NTD | Eurofins Clinical Diagnostics

Patient Information — Final Report

PATIENT NAME: [redacted]
DATE OF BIRTH: [redacted]
CROSS REFERENCE #: [redacted]
REFERRING DIAGNOSIS: [redacted]
PATIENT ID: [redacted]
LABORATORY #: [redacted]

TYPE OF SPECIMEN: [redacted]
DATE COLLECTED: [redacted]
DATE RECEIVED: [redacted]
FINAL REPORT: [redacted]

REFERRING PHYSICIAN: [redacted]
ADDRESS: [redacted]

TELEPHONE: [redacted]
FAX NUMBER: [redacted]
REFERRING PRACTICE: [redacted]

TELEPHONE: [redacted]
FAX NUMBER: [redacted]

Cystic Fibrosis: *CFTR* Common Mutation Panel

Results: One mutation detected. One copy of the delta F508 mutation was detected in the *CFTR* gene of this individual. A second mutation was not detected.

Interpretation

A sample from this individual was referred to our laboratory for targeted molecular analysis of 39 *CFTR* gene mutations associated with cystic fibrosis. CF is an autosomal recessive chronic condition involving multiple organ systems. Classical CF primarily involves the respiratory and digestive systems, and may have a range of clinical severity. Mutations in the *CFTR* gene cause CF. Two mutations within the *CFTR* gene, one inherited from each parent, are required to cause the disease.

One copy of the *CFTR* mutation delta F508 was detected in this individual. A second mutation was not identified. The detection rates in various racial/ethnic groups for this assay are listed below. This result does not rule out a diagnosis of cystic fibrosis and must be interpreted in the context of this patient's clinical presentation. Genetic counseling is recommended.

Sequence analysis was requested for this sample if both mutations were not identified by common mutation panel analysis. The sequencing of the *CFTR* gene will be initiated for this patient. For more information, please visit eglgenetics.com or call 855-831-7447 (toll free) to contact a laboratory genetic counselor or consult with a laboratory director. Genetic counseling is recommended.

Ethnic group	Detection rate	% of CF patients with at least one mutation
Ashkenazi Jewish	94%	99.6%
Non-Hispanic Caucasian	90%	99.0%
Hispanic American	74%	93.2%
African American	68%	89.8%
Asian American	49%	74.0%

Methodology: This panel of 39 pathogenic variants in the CFTR gene includes the recommended ACMG panel of 23 common pathogenic variants. These variants were screened for using the Luminex xTAG® Cystic Fibrosis 39 Kit v2 after multiplex polymerase chain reaction (PCR).

Note: All risk figures include approximations based on population prevalence studies. Direct detection of CF mutations is highly accurate. Possible diagnostic errors include sample mix-ups, genotyping errors, rare genetic variants that interfere with analysis, and other sources. Pursuant to the requirements of CLIA '88, this test was developed and its performance validated by EGL Genetic Diagnostics LLC. It has not been cleared or approved by the U.S. Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary. This test is used for clinical purposes.

Medical Consultant — Laboratory Director, Molecular Genetics

This case has been reviewed and electronically signed by a Laboratory Director.