Your Genetic Testing Options

These are optional tests that you can choose to do to get more information about your baby's risk for having certain medical conditions/complications

Maternal Carrier Testing	Non-invasive Screening	Invasive Diagnostic Testing	Fetal Sex Prediction Kits
Looks at *your own genetics* to determine if you are a carrier of certain conditions that can be passed on to your offspring. Tests for Cystic Fibrosis (CF) & Spinal Muscular Atrophy (SMA) Thalassemia and Sickle Cell Anemia One time test, does not need to be repeated each pregnancy If your test is Positive- the next step is having the biological father of the baby tested. If they are negative then the risk of your baby having these conditions is very low and no follow up is needed. If his test is positive as well you will be referred to a genetic counselor to discuss specific risks to your children. If your test is Negative- then your risk of low for passing these conditions on to your baby Get more info at: https://www.integratedgenetics.com/providers/tests/carrier-screening/inheritest	Screening tests that look for certain conditions in the fetus. *Non-invasive*- Done through blood draws from the mother's arm in our clinic. *Screening Test* (not diagnostic) - Not 100% accurate, abnormal results must be confirmed - Positive test means there is an increased chance of the baby having a condition - Negative test- the risk of the baby having these specific conditions are low, no follow up needed Several tests available- *see next page for more info* -Cell free DNA: analyses small fragments of the baby's DNA present in the mom's blood stream. Done by the Integrated Genetics company (Brand name MaterniT21 Plus). Might be costly, and in some situations only covered for women over 35 years old (check you cost at IntegratedGenetics.com) -California Prenatal Screening Program- compares hormone markers in the mother's blood in the first and second trimester. This test is also called the AFP test. It is run by the state, cost is subsidized and is generally more affordable and widely covered (about $225)	Comprehensive testing done out of the area by sampling either amniotic fluid or a placental tissue. *Extremely accurate* analysis for a wide variety of chromosomal abnormalities. Carries a small *risk of complications* including pain, miscarriage (about 1 in 500 pregnancies), infection and rupture of the membranes Two testing options include: - Chorionic Villus Sampling: done by obtaining a small amount of placental tissue through the cervix between 10-15 weeks - Amniocentesis: needle aspiration through the mother's abdomen between 15-24 weeks Tests are performed at the UCSF Perinatal Diagnostic Center (call for more information or for an appointment: (415) 476-4080	Test that can be done at home to predict the sex of the baby Can be done as early as 7 weeks 99% accurate Out of pocket cost (not covered by insurance) You collect a blood sample at home and mail it to the company. In some circumstances we can help you obtain the blood sample at our lab Two available brands: -Sneak Peek (sneakpeektest.com) -Peekaboo (DNACenter.com)

Non-Invasive Screening Options

	Cell Free Fetal DNA (One Blood Draw)	Sequential Screening (2 blood drawn + a extra ultrasound)	Serum Integrated Screening (2 blood draws)	Quad Marker Screening (1 blood draw)
What it tests for	Baby's risk for chromosomal abnormalities: -Trisomy 21 -Trisomy 18 -Trisomy 13 -Various sex chromosome aneuploidies - Various Deletion syndromes	-Trisomy 21 -Trisomy 18 - Neural tube defects -SLOS	-Trisomy 21 -Trisomy 18 - Neural tube defects -SLOS	- Neural tube defects - SLOS Can also test for Trisomy 21 and 18 if not previously done
How it tests for & When it's done	1 blood draw from mom's arm any time after 9 weeks gestation	Blood draw from mom's arm at our clinic between 10-13 weeks ---PLUS--- Nuchal ultrasound at 11-13 weeks at the hospital --PLUS— Another blood drawn at 15-20 weeks at our clinic	Blood draw from mom's arm at our clinic between 10-13 weeks ---PLUS --- A second blood draw from mom's arm at our clinic between 15-20 weeks	1 blood draw from mom's are at our clinic between 15-20 weeks
FYI	-Most accurate for women >35 -Can be combined with the Quad Marker Test to screen for spina bifida - Maybe costly if not covered by insurance. We recommend you check the MaterniT21 test website before having this test: **IntegratedGenetics.com** or 877-821-7266	This option includes the **Nuchal ultrasound**: a limited ultrasound done by a certified technician to measure the thickness of the back of the fetus' neck. An increased thickness may indicate an increased risk for Trisomy 21 or Trisomy 18. Done locally at St Joe's hospital and in Fortuna at Redwood Women's Heath, or out of the area	Slightly less accurate for detection of Trisomy 21 and 18 than the sequential screening testing (nuchal ultrasound increases accuracy of that test)	-Least accurate test for Trisomy 18 and 21 -Can be done as a standalone test or in combination with other screening

	Sequential Screening 2 blood draws and an extra ultrasound	Serum Integrated Screening 2 blood draws	Quad Marker Screening 1 blood draw	Prenatal Cell-free DNA Screening 1 blood draw	Diagnostic Testing (Chorionic Villus Sampling or Amniocentesis)	
How it is done	Blood draw from mother's arm at 10-14 weeks at our clinic -----PLUS----- Nuchal translucency ultrasound at 11 ½- 13 ½ weeks -----PLUS----- Another blood draw from mom's arm at 15-20 weeks at clinic	Blood draw from mother's arm at 10-14 weeks at our clinic -----PLUS----- Blood draw from mother's arm at 15-20 weeks at clinic	Blood draw from mother's arm between 15-20 weeks at our clinic	1 blood draw from mother's arm at our clinic after 10 weeks	Chorionic Villus Sampling: small amount of placental tissue obtained through the cervix between 10-15 weeks Amniocentesis: needle aspiration through mother's abdomen into the amniotic fluid between 15-24 weeks	
What it tests for	Down syndrome (90 in 100) Trisomy 18 (80 in 100) Anencephaly (97 in 100) Spina Bifida (80 in 100) Abd wall defects (85 in 100) SLOS (60 in 100)	Down syndrome (85 in 100) Trisomy 18 (80 in 100) Anencephaly (97 in 100) Spina Bifida (80 in 100) Abd wall defects (85 in 100) SLOS (60 in 100)	Down syndrome (80 in 100) Trisomy 18 (67 in 100) Anencephaly (97 in 100) Spina Bifida (80 in 100) Abd wall defects (85/100) SLOS (60 in 100)	Down Syndrome (99/ 100) Trisomy 18 (98 in 100) Trisomy 13 (98 in 100) Gender (99 in 100) Sex-linked conditions	Comprehensive analysis of numerous genetic and chromosomal abnormalities	
False Positive Rate (how many will get a positive test but not have an abnormality present)	1-5% depending on age	1-5% depending on age	1-5% depending on age	0.2-1 % depending on age	0% (100% accuracy)	
Considerations	**Nuchal translucency Ultrasound**: limited ultrasound done by a certified technician. Measures the fluid at the back of the baby's neck; babies with Down syndrome and Trisomy 18 often have an increased measurement. Offered in Eureka, Fortuna, Redding, Santa Rosa and the bay area	Slightly less accurate than the sequential screen (nuchal translucency ultrasound increased accuracy of screening for Down syndrome and Trisomy 18)	Least accurate test for down syndrome and trisomy 18	Most accurate for woman 35 or older Can be combined with the Quad marker to test for spina bifida and abdominal wall defects Can also be combined the nuchal translucency ultrasound, although this offers in increased accuracy for screening Maybe costly if not covered by your insurance provider	Invasive procedure that are performed at a hospital (out of area) Can cause miscarriage in 1 in 500 pregnancies Most comprehensive and accurate test available	