



Genetic Carrier Screening of Patients

Why should I undergo genetic carrier screening?

The doctors and staff at Rocky Mountain Fertility Center do our best to ensure that you receive optimal care and attention to improve your chances of having a healthy pregnancy and child. An important part of family planning is being informed about your testing options. Genetic carrier screening is one test that may help you better understand the chance to have a child with a genetic disease. Carriers of a certain genetic diseases are usually healthy individuals; but when both parents are carriers of the same genetic disease, they can have a child with that disease. Most people do not know they are carriers until they have a child born with the disease. If both you and your partner are carriers for the same disease, your child typically has a 1 in 4 (25%) chance of having that disease.

What diseases would I be tested for if I elect to undergo genetic carrier screening?

Rocky Mountain Fertility Center recommends all patients undergo genetic carrier screening through a single blood or saliva test that screens for over 200 genetic diseases – many of which you probably have never heard of before – each with its own unique features. Some disorders impact intellectual ability, while others impact physical ability. Some decrease life span, while others impact daily life. Some have treatment options, like lifelong medications or restrictive diets. For a full list of genetic conditions that are tested with Nxgen expanded carrier screening, please go to Nxgen's website. Like any other carrier screening test, some carriers will not be detected – so this test can reduce, but not eliminate, the chance for a genetic disease. As the United States population is becoming more ethnically blended, the ability to make appropriate recommendations based on ancestry alone is reduced. Thus, we recommend an inclusive approach to genetic testing and elected to offer patients expanded carrier screening through Nxgen. You may elect to use a different genetic testing company, of your wish. You **must** notify Rocky Mountain Fertility Center if you wish to use another company. Given the large number of diseases included in expanded carrier screening, we estimate that nearly half of our patients will be found to be a carrier for at least one of the conditions for which we are testing. So, if you are currently pregnant, you may want to consider having both you and your partner to be tested at the same time. Since many disorders require that both members of the couple be carriers for there to be an increased risk to have an affected child, testing at the same time will provide results more quickly.

Why should I be tested if there is nothing, I can do to change my genes? What are the limitations of testing?

If you are found to carry a genetic disease, you have options. You may decide to have preimplantation genetic diagnosis (a pre-pregnancy process that significantly reduces the risk that a child will inherit the genetic disease) or undergo testing during pregnancy to make informed reproductive decisions. Some individuals consider adoption or opt to not have children. Even if you would not choose any of these options, you can use the information to prepare for the birth of a child with a genetic disorder (ie. have a specialist at delivery to help increase the likelihood the baby does survive). You will have the opportunity to speak with your physician or a genetic counselor about the medical options available to you. Genetic carrier screening has limitations. Results are available only for the disease(s) and the mutations tested in the panel. Furthermore, not all mutations for each disease are known – novel mutations continued to be discovered for many diseases (e.g. cystic fibrosis). For this and many other technical reasons you should be aware that no carrier screening test is 100% reliable. Furthermore, negative or “normal” test results do not “guarantee” pregnancy or a healthy baby but rather they estimate your potential risk for the diseases tested in your future children.

What is my financial obligation?

Genetic testing is performed and billed through an independent company. For patients with active health insurance: your test will be submitted directly to your insurance company. The independent lab will communicate with you an expected out of pocket cost prior to running your tests. You may opt for a Self/cash pay price rather than bill your insurance if it is more cost effective.

Isn't genetic testing expensive?

Genetic carrier screening is covered by most insurance plans, however, copays, co-insurance, and/or deductibles may vary by health plan. To determine what your out-of-pocket expenses may be, please contact the billing specialist at Nxgen.

____ **YES**, I am interested in testing to determine if I am a carrier for the diseases screened with the Nxgen testing

____ **YES**, I am interested in testing to determine if I am a carrier for the diseases screened with another genetic company of my choosing

____ **NO**, I am not interested. I understand that, by choosing not to be screened, RMFC cannot determine whether or not I am a carrier for genetic diseases. Also choose this option if you have previously been screened and do not desire rescreening with the Nxgen panel.

Patient Name(Print) _____ Patient Signature _____ Date _____