



Procedure	Result	Units	Ref Interval	Accession	Collected	Received	Reported/Verified
Cystic Fibrosis, Allele 1	p.Phe508del *			17-354-900052	20-Dec-17 10:38:00	20-Dec-17 10:38:00	20-Dec-17 17:12:45
Cystic Fibrosis, Allele 2	Negative			17-354-900052	20-Dec-17 10:38:00	20-Dec-17 10:38:00	20-Dec-17 17:12:45
Cystic Fibrosis 5T Variant	Not Applicable			17-354-900052	20-Dec-17 10:38:00	20-Dec-17 10:38:00	20-Dec-17 17:12:45
Cystic Fibrosis, 165 Variants, Interp	0 variants f			17-354-900052	20-Dec-17 10:38:00	20-Dec-17 10:38:00	20-Dec-17 17:12:45

20-Dec-17 10:38:00 Cystic Fibrosis, 165 Variants, Interp:

One severe pathogenic cystic fibrosis (CF) variant was identified indicating this individual is a carrier of CF. Adult family members and this individual's reproductive partner should be offered CF carrier screening. Genetic consultation is recommended.

Specimen: Whole Blood
 Symptoms: No
 Ethnicity: Multi Ethnic
 Family History: No

This result has been reviewed and approved by Pinar Bayrak-Toydemir, M.D., Ph.D.

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from patients (or their legal guardians) prior to pursuing genetic testing. These forms must be kept on file by the ordering physician. Consent forms for genetic testing are available at www.aruplab.com. Incidental findings are not reported unless clinically significant but are available upon request.

20-Dec-17 10:38:00 Cystic Fibrosis, 165 Variants, Interp:

BACKGROUND INFORMATION: Cystic Fibrosis (CFTR),
 165 Pathogenic Variants

CHARACTERISTICS OF CLASSIC CYSTIC FIBROSIS (CF): Chronic sino-pulmonary disease, gastrointestinal malabsorption/pancreatic insufficiency, and obstructive azoospermia. Symptoms of a CFTR-related disorder are often limited to a single organ system such as isolated pancreatitis, bilateral absence of the vas deferens, nasal polyposis, or bronchiectasis.

INCIDENCE: 1 in 2,300 Ashkenazi Jewish, 1 in 2,500 Caucasians, 1 in 13,500 Hispanics, 1 in 15,100 African Americans, 1 in 35,100 Asians.

INHERITANCE: Autosomal recessive.

PENETRANCE: High for severe pathogenic variants, variable for moderate or mild pathogenic variants.

Cause of Classic CF: Two severe, or one severe and one moderate, pathogenic CFTR variants on opposite chromosomes.

Cause of CFTR-Related Disorder: Two pathogenic CFTR variants on opposite chromosomes; two mild, one mild and one severe or one mild and one moderate.

PATHOGENIC VARIANTS TESTED: Variants are listed by standard nomenclature. Legacy names are also provided for the 23 recommended ACMG variants: c.1A>G, p.Met1Val; c.54-5940_273+10250del, Exons 2-3del; c.115C>T, p.Gln39X; c.178G>T, p.Glu60X; c.200C>T, p.Pro67Leu; c.223C>T, p.Arg75X; c.254G>A (Legacy G85E), p.Gly85Glu; c.262_263delTT, p.Leu88IlefsX22 (aka p.Leu88fs); c.273+1G>A, Intronic; c.273+3A>C, Intronic; c.274-1G>A, Intronic; c.274G>A, p.Glu92Lys; c.274G>T, p.Glu92X; c.292C>T, p.Gln98X; c.313delA, p.Ile105SerfsX2 (aka p.Ile105fs); c.325_327delTATinsG, p.Tyr109GlyfsX4 (aka p.Tyr109fs); c.328G>C, p.Asp110His; c.349C>T, p.Arg117Cys; c.350G>A (Legacy R117H), p.Arg117His; c.366T>A, p.Tyr122X; c.442delA, p.Ile148LeufsX5 (aka p.Ile148fs); c.489+1G>T (Legacy 621+1G>T), Intronic; c.531delT, p.Ile177MetfsX12 (aka p.Ile177fs); c.532G>A, p.Gly178Arg; c.579+1G>T (Legacy 711+1G>T), Intronic; c.579+5G>A, Intronic; c.579+3A>G, Intronic; c.580-1G>T, Intronic; c.595C>T, p.His199Tyr; c.613C>T, p.Pro205Ser; c.617T>G, p.Leu206Trp; c.658C>T, p.Gln220X; c.680T>G; p.Leu227Arg; c.720_741delAGGGAGAATGATGATGAAGTAC, p.Gly241GluX13 (aka p.Gly241fs); c.803delA,

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p.Asn268IlefsX17 (aka p.Asn268fs); c.805_806delAT, p.Ile269ProfsX4 (aka p.Ile269fs); c.933_935delCTT, p.Phe312del; c.948delT, p.Phe316LeufsX12 (aka p.Phe316fs); c.988G>T, p.Gly330X; c.1000C>T (Legacy R334W), p.Arg334Trp; c.1007T>A, p.Ile336Lys; c.1021T>C, p.Ser341Pro; c.1022_1023insTC, p.Phe342HisfsX28 (aka p.Phe342fs); c.1040G>A, p.Arg347His; c.1040G>C (Legacy R347P), p.Arg347Pro; c.1055G>A, p.Arg352Gln; c.1081delT, p.Trp361GlyfsX8 (aka p.Trp361fs); c.1116+1G>A, Intronic; c.1127_1128insA, p.Gln378AlafsX4 (aka p.Gln378fs); c.1153_1154insAT, p.Asn386IlefsX3 (aka p.Asn386fs); c.1202G>A, p.Trp401X; c.1203G>A, p.Trp401X; c.1209+1G>A, Intronic; c.1329_1330insAGAT, p.Ile444ArgfsX3 (aka p.Ile444fs); c.1340delA; p.Lys447ArgfsX2 (aka p.Lys447fs); c.1364C>A (Legacy A455E), p.Ala455Glu; c.1393-1G>A, Intronic; c.1397C>A, p.Ser466X; c.1397C>G, p.Ser466X; c.1400T>C, p.Leu467Pro; c.1418delG, p.Gly473GluX54 (aka p.Gly473fs); c.1438G>T, p.Gly480Cys; c.1466C>A, p.Ser489X; c.1475C>T, p.Ser492Phe; c.1477C>T, p.Gln493X; c.1519_1521delATC (Legacy I507del), p.Ile507del; c.1521_1523delCTT (Legacy F508del), p.Phe508del; c.1545_1546delTA, p.Tyr515X; c.1558G>T, p.Val520Phe; c.1572C>A, p.Cys524X; c.1573C>T, p.Gln525X; c.1585-1G>A (Legacy 1717-1G>A), Intronic; c.1585-8G>A, Intronic; c.1624G>T (Legacy G542X), p.Gly542X; c.1645A>C, p.Ser549Arg; c.1646G>A, p.Ser549Asn; c.1647T>G, p.Ser549Arg; c.1651G>A, p.Gly551Ser; c.1652G>A (Legacy G551D), p.Gly551Asp; c.1654C>T, p.Gln552X; c.1657C>T (Legacy R553X), p.Arg553X; c.1675G>A, p.Ala559Thr; c.1679G>A, p.Arg560Lys; c.1679G>C (Legacy R560T), p.Arg560Thr; c.1679+1.6kbA>G, Intronic; c.1680-1G>A, Intronic; c.1703delT, p.Leu568CysfsX4 (aka p.Leu568fs); c.1705T>G, p.Tyr569Asp; c.1721C>A, p.Pro574His; c.1753G>T, p.Glu585X; c.1766+1G>A (Legacy 1898+1G>A), Intronic; c.1766+3A>G, Intronic; c.1792_1798delAAACTA, p.Lys598GlyfsX11 (aka p.Lys598fs); c.1911delG, p.Gln637HisfsX26 (aka p.Gln637fs); c.1923_1931del9insA, p.Ser641ArgfsX5 (aka p.Ser641fs); c.1973_1985del13insAGAAA, p.Arg658LysfsX4 (aka p.Arg658fs); c.1976delA; p.Asn659IlefsX4 (aka p.Asn659fs); c.2012delT, p.Leu671X; c.2051_2052del, p.Lys684ThrfsX4; c.2051_2052delinsG (aka c.2051_2delinsG), p.Lys684SerfsX38; c.2052delA (Legacy 2184delA), p.Lys684AsnfsX38; c.2125C>T, p.Arg709X; c.2128A>T, p.Lys710X; c.2175_2176insA, p.Glu726ArgfsX4 (aka p.Glu726fs); c.2195T>G, p.Leu732X; c.2215delG, p.Val739TyrfsX16 (aka p.Val739fs); c.2290C>T, p.Arg764Ter; c.2453delT, p.Leu818TrpfsX3 (aka p.Leu818fs); c.2464G>T, p.Glu822X; c.2490+1G>A, Intronic; c.2491G>T, p.Glu831X; c.2537G>A, p.Trp846X; c.2538G>A; p.Trp846X; c.2551C>T, p.Arg851X; c.2583delT, p.Phe861LeufsX3 (aka p.Phe861fs); c.2657+5G>A (Legacy 2789+5G>A), Intronic; c.2668C>T, p.Gln890X; c.2737_2738insG, p.Tyr913X; c.2780T>C, p.Leu927Pro; c.2810_2811insT, p.Val938GlyfsX37 (aka p.Val938fs); c.2834C>T, p.Ser945Leu; c.2875delG, p.Ala959HisfsX9 (aka p.Ala959fs); c.2908G>C, p.Gly970Arg; c.2988+1G>A (Legacy 3120+1G>A), Intronic; c.2988G>A, Intronic; c.2989-1G>A, Intronic; c.3039delC, p.Tyr1014ThrfsX9 (aka p.Tyr1014fs); c.3067_3072delATAGTG, p.Ile1023_Val1024del (aka I1023_V1024del); c.3140-26A>G, Intronic; c.3194T>C, p.Leu1065Pro; c.3196C>T, p.Arg1066Cys; c.3197G>A, p.Arg1066His; c.3230T>C, p.Leu1077Pro; c.3266G>A, p.Trp1089X; c.3276C>A, p.Tyr1092X; c.3276C>G, p.Tyr1092X; c.3302T>A, p.Met1101Lys; c.3310G>T, p.Glu1104X; c.3472C>T, p.Arg1158X; c.3484C>T (Legacy R1162X), p.Arg1162X; c.3528delC (Legacy 3659delC), p.Lys1177SerfsX15 (aka p.Lys1177fs); c.3536_3539del, p.Thr1179AsnfsX12 (aka p.Thr1179fs); c.3587C>G, p.Ser1196X; c.3611G>A, p.Trp1204X; c.3612G>A, p.Trp1204X; c.3659delC, p.Thr1220LysfsX8 (aka p.Thr1220fs); c.3691delT, p.Ser1231ProfsX4 (aka p.Ser1231fs); c.3712C>T, p.Gln1238X; c.3718+2477C>T (Legacy 3849+10kbC>T), Intronic; c.3731G>A, p.Gly1244Glu; c.3744delA, p.Lys1250ArgfsX9 (aka p.Lys1250fs); c.3752G>A, p.Ser1251Asn; c.3763T>C, p.Ser1255Pro; c.3764C>A, p.Ser1255X; c.3773_3774insT, p.Leu1258PhefsX7 (aka p.Leu1258fs); c.3846G>A (Legacy W1282X), p.Trp1282X; c.3873+1G>A, Intronic; c.3909C>G (Legacy N1303K), p.Asn1303Lys; c.3937C>T, p.Gln1313X; c.3964-78_4242+577del, Exons 22-23del; c.4028delG, p.Gly1343AlafsX4 (aka p.Gly1343fs); c.4046G>A, p.Gly1349Asp; c.4077_4080delTGTTinsAA, p.Val1360fsX3 (aka p.Val1360fs); c.4111G>T, p.Glu1371X; c.4251delA, p.Glu1418ArgfsX14 (aka p.Glu1418fs). The IVS-8 variant, c.1210-12[5], will be reported only when R117H is detected or in patients who are reported to be symptomatic.

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CLINICAL SENSITIVITY: Ashkenazi Jewish 96 percent; Caucasian 92 percent; Hispanic 80 percent; African American 78 percent; Asian American 55 percent.

METHODOLOGY: Polymerase chain reaction (PCR) and fluorescence monitoring.

Analytical Sensitivity & Specificity: 99 percent.

LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. Only the 165 pathogenic CFTR variants and 5T variant (listed above) will be interrogated.

See Compliance Statement C: www.aruplab.com/CS