



Sequential screening (SS) is a test that can tell the chance that a pregnancy has [Down syndrome](#) (trisomy 21), [trisomy 18](#), and [open neural tube defects \(ONTDs\)](#). The first part of the SS is generally performed in the first trimester and consists of a blood draw from mom and an [ultrasound](#). Part two consists of a second blood draw from mom.

The first part of the sequential screen is similar to the [first trimester screen \(FTS\)](#), and the second part is similar to [second trimester screening \(STS\)](#). The first part of the sequential measures two hormones and 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).

- 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](#).
- 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.

The results of the blood test is combined with the first trimester [ultrasound](#), which measures the fetus to verify dating as well as pocket of fluid behind the baby's neck called the nuchal translucency (NT). The lab uses a formula that includes the information from the blood work as well as the NT measurement to come up with a risk number. The second part of the sequential screen measures hCG again, as well as:

- Inhibin-A: Inhibin-A comes from the placenta. Levels of this hormone in mom's blood remain relatively constant through weeks 15-18 of the pregnancy. Pregnancies with [Down syndrome](#) tend to have higher than average levels of inhibin-A, while pregnancies with [trisomy 18](#) tend to have lower levels of inhibin-A.
- Unconjugated estriol (uE3): uE3 is made by both the baby's liver and the placenta. Levels of this hormone rise throughout the pregnancy. Pregnancies with [Down syndrome](#) or [trisomy 18](#) tend to have lower levels of uE3.
- Alpha-fetoprotein (AFP): AFP is a protein produced in the baby's liver during the second trimester of pregnancy. The levels of AFP generally increase the further into the pregnancy you are. AFP levels tend to be higher when the baby has an [open neural tube defect \(ONTD\)](#), such as spina bifida. With [ONTDs](#), there is an opening in the skin and AFP has another 'escape route' out. AFP levels tend to be lower in pregnancies



that have [Down syndrome](#) or [trisomy 18](#).

The lab uses information from the blood draws and 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.

The first part of the sequential screen will usually give the chance for [Down syndrome](#) and [trisomy 18](#). The results do not tell you 'yes' or 'no', but tell you the chance that the pregnancy has that condition. For example, the first part of the sequential screen 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](#).

The second part of the sequential screen will give you an updated risk for [Down syndrome](#) and [trisomy 18](#), as well as the risk for [ONTDs](#).

A low-risk, or negative, SS means the chances are reduced, but this test does not rule out any medical conditions. Similarly, a SS result that shows an increased risk 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' SS 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 SS 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](#) 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 90-96% of pregnancies that have [Down syndrome](#) and [trisomy 18](#) will have a positive SS, which means that approximately 4-10% will have low-risk SS 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 SS or not?**

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

- If the SS 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.

## **Related Articles**

- [Ultrasound](#)  
Prenatal ultrasound, also known as sonogram, is an imaging technique that is used to

look at a fetus during the pregnancy. Ultrasound involves using sound waves that reflect off of the different parts of the baby to generate a picture. This picture can be used to take measurements, which can...

- [First Trimester Screening \(FTS\)](#)

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...

- [Second Trimester Screening](#)

Second trimester screening (STS), also called quad screen or triple screen, is a test that can tell the chances that a pregnancy has Down syndrome (trisomy 21), trisomy 18, and open neural tube defects (ONTDs). STS is generally done between 15 and 22 weeks of the pregnancy, and consists of...

- [Cell-free DNA Screening](#)

Cell-free DNA (cfDNA) is a screening test that can be performed any time after 10 weeks of pregnancy that screens for common chromosome conditions (Down syndrome, trisomy 18, and trisomy 13), as well as sex chromosomes (XX and XY), which can also incidentally tell the sex of the baby. cfDNA...

- [Carrier Screening](#)

While most screening tests we talk about during pregnancy are designed to give us more information about the baby, genetic carrier screening is a test that gives us information about mom and dad. As you may recall from Genetics 101, we have two copies of most genes: one we get...