Follow up to Screening

The results from part one and part two of the sequential screen will be finished approximately 5-7 days after each blood draw.

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 it cannot guarantee a normal outcome.

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 villus 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 of approximately 1/1000.

Should I have Screening

The decision to have screening 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

Many pregnant women choose 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. Screening may be able to help women of all ages and backgrounds make important decisions about their diagnostic testing options during pregnancy.

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 cannot estimate a risk for all types of birth defects. For every pregnancy the background risk for birth defects is 3-5%.

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.

Sequential Screening

The sequential screen is completed in two parts, which are combined to determine a final risk estimate for Down syndrome, trisomy 18, and neural tube defects.

First Trimester

The first part of this screening is completed between the 10th week of pregnancy through the end of the 13th week. The screen includes a specialized ultrasound measurement at the back of the baby's neck, or the nuchal translucency. This is combined with a maternal blood sample. This blood sample analyzes two proteins produced by the pregnancy. The first part of the screen will estimate a preliminary risk for Down syndrome and trisomy 18.

The nuchal translucency (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, causing the NT space to be thicker or larger. Sonographers at Desert Perinatal Associates are certified to perform an NT measurement.

Second Trimester

The second part of screening can be completed between the 15th week through the end of the 21st week. The second maternal blood draw measures four chemicals produced by the placenta and the baby. The sequential screen is completed after information collected in the second part is combined with the information from the first part of the screen. The sequential screen will give a final risk estimate for Down syndrome, trisomy 18, and neural tube defects.

If the result from the first part is screen negative, or low risk, the second part of the blood work will be drawn. After the first part and second part of the sequential screen have been combined a final risk estimate will be calculated.

If the result from the first part of screening is screen positive, or high risk, it is not necessary to complete the second part of the sequential screen. At that time, you will be offered genetic counseling and diagnostic testing, such as CVS or amniocentesis.

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How accurate is the test?

The Sequential Screen alone detects approximately 80-90% of pregnancies affected with Down syndrome or trisomy 18. The detection rate for a neural tube defect is approximately 70-80%.

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 cannot guarantee that your baby will be healthy and have no birth defects.

Are there risks with screening?

There is no risk to you, or your unborn baby with screening. It simply requires a blood draw from your arm. The nuchal translucency is accomplished with ultrasound waves that are harmless to your baby.