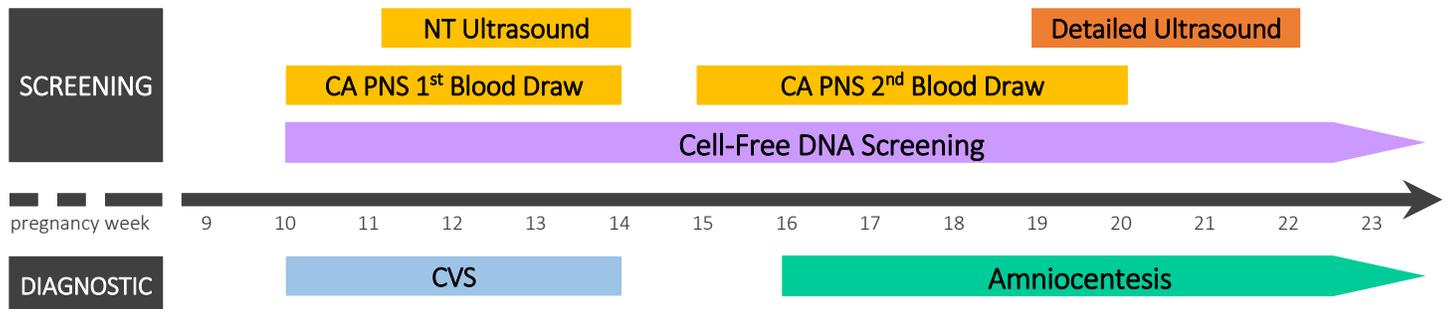


PRENATAL TESTING OPTIONS

There are many types of tests available during pregnancy. No test can detect every possible condition, however there are many tests that can provide more information. Patients may elect or decline any of these tests as they are all optional.



SCREENING TESTS

Screening tests provide probabilities (odds) of having a baby with certain conditions, however screening tests cannot detect all cases of these conditions. Screening tests are performed through maternal blood and ultrasound, so there is no risk for miscarriage from these tests. Patients will receive either “screen negative” or “screen positive” results from their screening tests. When results are screen negative, these are considered low-risk for that condition, however it is not impossible that the baby has that condition. When results are screen positive, these are considered high-risk for that condition, however most women with screen positive results are carrying a healthy baby! Follow up testing is available to confirm screening results.

■ California Prenatal Screening Program (CA PNS) (also called Sequential Integrated Screening)

- Combines blood draw(s) and the NT ultrasound with maternal age risk to screen for Down syndrome, trisomy 18, open neural tube defects (example: spina bifida), and Smith-Lemli-Opitz syndrome
- In a pregnant woman’s blood there are natural chemicals made by the pregnancy which may show different patterns or levels if a baby has certain conditions
- The nuchal translucency (NT) ultrasound measures the fluid in the back of the baby’s neck, which may be larger if a baby has certain conditions
- Preliminary risk assessment result is available after the 1st blood draw plus the NT ultrasound
- Final (more accurate) risk assessment is available after the 2nd blood draw is combined with the preliminary results

■ Cell-free DNA Screening (also called Non-invasive Prenatal Screening/Testing or NIPS/NIPT)

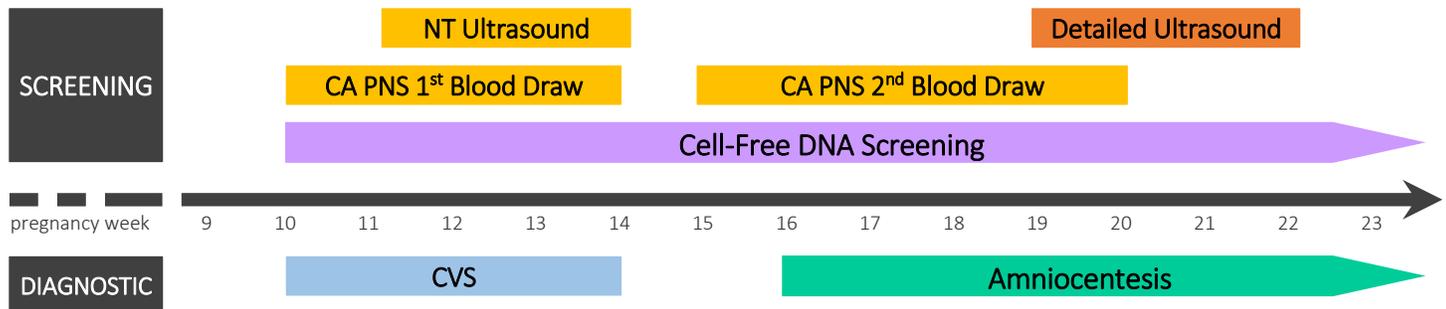
- Blood test measures the amount of DNA (maternal and placental) from chromosomes 13, 18, 21, X and Y
- Screens for Down syndrome, trisomy 18, trisomy 13, and missing or extra sex chromosomes
- May not be covered by insurance for low-risk pregnancies
 - PAMF orders this screening through a company called Counsyl, which is in-network with most insurance providers. Counsyl can perform an insurance check to provide an estimate of your out of pocket cost. You can call Counsyl directly to request this estimate at 888-COUNSYL (888-268-6795).
- Not recommended by PAMF for twin pregnancies at this time

■ Detailed Anatomy Ultrasound

- Screens for major structural birth defects, including open neural tube defects
- Can increase suspicion for conditions like Down syndrome however cannot provide a definite yes or no

PRENATAL TESTING OPTIONS

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DIAGNOSTIC TESTS

Diagnostic tests provide a definitive yes or no answer as to whether the baby has a known condition, and are the most accurate prenatal testing for chromosome disorders. They can test for types of conditions that would be missed by screening tests. Procedure related pregnancy loss is a rare occurrence at 1 in 1000 (0.1%).

■ CVS (chorionic villus sampling)

- Results obtained by analyzing a small sample of the placenta
- Performed either by using a catheter passing through the cervix, or by inserting a needle through the abdomen
- 1-2% risk for ambiguous (mosaic) result (amniocentesis recommended if this is found)

■ Amniocentesis

- Results obtained by analyzing a small sample of the amniotic fluid that surrounds the baby
- Performed by inserting a needle through the abdomen

When doing a diagnostic test, the chromosomes can be looked at in two different ways: A fetal **karyotype** looks at the number of chromosomes the baby has and looks for large structural rearrangements. A fetal **chromosomal microarray** looks for the number of chromosomes and for small missing or extra pieces of DNA (microdeletions or microduplications) which can cause birth defects and intellectual disabilities. Other testing can also be performed on the diagnostic testing sample as indicated (example: testing for a genetic condition when both parents are known carriers for the condition).

CONDITION	CHANCE FOR CONDITION	CHANCE TO <u>NOT</u> HAVE CONDITION
Down syndrome (chance changes with maternal age)	AGE 25 0.10% AGE 35 0.34%	AGE 25 99.90% AGE 35 99.66%
	AGE 30 0.14% AGE 40 1.15%	AGE 30 99.86% AGE 40 98.85%
Trisomy 18 (chance changes with maternal age)	AGE 25 0.02% AGE 35 0.09%	AGE 25 99.98% AGE 35 99.91%
	AGE 30 0.04% AGE 40 0.30%	AGE 30 99.96% AGE 40 99.70%
Microdeletions or microduplications	1-2%	98-99%
Open neural tube defects (example: spina bifida)	0.1%	99.9%
Overall chance for a birth defect or genetic condition	3-5%	95-97%