The California Prenatal Screening Program

Results for Screening in the First Trimester

The result of your blood screening 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

Positive T18—1T

As part of your prenatal care, you had a First Trimester Screening between 10 and 14 weeks. This included:

- a blood test and
- nuchal translucency ultrasound

The First Trimester Screening result tells 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 First Trimester Screening was "Screen Positive for Trisomy 18". The risk of your fetus having Trisomy 18 is _____.

You were also given a risk for Down syndrome. That risk is _____.

What is Trisomy 18?

This birth defect causes severe mental retardation and very serious health problems. Trisomy 18 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?

Choose one of the following options:

1. Another blood screening test between 15 and 20 weeks to get a revised risk *(see page 3)*, **or**
2. Follow-up testing to know for sure if the fetus has Trisomy 18 *(see page 4)*.

Either option is available at no additional charge.

You may ask for a referral for free genetic counseling at a State-approved Prenatal Diagnosis Center. The genetic counselor will explain your test results and help you decide which option is best for you.

Option 1: Another blood screening test at 15 to 20 weeks

This is an option for women who decide not to have follow-up testing right away. The results of this new blood screening test are combined with results from the First Trimester Screening blood test you had. Combining the results gives a new, revised risk for several birth defects, including Trisomy 18.

About half the time, the new results will again be "Screen Positive". In this case, you will once again be offered follow-up testing. About half the time, the new results will be "Screen Negative" (meaning low risk).

Please note that even if your result has changed to "Screen Negative", there is still a small chance (your new risk), that the fetus has Trisomy 18. However, this "Screen Negative" result will mean that diagnostic testing will not be offered by the Program. This means that a few cases of Trisomy 18 will not be identified by the Program after a second blood test.

Option 2: Follow-up testing . . . to know for sure

The follow-up services that are offered are:

- Genetic Counseling
- Chorionic Villus Sampling (CVS)
- Ultrasound
- Amiocentesis

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 ask the counselor any questions you may have.

The counselor will explain that you have several testing options to choose from after a "Screen Positive" result. CVS, ultrasound, and amniocentesis are discussed on the next pages.

CVS (Chorionic Villus Sampling)

This diagnostic test can only be done early in pregnancy, **between 10 and 14 weeks.** This test removes a few cells from the placenta (in the uterus), using a very thin needle or tube. The fetus is not touched. The cells contain the same chromosomes as the fetus. The chromosomes are counted and examined.

CVS can tell you if your fetus has Trisomy 18. CVS also detects 99% of other chromosomal birth defects. It is considered a safe test when performed by the medical experts at a State-approved Prenatal Diagnosis Center. The risk of miscarriage due to CVS is small – less than 1%.

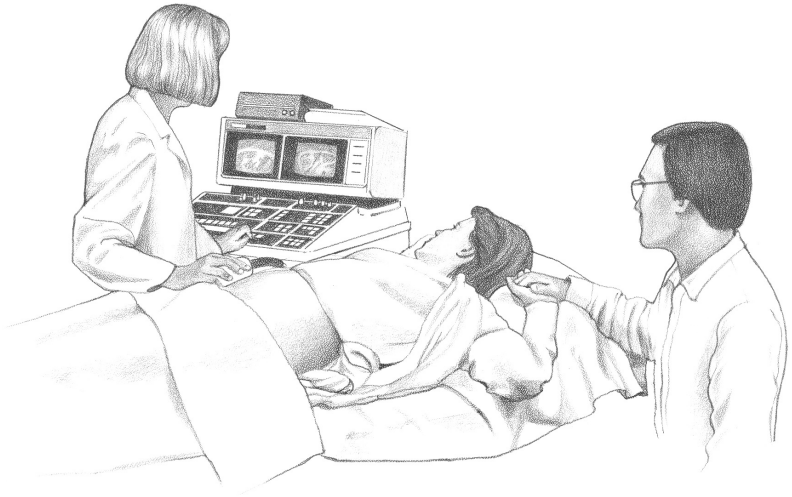
The results of CVS are ready in about two weeks. Most CVS results are normal.

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 CVS or Amniocentesis.



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 the 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.

What if Trisomy18 is found?

A doctor or genetic counselor would give you information about Trisomy 18. Infants with this birth defect have severe mental retardation 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

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For more information see our website:
www.cdph.ca.gov/programs/pns

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