Down Syndrome, hCG – Human Chorionic Gonadotropin, NIPT – Non-Invasive Prenatal Testing, NT – Nuchal Translucency, ONTD – Open Neural Tube Defect, PAPP-A – Pregnancy-Associated Placental Protein A, Pt – Patient, SPR – Screen Positive Rate, T18 – Trisomy 18, uE3 – Unconjugated Estriol

Last updated: May 2013