

# The California Prenatal Screening Program

## California Department of Public Health

### Genetic Disease Screening Program

Coming Soon: cfDNA as First-tier Screen for Chromosomal Abnormalities

Fall 2021

#### Breaking News!

The California Prenatal Screening (PNS) Program is moving to cell-free DNA (cfDNA) as the first-tier screen for select chromosomal aneuploidies. Maternal serum alpha-fetoprotein (MSAFP) screening for certain structural birth defects will still be available through the PNS Program in the second trimester.

The PNS Program will continue to provide high-quality prenatal screening for every pregnant individual in California. Visit our website to follow program changes, which will begin in summer 2022: <https://www.cdph.ca.gov/PNS>

#### Why the Changed Program?

cfDNA is a first-tier screen that is available universally, throughout the state. The high sensitivity and specificity of this new technology, as reported in the scientific literature, means that fewer people will be referred for follow-up testing at our state-approved Prenatal Diagnosis Centers. Many patients will avoid unnecessary invasive diagnostic procedures, thus reducing concerns to them and their families.

cfDNA screening is a form of noninvasive screening, also known as noninvasive prenatal testing (NIPT). It is used to determine risk for a fetus to have certain genetic abnormalities. The noninvasive term is used to contrast cfDNA screening with traditional diagnostic tests such as amniocentesis or chorionic villus sampling (CVS). cfDNA testing analyzes fragments of placental DNA in maternal blood. The PNS Program will use cfDNA in maternal blood samples to screen for trisomy 21, trisomy 18, and trisomy 13 in the fetus.

The American College of Obstetricians and Gynecologists (ACOG) and American College of Medical Genetics (ACMG) recommend offering cfDNA screening for all pregnancies, the removal of previous guidelines recommending cfDNA to only 35-and-older and limiting to other-known-risk factors. ACOG specifically states in the new guidelines that cfDNA, "offers superior sensitivity and specificity and lower false positive rates for some aneuploidies," (most significantly for Down's syndrome). ACOG has stressed that cfDNA screening has the potential for false-positive and false-negative

results and is "not equivalent to diagnostic testing."

#### How Is the New Program Different?

Two screening tests will be available through the PNS Program, both requiring a maternal blood sample. There will be two separate fees, one for each test.

cfDNA screening will replace the biochemical screening tests now used in the first and second trimesters to screen for trisomy 21, trisomy 18 and trisomy 13. Nuchal translucency (NT) ultrasounds will no longer be used as part of risk assessment for the PNS Program. cfDNA screening can be offered as early as 10 weeks and up to 21 weeks of pregnancy.

MSAFP screening will still be available through the PNS Program in the second trimester to screen for neural tube and abdominal wall defects. cfDNA does not screen for neural tube or abdominal wall defects.

A patient with a screen positive result on either of the tests will continue to be offered follow up services at a State-approved Prenatal Diagnosis Center up to 24 weeks of pregnancy

## The Prenatal Screening Results Portal has launched!

<https://www.cdph.ca.gov/Programs/CFH/DGDS/Pages/pns/PNS-Results-Portal.aspx>

The Genetic Disease Screening Program (GDSP) now provides a secure online portal where the prenatal screening results ordered through the PNS Program can be viewed and downloaded electronically. The PNS Program will still mail paper mailers to providers ordering screening, but now providers will have an alternative option to review, print, and chart their patients' prenatal screening results.

### Frequently Asked Questions

#### What do I need to view results on the portal?

Users will need the PNS form number that the specimen was originally submitted with and the patient's date of birth as submitted to the PNS Program (or updated by a case coordinator). To view an integrated result, the PNS form number of the valid second trimester specimen must be used.

#### Who has access to the portal?

Those who have access are medical doctors, doctors of osteopathy, licensed midwives, and genetic counselors, all of whom are authenticated by their respective state licensing boards or governing bodies. Medical doctors and doctors of osteopathy also can delegate their access to support staff, such as nurses, medical assistants, office staff, ultrasonographers, etc. Certified Nurse Midwives (CNMs), nurse practitioners (NPs), and registered nurses (RNs) must be made delegates at this time, although GDSP is working with the Board of Registered Nursing to obtain direct license verification for nurses. Patients will not have access to the portal.

#### What security measures will be in place regarding access to the portal?

CDPH adheres to standard state agency security rules and practices for protecting personal health information. Multiple firewall layers and multi factor authentication are used to secure access, and all transactions are recorded and auditable.

#### Why won't coordinators just fax me results anymore?

In the past, coordinators have shared results as a courtesy. Case coordinator workload is expected to increase because of upcoming changes to the PNS Program. These demands will prevent coordinators from continuing with this courtesy service.

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