The result of your Second Trimester blood test is: “Screen Positive for Trisomy 18”

This means there is an increased risk that the fetus may have a birth defect called Trisomy 18
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

Your Screening results include any of these tests that were done. The Screening results tell you the risk, or chance, that there is a certain birth defect. For example, a risk could be 1 in 40 or 1 in 5,000.

The result of your Screening test was “Screen Positive for Trisomy 18”. The risk of your fetus having Trisomy 18 is ________________.

**What is Trisomy 18?**

It is a birth defect which causes severe intellectual disabilities and very serious health problems. It is caused by an extra chromosome #18. Chromosomes are packages of genetic information found in every cell of the body. Birth defects can occur when there are too few or too many chromosomes.

**Does the “Screen Positive” result mean that the fetus really does have Trisomy 18?**

No… Most women with this test result have normal, healthy babies.

**What can you do next?**

Follow-up testing to know for sure if the fetus has Trisomy 18 *(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
- Non-invasive Prenatal Testing
- 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.

The counselor will explain that you have testing options to choose from after a “Screen Positive” result. Ultrasound, NIPT, 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 help determine the age of the fetus. It may identify some birth defects or abnormalities. However, it is not a diagnostic test for Trisomy 18.

You can have this high level ultrasound even if you decline NIPT and Amniocentesis.
NIPT (Non-invasive Prenatal Testing)

This is a test on fetal DNA that is found in the mother’s blood. NIPT is considered to be a very accurate screening test for certain chromosome abnormalities like Down syndrome, Trisomy 18, Trisomy 13, and some sex chromosome abnormalities. NIPT is offered in the first trimester (11-14 weeks) and second trimester (15-24 weeks) of pregnancy.

The results of NIPT are ready in about two weeks.

Amniocentesis at 15 – 24 weeks

This diagnostic test involves removing a small amount of the fluid around the fetus. A thin needle is used to remove a small quantity of the fluid. The fetus is not touched. This fluid contains cells from the fetus. The chromosomes in these cells are counted and examined.

Amniocentesis can tell if the fetus has Trisomy 18. It can also detect 99% of other chromosomal birth defects. 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 in 100.

The results of the amniocentesis are ready in about 2 weeks. Most results are normal.
What if Trisomy 18 is found?

A doctor or genetic counselor would give you information about Trisomy 18. Infants with this birth defect have severe intellectual disabilities and very serious health problems. They usually die before birth or in early infancy. Trisomy 18 occurs in about 3 out of every 10,000 births in the United States.

Options for continuing or ending the pregnancy will be discussed during counseling. The decision is entirely up to you.

The California Prenatal Screening Program does not pay for any other medical services after the follow-up tests and counseling. Referrals for medical care and support services are available.

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

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