California Prenatal Screening Program

Prenatal screening result and trisomy 18

What you should know when your blood screening result suggests there is an increased chance that your fetus may have a genetic condition called trisomy 18 or Edwards syndrome.

Trisomy 18
www.cdph.ca.gov/pns
Prenatal screening result and trisomy 18

The result of your screening suggests that your fetus may have an increased chance of a genetic condition called trisomy 18 or Edwards syndrome.

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 trisomy 18 or Edwards syndrome.

Does this screening result mean that the fetus really does have trisomy 18?
No. Prenatal screening finds fetuses at higher risk. For trisomy 18 it is fairly accurate, but not 100% accurate. Follow up testing can help give more information.

What is trisomy 18?
Trisomy 18 is a genetic condition that causes severe intellectual disability and very serious health problems.

Trisomy 18 is caused by a third copy of chromosome 18. 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 trisomy 18. You decide whether 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 trisomy 18.

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 exam
- Chorionic villus sampling or amniocentesis

Trisomy 18 is caused by three copies of chromosome 18 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 genetic 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 trisomy 18.

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 trisomy 18. CVS can be done from **10 to 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 trisomy 18. 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 trisomy 18?**

A doctor or genetic counselor will give you information about trisomy 18 and how it could affect your fetus’s health, and your family.

Trisomy 18 can affect fetuses differently. They usually die before birth or in infancy. Infants with this genetic condition have severe intellectual disabilities and very serious health problems. Trisomy 18 happens in about 1 out of every 3,000 births in the United States.

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. Medi-Cal and private insurance must cover all program fees, with only a few exceptions.
California Prenatal Screening Program
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