

Genetic Disease Screening Program: Prenatal and Newborn Screening



Maternal and Child Health Access

September 15, 2022

Greetings!

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California Newborn & Prenatal Screening Programs



Genetic Disease Screening Program

California Department of Public Health

Today's Presentation

Faith Raider

- Genetic Disease Screening Program Overview
 - California Newborn Screening Program
 - California Prenatal Screening Program
- Focus on California Prenatal Screening Program
- Website and Educational Materials

Dr. Lisa Feuchtbaum

- Questions?



Genetic Disease Screening Program

- Largest screening program in the world providing prenatal and newborn screening services for over 35 years
- Sets standards for high-quality, cost-effective genetic services
- Serves all pregnant individuals and their newborns in California

Mission: To reduce the emotional and financial burden of disability and death caused by disorders that are genetic (passed down in families) or congenital (appear at birth).

Terms

- **Screening** is a way to identify people in a large population who have an **increased chance** of having a problem
- Screening assesses for risk – it is never 100% accurate
 - Health screenings include mammograms, blood pressure
- Newborn screening checks **newborn baby** for increased chances of having 80 rare genetic disorders
- Prenatal screening checks **fetus** for increased chances of having certain birth defects
- **Testing** is needed to find out if the screening was right

California Newborn Screening Program

- Blood is taken from baby's heel at 12 - 48 hours after birth and put on lab paper for testing
- Program screens for over 80 genetic disorders; testing needed
- Goal is early treatment
- Parents can say "no" only for religious reasons
- Parents get lab results and then can request sample destruction



California Prenatal Screening Program

- Blood is taken from pregnant individual in 1st and 2nd trimesters
- Program screens fetus for certain birth defects; testing needed
- Goal is to find out about birth defects early
- Pregnant individual decides whether to get prenatal screening

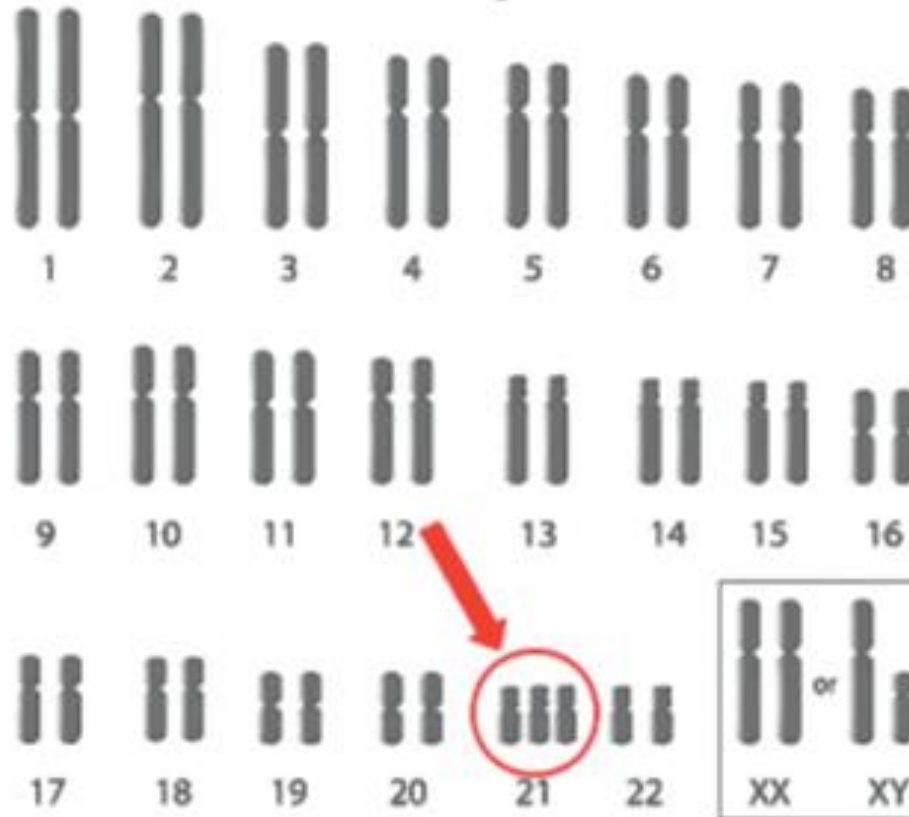


Prenatal Screening and Birth Defects

- Birth defects can happen in about 3 or 4 out of 100 pregnancies
- Birth defects are conditions in the fetus that can cause physical changes and intellectual disabilities
- Birth defects the state program screens for
 - Down syndrome and two other “genetic conditions”
 - caused by an extra chromosome in cells
 - Neural tube defects
 - happen during the development of the fetus’s nervous system

Genetic Condition Example: Down Syndrome

Trisomy 21



State Prenatal Program Screenings

- Two screenings (as of September 19, 2022)
 - **Cell-free DNA (cfDNA) screening for genetic conditions**
 - Recommended from 10 weeks to the first day of 21 weeks
 - Checks for Down syndrome (trisomy 21), trisomy 18, trisomy 13
 - **Maternal serum alpha-fetoprotein (MSAFP) screening for birth defects**
 - From 15 weeks to the first day of 21 weeks
 - Checks for neural tube defects, like spina bifida
- It is important to do both

What to Expect from the State Program

1. Providers must give information to pregnant individuals about prenatal screening through the state program
2. Pregnant individuals decide if they want to have the screenings
3. If so, there are consent forms for the two screenings
4. Blood is taken in a lab, clinic, or office – just like other blood draws
5. Prenatal care provider will give screening results

Possible Screening Results

- “No increased chance” of screened birth defects (most common result)
- “No call” result (unable to determine risk)
- “Increased chance” of screened birth defects



“No increased chance” of screened birth defects

- The chances of the fetus having one of the screened birth defects are not higher than usual
- The PNS Program does not offer follow-up testing and services
- The chances of the fetus having any of the screened birth defects are **low, but not zero**
- This result does not guarantee that there are no birth defects because no prenatal screening can detect 100% of birth defects

Screening Result of “No Call”



- “No call” means unable to determine risk
- Possible reasons include screening too early or too late
- Provider should share the reason for “no call” result
- Provider might offer to repeat screening
- Provider shares next steps

“Increased chance” of screened birth defects

- The chances of the fetus having one of the screened birth defects are higher than usual
- This result does not always mean that the fetus has a birth defect. **Testing is needed**
- Genetic counseling and follow-up testing and services at an approved Prenatal Diagnosis Center are offered
- Follow up services are provided at no additional cost

Program Participant Billing and Payment

- Two fees
 - Cell-free DNA (cfDNA) screening = \$232 fee
 - Maternal serum alpha-fetoprotein (MSAFP) screening = \$85 fee
- Medi-Cal and private insurance **must** cover program fees
- Exceptions: self-insured employers or out-of-state health plans
- If insurance does not cover fees, participant will receive bill
- Fees include follow-up services, if needed

Prenatal Screening Program Changes

- The Prenatal Screening Program is changing on September 19, 2022 – improved program
- cfDNA screening is more accurate than earlier method
- Screening options are simpler – first, second trimester
- Equity issues – Program gives all pregnant individuals access to a better screening option with price and quality regulated
- A parent who is pregnant may notice the program is a bit different than before

Prenatal Screening Program Website

- www.cdph.ca.gov/PNS
- Don't try to google us – use bookmarks

Home | Programs | Center for Family Health | Division of Genetic Disease Screening | Prenatal Screening Program

PRENATAL SCREENING PROGRAM

Genetic Disease Screening Program

Prenatal Screening Program

- About Us
- Communications and Education
- CalGenetic Portal
- Information for Individuals**
- Information for Providers
- Contact Us
- Patient Billing and

The PNS Program Is Changing!
Program changes will go into effect September 2022.
[Read More](#)

Healthcare provider using... TO

California Prenatal Screening Program



Scan QR code
to access the
PNS web page

Program Participant Information

- [Information for Individuals web page](https://bit.ly/PNS4Pregnancy)
(<https://bit.ly/PNS4Pregnancy>)
- Patient video in 3 languages (English, Spanish, Chinese)
- Patient Booklet in 6 languages (+Korean, Vietnamese, Tagalog)
 - 12 pages
 - Summary
- Patient FAQ
- Genetic Support Foundation videos



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access
the page

Prenatal Screening Patient Video

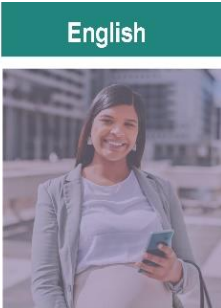



**Scan QR code to
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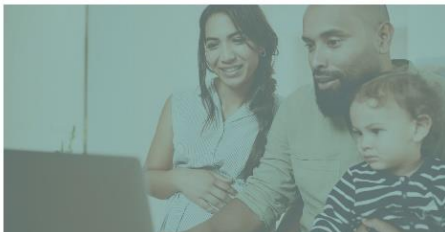


Prenatal Screening Patient Booklet

Prenatal Screening Patient Booklet

English



California Prenatal Screening Program
What Is Right for You?



www.cdph.ca.gov/pns



Scan QR code to
access the patient
booklet

Questions?



Evaluation

- Please **complete the evaluation survey online**
(<https://forms.office.com/g/CaETb25Lt0>)



Scan QR code to
access the evaluation

Thank You!

California Prenatal Screening Program

Send questions and comments:

GDSP.Communications@cdph.ca.gov

www.cdph.ca.gov/PNS

