MAYO CLINIC LABORATORIES

Targeted Variants Interrogated by Cystic Fibrosis Variant Panel

Associated Phenotype	Gene (Transcript)	Variants
Cystic fibrosis	*CFTR* (NM_000492)	c.-9_14del, c.1A>C, c.1A>G, c.2T>A, c.2T>C, c.2T>G, c.3G>A, c.3G>T, c.11C>A*, c.14C>T, c.19G>T, c.40A>T, c.42del, c.43del, c.50del, c.50dup, c.53+1G>T, c.54-2A>G, c.54-1G>A, c.57G>A, c.79G>T*, c.88C>T, c.112_113del, c.114C>G, c.115C>T*, c.137C>A, c.156del, c.164+1G>A, c.164+1G>C, c.164+1G>T, c.164+2T>A, c.164+2T>C, c.164+2T>G, c.164+12T>C*, c.165-2A>G, c.165-1G>A, c.166G>A*, c.168del*, c.170G>A, c.171G>A, c.175dup, c.178G>T*, c.200C>T*, c.206T>A, c.217del, c.220C>T*, c.223C>T*, c.233del, c.233dup, c.234del, c.236G>A, c.248dup, c.254G>A*, c.259T>A*, c.262_263del*, c.263T>A, c.263T>G, c.271G>A, c.273+1G>A, c.273+3A>C*, c.274-2A>C, c.274-2A>G, c.274-1G>A*, c.274-1G>C, c.274-1G>T, c.274G>A*, c.274G>T*, c.292C>T*, c.302T>G, c.307G>T*, c.310del*, c.313del, c.319_326del, c.325_327delinsG, c.326_327del, c.328del*, c.328G>C*, c.330C>A, c.340A>T, c.343G>T, c.349C>T*, c.350G>A*, c.357del, c.366T>A, c.376G>A, c.380T>G, c.409_412del, c.413_415dup, c.415_416insGA, c.415_416insTA, c.416A>T, c.422C>A, c.424del, c.429del, c.433del, c.442del, c.443T>C*, c.445G>T, c.470T>G, c.476dup, c.476T>A, c.487A>G, c.489+1G>T*, c.489+2T>C*, c.489+2T>G, c.489+3A>G, c.490-2A>C, c.490-2A>G, c.490-1G>A, c.494del, c.522_526del, c.526del, c.531del*, c.532G>A*, c.543_546del, c.547C>A, c.550del, c.567C>A, c.575A>G, c.577G>A, c.577G>T*, c.578_579+5del*, c.579+1G>T*, c.579+3A>G*, c.579+5G>A*, c.580-1G>T*, c.595C>T, c.601del, c.606G>A, c.613C>T*, c.617T>G*, c.619C>T, c.647G>A, c.650_659del, c.653T>A, c.658C>T*, c.675T>A, c.680T>G, c.708del, c.714del, c.722_743del*, c.741C>G, c.743+1G>A*, c.743+1G>C, c.744-2A>G, c.761del, c.773del, c.803del*, c.805_806del, c.825C>G, c.828C>A, c.850dup, c.860dup, c.861_865del*, c.868C>T, c.870-2A>G, c.881_882del, c.912C>G, c.927del, c.935_937del*, c.938C>A, c.948del*, c.959T>A, c.980del, c.987del, c.988G>T*, c.1000C>T*, c.1005_1006insG, c.1007T>A*, c.1013C>T*, c.1021T>C*, c.1021_1022dup*, c.1029del*, c.1040G>A*, c.1040G>C*, c.1053_1054del, c.1055G>A*, c.1057C>T, c.1068G>A, c.1069del, c.1075_1079delinsAAAAA*, c.1075C>A*, c.1081del, c.1083del, c.1086T>A, c.1090T>C, c.1093_1094del, c.1116+1G>A*, c.1116+1G>C*, c.1117-1G>A, c.1130dup, c.1135G>T*, c.1152del*, c.1155_1156dup*, c.1155_1156insTA*, c.1159_1160del, c.1162_1168del, c.1177del, c.1192dup, c.1202G>A, c.1203G>A, c.1209+1G>A, 5T (c.1210-7_1210-6del)*, 9T (c.1210-13G>T)*, c.1240C>T, c.1327_1330dup*, c.1340del*, c.1364C>A*, c.1365_1366del*, c.1393-2A>G, c.1393-1G>A*, c.1397C>A, c.1397C>G, c.1400T>C, c.1418del*, c.1433_1434del, c.1435G>T, c.1438G>T*, c.1439del, c.1446dup, c.1456G>T, c.1466C>A*, c.1470_1471del, c.1475C>T*, c.1477C>T*, c.1477_1478del, c.1478A>T, c.1482_1483del, c.1487G>A, c.1510G>T, c.1519_1521del*, c.1521_1523del*, c.1528del, c.1545_1546del*, c.1550A>G*, c.1558G>A*, c.1558G>T*, c.1572C>A, c.1573C>T, c.1584+1G>A*, c.1584+2T>C, c.1585-8G>A*, c.1585-2A>G, c.1585-1G>A*, c.1588A>C, c.1606A>T, c.1611_1612del, c.1624G>T*, c.1628A>C, c.1642_1643del, c.1645A>C, c.1645_1648del,

Targeted Variants Interrogated by Cystic Fibrosis Variant Panel (continued)

Associated Phenotype	Gene (Transcript)	Variants
Cystic fibrosis (continued)	*CFTR* (NM_000492) (continued)	c.1646G>A*, c.1646G>T, c.1647T>G*, c.1648G>T, c.1650del, c.1651G>A, c.1652del, c.1652G>A*, c.1654C>T, c.1656del, c.1657C>T*, c.1670del, c.1673T>C, c.1674del, c.1675G>A*, c.1679G>A*, c.1679G>C*, c.1679+1G>A*, c.1679+1G>C*, c.1679+2T>C, c.1680-886A>G, c.1680-877G>T*, c.1680-1G>A*, c.1682C>A*, c.1692del, c.1705T>G, c.1707T>A, c.1714_1715del, c.1721C>A*, c.1731C>A, c.1736A>G, c.1738del, c.1742dup, c.1753G>T*, c.1766+1G>A*, c.1766+1G>C*, c.1766+1G>T*, c.1766+2T>C, c.1766+3A>G*, c.1766+5G>T*, c.1786_1787del, c.1792A>T, c.1792_1798del, c.1800del, c.1820_1903del*, c.1826A>G, c.1865G>A*, c.1871_1878del, c.1900C>T, c.1909C>T, c.1911del, c.1919_1920del, c.1923_1931delinsA*, c.1966G>T*, c.1973_1985delinsAGAAA*, c.1976del, c.1981del, c.1986_1989del*, c.1990G>T, c.2012del, c.2017G>T, c.2036G>A, c.2044del, c.2051_2052delinsG*, c.2052del*, c.2052dup*, c.2053C>T, c.2053dup, c.2058_2061dup*, c.2062A>T, c.2065C>T, c.2074G>T, c.2083dup, c.2089dup, c.2108del, c.2125C>T*, c.2128A>T*, c.2143C>T, c.2156T>A, c.2158C>T*, c.2175dup*, c.2188G>T*, c.2195T>G*, c.2203del, c.2215del*, c.2233G>T, c.2249_2256del, c.2276_2277del, c.2277del, c.2324_2325del, c.2327C>G, c.2341C>T, c.2353C>T*, c.2374C>T, c.2380del, c.2393dup, c.2421A>G*, c.2423_2424dup, c.2435dup, c.2440C>T, c.2453del, c.2463_2464del, c.2464G>T, c.2467G>T, c.2472del, c.2479G>T, c.2488A>T, c.2490+1G>A*, c.2490+1G>T, c.2490+2T>C, c.2491-2A>G, c.2491G>T*, c.2502del, c.2502dup, c.2508del, c.2537G>A*, c.2538G>A, c.2547C>A, c.2551C>T, c.2555A>T, c.2566_2567insT, c.2583del, c.2589_2599del, c.2592_2593del, c.2600T>A, c.2601dup, c.2615del, c.2619+1G>A, c.2619+2T>A, c.2619+4dup, c.2620-26A>G*, c.2620-2A>G, c.2620-1G>C, c.2620-1G>T, c.2645G>A, c.2657+2_2657+3insA, c.2657+5G>A*, c.2658-2A>G, c.2658-1G>C, c.2658-1G>T, c.2668C>T*, c.2735C>A*, c.2737_2738insG*, c.2739T>A*, c.2763_2764dup, c.2776_2777del, c.2777del, c.2780T>C, c.2810dup*, c.2825del, c.2834C>T*, c.2836A>T, c.2856G>C, c.2859_2890del, c.2875del*, c.2876del, c.2896del, c.2908G>C, c.2908+2T>C, c.2909-1G>A, c.2909del, c.2924_2925del, c.2930C>T, c.2932A>T*, c.2988G>A*, c.2988+1G>A*, c.2988+2T>C, c.2989-2A>G, c.2989-1G>A, c.2997_3000del, c.2998del, c.3002_3003del, c.3007G>T, c.3017C>A, c.3021del, c.3022del, c.3039del, c.3039dup, c.3041A>G, c.3042_3043del, c.3061C>T, c.3067_3072del*, c.3068_3072del, c.3103C>T, c.3106del, c.3124C>T, c.3139_3139+1del, c.3140-26A>G*, c.3140-1G>A*, c.3154T>G*, c.3160C>G, c.3161del, c.3176T>G, c.3179A>C*, c.3181G>C*, c.3189G>A, c.3194T>C, c.3196C>T*, c.3197G>A, c.3199G>A*, c.3205G>A*, c.3208C>T*, c.3209G>A, c.3211C>T, c.3222T>A, c.3229_3230del, c.3230T>C, c.3263dup, c.3264del, c.3266G>A*, c.3276C>A*, c.3276C>G, c.3287del, c.3291del, c.3292T>C, c.3293G>A, c.3294del, c.3294G>A, c.3299A>C*, c.3302T>A*, c.3304A>T, c.3310G>T*, c.3315del, c.3364del*, c.3368-2A>G, c.3368-1G>A, c.3382A>T*, c.3430C>T, c.3434G>A, c.3435G>A, c.3445del, c.3454G>C*, c.3468+2T>C*, c.3469-2A>G, c.3472C>T*, c.3484C>T*, c.3486_3487del, c.3492dup, c.3497del, c.3528del*, c.3529A>T, c.3530del*, c.3534_3535insTCAA, c.3536_3539del, c.3540del, c.3546C>G, c.3556C>T, c.3587C>G,

Targeted Variants Interrogated by Cystic Fibrosis Variant Panel (continued)

Associated Phenotype	Gene (Transcript)	Variants
Cystic fibrosis (continued)	*CFTR* (NM_000492) (continued)	c.3592del, c.3600del, c.3605del, c.3611G>A, c.3612G>A*, c.3617C>A, c.3618_3619del, c.3659del*, c.3691del*, c.3712C>T, c.3717+1G>A*, c.3717+4A>G, c.3717+40A>G, c.3718-2477C>T*, c.3718-3T>G*, c.3718-1G>A*, c.3731G>A*, c.3744del*, c.3747del, c.3752G>A*, c.3761T>G*, c.3763T>C, c.3764C>A*, c.3764C>G*, c.3764C>T*, c.3773dup*, c.3808del, c.3808G>A, c.3810T>A, c.3816_3817del, c.3822G>A, c.3829del, c.3835_3836del, c.3841C>T, c.3846G>A*, c.3848G>T, c.3855del, c.3871C>T, c.3873+1G>A, c.3873+2T>C*, c.3883del, c.3883_3884insG, c.3883_3886del, c.3908del*, c.3908dup, c.3909C>G*, c.3921T>A, c.3922G>T, c.3929G>A, c.3937C>T*, c.3947G>A, c.3963+2T>A, c.3964-1G>A, c.3976del, c.3985G>C, c.4025_4028dup, c.4039dup, c.4040_4041del, c.4042del, c.4046G>A*, c.4077_4080delinsAA*, c.4086dup, c.4111G>T, c.4139del, c.4140del*, c.4141T>C, c.4143C>A*, c.4144C>T*, c.4147dup, c.4168C>T*, c.4170del*, c.4197_4198del, c.4201dup, c.4201G>T, c.4231C>T, c.4234C>T, c.4242+1G>A, c.4242+1G>T, c.4243-2A>C, c.4243-1G>C, c.4251del, c.4252G>T, c.4297G>A, c.4300_4301insAG, c.4364C>G, c.4417G>T, c.4426C>T*, Deletion and duplication analysis of exon 1, exon 2, exon 3, exon 4, exon 5, exon 6, exon 7, exon 8, exon 9, exon 10, exon 11, exon 12, exon 13, exon 14, exon 15, exon 16, exon 17, exon 18, exon 19–20, exon 21, exon 22, exon 23, exon 24, exon 25, exon 26, exon 27

* Previously detected in a known positive sample