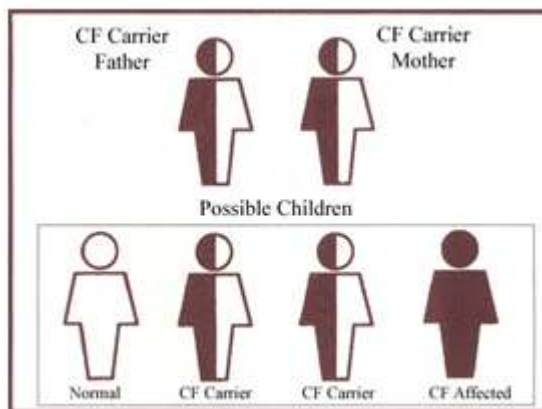


Preconception Genetic and Immunity Testing

The American College of Obstetrics and Gynecologists (ACOG) and the American College of Medical Genetics (ACMG) recommends testing for certain genetic diseases based on ethnicity, family history, or other known risk factors and testing for certain contagious diseases that can be harmful to a fetus, prior to pregnancy.

Genetic Disorders

Every normal person can carry genes that may give rise to a child with a genetic disorder if the child inherits a copy of the abnormal gene from both parents. These are called recessive genes and their prevalence varies among ethnic groups. When both parents are carriers of the same recessive disease, there is a 1 in 4 (25%) chance that the child will inherit the trait from both parents and have the disease, a 1 in 2 (50%) chance that the child will inherit the trait from only one parent and be a carrier, like the parents, or a 1 in 4 (25%) chance that the child will not inherit the trait from either parent and will not be a carrier or have the disease.



Carrier status for a recessive disease is passed silently from generation to generation. Carriers can only be identified by a specific genetic test.

There are a number of specific genetic disorders that could be passed on to a child. A parent can carry a gene for a disorder but not have the disease themselves; therefore they are completely healthy. If an individual carries a genetic defect, the resulting child has a certain risk of actually having the disease or of being a genetic carrier themselves. This risk to the child is high if both parents carry the same genetic defect. Most of these illnesses are substantial, generally associated with a shorter life span, and have serious ongoing medical problems. A good example of one of these diseases is cystic fibrosis. An individual with cystic fibrosis can have breathing and gastrointestinal problems throughout their life.

The risk for these genetic diseases is based on a person's ethnicity. For instance, Caucasians are more at risk for cystic fibrosis while Asians are more at risk for a serious anemia called thalassemia. People of Jewish ancestry are more at risk for a series of specific metabolic disorders. For individuals of Asian or African ethnicity a complete blood count (CBC) is recommended in addition to the genetic tests. While various laboratories offer genetic carrier testing, SRM recommends the Counsyl genetic screening tool because it is more cost effective.

The thirteen 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 13 disorders. The other 80+ disorders, as included on the full Counsyl panel, are more rare 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 13 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 13 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: Beta Thalassemia, Bloom Syndrome, Canavan Disease, Cystic Fibrosis, Familial Dysautonomia, Fanconi Anemia, Gaucher Disease, Hexosaminidase A Deficiency, Mucopolidosis, Niemann-Pick Disease, Sickle Cell Disease, Spinal Muscular Atrophy, Tay-Sachs Disease.

The Counsyl tests can be run on either saliva or a blood sample. The results are usually available in 12-14 days. Please visit www.counsyl.com or www.mytestingoptions.com for more information.

Contagious Infectious Diseases

There are certain contagious diseases that can be harmful to a fetus if a woman is exposed during pregnancy. These diseases can be avoided or be less harmful if the mother receives an immunization prior to pregnancy. It is recommended that all women wanting to become pregnant have their immunity to *rubella* and *varicella* tested. If she is found to be susceptible to either of these diseases, it is recommended that she have an immunization prior to pregnancy. It may be also recommended to wait 30 days after receiving an immunization before attempting conception.

Women are also encouraged to receive a single Tdap (*tetanus, diphtheria, and acellular pertussis*) booster once as an adult prior to pregnancy. Td vaccine (*tetanus and diphtheria*) is recommended for women who are already immune to pertussis and have had 10 or more years elapse since a previous Td booster.

In addition, an *influenza* (flu) vaccine is recommended prior to pregnancy.

Acknowledgement

Preconception genetic and immunity testing is **recommended** but not **required** prior to fertility therapy. The choice to proceed with some or all of the recommended testing is yours. Your signature acknowledges that you realize genetic testing and immunity screening has been recommended to you prior to initiating fertility therapy.

Female Patient

Partner (if applicable)

Date

Signature

Signature

Print Name

Print Name