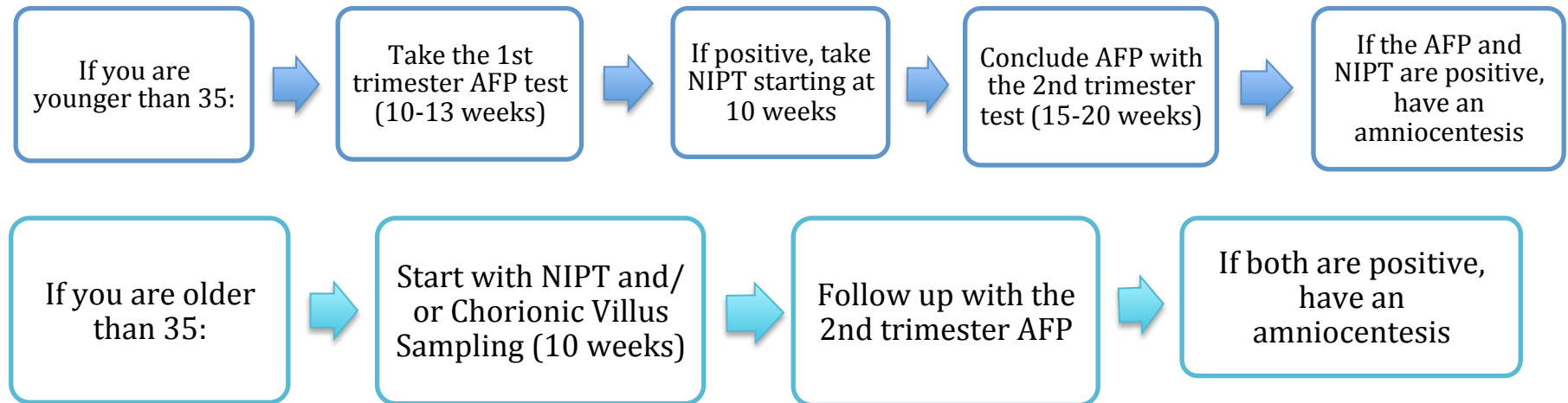


Maternal serum AFP testing and Non-invasive Prenatal Test



Maternal Serum AFP test:

Three types of screening tests identify pregnant women who are at increased risk for carrying a fetus with a specific birth defect.

1. Quad Marker Screening (2nd trimester test): One blood specimen drawn at 15-20 weeks of pregnancy (second trimester test).
2. Serum Integrated Screening: Combines a first trimester blood test screening result (10 weeks-13 weeks, 6 days) with a second trimester blood test screening result (15-20 weeks).
3. Sequential Integrated Screening: Combines first and second blood test results with Nuchal Translucency (NT) ultrasound results, done by clinicians with special training. It measures the back of the fetus' neck to screen for Down syndrome (trisomy 21). (Note: the Screening Program does not pay for NT ultrasounds).

The Prenatal Screening Program provides pregnant women with a risk assessment for open neural tube defects (NTD), Down syndrome (trisomy 21), trisomy 18 and SLOS (Smith-Lemli-Opitz Syndrome) through one or two blood tests. **The screening test indicates risk, but does not diagnose fetal birth defects.**

Non-invasive pregnancy testing (NIPT):

NIPT looks for Trisomy 13, 18, 21 in your baby's chromosomes found in your blood. It can also determine sex chromosomes. Some companies offer additional testing for other chromosomal abnormalities including micro-deletion syndromes like cri-du-Chat, Angelman, Prader-Willi, and DiGeorge syndromes. Micro-deletion tests have not been validated in clinical studies and accuracy has not been determined; therefore ACOG does not recommend testing for micro-deletions via NIPT at this time.

NIPT testing can be done any time after 9-10 weeks of pregnancy. However, it is more accurate the further past 10 weeks you are but false positives remain a slight possibility ($\leq 1\%$). No pregnancy should be terminated based on the NIPT results—an amniocentesis is required to confirm results.

How accurate are these tests?

Maternal serum AFP testing has a high false positive rate, meaning you can get an abnormal test result but your baby is perfectly healthy. It can also miss a baby this is affected. This is frustrating to patients and is a common reason given for not doing the test to avoid anxiety and extra testing unnecessarily. NIPT is 99% accurate, but no pregnancies should be terminated based on the results of an NIPT: an amniocentesis is required to confirm results.

Our recommendations depend on your risks:

Consult our flow chart regarding which path of testing you should take, which depends on your age and risk factor. If the AFP test comes back abnormal, we suggest that you be referred to a perinatologist (high risk pregnancy specialist). Genetic counseling will take place, and an NIPT testing and a detailed ultrasound will likely be ordered. If the ultrasound and NIPT are normal then you can assume the maternal serum AFP test was a false positive and feel confident that your baby does not have any of the above disorders.

You are high risk if you are older than 35 years of age; have had a previous pregnancy with a trisomy that can be screened (13, 18, or 21); or an ultrasound showed increased risk for abnormal chromosomes; a parent carried a balanced translocation with an increased risk of trisomy 13 or 21. If you are high risk and over 35, we recommend starting with the NIPT for the most accurate risk assessment for your pregnancy.

Why do testing?

For some, this may determine whether they continue or elect to terminate a pregnancy at risk for the above disorders. For others, it may be a way to prepare for a special needs baby and the special considerations at the time of delivery. For most it will be a reassurance that their baby is healthy.

How long does it take to get results back?

We are called with the results of an abnormal test a week after the maternal serum AFP test is administered. Normal results can take up to 3-4 weeks to get back. The NIPT testing takes 1-2 weeks to get the results back.

What is the cost?

The maternal serum AFP blood test in the first and second trimester without an ultrasound is \$207. This may be covered by insurance, but if not any further testing will be covered through the California Prenatal Screening Program. NIPT testing is typically \$1200 to \$5000 depending on the test. Some patients may end up only paying a small portion of this because it may be covered by insurance. Others will have to pay the entire cost, but it may apply towards a deductible. Most NIPT companies strive to keep the cost reasonable for you by offering special programs or payment plans.