HerediT™ CF Lab Report

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Final Report

Ordering Provider:	**Doe, John, MD**	Patient:	**Sample, Jane**
Provider Location:	**Grand Rapids**	DOB:	**09/13/1970**
Provider Phone:	**555-555-5555**	Patient Ethnicity:	**Other**
Provider Fax:	**555-555-5556**	Patient ID:	**12345-01234**
Date Ordered:	**1/28/2015**	Specimen:	**1035600024**
Date Collected:	**1/29/2015**	Referral Clinician:	**Smith, Jane, GC**
Date Received:	**1/30/2015**	Lab Director:	**Thomas J. Monroe, PhD**
Order ID:	**ORD12345-01234**	Date Reported:	**2/10/2015 2:00 PM PT**

Cystic Fibrosis Carrier Screen Lab Report

Test Result	
Negative	Negative for the mutations and variants analyzed. Residual Risk: 1 in 344

About the Test

The HerediT Cystic Fibrosis (CF) Carrier Screen test is a laboratory-developed test (LDT) that analyzes 136 mutations and 5 variants. The test interrogates phenotypically relevant CF mutations that were selected from the Johns Hopkins CFTR2 database (http://www.cftr2.org). CF causing mutations were identified by collecting data from North American and European patient registries.

Interpretation

This patient is negative for all mutations and variants analyzed. Residual risk is based on reported ethnicity and personal/family CF history. For questions about this result, genetic consultation and clinical correlation are recommended.[1]

Test Method

A [blood specimen][buccal swab] was collected from the patient and genomic DNA was purified and subjected to polymerase chain reaction-based amplification. The cystic fibrosis transmembrane regulator (CFTR) gene was interrogated for 136 mutations and 5 variants. Primer extension products were analyzed using matrix-assisted laser desorption/ionization mass spectrometry, and the specific genotypes assigned. The CFTR gene mutation panel analyzed in this test may be viewed online at www.Sequenom.com/laboratories/HerediTCFpanel.

Performance

The performance characteristics of the HerediT CF test have been analytically validated using CFTR gene mutations shown to be phenotypically relevant.[2]

Additional Perspective

DNA studies do not provide a definitive genetic risk in all individuals. While results of this testing are highly accurate, infrequent errors may be due to unusual DNA sequences in the DNA analyzed, or other causes. A negative HerediT CF test result does not remove all risk of having a child with CF because undetected rare mutations may influence the risk of developing disease.

Note

This test was developed and its performance characteristics determined by Sequenom Laboratories. It has not been cleared or approved by the U.S. Food and Drug Administration (FDA). FDA does not require this test to go through premarket FDA review. This test is used for clinical purposes. It should not be regarded as investigational or for research. This laboratory is certified under the Clinical Laboratory Improvement Amendments (CLIA) as qualified to perform high complexity clinical laboratory testing and accredited by the College of American Pathologists.

References

1. Update on Carrier Screening for Cystic Fibrosis, ACOG Committee Opinion No. 486. American College of Obstetricians and Gynecologists. *Obstet Gynecol* 2011;117(4):1028-1031.
2. Clinical and Functional Translation of CFTR. US CF Foundation, Johns Hopkins University, The Hospital for Sick Children Web site. http://www.CFTR2.org. Accessed Oct 4, 2012.

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[2/10/2015]