PATIENT INFORMED CONSENT FOR GENETIC TESTING FOR CYSTIC FIBROSIS

Illumina MiSeqDx™ Cystic Fibrosis Clinical Sequencing Assay

You should review the information provided below and discuss any questions or concerns with your healthcare provider or genetic counselor before signing this form. Genetic counseling should be offered prior to signing informed consent and will be available after the test has been completed in order to facilitate the understanding of the clinical significance of the findings. In addition, further testing or additional physician consultation may be warranted.

Disease Details: Cystic Fibrosis (CF) is one of the most common inherited genetic diseases. It is a multisystem disease that can affect the lungs, pancreas, gastrointestinal track as well as the reproductive system. CF has been reported to affect 70,000 people worldwide, approximately 30,000 people in the United States and one in 2,500 Caucasian newborns has CF. The incidence of CF in other ethnic groups varies from 1:8000 in Hispanics, 1:15,000 in African Americans, to 1:32,000 in Asian Americans.

Test Details: the Illumina MiSeqDx™ Cystic Fibrosis Clinical Sequencing Assay is an FDA-cleared next generation sequencing based assay that re-sequences the protein coding regions and intron/exon boundaries of the cystic fibrosis transmembrane conductance regulator (CFTR) gene using genomic DNA isolated from human peripheral blood specimens. This test can detect single nucleotide variants and small insertions and deletions of up to 3bp in size across 27 exons of the CFTR gene, as well as two deep intronic mutations, two large deletions, and the PolyTG/PolyT region. This test can be used to aid in the diagnosis of individuals with suspected CF and it is more appropriate for atypical or non-classic CF presentations or for patients that have a negative result from CF disease-causing variant panels.

I hereby attest that the principle, benefits, risks, results and limitations of this test have been explained to by a genetic counselor or healthcare professional, and that I understand that:

1. There is a small risk associated with the collection of the peripheral blood specimens such as hematomas or infections during blood collection.
2. I can request genetic counseling before signing this form and I may ask questions about the collection, testing or reporting process.
3. My specimen, both peripheral blood and extracted DNA will be stored at the laboratory for maximum 60 days and destroyed after that, unless I (parent/or legal guardian) agrees with the usage and storage of the specimen and/or extracted DNA for medical research, test validation, control material or education, as long as patient privacy and confidentiality are maintained. A separate signature is required for this consent.
4. That the U.S. Food and Drug Administration (FDA) has approved this test to aim in the diagnosis and carrier status for CF.
5. A positive result may diagnose the condition, indicate status as a carrier of the condition, and/or disclose a risk that a family member may develop or be a carrier of this condition. Genetics counseling and other recommendations will be provided.
6. A negative result only applies to the regions that were re-sequenced in the CFTR gene by this test and does not exclude the possibility of the presence of a rare variant in the CFTR intronic regions or large deletions not interrogated by this test. Therefore, a “wild-type” result does not guarantee that other CFTR variants are not present in the specimen analyzed.
7. Clinical misdiagnosis may occur due to sample misidentification, inaccurate family history, presence of the variant in a small fraction of cells not detectable by this technology (mosaicism).
8. I understand the significance of the possible test results based on family history based on the healthcare professional explanation and that further consultation is available.
9. Results will be released only to the ordering physician. Results will be released to a third party only if permission has been submitted in writing by the patient.

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Patient Authorization

My signature on this document constitutes my authorization to collect and test samples for the above-designated genetic test and my acknowledgement that I understand the purpose of this test and have the opportunity to clarify all my questions or concerns with my healthcare provider or genetic counselor. This signature also authorizes the laboratory to use the information on this form for reimbursement purposes and that I am responsible for any amount not covered by the insurance. The signature of the mother, or other legally authorized individual, provides authorization to collect and test samples from a child.

Patient's Name_____ Date_____/_____/_____

Patient's Signature_____

Parent/Guardian's Signature if patient is a minor_____

I (Patient Name) consent to the use of my sample for research purposes as long as my privacy and confidentiality are maintained: YES ☐ NO ☐

Physician Authorization (genetic counselor or healthcare provider administering the form)

My signature on this document, authorizes the laboratory to perform the above-designated genetic test and attest that I have discuss the purpose of this test with the patient and/or parent/guardian.

Physician's Name_____ Date_____/_____/_____

Physician's Signature_____

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