

MOLECULAR PATHOLOGY BRIEFS

MOLECULAR TEST NAME: **CYSTIC FIBROSIS (CF) CARRIER TESTING**

LABORATORY LOCATION: Molecular Pathology Laboratory

LABORATORY DIRECTOR: Felicitas L. Lacbawan, MD, FCAP, FACMG

ACCOMPANYING DOCUMENT/FORM:

Patient Information Sheet on "Carrier Testing for Cystic Fibrosis"
Informed Consent/Decline for Cystic Fibrosis (CF) Carrier Testing

PRE-TEST PREREQUISITES:

Genetic counseling and proper documentation of patient's test consent and proper test ordering on Cerner.

POST-TEST REQUIREMENT:

Genetic counseling after receipt of test results and further testing or additional physician consults, if warranted.

TARGET POPULATION:

Available to all pregnant women and when appropriate, the biological father and other family members.

TEST INDICATION:

Determination of CF carrier status of one of the 39 most common mutations in the *CFTR* gene, which includes the panel of 23 disease-causing mutations recommended by the American College of Medical Genetics-American College of Obstetricians and Gynecologists. This means that not all mutations in the *CFTR* gene are being detected.

TEST DETECTION RATE:

The test for CF offered at University Hospital Brooklyn checks for the 39 most common *CFTR* gene mutations and 4 variants. It detects about 94% of *CFTR* gene mutations in Ashkenazi Jewish, 91% in North American Caucasians, 74% in Hispanic Americans, 68% in African American population, and 49% in Asian Americans.

SPECIMEN TYPE:

Whole blood in lavender top (EDTA) tube, 5 mL, room temperature.

TEST METHODOLOGY:

This test involves examining the genetic material in a blood sample of at least 5 ml using the FDA-approved xTAG Cystic Fibrosis mutation detection method by Luminex 200 xMAP. This **qualitative** test uses a liquid bead array to provide simple and high throughput mutation analysis of the *CFTR* gene after multiplex PCR amplification using allele-specific primer extension. This detects the 39 mutations in the test panel simultaneously.

TEST ORDERING PROCEDURE:

Following the general procedure for ordering laboratory test, under the "Orderable", enter "CF Carrier Screen" then Enter. All required fields must be filled in properly before submission. If entry is incomplete, no specimen can be collected from the patient.

TEST TURN-AROUND TIME:

Results are made available in 2 to 3 weeks after specimen submission.

FOR ADDITIONAL TEST INFORMATION:

Contact the Molecular Pathology Laboratory: 718-270-1837, or ext 8623, or ext 2648

Visit the website: <http://www.downstate.edu/pnp/lab/molecular%20diagnostics.html>