Integrated GENETICS
LabCorp Specialty Testing Group

Sample Report

Cystic Fibrosis Mutation Analysis

Patient Name:
Referring Physician: Ordering Doctor, MD
Specimen #: 12345678
Patient ID: 12345678-9

Client #: 123456
Case #: 1234567

Any Hospital
100 Main Street
Anytown, ST 12345
USA

DOB: /1978
Sex: F
SSN:

Date Collected: 05/28/2014
Date Received: 05/29/2014
LAB ID:
Hospital ID:
Specimen Type: **BLDPER**

Ethnicity: Caucasian

Indication: Carrier Test / No family history

RESULTS: Negative for the 97 mutations analyzed

INTERPRETATION:

This individual is negative for the mutations analyzed. This result reduces but does not eliminate the risk to be a CF carrier. See Comments for ethnic-specific risk reductions based on a negative family history.

COMMENTS:

Mutations Detection Rates among Ethnic Groups Detection Rates are based on mutation frequencies in patients affected with cystic fibrosis. Among individuals with an atypical or mild presentation (e.g. congential absence of the vas deferens, pancreatitis) detection rates may vary from those provided here.

Ethnicity	Carrier risk reduction when no family history	Detection rate	References
African American	1/61 to 1/316	81%	ACOG Committee Opinion 486 PMID: 21422883; Heim PMID: 11388756
Ashkenazi Jewish	1/24 to 1/767	97%	ACOG Committee Opinion 486 PMID: 21422883
Asian American	1/94 to <1/183	49-55%	ACOG Committee Opinion 486 PMID: 21422883; Watson PMID: 1384328
Caucasian	1/25 to 1/343	93%	ACOG Committee Opinion 486 PMID: 21422883; Heim PMID: 11388756; Palomaki PMID: 11882786
Hispanic	1/58 to 1/260	78%	ACOG Committee Opinion 486 PMID: 21422883; Heim PMID: 11388756; California Database: (http://www.cdph.ca.gov/programs/GDSP/Documents/CFTabelCurrent.pdf)
Jewish, non-Ashkenazi		Varies by country of origin	Orgad PMID: 11336401; Kerem PMID:10464623
Mixed or Other		Not Provided	For counseling, consider using the ethnic background with the most conservative risk estimates.

This interpretation is based on the clinical and family relationship information provided and the current understanding of the molecular genetics of this condition.

METHOD / LIMITATIONS:

CFTR gene regions are amplified enzymatically. The 97 CF mutations are tested by multiplex allele-specific primer extension, bead array hybridization, and fluorescence detection. The test discriminates between p.F508del and three polymorphisms (p.I506V, p.I507V and p.F508C). Numbering and nomenclature follow Human Genome Variation Society recommendations. Mutations and their legacy names are listed at www.integratedgenetics.com/CFplus. The DNA reference sequence is NG_016465.1. False positive or negative results may occur for reasons that include genetic variants, blood transfusions, bone marrow transplantation, erroneous representation of family relationships, or maternal contamination of a fetal sample.

Electronically Signed By: _____, PhD, FACMG on 06/02/2014

MUTATIONS ANALYZED			
c.54-5940_273+10250del21kb	c.1973_1985del13insAGAAA	p.R117H	p.R553*
c.262_263delTT	c.1976delA	p.Y122*	p.A559T
c.273+1G>A	c.2012delT	p.G178R	p.R560T
c.273+3A>C	c.2051_2052delAAinsG	p.L206W	p.P574H
c.274-1G>A	c.2052delA	p.F312del	p.R709*
c.313delA	c.2052dupA	p.G330*	p.K710*
c.325_327delTATinsG	c.2175dupA	p.R334W	p.R764*
c.442delA	c.2657+5G>A	p.T338I	p.Q890*
c.489+1G>T	c.2737_2738insG	p.R347H	p.R1066C
c.531delT	c.2988G>A	p.R347P	p.W1089*
c.579+1G>T	c.2988+1G>A	p.R352Q	p.Y1092*
c.579+5G>A	c.3039delC	p.[Q359K;T360K]	p.M1101K
c.580-1G>T	c.3067_3072delATAGTG	p.S364P	p.D1152H
c.803delA	c.3528delC	p.A455E	p.R1158*
c.805_806delAT	c.3536_3539delCCAA	p.G480C	p.R1162*
c.948delT	c.3659delC	p.Q493*	p.S1196*
c.1155_1156dupTA	c.3717+12191C>T	p.I507del	p.W1204*
c.1545_1546delTA	c.3744delA	p.F508del	p.Q1238*
c.1585-1G>A	c.3773dupT	p.V520F	p.S1251N
c.1680-1G>A	c.3889dupT	p.C524*	p.S1255*
c.1766+1G>A	p.E60*	p.G542*	p.W1282*
c.1766+5G>T	p.R75*	p.S549N	p.N1303K
c.1820_1903del84	p.G85E	p.S549R	
c.1911delG	p.E92*	p.G551D	
c.1923_1931del9insA	p.R117C	p.Q552*	

This test was developed and its performance characteristics determined by Esoterix Genetic Laboratories, LLC. It has not been cleared or approved by the Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary.