

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



Quad Marker Screening, or  
Serum Integrated Screening, or  
Full Integrated Screening

**The result of your Second Trimester blood test is:**

**“Screen Positive for  
Smith-Lemli-Opitz Syndrome  
or Other Birth Defects”**

This means there is an increased risk that the fetus may have a birth defect called Smith-Lemli-Opitz Syndrome.

**A**s 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

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 Smith-Lemli-Opitz Syndrome (SLOS). The risk of your fetus having SLOS is \_\_\_\_\_.

### **What is SLOS?**

This is a very rare birth defect. Babies born with SLOS cannot make cholesterol normally. “Screen Positive” results for SLOS can also indicate increased risk for **C**ongenital abnormalities or Fetal **D**emise (fetal death). This is why this screening is also called **SCD** screening. *(See page 5 for more information.)*

### **Does the “Screen Positive” result mean that the fetus really does have SLOS?**

*No*...Many women with this test result have normal, healthy babies.

### **What can you do next?**

Consider follow-up testing to know for sure if the fetus has SLOS, a congenital abnormality or fetal demise (*see page 2*).

## Follow-up testing . . .

There are no additional charges for these authorized services at a State-approved Prenatal Diagnosis Center.

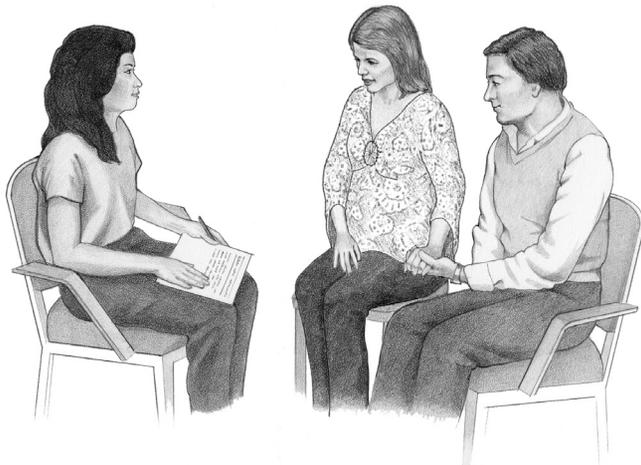
The follow-up services that are offered are:

- Genetic Counseling
- Ultrasound
- 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.

You have testing options to choose from after a "Screen Positive" result. Ultrasound and amniocentesis are discussed on the next pages.

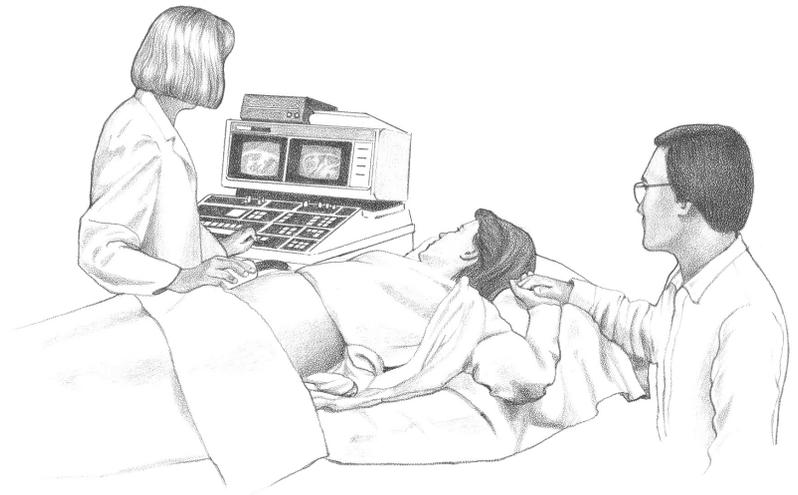


## Ultrasound at a State-approved Prenatal Diagnosis Center

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 show how many fetuses you are carrying. Ultrasound may help identify some congenital abnormalities such as fetal heart and bone abnormalities (*see page 5*).

You can have this high level ultrasound even if you decline amniocentesis.



## **Amniocentesis at 15 – 24 weeks**

This diagnostic test involves removing a small amount of fluid from around the fetus. A thin needle is used to remove a small quantity of the fluid. The fetus is not touched. Some of the fluid is used to examine the chromosomes in the fetal cells. Some of the fluid is also used to test for SLOS by measuring cholesterol production in the fetus.

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%.

The results of the amniocentesis are ready in about 2 weeks. Most results are normal.

## **What is SLOS (Smith-Lemli-Opitz Syndrome)?**

This is a very rare birth defect. Babies born with SLOS cannot make cholesterol normally. Babies born with this condition are mentally retarded and have many serious physical defects.

## **What congenital abnormalities (birth defects) may be found?**

Babies may have skeletal abnormalities such as microcephaly (small head size) or polydactyly (an extra little finger or toe). Cardiac (heart) defects are also common.

## **What is fetal demise?**

Fetal demise means the fetus is no longer alive.

## **What if abnormalities are diagnosed?**

A doctor or genetic counselor will give you information about the diagnosis. 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.

# The California Prenatal Screening Program

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

