First trimester screening (FTS) is a test that can give more information on the chances that a pregnancy has Down syndrome (trisomy 21) and trisomy 18 (some labs also offer screening for trisomy 13). FTS is generally performed between 11 and 14 weeks of the pregnancy, and is made up of two parts: a blood draw from mom and an ultrasound. The blood draw measures two proteins that are in mom's blood that are coming from the pregnancy: pregnancy-associated plasma protein-A (PAPP-A), and human chorionic gonadotropin (hCG).

- Pregnancy-associated plasma protein-A (PAPP-A): PAPP-A is produced by the placenta and crosses over into mom's blood stream. Lower levels of PAPP-A increase the chances of Down syndrome or trisomy 18, and can also be associated with poor pregnancy outcomes (miscarriage, low birth weight, and high blood pressure/preeclampsia). However, keep in mind that most pregnancies that have low PAPP-A levels go on to produce healthy babies.
- Human chorionic gonadotropin (hCG): hCG is also produced by the placenta and crosses over into mom's blood stream. hCG tends to be higher in pregnancies with Down syndrome and lower in pregnancies with trisomy 18.

The first trimester ultrasound measures the fetus to verify dating, as well as pocket of fluid behind the baby's neck called the nuchal translucency (NT). There should be some fluid in this area, but if the fluid measures larger than expected, it increases the chance for Down syndrome, trisomy 18, and other birth defects.

The lab uses the measurements from the ultrasound and the blood draw, combined with information about the mom (age, weight, ethnicity, etc) to calculate a risk number. The results will be broken down by each condition that was screened for. For example, a first trimester screen result may say that the chance for Down syndrome is 1 in 5,000, and the chance for trisomy 18 is 1 in 10,000. This means that if there were 5,000 women that had the exact same test result, only 1 would have a child with Down syndrome. Likewise, if there were 10,000 women with the exact same test result, only 1 would have a child with trisomy 18.

A low-risk, or negative, FTS means the chances are reduced, but this test does not rule out any medical conditions. Similarly, a FTS result that shows an increased chance does not mean that the pregnancy has that condition. If your results show an elevated risk, you may be offered further testing, such as a diagnostic prenatal test. It's important to remember that most people who have a 'positive' or 'high-risk' FTS will go on to have healthy babies.

People choose either to do or not to do prenatal genetic screening for many reasons. It is

important to remember that this testing is voluntary and is not required.

Your results are high-risk. Now what?

If you had a FTS done with your primary OB-GYN, they may refer you to a high-risk pregnancy doctor called a perinatologist. You may also be referred to meet with a genetic counselor. These visits are designed to gather more information, but also to make sure you have a clear understanding of everything that is going on. It is also important during these visits to talk about the risks, benefits, and limitations of different options for further testing, including cell-free DNA, chorionic villus sampling (CVS), or amniocentesis. The choice of whether or not to undergo further testing is yours, and further testing is always voluntary.

Your results are low-risk. Now what?

A low-risk test result means the chance that the pregnancy has the conditions that were screened for is reduced, but not eliminated. Depending on the lab, between 80-95% of pregnancies that have Down syndrome and trisomy 18 will have a positive FTS, which means that approximately 5-20% will have low-risk FTS results. The chance that a pregnancy that does not have Down syndrome or trisomy 18 will have a high-risk test result, or a false positive, is usually between 3-5% (depending on the lab).

How do I decide whether to get FTS or not?

If you're on the fence about whether or not to pursue FTS, it may be helpful to consider the following:

- If the FTS shows a high risk for a condition, would you pursue additional testing, such as CVS or amniocentesis?
    - If not, would you be ok waiting until the baby is born to know for sure if they are affected?
- Would you want this kind of information to be better prepared?
- Would you consider doing anything differently if you knew a pregnancy was affected with one of these conditions, such as placing the baby up for adoption or not continuing the pregnancy?

- Does more information with the possibility of uncertainty make you nervous or anxious?

It is important to talk with your medical provider or a genetic counselor if you have further questions or concerns.

Click here to learn more about scheduling a genetic counseling appointment for pregnancy-related questions.

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