Carrier Testing Informed Consent Overview

Notice to Health Care Provider

This document is a consent form for Praxis Genomics Optical Genome Mapping and Illumina Whole Genome Sequencing Carrier Testing. If you are ordering a test for diagnostic purposes based on a symptom already manifesting, please use the consent form specific for diagnostic tests found at www.praxisgenomics.com/forms. Currently, the laboratory will only accept requests for carrier testing after the individual(s) to be tested have received genetic counseling from a healthcare provider with experience in counseling patients for such a test. Please explain this consent to the individual(s) for whom the test is ordered and obtain their informed consent. Alternatively, contact Praxis Genomics for consenting services available by appointment booked through our website at www.praxisgenomics.com.

Note: Be aware of state laws applicable to carrier testing and the effects that discovery of certain disease associated variants might have on health or life insurance policies of your patients.

Couple Carrier Testing Informed Consent

Because results of carrier testing have important implications for the individual(s) tested as well as their families, we recommend that the consenting and ordering process be performed with the assistance of a genetic counselor and/or the ordering physician.

About the test:
Praxis Genomics offers the most comprehensive carrier testing currently available anywhere in the world. We use Optical Genome Mapping (OGM) and/or Whole Genome Sequencing (WGS) to analyze the customers' genetic composition at a previously unavailable resolution. These tests are designed to detect rare, clan specific variants that are not screened for and cannot be identified by other currently available carrier testing methods. OGM focuses on detecting structural variants in the genome, such as deletions, insertions, inversions, and translocations that may have pathogenic associations. Whole Genome Sequencing
(WGS) is primarily used for detection of small variants, such as single nucleotide variants (SNVs) or small insertions and deletions (indels) less than 50bp long. WGS can also detect repeat expansions associated with several neurological conditions and is useful for confirmation of structural variants detected by OGM.

Sample requirements:
Both OGM and WGS are performed on 3 ml of blood each.

Who should get tested?
The test is offered in an "individual" and a "couple" testing version.
The "individual test" reports all variants that would lead to severe chronic or life-threatening conditions during the person's lifetime or could have adverse effects on his or her children with partners carrying similar changes.
The "couple test" will test both partners planning to have children together and will only report variants that are present in both individuals and could affect their child.

What kind of results are reported?
1. In a positive report we will list variants that are known pathogenic or are deemed likely pathogenic based on well-established scientific evidence (ACMG) that are present in both partners and would cause a severe medical condition in the couple's offspring in the homozygous or compound heterozygous form. This means that there would be a 1 in 4 chance that their child would be affected.
2. In a negative report we will state the absence of such variants.

What implications do positive and negative results have?
When a known or predicted disease causing variant is detected, the test result is highly accurate. A positive result will help your clinician to prepare for, and potentially prevent the birth of an affected child. This could be done through preimplantation testing of the embryo. A negative result does not guarantee the absence of carrier status for all disease-causing mutations, only those that based on current medical and scientific knowledge can be categorized as such.

Are there limitations to carrier testing?
There is a possibility that genetic variants that cause a condition are not identified by testing either because of the technical limitations of the assay or because of incomplete understanding of the significance of variants detected.

Are there results that will not be reported?
Variants in genes that affect susceptibility to a condition, but do not definitively cause it, will not be reported.

Who will have access to the results?
Test results are maintained electronically by the laboratory. The results are provided to the ordering physician and/or health care facility that ordered the test. Results may also be made available to individuals/organizations with a legal right of access under applicable Federal and/or State law, or as authorized by the patient or the patient's representative. Patient privacy is of utmost concern to us, and we adhere to HIPAA privacy and security requirements.

How long are carrier testing results kept in the testing lab?
The laboratory may keep the identified carrier testing raw data in the lab indefinitely. This helps us improve our diagnostic capabilities. To advance the understanding of genetic disorders, your results might be analyzed and published in scientific articles in a de-identified manner consistent with HIPAA guidelines.

Carrier Testing Couple Informed Consent

What will happen to my DNA sample?
No additional tests will be performed on these samples without specific signed authorization by the individual(s) tested. However, the samples will be kept indefinitely for possible additional testing unless you instruct us to destroy the samples. In this case the samples will be destroyed within 60 days from the test order date.

What are the risks of testing?
1. Carrier testing may identify carrier status for serious and/or untreatable genetic conditions. It can result in unexpected psychological trauma, both for you and your family. The detection of such a condition or conditions could also affect the health or health care needs of your siblings, children, or other close relatives.
2. Although carrier testing is highly accurate, the interpretation of the report is based on current medical knowledge which is incomplete. This may result in failure to predict the manifestation or severity of certain conditions that are not yet described or not well characterized.
3. Genetic non-discrimination law prevents insurance companies from using your genetic information to deny health insurance coverage, but the law does not cover life insurance, disability insurance or long- term care insurance. Please be aware of any applicable State laws and applicable terms of any active insurance policies.

Circle Yes/No. Leaving these lines blank is interpreted as agreement.

Partner 1		Partner 2		
Y / N	_____ Initial	**Y / N**	_____ Initial	**I agree that my sample may be kept beyond 60 days for future testing or research participation. I understand that my sample may be used for quality control purposes. I understand that there is no guarantee of availability for my sample beyond 60 days.**
Y / N	_____ Initial	**Y / N**	_____ Initial	**I agree that my carrier testing data may be retained indefinitely to help improve Praxis Genomics' diagnostic capabilities.**
Y / N	_____ Initial	**Y / N**	_____ Initial	**I agree that my carrier testing data may be used for scientific publications in a de-identified manner.**

Consent for Couples Carrier Testing

All of the above has been explained to me to my satisfaction. My signature below confirms this. I understand this testing is voluntary, and I have had the opportunity to ask questions about alternative testing. I authorize my Health Care Provider to have access to my results.

_____ Partner 1 Signature

_____ Partner 1 Name

_____ Date of Birth

_____ Signature Date

_____ Partner 2 Signature

_____ Partner 2 Printed Name

_____ Date of Birth

_____ Signature Date

I have provided genetic counseling and have explained the risks, benefits and limitations of carrier testing to the individual

_____ Provider Signature

_____ Provider Printed Name

_____ NPI#

_____ Signature Date