El Camino Women's Medical Group
Obstetrics, Gynecology, Infertility & Minimally Invasive Surgery

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Prenatal Genetic Screening

The purpose of genetic screening is to identify risk for birth defects. These defects are often caused by chromosomal abnormalities, in which there can be an absent or extra chromosome. Birth defects can affect a baby's appearance, body function, and development. **Screening tests** can assess the baby's risk of having Down syndrome and other chromosomal problems, as well as neural tube defects. If a screening test shows an increased risk of a birth defect, **diagnostic tests** may be done to determine if a specific birth defect is present.

Although screening tests are offered to all pregnant women, it is your choice to have them done. Knowing whether you baby is at risk of or has a birth defect beforehand allows you to prepare for having a child with a particular disorder and organize the medical care your child may need. At El Camino Women's Medical Group, our goal is to help you determine if your baby is at increased risk of birth defects, and to offer you the appropriate testing with which you are comfortable.

Routine screening:

Sequential Integrated Screening:
This combines results from a first trimester and second trimester screen. The overall detection rate for Down Syndrome is 94-96%.

First trimester screen: combines a 1st trimester blood test (10 weeks – 13 weeks 6 days) and ultrasound (Nuchal translucency, NT at 11-14 weeks). This is a special ultrasound performed by clinicians with special training. It measures the back of the baby's neck. 1st trimester screen alone has a detection rate of 82-87%.

Second trimester screen (Quad screen): blood test drawn at 15- 20 weeks. If the 2nd trimester screen is done alone (no first trimester screen) it has a detection rate of 81%.

Ultrasound:

With high quality ultrasound we are able to identify birth defects that my require attention before or after birth. The second trimester anatomy scan, typically done between 18-20 weeks, is a detailed ultrasound examining fetal anatomy. Views obtained include the fetal head and spine, chest, heart, abdomen, and urinary tract. Fetal gender is also evaluated.

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Non-invasive Prenatal Testing (NIPT):

Also known as cell-free DNA, fetal DNA can be isolated from the **mother's blood** as early as 10 weeks during pregnancy. With good accuracy it can test for Trisomy 21 (99%), Trisomy 18 (99%), Trisomy 13 (92%), and sex chromosomes (99%) to determine sex-chromosome related disorders and also to report gender. While results of this testing are highly accurate (99%), a negative test result does not ensure an unaffected pregnancy, and not all chromosomal abnormalities can be tested. Patients who receive a positive result will be referred for genetic counseling and diagnostic testing. Lab names for NIPT include MaterniT21, Counsyl, Panorama, and Harmony.

By 2015, NIPT became standard testing for all pregnancies in women over the age of 35. In 2016, NIPT has become nearly standard testing for all pregnancies. Most major insurers cover NIPT.

ECWMG routinely orders NIPT testing on all patients, it is the most accurate genetic screening test available.

We partner with Natera (the Panorama test) to make the financial aspects of this test as smooth as possible. When covered, if the cost of your test goes to your deductible, Panorama will arrange interest free payment plans as low as $25/mo when requested. If your insurance denies coverage, Natera will not charge you more than $150.

Diagnostic Tests (invasive):

Amniocentesis (15-20 weeks): A procedure in which a needle is used to withdraw and test a small amount of amniotic fluid and cells from the sac surrounding the fetus. There is a small chance of miscarriage (1 in every 400 procedures)

Chorionic villus sampling, CVS (10-14 weeks): A procedure in which a small sample of cells is collected from the placenta. There is a similar chance of miscarriage compared to amniocentesis when performed by an experienced provider.

Diagnostic tests are 100% accurate for the chromosomal tests they perform.