ARUP Consult

Prenatal Screening and Diagnosis

Click here for topics associated with this algorithm

PRENATAL SCREENING
LOW-RISK PREGNANCY

- No prior pregnancy with a chromosome disorder, ie, Down syndrome (DS) or trisomy 18 (T18)
- No family history of open neural tube defect (ONTD)
- Maternal age <35 at estimated date of delivery (EDD)

PRENATAL DIAGNOSIS
HIGH-RISK PREGNANCY

- Previous pregnancy with chromosome disorder
- Family history of ONTD
- Maternal age ≥35 at EDD
- Patient taking valproic acid or carbamazepine
- Patient is an insulin-dependent diabetic

If patient presents during second trimester or declined screening during first trimester

FIRST TRIMESTER:

Choice of testing depends on patient preference and availability of follow-up testing

First Trimester Screen

- Nuchal translucency* (NT) and blood draw between 11w0d and 13w6d (CRL of 42-79mm)
- Maternal Serum Screen, First Trimester test

Note: Regardless of outcome, perform Maternal Serum Screen, Alpha Fetoprotein (Only) in the 2nd trimester

Sequential Screen 1st Draw

- NT and blood draw between 11w0d and 13w6d (CRL of 42-79mm)
- Maternal Screening, Sequential, Specimen #1 (PAPP-A and hCG) test

Integrated Screen 1st Draw

- NT* (optional but recommended) and blood draw between 10w3d and 13w6d (CRL of 36-79mm)
- Maternal Serum Screening, Integrated, Specimen #1 (PAPP-A) test

normal → Low risk for DS or T18

abnormal → Increased risk for DS or T18:
- Offer genetic counseling and diagnostic testing (CVS or amniocentesis)

ORDER
- Maternal Serum Screen, Alpha Fetoprotein (Only) in the 2nd trimester

risk <1 in 25 for DS or T18; second sample required for interpretation

*NOTE: If nuchal translucency >3.5mm and aneuploidy screens are negative, offer patient targeted US or fetal echo or both

SECOND TRIMESTER:

Quad Screen

ORDER
Maternal Serum Screen, Alpha Fetoprotein, hCG, Estriol, & Inhibin A (blood draw between 14w0d and 24w6d)

Sequential Screen 2nd Draw

ORDER
Maternal Screening, Sequential, Specimen #2 (AFP, hCG, Estriol, Inhibin A with previous PAPP-A)
(blood draw between 15w0d and 22w6d)

Integrated Screen 2nd Draw

ORDER
Maternal Serum Screening, Integrated, Specimen #2 (AFP, hCG, Estriol, Inhibin A with previous PAPP-A)
(blood draw between 15w0d and 22w6d)

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

high AFP / abnormal → Level II US to check dating and look for twins and/or fetal/ placental abnormalites

If EDD changes by more than 10 days based on US, recalculate
- normal → Low risk for DS, T18, and ONTD; no further testing recommended
- abnormal → Offer genetic counseling and diagnostic testing (amniocentesis)

EDD is correct within 10 days → Offer genetic counseling and diagnostic testing (amniocentesis)

© 2006-2010 ARUP Laboratories. All Rights Reserved. Revised 11/24/2008