

PATIENT INFORMED CONSENT FOR GENETIC TESTING FOR CYSTIC FIBROSIS

Luminex Molecular Diagnostics xTAG® Cystic Fibrosis (CFTR) 39 kit v2 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 Luminex Molecular Diagnostics xTAG® Cystic Fibrosis (CFTR) 39 kit v2 Assay is an FDA-cleared multiplex PCR/multiplex allele specific primer extension (ASPE) based assay that enables the detection of 39 clinically relevant CF disease-causing variants of the cystic fibrosis transmembrane conductance regulator (CFTR) gene using genomic DNA isolated from human peripheral blood specimens. This test includes the 23 variants recommended by the American College of Medical Genetics and Genomics (ACMG) and by the American College of Obstetricians and Gynecologists (ACOG), 4 polymorphisms [5T/7T/9T, F508C, I507V, I506V], and the 16 world's most common and North American-prevalent variants. This test can be used for carrier screening in adults of reproductive age, as an aid in newborn screening, and in confirmatory diagnostic testing in newborns and children.

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 32 variants analyzed and does not exclude the possibility of the presence of a variant in the CFTR gene not interrogated by this test. The CF carrier risk after a negative result will be provided in the report.
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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