WMC Health | Westchester Medical Center

Westchester Medical Center Health Network

INFORMED CONSENT FOR CYSTIC FIBROSIS MUTATION TESTING

IF NO LABEL, PRINT PATIENT'S NAME, MR NO., GENDER, ROOM, DOB

1. I (or the person for whom I am signing) am consenting to genetic (DNA-based) tests for specific mutations in the cystic fibrosis transmembrane receptor (CFTR) gene, using methods that can distinguish different DNA sequences.
2. The following has been explained to me:
   Cystic fibrosis is one of the most common life-threatening genetic disorders. It has a disease incidence estimated at one of 2,000 to 4,000 live births in the US population. The disease appears when an individual inherits two mutated copies of the CFTR gene. Currently more than 2,000 mutations in the CFTR gene have been reported; approximately 200 of these are determined to be cystic fibrosis causing mutations.
3. I understand that the purpose of this analysis is for screening of cystic fibrosis carrier in adults of reproductive age, for confirmatory diagnosis of cystic fibrosis in newborns and children, and as an initial test to aid in the diagnosis of individuals with suspected cystic fibrosis. I have been advised that I (or the person for whom I am signing) may want to obtain professional genetic counseling prior to signing this informed consent. I have obtained that counseling or have decided, after a reasonable opportunity to do so, to forgo genetic counseling.
4. I understand that I (or the person for whom I am signing) am free to refuse this test without penalty or loss of benefits to which I am otherwise entitled, and that the consequences of denying this test may include, but are not limited to, unknown knowledge of the risk of having an affected child, or delayed diagnosis and access timely medical service for the best outcome in individuals with the disease.
5. I understand that a positive result is an indication that I (or the person for whom I am signing) may be predisposed to or have the specific disease or condition tested for and may want to consider further independent testing, consult the physician or pursue genetic counseling.
6. The condition I am consenting to being tested for is a subset of known variants causing cystic fibrosis. I understand that this test does not include all variants identified in the CFTR gene. Therefore, the failure to identify a variant does not guarantee that other CFTR variants are not present in the samples being analyzed. If no mutation is found, I (or the person for whom I am signing) may still develop cystic fibrosis due to mutations in other genes, other genetic factors, and on environmental conditions.
7. I understand that the results of the above test become a part of my (or the person for whom I am signing) medical record, and may be made available to individuals/organizations with legal access to the medical record, including, but not limited to the physicians and nursing staff directly involved in my (or the person from whom I am signing) care, my (or the person for whom I am signing) current and future insurance carriers for the purpose of claims administration, and others specifically authorized by me or the patient/authorized representative to gain access to the medical records.
8. No tests other than that authorized shall be performed on the biological sample and that the original blood sample shall be destroyed at the end of the testing process or not more than sixty (60) days after the sample was taken. The DNA will be retained for up to 6 months. In some circumstances, a patient's DNA may be used anonymously as a quality control sample in future testing, but, in this circumstance, all identifiers will be removed and the DNA sample and results obtained will remain anonymous.

   ☐ I understand and agree that my DNA remaining after testing may be stored for up to 6 months should additional testing be required. *Please check the box*.

I hereby give my consent to perform genetic testing of cystic fibrosis. A phlebotomist will take 1 vial of blood for this test.

Name of Person Obtaining Consent: _____ **Title:** _____

Signature: _____ **Date:** _____

☐ **YES:** I have read and fully understood the above, and give my consent for testing.

☐ **NO:** I DECLINE the above described test. I understand and accept the consequences of this decision.

Patient (parent or legally authorized representative) name: _____

Relationship: _____ **Signature:** _____ **Date:** _____

Witnessed by: _____