Sutter Health
Palo Alto Medical Foundation

Introduction to 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. Insurance companies provide various levels of coverage for the different options explained below, and out-of-pocket costs can range significantly. PAMF provides the CPT or billing codes so you can check for coverage and any share of cost with your insurance company.

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 blood draws and ultrasound, so there is no risk for miscarriage from these tests. We provide two options for screening between 10 weeks to 20 weeks gestational age.

EXPANDED CARRIER SCREENING

Expanded carrier screening is a blood test which looks to see if someone is a "carrier" for a genetic condition. The expanded carrier screening test currently offered by PAMF looks for approximately 175 genetic conditions, including Cystic Fibrosis, Tay-Sachs disease, common types of Thalassemia, Spinal Muscular Atrophy, and Fragile X syndrome.

DIAGNOSTIC TESTS

Diagnostic tests provide a definitive yes or no answer as to whether the baby has a known condition. They can test for types of conditions that would be missed by screening tests. Two diagnostic tests are performed through obtaining very small samples of the placenta or amniotic fluid.

SCREENING TESTS

Option 1: California Prenatal Screening Program (CA PNS)

Sequential Integrated Screening (3 steps) Combines blood draw(s) and the Nuchal Translucency ultrasound with a person's age-related risk to screen for Down syndrome, Trisomy 18, open neural tube defects (example: spina bifida), and Smith-Lemli-Opitz syndrome.

- First blood draw – between 10 weeks to 13 weeks 6 days of pregnancy
- Nuchal Translucency Ultrasound – between 11 weeks 2 days to 14 weeks 2 days of pregnancy
- Second blood draw – between 15 to 20 weeks of pregnancy

Serum integrated screening (2 steps) Two blood draws only

- First blood draw – between 10 weeks to 13 weeks 6 days of pregnancy
- Second blood draw – between 15 to 20 weeks of pregnancy

Note: NIPT and/or diagnostic testing is available (at no out of pocket cost) if there are abnormal results from the tests covered under the CA PNS.

Option 2: Cell-free DNA Screening (also called Non-invasive Prenatal Screening/Testing or NIPS/NIPT)

- Blood draw screens small pieces of DNA from the pregnant person and the placenta for Down syndrome, trisomy 18, trisomy 13, and missing or extra sex chromosomes
- PAMF orders this screening through a company called Myriad, which is in-network with most insurance providers.

DIAGNOSTIC TESTS

Diagnostic Test: 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)

Diagnostic Test: Amniocentesis

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

Note: while Diagnostic tests are invasive, the risk of a procedure-related pregnancy loss is a rare occurrence at 1 in 1000 (0.1%).

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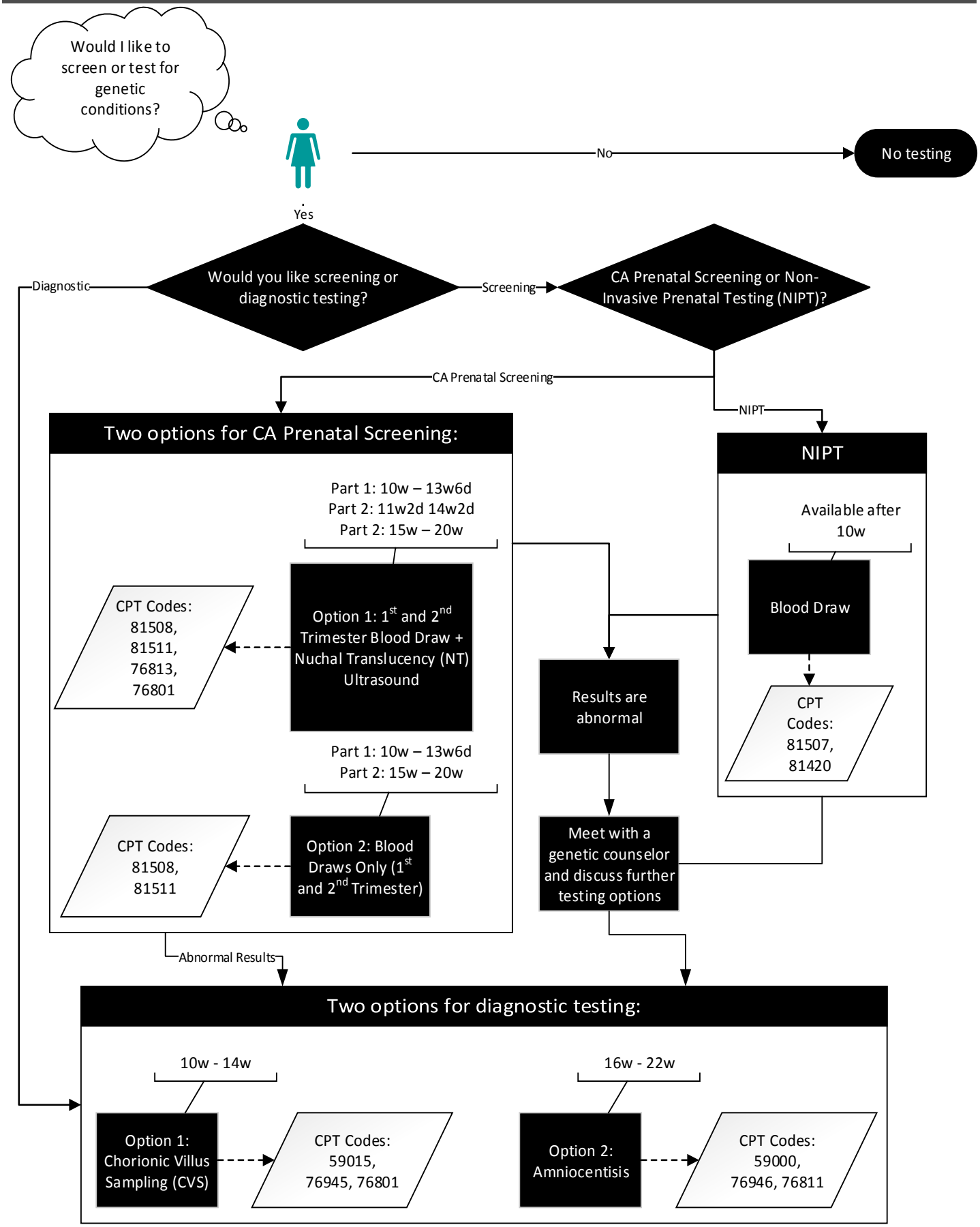

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