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

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.

Non-Invasive Perinatal 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 gender determination (99%). This state is ordered and run through the state of California.

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.

There are additional capabilities of NIPT, including screening for sex chromosome aneuploidies, triploidy, and microdeletions. ECWMG has always recommended testing for sex chromosome aneuploidies and triploidy, but not for microdeletions. These are all offered as add-on testing through private labs that run NIPT. Our physicians will talk to you about adding on sex chromosome aneuploidies and triploidy screening. This may add an up to an additional $99 to your testing costs. Though we do not recommend testing for microdeletions, that option remains open to all patients if requested.

Ultrasound:

With high quality ultrasound we are able to identify most birth defects that will require attention before or after birth. The second trimester anatomy scan, typically done between 18-22 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.

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.