

**The California Prenatal Screening Program  
Results for Screening in the First Trimester**



**The result of your blood screening test is:  
“Screen Positive for Down Syndrome”**

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

**A**s 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 Down syndrome”. The risk of your fetus having Down syndrome is \_\_\_\_\_

You were also given a risk for Trisomy 18.  
That risk is \_\_\_\_\_ .

### **What is Down syndrome?**

This birth defect causes mental retardation and some serious health problems. Down syndrome is caused by an extra chromosome #21. 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 Down syndrome?

*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 Down syndrome (*see page 4*)

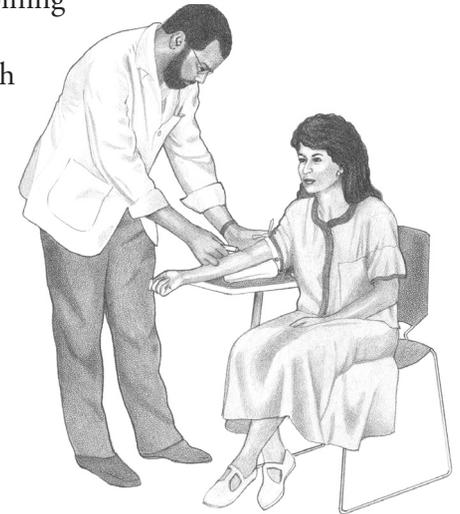
Either option is available at no additional charge.

**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 Down syndrome.

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 Down syndrome. However, this “Screen Negative” result will mean that diagnostic testing will not be offered by the Program. This means that a few cases of Down syndrome 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
- 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 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 Down syndrome. 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%.

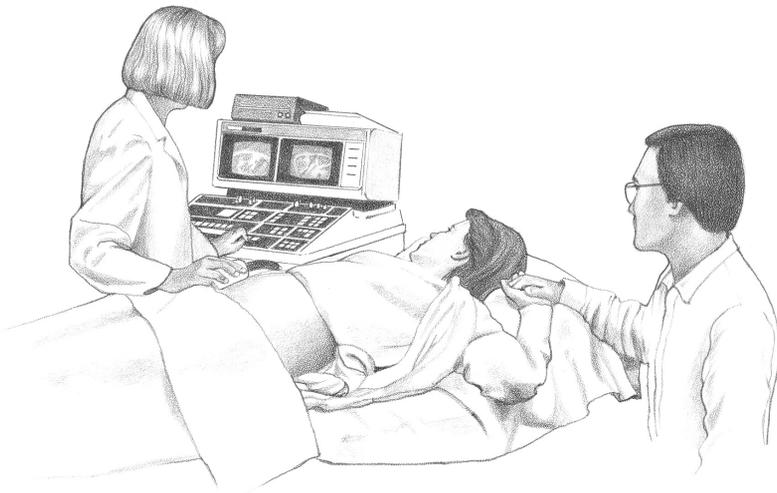
The results of CVS are ready in about two weeks. Most CVS results are normal.

## 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 Down syndrome.

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 Down syndrome. 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 Down syndrome is found?

A doctor or genetic counselor would give you information about Down syndrome. Infants with this birth defect are moderately retarded; a few are mildly or severely retarded. Heart defects are common. These heart defects can usually be treated with surgery and medications. Other serious health problems often exist with Down syndrome. Medical treatment can help some of these problems.

Special programs are available throughout California to help children and adults with Down syndrome to achieve their full potential.

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. Referral 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  
850 Marina Bay Parkway, F175  
Richmond, CA 94804  
866-718-7915 (Toll Free)

For more information see our website:  
[www.cdph.ca.gov/programs/pns](http://www.cdph.ca.gov/programs/pns)

