HerediT

Phenotypically relevant mutations.
Nothing more, nothing less.

CYSTIC FIBROSIS CARRIER SCREENING TEST

The large mutation panel contained in the HerediT carrier screening panel for CF was selected from the Johns Hopkins CFTR2 database (http://cftr2.org).

Legacy name	New nomenclature (nucleotide)
1078delT	c.948delT
1154insTC	c.1022_1023insTC
1213delT	c.1081delT
1248+1G->A	c.1116+1G>A
1259insA	c.1127_1128insA
1341+1G->A	c.1209+1G>A
1461ins4	c.1329_1330insAGAT
1525-1G->A	c.1393-1G>A
1548delG	c.1418delG
1677delTA	c.1545_1546delTA
1717-1G->A	c.1585-1G>A
1811+1c.6kbA->G	c.1679+1c.6kbA>G
1812-1G->A	c.1680-1G>A
1898+1G->A	c.1766+1G>A
2143delT	c.2012delT
2183AA->G	c.2051_2052delAAinsG
2184delA	c.2052delA
2184insA	c.2052_2053insA
2307insA	c.2175_2176insA
2347delG	c.2215delG
2585delT	c.2453delT
2622+1G->A	c.2490+1G>A
2711delT	c.2583delT
2789+5G->A	c.2657+5G>A
3007delG	c.2875delG
3120+1G->A	c.2988G>A
3121-1G->A	c.2989-1G>A
3199del6	c.3067_3072delATAGTG
3272-26A->G	c.3140-26A>G
3659delC	c.3528delC
3791delC	c.3659delC
3849+10kbC->T	c.3717+12191C>T
3876delA	c.3744delA
3905insT	c.3773_3774insT
394delTT	c.262_263delTT
4005+1G->A	c.3873+1G>A

Legacy name	New nomenclature (nucleotide)
4016insT	c.3884_3885insT
405+1G->A	c.273+1G>A
406-1G->A	c.274-1G>A
4209TGTT->AA	c.4077_4080delTGT-TinsAA
4382delA	c.4251delA
457TAT->G	c.325_327delTATinsG
574delA	c.442delA
621+1G->T	c.489+1G>T
663delT	c.531delT
711+1G->T	c.579+1G>T
712-1G->T	c.580-1G>T
852del22	c.720_741delAGGGAGAAT-GATGATGAAGTAC
A455E	c.1364C>A
A559T	c.1675G>A
CFTRdele2,3	c.54-5940_273+10250del21kb
CFTR-dele22,23	c.3964-78_4242+577del
D110H	c.328G>C
D1152H	c.3454G>C
D1270N	c.3808G>A
D579G	c.1736A>G
DF508 (F508del)	c.1521_1523delCTT
DI507 (I507del)	c.1519_1521delATC
E1104X	c.3310G>T
E585X	c.1753G>T
E60X	c.178G>T
E822X	c.2464G>T
E831X	c.2491G>T
E92K	c.274G>A
E92X	c.274G>T
F1052V	c.3154T>G
G1069R	c.3205G>A
G1244E	c.3731G>A
G178R	c.532G>A
G330X	c.988G>T
G542X	c.1624G>T
G551D	c.1652G>A

Legacy name	New nomenclature (nucleotide)
G85E	c.254G>A
G970R	c.2908G>C
I1234V	c.3700A>G
I336K	c.1007T>A
K710X	c.2128A>T
L1065P	c.3194T>C
L1077P	c.3230T>C
L206W	c.617T>G
L732X	c.2195T>G
L927P	c.2780T>C
M1101K	c.3302T>A
M1V	c.1A>G
N1303K	c.3909C>G
P67L	c.200C>T
Q1313X	c.3937C>T
Q220X	c.658C>T
Q39X	c.115C>T
Q493X	c.1477C>T
Q525X	c.1573C>T
Q552X	c.1654C>T
Q890X	c.2668C>T
Q98X	c.292C>T
R1066C	c.3196C>T
R1066H	c.3197G>A
R1070Q	c.3209G>A
R1070W	c.3208C>T
R1158X	c.3472C>T
R1162X	c.3484C>T
R117C	c.349C>T
R117H	c.350G>A
R334W	c.1000C>T
R347H	c.1040G>A
R347P	c.1040G>C
R352Q	c.1055G>A
R553X	c.1657C>T
R560K	c.1679G>A

Legacy name	New nomenclature (nucleotide)
R560T	c.1679G>C
R709X	c.2125C>T
R74W	c.220C>T
R75X	c.223C>T
R764X	c.2290C>T
R851X	c.2551C>T
S1196X	c.3587C>G
S1251N	c.3752G>A
S466X	c.1397C>A or
S489X	c.1466C>A
S492F	c.1475C>T
S549N	c.1646G>A
S549R A->C	c.1645A>C
S549R T->G	c.1647T>G
S945L	c.2834C>T
T338I	c.1013C>T
V520F	c.1558G>T
W1089X	c.3266G>A
W1204X 3743G->A	c.3611G>A
W1204X 3744G->A	c.3612G>A
W1282X	c.3846G>A
W401X_TAG	c.1202G>A
W401X_TGA	c.1203G>A
W846X	c.2537G>A
W846X 2670TGG>TGA	c.2538G>A
Y1092X C>A	c.3276C>A
Y1092X C>G	c.3276C>G
Y122X	c.366T>
Variants	
5T/non 5T	c.1210?12[5] (AJ574948.1:g.152T[5])
F508C	c.1523T>G
I506T	c.1517T>C
I506V	c.1516A>G
I507V	c.1519A>G

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HerediT®

CYSTIC FIBROSIS CARRIER SCREENING TEST

The performance characteristics of the HerediT carrier screening test for CF have been analytically validated using CFTR gene mutations shown to be phenotypically relevant.[1,2,3]

Ethnicity	Detection rate	Population risk	Residual risk after a negative screen
Caucaisan	93%	1/25	1/344
Ashkenazi Jewish	95%	1/24	1/461
Hispanic	82%	1/58	1/318
African American	76%	1/61	1/251
Asian	55%	1/94	1/208

1. Clinical and functional translation of CFTR. US CF Foundation, Johns Hopkins University, The Hospital for Sick Children Web site. http://CFTR2.org. Accessed Oct 4, 2012.
2. Update on Carrier Screening for Cystic Fibrosis, ACOG Committee Opinion No. 486. Obstet Gynecol. 2011;117(4):1028-1031.
3. Paladino T, et al. Comprehensive Cystic Fibrosis Screening Panel for the Detection of Clinically Relevant Mutations. Poster session presented at: American College of Medical Genetics and Genomics Annual Clinical Genetics Meeting; 2013 June 20-23; Phoenix, AZ.

About the test

HerediT CF is a laboratory-developed test that was developed and its performance characteristics determined by Sequenom Laboratories. This test is performed exclusively by Sequenom Laboratories. This test is used for clinical purposes; it should not be regarded as investigational or for research. This laboratory-developed test has not been cleared or approved by the U.S. Food and Drug Administration (FDA). Although laboratory-developed tests to date have not been subject to U.S. FDA regulation, certification of the laboratory is required under the Clinical Laboratory Improvement Amendment (CLIA) to ensure the quality and validity of test results. Sequenom Laboratories is CAP accredited and certified under CLIA to perform high complexity clinical laboratory testing.

Limitations

In an unknown number of cases, nearby genetic variants may interfere with mutation detection. Other possible sources of diagnostic error include sample mix-up, trace contamination, bone marrow transplantation, blood transfusions or other causes. If more than one variant is detected in a gene, additional studies may be necessary to determine if those variants lie on the same chromosome or different chromosomes. All results should be interpreted in the context of family history, and additional evaluation may be warranted if there is history of any genetic conditions. Reproductive risk assessment depends on accurate knowledge of paternity. Furthermore, not all mutations will be identified in the genes analyzed and additional testing may be beneficial for some patients. While results of this testing are highly accurate, a negative test significantly reduces, but does not eliminate, the chance of being a carrier. A patient with a positive test result should be referred for genetic counseling and further evaluation.

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