PsiGenex™

Informed Consent for Carrier Screening

Introduction

This Informed Consent Form is intended to provide you with information about Carrier screening. Carrier screening is a genetic test that is used to identify people who have a change, or variant, in one or more of their genes that can increase the risk of having a child with certain genetic disorders.

This form will review benefits, possible results, risks, and limitations of the genetic test that is being ordered by your healthcare provider. Further testing may be warranted for you or your reproductive partner to determine the risk of having a child with any of the genetic disorders tested for. If you agree to testing, you will need to sign this consent. You may wish to obtain genetic counseling prior to signing. Genetic counseling can be provided to you upon your request. Please read carefully and discuss any questions you may have with your healthcare provider before signing below.

Purpose of Testing

Certain genetic disorders are caused by changes, or variants, in a specific gene or genes. This test analyzes those genes in your test sample to determine if there are genetic changes that may increase your chance of being a carrier of a genetic disorder. Carriers do not typically show symptoms but are at risk of having a child who is affected with the disorder they carry. These results may also inform you about your own health and possible susceptibility of developing a genetic disease or medical condition.

Test Procedure

Your healthcare provider will obtain a blood or saliva sample and send it to PsiGenex (a Cairo Diagnostics Laboratory). The specific genes related to certain genetic diseases are analyzed using Next Generation Sequencing (NGS).

Test Results and Interpretation

Your test results should be explained in support with your personal and family health history, and other laboratory tests.

A Positive Result

A variant was identified in one or more of the genes analyzed that is associated with being a carrier of a certain genetic condition. A positive result may indicate that you are at increased risk of having a child affected with that genetic condition. It also may reveal that you are at increased risk of having genetic condition. The results should be interpreted in the context of your clinical findings, biochemical profile, and family history.

A Negative Result

No mutations were identified in any of the genes that were analyzed using the test method specified. This result significantly reduces the likelihood that you have a variant in the genes that were analyzed. It does not, however, eliminate your risk entirely. Also, it does not rule out variants that are not analyzed by this test or guarantee the birth of a healthy child.

Benefits of the Test

The results of your test will be sent to your ordering physician, may be sent to a third-party genetic counseling company if you wish to receive genetic counseling services, and will become part of your medical record. Your genetic test results may help you and your healthcare provider make more informed decisions about your personal and reproductive health. Results may also have clinical or reproductive implications for other family members. If you are found to carry a variant in any of the genes analyzed, you may want to consider sharing that information with your family members as they may also be at risk of having the same gene variant.

Risks of the Test

Genetic testing is performed on DNA obtained from a blood or saliva sample. Side effects from having blood drawn are uncommon, but may include dizziness, fainting, soreness, bleeding, bruising and rarely, infection. Saliva collection is not invasive. Genetic testing may cause you to discover sensitive information about your health or disease risks, including disease risks other than the one you are testing for, or for diseases that currently have no treatment.

The US Genetic Information Discrimination Act-GINA of 2008 prohibits discrimination based on genetic information in regard to health insurance and employment. The results of genetic testing are considered Protected Health Information, PHI, as described in the Health Insurance Portability and Accountability Act, HIPPA of 1996 (Public Law 104.191). Federal legislation prohibits unauthorized disclosure of confidential personal information.

Limitations of the Test

This test only analyzes specific genetic changes that are associated with an increased risk for developing certain genetic conditions (please refer to gene table included with your report). Genetic testing provides a risk assessment only for those genes being analyzed. While this testing is highly accurate, rare testing errors may occur. Accurate results may not be obtained for reasons including but not limited to sample mix-up, bone marrow transplant, recent blood transfusion, or technical problems. PsiGenex will consult with your healthcare provider for “incidental findings.” when there are accepted medical interventions available. PsiGenex will only report these findings if necessary, to provide correct test results.

I have read (or have had read to me) all of the above and have had the opportunity to ask questions I might have about the testing, procedure, risks, and alternatives before consenting. My signature below acknowledges my consent to having this testing performed.

Signature of Patient or Authorized Representative

Relationship to Patient (If representative)

Date / /