

# California Prenatal Screening Program

## Prenatal screening result and Down syndrome

What you should know when your blood screening result suggests there is **an increased chance** that your fetus may have a genetic condition called Down syndrome, or trisomy 21



**Down Syndrome**

[www.cdph.ca.gov/pns](http://www.cdph.ca.gov/pns)





# Prenatal screening result and Down syndrome

The result of your screening suggests that your fetus may have an increased chance of a genetic condition called Down syndrome, or trisomy 21.

## Why have you been given this booklet to read?

As part of your prenatal care, you consented to prenatal screening. Your blood was sent to a laboratory to screen for certain genetic conditions and birth defects in your fetus. The result of your screening suggests that your fetus **may have an increased chance** of a genetic condition called Down syndrome, or trisomy 21.

## Does this screening result mean that the fetus really does have Down syndrome?

No. Prenatal screening finds fetuses at higher risk. For Down syndrome it is accurate, but not 100% accurate. Follow-up testing can help give more information.

## What is Down syndrome?

Down syndrome is a genetic condition that causes intellectual disabilities and some serious health problems.

Down syndrome is caused by a third copy of chromosome 21. Usually there are only two copies.

Chromosomes help the fetus develop and are found in every cell in the body. If there are missing or extra chromosomes, birth defects can happen.

## What can you do next?

There are several tests that can tell with more certainty if the fetus does have Down syndrome. You decide if you want to have one of these tests.

## What follow-up services are available?

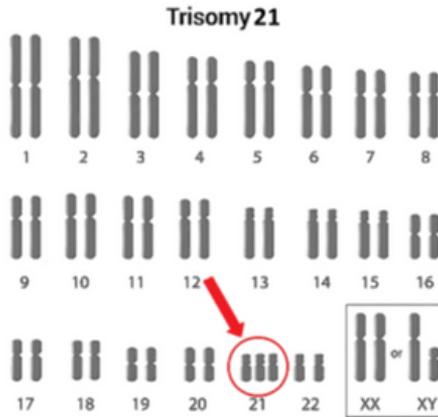
Your prenatal care provider will offer follow-up services, including genetic counseling and ways to tell with more certainty if the fetus does have Down syndrome.

Follow-up services are offered at locations that specialize in these services. These locations are called state-approved Prenatal Diagnosis Centers.

Follow-up services are available at no additional charge. You decide if you want follow-up services.

## Follow-up services include:

- Genetic Counseling
- Ultrasound
- Chorionic villus sampling or amniocentesis



Down syndrome is caused by three copies of chromosome 21 in cells

## What is genetic counseling?

Genetic counseling is the first service provided at a state-approved Prenatal Diagnosis Center. A genetic counselor discusses your screening result and what it means. You will also review your family's health history. The genetic counselor will give you information about the other follow-up services available to you.

The counselor will explain that you have several options that include ultrasound and chorionic villus sampling or amniocentesis. Be sure to ask the genetic counselor any questions you may have.

You decide if you want any additional follow-up services after genetic counseling.

## Ultrasound (sonogram)

Doctors with special training can do a high-level ultrasound, in general starting from **12 weeks of pregnancy**. It gives a very detailed picture of the fetus.

An ultrasound checks the age of the fetus. It may identify some birth defects. However, it is not a way to find out for certain if your fetus does have Down syndrome.

You can have this ultrasound even if you decide not to have chorionic villus sampling or amniocentesis.

## Chorionic villus sampling

Chorionic villus sampling (CVS) is a test that can tell if the fetus has Down syndrome. CVS can be done from **10 through the end of 13 weeks of pregnancy**.

For this test, an experienced doctor removes a small amount of tissue using a very thin needle or tube. The fetus is not touched. The tissue contains the same chromosomes as the fetus. The chromosomes are counted and examined.

It is considered a safe test when performed by a medical expert at a state-approved Prenatal Diagnosis Center. The risk of miscarriage due to CVS is small – less than 1 in 450. The CVS result is usually ready in two weeks.

## **Amniocentesis**

Amniocentesis is a test that can tell if the fetus has Down syndrome. This test can be done starting from **15 weeks of pregnancy**.

For this test, an experienced doctor takes a small amount of the fluid around the fetus with a thin needle. The fetus is not touched. This fluid contains cells from the fetus. The chromosomes in these cells are counted and examined.

Amniocentesis is considered a safe test when done by a medical expert at a state-approved Prenatal Diagnosis Center. The risk of miscarriage due to amniocentesis is small – less than 1 in 900. The amniocentesis result is usually ready in two weeks.

## **What if a test confirms your fetus has Down syndrome?**

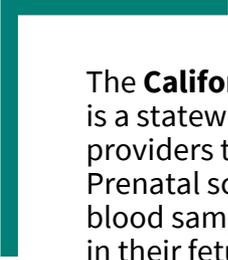
A doctor or genetic counselor will give you information about Down syndrome and how it could affect your fetus's health and your family.

Infants with this genetic conditions have intellectual disabilities that could be mild, moderate, or severe since Down syndrome can affect people differently. Heart defects are common. These heart defects can usually be treated with surgery and medications. Down syndrome often causes other serious health problems. Medical treatment can help some of these problems.

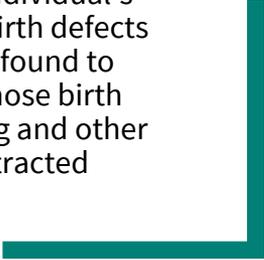
Special programs are available throughout California to help children and adults with Down syndrome to thrive. They teach the importance of supporting and valuing all people of differing abilities.

Your health care provider can discuss your options for the pregnancy with you. When reviewing your options, please remember that the decision on what to do is entirely up to you.

Medi-Cal and private insurance must cover all PNS Program fees, with only a few exceptions. Referral for additional medical care and support services are available, but these costs are not covered by the PNS Program.



The **California Prenatal Screening Program** is a statewide program offered by prenatal care providers to all pregnant individuals in California. Prenatal screening uses a pregnant individual's blood samples to screen for certain birth defects in their fetus. Individuals with a fetus found to have an increased chance of one of those birth defects are offered genetic counseling and other follow-up services through state-contracted Prenatal Diagnosis Centers.





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[www.cdph.ca.gov/PNS](http://www.cdph.ca.gov/PNS)

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