Ambry Genetics®

Patient Consent
Idiopathic Pulmonary Fibrosis Carrier Test

Test Purpose: I __________________ desire to have molecular genetic testing to ascertain if I carry the mutation in one of the genes thought to be responsible for development idiopathic pulmonary fibrosis (IPF) which was identified previously in a member of my family. 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 early research studies have shown gene testing may reveal disease-causing mutations in individuals with familial IPF. Such a mutation has been identified in a member of my family who carries the diagnosis of IPF. The results of the molecular genetics test may be one of the following:

Positive Testing revealed the mutation that was found previously in my family. This mutation is either clearly deleterious to gene function, or has been reported in the medical literature to be disease causing.

No abnormality Testing did not identify the mutation found previously in my family.

I understand that Familial IPF 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 as yet unidentified genetic and environmental factors as well as known risk factors such as cigarette smoking. Therefore a positive result on testing does not insure that I will develop IPF. Likewise a negative result, though reassuring, does not unconditionally guarantee that I will not develop IPF.

I understand that there are psychological risks of this testing. A negative result could generate feelings of guilt as well as joy, or relief. A positive test could lead to feelings of sadness, depression, despair or futility. Due to the potential psychological stress related to this testing I have identified individual(s) to whom I can look for support and I have informed them of my intent to undergo testing.

I confirm that I have been counseled by my physician/genetic counselor_________________________ (name) with regard to implications of either a positive or negative genetic test, and the impact that such a test may have on my family and myself. I have had the chance to ask questions and had them answered.

I understand that there is a potential risk for insurance discrimination. Test results are confidential and will be released only to the ordering physician or genetic counselor, unless I request that insurance be billed for my test. My insurance company may require test results for reimbursement purposes.

I understand the accuracy of these results depends on the accuracy of relationship and mutation information provided with this testing request. 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, 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):____

Physician / Genetic Counselor: I confirm that I have explained IPF molecular genetic testing, its limitations and implications to the above patient and answered all their questions.

Physician / Genetic Counselor Signature: ______________________________ **Date:** ____________________

Physician / Genetic Counselor Name (please print):_______________________________________