Genetic Carrier Screening Information

We offer a panel of genetic tests for the most common hereditary recessive diseases which could potentially affect your baby. At CWCC we follow the guidelines of The American College of Ob/GYN and the American College of Medical Genetics who recommend genetic testing for ALL pregnant patients or those planning pregnancy. These are new recommendations, and in doing these tests you are staying up to date with the latest benefits that medical science has to offer.

Recessive diseases require both parents to carry the affected gene so the disease will frequently skip generations and may appear in families with no known prior family history. There are 11 diseases tested in this panel, the most common being cystic fibrosis, spinal muscular atrophy, fragile X, and Tay-Sachs disease. The complete list is included in your patient information. It is important to do these tests early because babies with these diseases will be severely affected and in some cases the disease will not be compatible with life. Knowing about this early allows families to plan, seek appropriate care and support, and make informed decisions. Most importantly, finding out that you do NOT carry these genes adds enormous peace of mind and is certainly the most likely outcome of the test.

These diseases cannot be diagnosed by ultrasound or through any other non-invasive means prior to delivery since an affected baby will appear totally normal, sometimes even for months or years after birth.

All 11 genes can be tested from a single cheek swab sample and the results take about 2-3 weeks to be completed. If you have a positive test we will recommend testing your partner and then schedule you for a consultation with a genetic counselor if both you and your partner are positive. Since these genes do not change throughout your life the test only needs to be done once. Since it is a new test in 2017, we recommend testing even if this is not your first pregnancy. Thank you for entrusting us with your healthcare!