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Facts about Non-Invasive Prenatal Testing or Cell Free Fetal DNA

All pregnant women in California are offered testing for certain birth defects through the California Prenatal Screening Program. This program is considered the current standard of care in this state for women who want to be screened for chromosome problems and other birth defects during pregnancy. The information about who should be offered NIPT and which chromosome problems it detects is changing rapidly. Your health care provider or a genetic counselor would have more up-to-date and complete NIPT information.

What is NIPT or Cell Free Fetal DNA Testing?

Non-invasive prenatal testing (NIPT) or cell free fetal DNA is a newer prenatal blood test. NIPT can detect Down syndrome (trisomy 21), two other chromosome problems (trisomy 18 and trisomy 13), and sex chromosome abnormalities after 10 weeks of pregnancy. The test finds extra DNA pieces from the developing baby in a pregnant woman's blood. Extra DNA is a sign that the pregnancy has a much higher chance for certain birth defects.

If you decide to have the California Prenatal Screening Program test first, and the results are positive (a higher risk), NIPT is offered as an option and paid for by the State Program. If you qualify for NIPT testing (see When is NIPT Discussed?), and decide to skip the California Prenatal Screening test and have NIPT instead, your insurance will cover the cost based on your deductible plan.

How accurate is NIPT?

For pregnancies with a high risk for chromosome problems, NIPT finds almost all pregnancies with Down syndrome and trisomy 18, most pregnancies with trisomy 13, and many sex chromosome abnormalities. The ability of NIPT to correctly screen for these chromosome problems in women under 35 years of age is less than for women over 35.

Sometimes, the NIPT result is negative, but the baby does have Down syndrome, trisomy 18, or trisomy 13 or a sex chromosome abnormality. This is called a false-negative result. NIPT may also give a false-positive result, which means it may show that a healthy pregnancy is a high-risk pregnancy.

Because NIPT can show a false-positive or false-negative result, it is a screening test, not a diagnostic test. An amniocentesis or chorionic villus sampling (CVS) diagnostic procedure is always recommended as follow-up for a positive NIPT result.

When is NIPT Discussed?

You and your health care team can discuss whether NIPT is right for you. It is usually discussed for these situations:

- Mother will be 35 or older at delivery
- Some types of positive California Prenatal Screening Program test results
- Some types of ultrasound abnormalities
- Chromosome problems in a prior pregnancy or certain very specific family history
- Physician's suggestion because of other pregnancy-related complications

Important Things to Consider

Non-invasive prenatal testing:

- May give false-positive or false-negative results
- Does not test for all chromosome problems
- Does not test for other genetic conditions or other birth defects
- Requires more testing co confirm a diagnosis of Down syndrome, trisomy 18, trisomy 13, or sex chromosome abnormalities.
- Can be done after 10 weeks of pregnancy

If you need more information about NIPT, ask your physician for other resources.