Ambry Genetics®

Patient Consent for Molecular Genetic Testing

Test Purpose: The purpose of this molecular genetic test is to ascertain if I am, my child is, or my unborn child is [please circle appropriate] carrying mutation(s) predisposing to or causing the specific disease or condition:_____ .

A supplemental disease description sheet is available from Ambry Genetics.

Test Method: The blood, body fluid, or tissue specimen submitted is required for isolation and purification of DNA for molecular genetic testing. The test will cover all disorders requested on the Ambry Genetics requisition form.

Test Results: I understand that due to the complexity of DNA based testing and the important implications of the test results, these results will be reported only through the patient's designated physician(s) or genetic counselor (where allowed) and that I must contact my provider to obtain the results of the test. The test results, in addition, could be released to all who, by law, may have access to such data.

I understand that if results of the molecular genetics tests are positive, I may be a carrier of, predisposed to, or have the specific disease or condition tested for and I may want to consider further independent testing, consult with my physician, or pursue genetic counseling. I understand that if results of the molecular genetics tests are negative, I may not be a carrier of, predisposed to, or have the specific disease or condition tested for and I may want to consider further independent testing, consult with my physician, or pursue genetic counseling. I understand the limitations of these results: the test results could be based upon probabilities, and may not provide a 100% definitive conclusion to either genetic disease predisposition or manifestations. I understand that the molecular genetic test may not generate results and that an additional blood, body fluid, or tissue sample may be needed to obtain accurate results. I understand that the molecular genetic test may not generate accurate results for the following reasons: sample mix-up, samples unavailable from critical family members, maternal contamination of prenatal samples, inaccurate reporting of family relationships, or technical problems, but not limited to these. In rare circumstances, a clinically significant finding may be identified in your sample following initial testing as a result of improvements to current technology or discovery of a diagnostic error. Should this occur, your healthcare provider will be re-contacted and provided with the additional results.

Ambry's Rights: Ambry reserves the right to: 1) suggest additional molecular testing if it would help in resolving the patient's clinical genotyping, 2) report additional testing results (other than requested) if they are clinically relevant to the patients and their families (e.g. The methodologies for evaluating specific gene(s) of interest may rarely identify incidental findings related or unrelated to the reason I/my child have been offered testing. In such instances, these results will be discussed with my healthcare provider and additional testing may be recommended.), and 3) refuse testing if one of the conditions in the Patient Consent form is not met.

Use of Specimens: Ambry Genetics is committed to research efforts with the goal of improving testing for future patients. Your sample or test results could be used in the validation new genetic testing methods and/or other test-related research and education efforts. All research testing is anonymized and you will not receive results of any research testing done on your sample. You have the option to opt-out of research testing use on this form or on the test requisition form. After testing is completed, I understand that my blood, body fluid or tissue specimens may be disposed of or retained indefinitely by Ambry Genetics for these purposes, as long as my privacy is maintained. I understand that no compensation will be given nor will funds be forthcoming due to any invention(s) resulting from research and development using the specimens submitted. I understand that I may refuse to submit my specimen for use in this way and may withdraw my consent at anytime by contacting the medical director. I understand that my refusal to consent to medical research will not affect my results. Indicate consent or denial below.

_____ Initials I consent to the use of my sample for research. ☐ YES ☐ NO

I have read or have had read to me all of the above statements and understand the information regarding molecular genetics testing and have had the opportunity to ask questions I might have about the testing, the procedure, the risks, and the alternatives prior to my informed consent. I agree to have the molecular genetic testing described within or above.

_____ _____

Patient (or authorized individual) Signature Date

_____ _____

Patient Name *(please print)* Authorized Individual Name and Relationship *(please print)*