

FIRST TRIMESTER SCREENING FOR CHROMOSOME ABNORMALITIES

Just like triple and quad screens done in the second trimester, first trimester screening is done to help us estimate the chance that your baby has Down syndrome or another chromosome abnormality, trisomy 18. The measurements of certain proteins from the baby in the mother's blood can be combined with a first trimester ultrasound measurement called a nuchal translucency (NT) to better estimate the risk to your baby. Down syndrome and trisomy 18 are two examples of chromosome problems. The risk of these conditions increases with a woman's age. However, they can occur to anyone and often occur to couples with no obvious risk factors.

The two proteins we measure are PAPP-A and free beta-HCG. Babies with Down syndrome make lower amounts of PAPP-A but higher amounts of HCG than babies without these conditions. Babies with trisomy 18 make decreased amounts of both proteins.

Early ultrasound measurements of your baby's NT (fluid area behind the neck) and nasal bone are potentially some of the most powerful screens to identify a baby who may have certain chromosome problems. Babies with certain chromosome problems have larger nuchal translucencies than usual. Babies with Down syndrome can have a small or absent nasal bone. Early data have suggested that the NT and nasal bone measurements may alert us to most babies with trisomy 21 and 18, but not all. In addition, about 40% of babies with heart defects will have an increased NT measurement.

The measurement of these proteins and the ultrasound measurements will be **combined** with the mother's age-related risk for a chromosome problem and a "new number" on the odds of these two conditions in your baby will be generated. If the new risk number is higher than that faced by a 35-year-old woman, your test will be considered "positive." Women with a positive test are offered diagnostic tests for a yes or no answer about these chromosome problems. Only these diagnostic tests (CVS or amniocentesis) will determine with certainty if your baby has a chromosome problem.

Combined First Trimester Screening

- The serum + NT in the first trimester is called **combined first trimester screening**. (The best time is at 11 weeks 2 days, but it can be done up to 13 weeks 6 days. The serum alone can be done earlier [9 weeks] and later combined with the NT).
- Combined testing will allow us to detect the majority of babies with chromosome problems. (Detection rates for trisomy 21 may be about 90% and be even higher for trisomy 18.) Neither the ultrasound nor the blood test will rule out the risk for all chromosome abnormalities nor comment on the risk for all the age-related trisomies.
- In order to provide a risk statistic for Down syndrome and trisomy 18 based on both the NT and first trimester blood test (combined screening), a center must complete an appropriate accreditation process to take the measurements. The new risk figure provided will be based on age, serum, and NT if measured by a certified sonographer. In addition, our center will obtain outcome data on all pregnancies to ensure accurate delivery of information for our patients.
- It is best to do both the blood screening and the NT measurement together. Providing a risk based on first trimester blood screening alone is not as effective as midtrimester quad

screening, and risk information based on NT alone is not recommended due to the high rate of false positives.

- Unbiased trials comparing first trimester combined screening (serum + NT) with second trimester quad screening suggest they are about the same at detecting Down syndrome.
- Those who have first trimester serum screening will still need a midtrimester (about 15 weeks) alpha fetoprotein (AFP) alone to screen for spina bifida.

If you have a combined first trimester screen, you do not need to do the quad screen. Giving a risk assessment after combined screening in the first trimester and then a separate risk after second trimester screening can be confusing and misleading.

First trimester combined screening is best for those who will have an early prenatal diagnostic test if the result is positive.

Integrated Screening

- In addition, midtrimester quad serum tests may be of additional value to those who have a first trimester combined test if the information is “integrated” to give one risk assessment. This option is called **integrated screening**.
- The integrated screen has the highest detection rate and the lowest false positive rate. It involves combining data from first trimester serum + NT with the second trimester quad and giving a single risk assessment. Risk information, however, is not given until the second trimester.
- Integrated screening is best for those who want the smallest chance of having an invasive test.

If the NT is increased, but combined or integrated screening does not suggest a high risk for Down syndrome, a diagnostic test for chromosome abnormalities will not be recommended. However, a targeted ultrasound in the second trimester will be offered to rule out heart defects or other problems also associated with an increased NT.

Both Combined and Integrated screening are possible on twins (not triplets), but it is less effective than for a singleton.

The goal of screening is to decide who would best benefit from diagnostic testing. If a screen is “positive,” the best offered option is a diagnostic test, not additional screening. Remember: diagnostic testing is an available option if you face an increased risk and want a yes or no answer about these problems, even if you do not seek screening first.

Genetic counseling is the first step of the screening process. You will meet with a genetic counselor before your serum test and/or ultrasound. All options will be discussed, your personal risks will be assessed, and all of your questions addressed.