Results for Screening in the Second Trimester

Quad Marker Screening, or Serum Integrated Screening, or Full Integrated Screening

The result of your Second Trimester blood test is:

“Screen Positive for Neural Tube Defects or Abdominal Wall Defects”

This means there is an increased risk that the fetus may have a birth defect called neural tube defect or abdominal wall defect.
As part of your prenatal care, you had a blood test between 15 and 20 weeks. You may also have had:
• a first trimester blood test and/or
• nuchal translucency ultrasound

The Screening results tell you the risk, or chance, that there is a certain birth defect.

The result of your Screening test was “Screen Positive” for a Neural Tube Defect (NTD) or Abdominal Wall Defect (AWD).

What is the neural tube?
The neural tube starts at the top of the head and ends at the bottom of the spine. The neural tube becomes the baby's brain and spinal cord. (See pages 4 and 5 for information on NTDs and AWDs.)

Does the “Screen Positive” result mean that the fetus really does have an NTD or AWD?
No…Most women with this test result have normal, healthy babies.

What can you do next?
Consider follow-up testing to know for sure if the fetus has an NTD or AWD (see page 2).
Follow-up testing . . .
There are no additional charges for these authorized services at a State-approved Prenatal Diagnosis Center.

The follow-up services that are offered are:
• Genetic Counseling
• Ultrasound
• Amniocentesis

Genetic counseling is the first service
A genetic counselor discusses your screening results and what they mean. Your family’s health history is also reviewed. The counselor will give you information about the follow-up testing available to you. You may decline any services or tests at any time. Be sure to talk with the counselor about any questions you may have.

You have testing options to choose from after a “Screen Positive” result. Ultrasound and amniocentesis are discussed on the next pages.

Ultrasound at a State-approved Prenatal Diagnosis Center
This test is also called a sonogram. This high level ultrasound is done between 15 and 24 weeks. It is a very detailed picture of the fetus done by doctors with special training.

An ultrasound can show how many fetuses you are carrying. It will also tell you how far along you are in your pregnancy. Ultrasound identifies some birth defects and abnormalities such as NTDs and AWDs (see page 5).

You can have this high level ultrasound even if you decline amniocentesis.
What is a Neural Tube Defect?
The neural tube is completely formed by 5 weeks after conception. Birth defects occur when the neural tube does not develop properly. The two most common neural tube defects are spina bifida and anencephaly. These defects occur in about 1 in every 1,000 births.

Infants born with spina bifida have an opening in the bones of the spine which exposes the nerves. This causes leg paralysis and loss of bowel and bladder control.

Anencephaly occurs when a large part of the skull is missing and most of the brain does not develop. This defect causes the death of the fetus or newborn.

What is an Abdominal Wall Defect?
This means there is an abnormal opening on the abdomen. This opening causes the intestines to form outside the body. Omphalocele is an opening on the infant's abdomen where the umbilical cord is attached. This occurs in about 1 in 4,000 births. Gastrochisis is an opening next to the umbilical cord. This occurs in 1 in 10,000 births. There may be chromosomal abnormalities as well. Surgery soon after birth can often repair an abdominal wall defect.

Amniocentesis at 15 – 24 weeks
This diagnostic test involves removing a small amount of fluid from around the fetus. A thin needle is used to remove a small quantity of the fluid. The fetus is not touched. Tests on this fluid show the presence of NTDs and AWDs. Some of the fluid is also used to examine the chromosomes in the fetal cells.

Amniocentesis is considered a safe test when done by medical experts at a State-approved Prenatal Diagnosis Center. The risk of miscarriage due to amniocentesis is small – less than 1%.

The results of the amniocentesis are ready in about 2 weeks. Most results are normal.

Please Remember:
Most women who have “Screen Positive” results have normal, healthy babies.
The California Prenatal Screening Program
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Genetic Disease Screening Program
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For more information see our website:
www.cdph.ca.gov/programs/pns