Sutter Health
Sutter East Bay
Medical Foundation

Prenatal Testing for Chromosomal Abnormalities: An Overview

Deciding whether or not to have screening or testing for fetal chromosomal abnormalities during pregnancy and deciding which test is right for you can be overwhelming. We hope that the information provided here will help guide you through this decision process.

SCREENING TESTS: These tests use information such as maternal blood samples, and ultrasound to determine which pregnancies are at increased risk for fetal chromosomal abnormalities. These tests do not increase the risk of miscarriage. Available options for screening include:

CA Prenatal Screening Program (CA PSP) cdph.ca.gov/programs/PNS/pages/default.aspx (also known as the "state screen") offers three screening tests. The most accurate is **Sequential Integrated Screening test.** This test is actually three separate tests integrated to give you a result for fetal chromosomal risk. The three tests include a blood test at 10 to 13 weeks, a nuchal translucency (NT) ultrasound (measurement of the fetal neck thickness) between 11 and 14 weeks, and a second blood test at 15 to 20 weeks. The other two screening tests offered by CA PSP are 1) **Quad marker screening** (second trimester blood test only, usually done for women who missed the window for integrated screening); and 2) **serum integrated screening,** which includes blood tests in the first and second trimester, but no nuchal translucency measurement.

Non-invasive prenatal testing (NIPT) or Cell-Free DNA (cfDNA) test analyzes cell-free fetal DNA that is circulating in maternal blood. This is a relatively new option for prenatal screening that can detect trisomy 21 and trisomy 18 with 99 percent accuracy, but may not be as accurate for trisomy 13 or sex chromosome abnormalities. The NIPT/cfDNA blood test can be drawn as early as 10 weeks. Abnormal NIPT/cfDNA tests must be validated with diagnostic tests (see below).

We recommend that the NIPT/cfDNA test be used as a secondary test if initial results of the state screen are abnormal rather than as a primary test.

DIAGNOSTIC TESTS: These tests directly sample fetal cells (from placenta or amniotic fluid) to determine whether a pregnancy is affected by a chromosomal abnormality. These tests may increase the risk of miscarriage. They can be used for primary testing or for confirming an abnormal screening test. Diagnostic testing options include:

Chorionic villus sampling (CVS) is done between 10 and 14 weeks. This test includes an ultrasound and a sample of the placenta with a needle. Miscarriage risk is about 1 in 300.

Amniocentesis is done between 16 and 18 weeks. This test involves an ultrasound and a sampling of the amniotic fluid with a needle. Miscarriage risk is likely lower than that of the CVS, but it has been difficult to estimate.

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	Screening Tests		Diagnostic Tests	
	CA State Screen (Sequential Integrated Screening)	NIPT/cfDNA***	CVS	Amniocentesis
Timing of test	10-20 weeks	Over 10 weeks	10-14 weeks	16-20 weeks
Population	All women	35+ or abnormal serum test	Usually age 35+ or abnormal serum test	Usually age 35+ or abnormal serum test
Excluded populations	None	Multiple gestation Donor egg	None	None
How is test performed?	Maternal blood and ultrasound	Maternal blood	Placental tissue	Amniotic fluid sample
Miscarriage risk	None	None	1:300	1:300 to 1:1,500
Detection rates*	T 21: 80-95% T 18: 80-95% T 13: uncertain	T 21: >99% T 18: 99% T 13: 72-92%	T 21: >99% T 18: >99% T 13: >99%	T 21: >99% T 18: >99% T 13: >99%
False positive rate	3-5%	<1%	<1%	<1%
Other screening opportunities** Results available	ONTD	-some sex chromosome abnormalities -22q11.2 deletion syndrome	-sex chromosome abnormalities -other numerical chromosomal abnormalities	-ONTD -sex chromosome abnormalities -other numerical chromosomal abnormalities
Results available	10 days	7-10 days	8-14 days	8-14 days
How are results reported?	Age-related risk reported based on blood and NT results	Reported as positive or negative risk, may include a likelihood ratio of risk	Reported as a normal or abnormal karyotype	Reported as a normal or abnormal karyotype
How are results followed up if abnormal?	NIPT, CVS or amniocentesis	CVS or amniocentesis	None (this is final result)	None (this is final result)
Additional testing needed if results normal?	No	State screening recommended in tandem	ONTD (blood test at 16-18 weeks)	No

* T21=trisomy 21 (Down's syndrome), T18=trisomy 18 (Edward's syndrome), T13=trisomy 13 (Patau syndrome)
**ONTD=open neural tube defects such as spina bifida and anencephaly
*** Companies offering this test include:
Roche (Harmony), Counsyl (Informed Pregnancy Screen), and Natera (Panorama)

ACOG (American College of Obstetrician and Gynecologists) statement on NIPT:
NIPT/cfDNA using cell free fetal DNA offers potential as a screening tool. Cell free fetal DNA testing should be an informed patient choice after pretest counseling and should not be part of routine prenatal assessment. NIPT/cfDNA should not be offered to low-risk women or women with multiple gestations because it has not been sufficiently evaluated in these groups. A negative NIPT/cfDNA test result does not ensure an unaffected pregnancy. A patient with a positive test result should be referred for genetic counseling and should be offered invasive prenatal diagnosis for confirmation of test results.

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