

Patient Consent

Pulmonary Arterial Hypertension Diagnostic Test

Test Purpose: I desire that I or **my child** (please circle appropriate) _____ have molecular genetic testing to ascertain if I/they carry mutation(s) in one of the genes thought to be responsible for development of pulmonary arterial hypertension (PAH). This testing is being pursued because I/my child have (has) symptoms and/or clinical testing which according to my physician suggests a diagnosis of PAH. 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.

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 research studies have shown this gene testing may reveal disease-causing mutations in approximately 70% of individuals with familial PAH and 11-40% of non-familial cases. Other, as yet to be identified genes may also be involved in the pathogenesis of PAH. Therefore, the results of the molecular genetics test may be one of the following:

Positive Testing revealed a mutation that is either clearly deleterious to gene function, or has been reported in the medical literature to be disease causing.

No abnormality Testing failed to find any significant abnormality in the genes. This does not preclude the possibility that a mutation does exist in this gene, which was not identified by the method used. Neither does it indicate that the clinical diagnosis of PAH is incorrect.

Novel variant Testing revealed a change in one of the genes tested. However, it is not known whether this change causes decreased function of the gene leading to disease. It is possible that, as new information becomes available, certain novel variants will be determined to be either disease causing, or a benign normal variant.

I understand that the results of this testing could have implications to other members of my family as to their risk of developing PAH. Familial PAH is believed to be incompletely penetrant, meaning that not all individuals who carry a deleterious mutation will develop the disease. Development of disease may also depend on additional factors. My family members or I may wish to seek genetic counseling and/or further discussions with my physician with regard to these implications.

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.

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, and 3) refuse testing if one of the conditions in the Patient Consent form is not met.

Use of Specimens: After testing is completed, I understand that my blood, body fluid or tissue specimens may be disposed of or retained indefinitely for research, test validation, and/or education by Ambry Genetics, 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. If a box is not marked consent is implied.

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

Financial Responsibility: I understand that if test cancellations are received prior to test set-up, processing will be honored at no charge. I understand that when requests for test cancellation are received after set-up, a cancellation report will be generated and a set-up fee will be charged. Once testing is initiated cancellation is not possible. I understand that I am responsible for all charges for testing and will be contacted for payment in the event my health plan does not reimburse for the test or Ambry Genetics does not receive a response from my health plan in a reasonable length of time.

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.

Patient Signature: _____ **Date:** _____

Patient Name (please print): _____