**Genetic Screening and Testing**
Starting in the early stages of your pregnancy, you have the option to screen and test for certain birth defects in your baby, including the following:

- Down syndrome (trisomy 21)
- Trisomy 13
- Trisomy 18
- Neural tube defects

Decisions about genetic screening and prenatal testing options are personal in nature. Any additional questions can be directed your provider or genetic counselor.

**Screening Options:**
Screening tests are non-invasive and pose no immediate risk to you or your pregnancy. Screening tests determine whether or not you are at average risk or higher than expected risk for a given condition. Screening tests *cannot* provide a definitive answer about your baby having a specific disorder or birth defect.

**Non-invasive prenatal screening or the cell free fetal DNA screen** is available anytime after 10 weeks gestation. It requires a single maternal blood draw. This screen provides risk information on Down syndrome, trisomy 18, trisomy 13, and the sex chromosome disorders, such as Turner syndrome and Kleinfelter syndrome. This risk is determined through analysis of small fragments of fetal DNA released from placental cells. Compared to other screening tools, the cell free fetal DNA screen has a higher detection rate for these chromosome disorders with fewer false positives.

**Sequential screening** provides risk information for Down syndrome, trisomy 18 and open neural tube defects. It is a method of combining both First Trimester and Second Trimester screens, which are described below.

**First Trimester Screening** is a combination of maternal blood and fetal ultrasound, which is performed between 10 to 13 weeks gestation. It can provide a preliminary risk for Down syndrome and trisomy 18. Specific risk estimates are calculated by taking into account your age and the levels of two biochemical markers in your blood that are produced by the placenta. In the first trimester, an ultrasound measurement of the thin layer of fluid found at the back of the fetal neck (nuchal translucency) is also used to calculate risk.

**Second Trimester Screening** involves a blood draw, collected between 15 and 21 weeks. Second trimester risk levels are calculated by taking into account your age and the levels of four hormones produced by the pregnancy.

With the sequential screen, the first trimester and second trimester portions of the screen are combined in order to finalize risk information for Down syndrome, trisomy 18 and open neural tube defects in the second trimester. If either the first trimester or the second trimester portion of screening provides an increased risk estimate, further genetic counseling would be available to discuss additional screening or diagnostic testing options.

**Ultrasound** (sonogram) can be used to evaluate growth/development of your baby and look for problem in the pregnancy. Routinely a detailed ultrasound (level II) is preformed between 18 and 20 weeks of pregnancy, when the baby is large enough to do an anatomical review.

**Carrier screening** is available to help couples learn more about their risk of having a child with certain genetic conditions, like cystic fibrosis, Tay Sachs disease, or sickle cell anemia. Expanded carrier screening is available to determine risk for over 100 genetic conditions. In certain ethnic groups some genetic conditions are more common. However, many couples are at risk of having a child with a genetic disorder and there is no prior family history.

**Diagnostic Options:**
Diagnostic tests are offered to patients believed to be at elevated risk for certain birth defects and would benefit from more direct testing of the pregnancy. Diagnostic tests determine whether or not the condition you are testing for is present. They are invasive tests and have a small risk for miscarriage.

**Chorionic Villus Sampling (CVS)** is a diagnostic test which involves sampling cells from the placenta. It is performed between 10 and 13 weeks gestation. Using ultrasound guidance, a needle is placed through the abdomen into the placenta. Alternatively, a catheter is guided through the cervix to obtain a biopsy of the placenta. Final results are typically available 7 to 10 days.

**Amniocentesis** is a diagnostic test which analyzes the amniotic fluid surrounding the fetus. It is performed after 15 weeks gestation. Using ultrasound guidance, a needle is placed through the abdominal wall into the uterus and a small amount of amniotic fluid is removed. The fetal cells in the fluid can be analyzed for Down syndrome or other genetic diseases. It can also test for certain birth defects, like an open neural tube defect. Final results are typically available in 10 to 14 days.

**Insurance Coverage for Genetic Testing**
While insurance coverage for genetic testing and other genetic services is common and improving, some insurance policies do not cover certain testing. If you know which testing you are expecting to have, please contact your insurance company prior to your visit using the CPT codes below as a reference. The Division of Clinical Genetics cannot guarantee insurance coverage of any testing or procedure that is performed. Patients are financially responsible for any costs not paid for by insurance, in addition to applicable co-pays.

Amniocentesis – 88235, 88269, 88280, 88285

CVS – 88235, 88269, 88280, 88285

MaterniT21 screen - 81420

Sequential screen –
Part I: 84163, 84702, 76801, 76813
Part II: 82105, 82677,84702, 86336

Carrier screening- 81479, 81401, 81407, 81406,
81405, 81408, 81404, 81209, 81220, 81221, 81222,
81223, 81400, 81242, 81243, 81244, 81251, 81257,
81403, 81255, 81260,81330

The **non-invasive prenatal screen** offered through the Division of Clinical Genetics is the MaterniT21 PLUS screening. Billing for this screen is done by Sequenom, the laboratory that performs the testing. If you have questions about billing for the MaterniT21 PLUS, Sequenom can be contacted at 877-821-7266, option 1. You can contact them prior to your visit, if desired. Sequenom also has a calculator to estimate potential out of pocket cost, which can be found at https://www.sequenom.com/billing-and-insurance#cost-estimate

For carrier **screening** the billing is done by Counsyl, the laboratory that performs this testing. If you have questions about billing prior to your appointment, Counsyl can be contacted at 1-888-COUNSYL (268-6795) or by email at billing@counsyl.com. If you are interested, the lab can contact prior to your visit.