

# Follow up to Screening

It is recommended that all pregnant women have a detailed ultrasound to screen for structural birth defects. Patients are informed that a normal or reassuring ultrasound reduces the risk of abnormalities in the baby, but is never a guarantee of a normal outcome. For women who have completed FTS and do not go on to have amniocentesis, a blood screen should be done for NTD risk only.

It is not recommended that a screen positive result be repeated or followed up by any other blood test for Down syndrome or trisomies 18 and 13. Women who receive screen positive or "at risk" results are offered genetic counseling. After counseling, they have the option to proceed with a diagnostic test, such as chorionic villi sampling (CVS) or an amniocentesis. Diagnostic testing is not completely risk free, but in the hands of an experienced physician, such as the Perinatologists at Desert Perinatal Associates, the risk for causing a miscarriage is very small. CVS is performed in the first trimester and is associated with a miscarriage risk of 1/200. Amniocentesis may be completed in the second trimester and has a miscarriage risk in the range of 1/300 to 1/1600.

## Should I have FTS?

The decision to have FTS is a personal one based on your beliefs and concerns. We hope this brochure has helped you to decide whether this option is for you. If you have additional questions please feel free to contact a genetic counselor at Desert Perinatal Associates.

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A  
helpful  
guide  
to  
Screening

Traditionally women over the age of 35 were offered amniocentesis and those under 35 were offered a form of blood screening. The boundaries are now blurring substantially. Many women over 35 are choosing to complete a serum screen to better estimate their personal risk for certain birth defects including Down syndrome, trisomies 18 and 13, and neural tube defects, before deciding about amniocentesis.

## Down syndrome

Down syndrome occurs when a baby has an extra copy of chromosome number 21 (three copies of chromosome #21 instead of the expected two). Individuals with Down syndrome have mild to severe mental retardation, characteristic facial features, and low muscle tone. They are also at an increased risk for heart defects. Approximately 1 in 800 babies are born with Down syndrome. Women of any age can have a child with Down syndrome. However, mothers age 35 or older are at a greater risk of having a baby with Down syndrome.

## Trisomy 18 and Trisomy 13

Trisomy 18 and trisomy 13 are also caused by extra chromosomes (three copies of either chromosome #18 or #13 instead of the expected two). Like Down syndrome, the chance of having a baby with either trisomy 18 or trisomy 13 increases in mothers over age 35. Babies born with trisomy 18 or trisomy 13 have severe mental retardation. They may also have heart and brain defects. The majority of these babies (90%) die within one year after birth. Trisomies 18 and 13 occur in about 1 in 6,000 to 1 in 8,000 births.

## Neural Tube Defects

Neural tube defects (NTD) occur when the spinal cord or brain does not develop correctly. NTDs occur early in development, between the 3rd and 4th weeks of pregnancy. One in every 1,000 babies is born with an NTD in the United States. Spina bifida is the most common type of NTD. Spina bifida occurs when the spinal cord bulges out of an opening in the vertebrae. Affected children usually have paralysis and loss of bowel and bladder control. Occasionally, they have hydrocephalus (a build up of fluid in the brain) and learning disabilities. Anencephaly is another common type of NTD, resulting in a very severe brain and skull malformation. Infants with anencephaly are either stillborn or die shortly after birth. Folic acid supplementation is known to reduce the risk for NTDs.

## Screening Test Options

Screening tests estimate a risk (or statistical probability) for certain birth defects. They provide a more personalized risk assessment for a woman's pregnancy than her age risk or general population risk alone. If a pregnancy is identified as high risk, additional testing such as ultrasound or amniocentesis, is needed to learn more. It is important to note that these screening tests can not estimate a risk for all types of birth defects. For every pregnancy the background risk for birth defects is 3-5%.

### First Trimester Screen (FTS)

The first trimester screen (FTS) estimates a risk for Down syndrome and trisomies 18 and 13. The FTS includes two kinds of information which are combined to determine the final risk. First, a maternal blood sample is collected which analyzes two proteins produced by the pregnancy. This blood sample can be obtained from the beginning of the 9th week of pregnancy through the end of the 13th week. The second part of the test is a specialized ultrasound measurement at the back of the baby's neck. This can be completed between the beginning of the 10th week and the end of the 13th week of pregnancy. The FTS results are generally available about one week after the blood sample is obtained.

The NT is the fluid filled space between the back of the baby's neck and the overlying skin. Babies with Down syndrome, certain other chromosomal diseases, and congenital heart defects tend to accumulate more fluid at the back of the neck, causing the space to be thicker or larger. Sonographers at Desert Perinatal Associates are certified to perform an NT measurement.

### Second Trimester AFP4 Screen

The AFP4 or quad screen is performed in the second trimester and calculates a risk for Down syndrome, trisomy 18, and neural tube defects. The AFP4 screen is drawn between the start of week 15 and the end of week 21. A maternal blood sample is analyzed for four proteins produced by the placenta and the baby. The pattern these proteins make is used to determine a risk estimate for Down syndrome, trisomy 18, and neural tube defects. The AFP4 screen results are finished approximately 3-5 days after blood draw.

## How accurate is the test?

The FTS detects approximately 85-90% of pregnancies affected with Down syndrome. The AFP4 screen detects about 70-80% of pregnancies with Down syndrome. Approximately 90% of pregnancies with a neural tube defect are identified as high risk by a second trimester AFP or AFP4. **Because of the higher detection rate and earlier results, we at Desert Perinatal Associates, believe that FTS is a better option than AFP4 screening.**

For accuracy of the screen test calculations it is important to include the correct gestational age, meaning the number of weeks and days completed in the pregnancy. It is determined by a woman's last menstrual period or an early ultrasound. The screen tests also use information on maternal age, weight, and race to determine the final risk estimate.

## What does a positive screen result mean?

A positive screen result does NOT mean that the baby definitely has a problem. It simply means that the pregnancy is at an increased risk for Down syndrome, neural tube defect or trisomy 18. For example, the test may indicate a 1 in 200 risk or a 0.5% chance for Down syndrome, which is considered screen positive. However, there is still a 99.5% chance the baby does not have Down syndrome. Even with a high risk estimate there is still a good chance the baby is unaffected.

A screen positive result identifies women at high risk so they can be offered further genetic counseling, detailed ultrasound, and diagnostic testing. The doctors and genetic counselors at Desert Perinatal Associates will help you understand your risk estimate and explain how ultrasound and diagnostic tests can help you learn more.

## What does a negative screen result mean?

A negative screen result means that the baby is at low risk to have Down syndrome, neural tube defect or trisomy 18. However, even with a low risk estimate there is still a small chance the baby could be affected. A low risk result does not guarantee that your baby will not have Down syndrome or some form of birth defect.

## Are there risks with screening?

There is no risk to you, or your unborn baby with first trimester screening. The blood sample will be obtained either by a fingerstick or simple blood draw from your arm. The nuchal translucency is accomplished with ultrasound waves that are harmless to your baby.