# California Prenatal Screening Program

## Screening for more genetic conditions

**As of April 1, 2024, the California Prenatal Screening (PNS) Program has added X and Y chromosome variations to the list of conditions screened by the program**.

The California Prenatal Screening (PNS) Program is a voluntary program offered by your prenatal care provider. Prenatal screening checks for genetic conditions and birth defects in your fetus (developing baby).

The PNS Program will continue screening for Down syndrome (trisomy 21), trisomy 18, trisomy 13, and neural tube defects - such as spina bifida.

### What are the new genetic conditions added to the PNS Program screening panel?

The PNS Program is now screening for the four most common types of X and Y chromosome variations:

- **Turner syndrome**
- **Trisomy X**
- **XXY (Klinefelter)**
- **XYY**

Your provider may refer to "X and Y chromosome variations" as "sex chromosome aneuploidies (SCAs)"

| X and Y chromosome variations | Description |
|---|---|
| • **Turner syndrome** | This genetic condition can cause mild developmental delays, learning disabilities, short stature, infertility, and heart defects. |
| • **XXY (Klinefelter)**<br>• **Trisomy X**<br>• **XYY** | These genetic conditions cause symptoms that vary from one person to another. Some individuals have no noticeable symptoms. In others, effects might include developmental delays and learning disabilities. Individuals often respond well to early intervention services like speech therapy. |

To learn more about these X and Y chromosome variations, please speak to your prenatal care provider and visit go.cdph.ca.gov/MyScreening.

### What causes X and Y chromosome variations?

Chromosomes guide the development of the fetus and are found in every cell in the body. Usually there are two copies of each chromosome. However, in rare situations, the number of chromosomes varies. In terms of X and Y chromosomes, the usual pattern is XX or XY. A fetus with an X and Y chromosome variation has a pattern of chromosomes other than XX or XY. X and Y chromosome variations usually do not run in families and most often happen by chance.

### Do you have to get your fetus screened for X and Y chromosome variations?

**No.** Whether you get prenatal screening or not is **your choice**. If you choose prenatal screening, we recommend getting screening through the PNS Program. The PNS Program panel includes screening for X and Y chromosome variations.

## How do you get your fetus screened for X and Y chromosome variations?

This is done through one of the screenings offered by the PNS Program called cell-free DNA (cfDNA) screening. The screening will tell you whether there is an increased chance of the fetus having an X and Y chromosome variation, as well as Down syndrome (trisomy 21), trisomy 18, or trisomy 13. The maternal serum alpha-fetoprotein (MSAFP) screening checks for neural tube defects. We recommend getting both screenings – cfDNA screening and MSAFP screening.

## What happens if your screening shows an increased chance for an X and Y chromosome variation?

Prenatal screening for X and Y chromosome variations may not be as accurate as screening for the other genetic conditions and birth defects in the PNS Program. If screening through the PNS Program finds an increased chance of an X and Y variation, diagnostic testing is needed to confirm the result. The PNS Program offers follow-up services at no additional cost.

## X and Y chromosome variation screening could reveal fetal sex

If you decide to get prenatal screening, you can choose to learn the predicted fetal sex or not. If you get a screening result showing an increased chance for X and Y chromosome variations, it will probably be necessary to disclose the predicted sex of the fetus when discussing the screening results. Talk to your prenatal care provider about this.

## Why should I get prenatal screening through the PNS Program?

Some expecting parents want to know as much as they can about their fetus during pregnancy.

- The PNS Program gives pregnant individuals the option to get high-quality prenatal screening **at a low or no cost**.

- If individuals participate in the PNS Program, they can access the follow-up services at **no additional cost**. The follow-up services are available for individuals screened through the program who have received results of an increased chance for one (or more) of the conditions screened. These follow-up services include genetic counseling, an ultrasound exam, and diagnostic testing (chorionic villus sampling [CVS] or amniocentesis).

## What's the cost for screening through the PNS Program?

The fees are $232 for cfDNA screening and $85 for MSAFP screening. The cfDNA screening fee will increase to $344 on July 1, 2024.

**Almost all program participants do not pay these fees themselves.** Medi-Cal or private health insurance must cover the program fees with only a few exceptions for self-insured employers and out-of-state health plans. There is no co-payment, co-insurance, deductible, or any other form of cost sharing required for families with insurance coverage.

To learn more about X and Y chromosome variations and the recent California PNS Program changes, please visit go.cdph.ca.gov/MyScreening