English

California Prenatal Screening Program

Prenatal screening and Klinefelter syndrome

What you should know when your blood screening result suggests there is **an increased chance** that your fetus has a genetic condition called Klinefelter syndrome.

Klinefelter syndrome
go.cdph.ca.gov/MyUnexpectedResults

California Department of PublicHealth

Prenatal screening result and Klinefelter syndrome

The result of your screening **suggests** that your fetus has **an increased chance** of a genetic condition called Klinefelter syndrome. With this screening result, follow-up testing can give you more information.

Why have you been given this booklet to read?

As part of your prenatal care, you consented to a cell-free DNA (cfDNA) prenatal screening through the California Prenatal (PNS) Screening Program. Your blood was sent to a laboratory to check for certain genetic conditions in your fetus. Your provider likely shared your screening results and this booklet so that you can learn about the follow-up services available to you.

Does this screening result mean that the fetus has Klinefelter syndrome?

No. Prenatal screening finds fetuses with **an increased chance** for Klinefelter syndrome. More testing is needed to know for certain.

What is Klinefelter syndrome?

Klinefelter syndrome, also called XXY, can cause mild developmental delays, learning disabilities, and infertility. The signs of Klinefelter syndrome are different from one person to another. Some people have no obvious signs.

Klinefelter syndrome occurs when there is an extra X chromosome (XXY). Most people either have two X chromosomes (XX) or one X and one Y chromosome (XY).

Your provider might refer to these as “sex chromosomes.” Chromosomes help the fetus develop and are found in nearly every cell in the body.

What can you do next?

There are follow-up tests that can tell you with more certainty if the fetus has Klinefelter 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 to tell you with more certainty if the fetus has Klinefelter syndrome.

Follow-up services include:

- Genetic Counseling
- Ultrasound
- Diagnostic tests: chorionic villus sampling or amniocentesis

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 if you get screening through the California PNS Program. You decide if you want the follow-up services.

What is genetic counseling?

Genetic counseling is the first service provided at a state-approved Prenatal Diagnosis Center. A genetic counselor will discuss 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. These include an ultrasound and chorionic villus sampling or amniocentesis.

Be sure to ask the genetic counselor any questions you might have. You decide if you want any additional follow-up services after genetic counseling.

Ultrasound (sonogram)

Providers with special training can do a high-level ultrasound as early as **12 weeks of pregnancy**. It gives a detailed picture of the fetus.

An ultrasound checks the age of the fetus. It might identify some birth defects. However, it is not a way to find out for certain if your fetus has Klinefelter 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 diagnostic test that can tell if the fetus has Klinefelter syndrome. CVS can be done from **10 weeks through the end of 14 weeks of pregnancy**.

For this test, an experienced provider removes a small amount of placental 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.

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

Amniocentesis

Amniocentesis is a diagnostic test that can tell if the fetus has Klinefelter syndrome.

This test can be done through the PNS Program starting from **15 weeks until 24 weeks of pregnancy.**

For this test, an experienced provider 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 a test done by a medical expert at a state-approved Prenatal Diagnosis Center. The risk of miscarriage due to amniocentesis is less than 1 in 900.The amniocentesis result is usually ready in two weeks.

What if a test confirms your fetus has Klinefelter syndrome?

A provider or genetic counselor will give you information about Klinefelter syndrome and how it could affect your family.

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.

For more information, please visit:
Go.cdph.ca.gov/MyUnexpectedResults

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 genetic conditions and birth defects in their fetus. Individuals with a fetus found to have an increased chance of one of those genetic conditions or birth defects are offered genetic counseling and other follow-up services through state-approved Prenatal Diagnosis Centers.

California Prenatal Screening Program
www.cdph.ca.gov/PNS

California Department of Public Health
Genetic Disease Screening Program
Prenatal Screening Program
850 Marina Bay Parkway,
F175 Richmond, CA 94804
866-718-7915 (toll free)
pns@cdph.ca.gov