

PRENATAL SCREENING LOW-RISK PREGNANCY

- No prior pregnancy with a chromosome disorder, i.e., Down syndrome (DS) or trisomy 18 (T18)
- No family history of open neural tube defect (ONTD)
- Maternal age <35 at 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 0081150

Note: Regardless of outcome, perform Maternal Serum Screen, Alpha Fetoprotein (Only) 0080434 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 0081293 (PAPP-A and hCG)

Integrated Screen 1st Draw

- NT (optional but recommended) and blood draw between 10w3d and 13w6d (CRL of 36-79mm)
- Maternal Screening, Integrated, Specimen #1 0081062 (PAPP-A)

normal

Low risk for DS or T18

abnormal

Increased risk for DS or T18:

- Offer genetic counseling and diagnostic testing (CVS or amniocentesis)
- Maternal Serum Screen, Alpha Fetoprotein (Only) 0080434 in the 2nd trimester

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

SECOND TRIMESTER:

Quad Screen

Maternal Serum Screen—Alpha Fetoprotein, hCG, Estriol & Inhibin A 0080269 (blood draw between 14w0d and 24w6d)

Sequential Screen 2nd Draw

Maternal Screening, Sequential, Specimen #2 0081294 (AFP, hCG, Estriol, Inhibin A with previous PAPP-A) (blood draw between 15w0d and 22w6d)

Integrated Screen 2nd Draw

Maternal Serum Screen, Integrated, Specimen #2 0081064 (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

normal

high AFP

Level II US to check dating and look for twins and/or fetal/placental abnormalities

If EDD changes by more than 10 days based on US, recalculate

abnormal

EDD is correct within 10 days

Offer genetic counseling and diagnostic testing (amniocentesis)