



EXPANDED CARRIER SCREEN CONSENT

Expanded carrier screening is a blood test of the parents that tests their genetic carrier status for some severe medical problems, which may affect their baby. A “carrier” has no symptoms of the disease, however if both parents are a carrier for the same disease, the child can be severely affected. This occurs in about 1:550 births, with 1:280 births affected by a less severe genetic condition caused by both parents being carriers. These inherited disorders account for 10% of all pediatric admissions to the hospital and 20% of infant mortality. An example of such a condition is spinal muscular atrophy, which is a neurodegenerative disorder that causes death in childhood. Other examples include cystic fibrosis, Duchene muscular dystrophy, Tay-sachs disease, acute infantile liver failure, severe combined immunodeficiency (SCID), and Leber congenital amaurosis. For some of these conditions, there is no known effective treatment. Some of these conditions can cause intellectual disability, congenital blindness or deafness. This test has no risk for your baby.

Expanded carrier screening through Sema4 (the company that performs the test) looks for 152 conditions known to cause severe disease or death. If one partner is found to be a carrier of a condition, the other parent needs to be tested. Results can take up to 2 weeks to return, so it makes sense to test both parents at the same time. This testing should be performed as early in pregnancy as possible, or prior to pregnancy. If an abnormality is found, a genetic counselor from Sema4 will call you to discuss the results. If you would like more information, a brochure is available at the front desk. We can also arrange for you to speak with a genetic counselor prior to testing.

Unfortunately, this test is not always covered by insurance because it is a new test. You should check with your insurance to see if this test is covered. If it is not covered, the out-of-pocket cost for you will be about 250\$. To determine if this test is covered by insurance, you may call Sema4 directly at 1 800 298 6470.

I consent to have Expanded Carrier Screening performed by Dr. Minior (or the covering doctor). All of my questions have been answered.

Signature: _____ Date: _____

Print name: _____