

Counsyl Genetic Testing

SRM has recommended that you or your partner undergo genetic testing using the Counsyl genetic screening tool. There are a number of serious genetic disorders for which you may be a carrier. A carrier is someone that carries a single abnormal recessive gene wherein if they have a child by a partner that carries the same recessive disorder their offspring can have a serious medical disorder.

Most of these disorders are serious metabolic diseases where an affected child is sick for life and usually does not have a normal lifespan. In other words, these disorders are serious and it would be very beneficial to avoid them. Your risk for each of the recessive genetic disorders as tested by Counsyl varies with you and your partner's ethnicity.

The seventeen recessive disorders that are included in the selected SRM Counsyl panel are relatively common. Several professional organizations such as the American College of Obstetrics and Gynecology (ACOG) and the American College of Medical Genetics (ACMG) have recommended routine screening of the pre-pregnant population for only these 17 disorders. The other 80+ disorders, as included on the full Counsyl panel, are rarer to the extent that the risk of you and your partner both testing positive is extremely low. Yet, if one of you tests positive for one of these rare disorders, we are obligated to also test the other partner which adds time and expense to the process.

The cost per test to you for screening for the 17 selected disorders or the full panel of over 100 disorders is the same. However, the cost in terms of needing to frequently run additional tests makes the full panel overall more expensive (the full panel is less cost effective).

SRM recommends that you be tested with the limited Counsyl panel (the 17 selected disorders). However, if you wish to be tested for the full 100+ disorders (full Counsyl panel) that is an option. Unless you specifically request the full panel, SRM will order the selected panel for you. Please feel free to ask questions if this explanation of the difference between the two panels (selected and full) is not clear.

The following tests are included in the selected Counsyl panel: Autosomal Recessive Polycystic Kidney Disease, Beta Thalassemia, Bloom Syndrome, Canavan Disease, **Cystic Fibrosis**, Familial Dysautonomia, Fanconi Anemia Type C, Gaucher Disease, Glycogen Storage Disease Type Ia, Mucopolidosis IV, Niemann-Pick Disease Type A, Sickle Cell Disease, **Spinal Muscular Atrophy**, Tay-Sachs Disease, Usher Syndrome Type 1F, Usher Syndrome Type 3.

Please visit www.counsyl.com or www.mytestingoptions.com for more information