Ambry Genetics®

Patient Consent for Carrier Screening Test - Page 1 of 2

Genetic Testing Introduction:
Everyone is a carrier for several genetic conditions but may not know their risk, especially if there are no affected family members. Screening for genetic conditions is complex. Pre- and post-test genetic education is recommended for all individuals undergoing a genetic screening test.

Purpose:
Reproductive carrier screening is a test to determine if you are a carrier of a pathogenic or likely pathogenic variant that could result in an increased risk to have a child with a genetic condition. This test will involve analysis of all genes included on the carrier screening panel that was ordered by your healthcare provider. Ambry reports only pathogenic or likely pathogenic variants and does not report all DNA variants present, including benign variants or variants of unknown clinical significance.

Specimen:
The blood or saliva specimen submitted is required for isolation and purification of DNA for molecular genetic testing. This specimen will be used for the purposes of determining whether there is a pathogenic or likely pathogenic variant present which may increase the risk of having a child with a genetic condition. In some cases, this genetic test may require an additional blood or saliva sample to obtain accurate results.

Ambry Genetics' Results Disclosure Policy:
Due to the complexity of genetic testing for carrier screening and the important implications of the test results, these results will be reported through your ordering provider. Your results report may be available to you after it has been released to you by your healthcare provider, upon your request, or as required to comply with local, state and/or federal regulations and laws. You should contact your provider to obtain and discuss the results of the test and additional testing and/or family planning options. Additionally, the test results could be released to all who, by law, may have access to such data.

See Ambry's Privacy Policy for more details: https://www.ambrygen.com/legal/notice-of-privacy-practices.

Genetic Testing Results and Risk Information:
Genes included on this test may be associated with multiple types of genetic conditions. Your healthcare provider's recommendations for additional testing and/or family planning options could differ depending upon the test results. Identification of a pathogenic or likely pathogenic variant in any gene does not imply that additional testing and/or family planning options will be covered by health insurance. Depending on the test results, there may not be enough information available to determine your precise risk. Further diagnostic or confirmatory testing may be needed in the future at the discretion of your healthcare provider as more information and data becomes available or if there are changes to your personal or family history.

Most of the genes on the carrier screening panels have been implicated in genetic conditions which are inherited in a recessive manner. If you are identified to carry a pathogenic or likely pathogenic variant in one of these genes, and your reproductive partner also carries a pathogenic or likely pathogenic variant in the same gene, there is a 25% chance of a child being affected by a recessive condition. The overall risk of having a child affected with one of these conditions is low. Further testing of you or your reproductive partner may be recommended based on the results of this test. If a patient is found to be a carrier of an X-linked condition, each child is at an increased risk for the condition regardless of your reproductive partner's carrier status.

Genetic test results have implications for your blood relatives. If you are found to carry a pathogenic or likely pathogenic variant in any of the genes analyzed, your family members may also have a risk to carry the same variant identified in you. This should be discussed with your healthcare provider.

There are several types of genetic test results, including:

- **Positive, pathogenic variant/s detected:** A pathogenic or likely pathogenic variant was identified in one or more genes associated with a genetic condition. This may be indicative that you are at increased risk of having a child with a genetic condition. The specific condition(s) depend on the gene(s) involved. Your healthcare provider will make recommendations for additional testing and/or family planning options based on what is known about the gene(s) in which a pathogenic or likely pathogenic variant was found.

- **Positive, two pathogenic variants detected in the same gene:** Two pathogenic or likely pathogenic variants were identified in a gene associated with a genetic condition. The two variants may be in the same copy of the gene, or one of the variants may reside in one copy of the gene and the other variant may reside in the other copy of the gene. The configuration of the variants affects the reproductive risk calculation. If the variants are in opposite copies of the gene, it is possible that you could have clinical symptoms of the disorder or could be at risk to develop symptoms of the disorder. It is usually necessary to test additional family members in order to establish the configuration of variants.

- **Possible Carrier:** A variant or variant(s) were identified in a gene associated with a genetic condition; however it is unclear if the result is indicative that you are at increased risk of having a child with a genetic condition. The configuration of the specific variant(s) affects the reproductive risk calculation and cannot be determined from this screening test alone. Your healthcare provider will make recommendations for additional testing/and or family planning options based on what is known about the gene(s) and/or variant(s) involved.

- **Negative:** No pathogenic or likely pathogenic variants were identified in any of the genes tested. This may be indicative of a reduced likelihood that you have a pathogenic or likely pathogenic variant in the genes tested (see limitations of testing). A negative result reduces but does not completely eliminate your risk to be a carrier for the tested condition(s). Your healthcare provider will make additional testing and/or family planning recommendations based on your personal and/or family history.

Genetic Discrimination:
There are federal laws in place that prohibit health insurers and employers from discriminating based on genetic information (for example, the Genetic Information Nondiscrimination Act (GINA) of 2008 (Public Law 110-233)). There are currently no federal laws that prohibit life insurance, long term care, or disability insurance companies from discriminating based on genetic information. Your state may have more comprehensive laws in this area. The results of genetic testing are considered protected health information and are confidential to the extent allowed by state and federal law. Release of test results is limited to authorized personnel, such as the ordering physician, and to other parties as required by law.

Technical Testing Limitations:
While this test is designed to identify most detectable pathogenic variants in the genes analyzed, there are pathogenic variants that this testing technology is unable to detect. The detection rate varies by gene and ethnicity. Some ethnicities or subpopulations may have an increased frequency for a specific pathogenic variant that cannot be detected by this assay. The chance that you have a pathogenic variant that was not identified by this carrier screening panel is called your "residual risk". In addition, there are other genes associated with carrier status for genetic conditions that are not included on this panel or that are not known at this time.

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The test is not for prenatal diagnostic purposes. This test is only meant to determine pathogenic variant carrier status for you. If you are determined to be at high risk of having an affected child, then gene specific prenatal diagnostic testing may be considered.

Standard Laboratory Limitations:
Inaccurate results may occur as a result of (but not limited to) the following reasons: sample mix-up, inaccurate or misleading medical information about your clinical condition or that of your family members, or technical problems. Due to limitations in technology and incomplete knowledge of genes, some changes in DNA or protein products that cause disease may not be detected by the test. There is a possibility that the result findings will be uninterpretable or of unknown significance. In rare circumstances, results may be suggestive of a condition different than that which was originally considered for purpose of consenting to this testing.

Ambry Reserves the Right to:
- Contact you to share additional genetic testing or clinical trial opportunities that may help understand your result.
- Retain your deidentified sample.
- Utilize the deidentified sample for laboratory quality control or for laboratory test validation optimization purposes.
- Utilize the deidentified sample for research to improve genetic testing for all patients and contribute to scientific research.

Patient Acknowledgement:
- I understand that genetic testing is voluntary, and I may choose not to have my sample tested.
- I have read and I understand the information provided in this consent, and all my questions have been answered.
- I acknowledge that the information provided by me on the test requisition form (TRF) is true and correct.
- I have had the opportunity to ask questions about the purpose of testing, about the test procedure, the test results, the risks, the limitations to testing, and my rights prior to me signing this informed consent.

Terms and Conditions:
Patient Acknowledgement: I acknowledge that the information provided by me is true and correct. For direct insurance billing: I authorize my insurance benefits to be paid directly to Ambry Genetics Corporation (Ambry), authorize Ambry to release medical information concerning my testing to my insurer, to be my designated representative for purposes of appealing any denial of benefits as needed and to request additional medical records for this purpose. I understand that I am financially responsible for any amounts not covered by my insurer and responsible for sending Ambry money received from my health insurance company.

For patient payment by credit card: I hereby authorize Ambry Genetics Corporation to bill my credit card as indicated above. In order to expedite consideration for eligibility for Ambry's Patient Assistance Program, please provide the total annual gross household income: $_____ and the number of family members in the household supported by the listed income: _____. I authorize Ambry Genetics Corporation to verify the above information for the sole purpose of assessing financial need, including the right to seek supporting documentation.

For NY Residents: I understand that under New York State law, Ambry Genetics must discard my sample after the longer of (a) testing completion and (b) 60 days after the Date of Collection above.

☐ I agree that Ambry Genetics may retain my sample for 6 months after the completion of all testing, including the testing above, any additional testing of my sample that I authorize within the initial 6-month retention period, and any extended or additional testing of the sample necessary and required to demonstrate the integrity of the sample tested or to resolve the analysis of a test with a previously indeterminate result.

☐ I do not agree that Ambry Genetics may retain my sample for 6 months after the testing above has been completed.

My signature below acknowledges my voluntary participation in this genetic screening test and such genetic analysis in no way guarantees my health, the health of an unborn child, or the health of other family members.

_____ _____

Patient Signature (or Parent/Guardian if patient is a minor) Date

_____ _____

Patient Name (Print) Name/Relationship (if patient is a minor)

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