MAYO CLINIC LABORATORIES

Targeted Variants Detected by Focused Carrier Screening Tests

This document provides the genes and variants included on the following panels:

- CFSMN / Cystic Fibrosis and Spinal Muscular Atrophy Carrier Screen Panel, Varies
- CSFP / Carrier Screen, Focused Panel, Varies

See footnotes for details on which genes are included on which panel.

Associated Phenotype	Gene (Transcript)	Variants
Cystic fibrosis	*CFTR* (NM_000492)[1,2]	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,

Targeted Variants Detected by Focused Carrier Screening Tests (continued)

Associated Phenotype	Gene (Transcript)	Variants
Cystic fibrosis (continued)	*CFTR* (NM_000492)[1,2] (continued)	c.1642_1643del, c.1645A>C, c.1645_1648del, 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, 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*,

Targeted Variants Detected by Focused Carrier Screening Tests (continued)

Associated Phenotype	Gene (Transcript)	Variants
Cystic fibrosis (continued)	*CFTR* (NM_000492)[1,2] (continued)	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.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
Fragile X	*FMR1*[2]	Trinucleotide repeat
Hemoglobinopathy, Alpha thalassemia	*HBA1* (NM_000558)[2]	Deletion analysis of exon 3
	HBA2 (NM_000517)[2]	c.427T>C*, c.*94A>G* Deletion analysis of exon 3
Hemoglobinopathy, Beta thalassemia	*HBB* (NM_000518)[2]	c.-140C>T, c.-138C>T, c.-137C>G, c.-81A>G*, c.-80T>A, c.-79A>G*, c.-78A>C, c.-78A>G*, c.1A>G, c.2T>G*, c.17_18del, c.20del*, c.25_26del, c.26A>G, c.27dup, c.36del, c.45dup*, c.46del, c.48G>A, c.51del*, c.52A>T*, c.59A>G*, c.75T>A*, c.79G>T*, c.85dup*, c.92G>C, c.92+1G>A*, c.92+5G>C*, c.92+6T>C*, c.93-21G>A*, c.112del, c.114G>A, c.118C>T*, c.126_129del*, c.135del*, c.155del, c.203_204del, c.217dup, c.271G>T, c.287dup, c.315+1G>A*, c.316-197C>T*, c.316-106C>G*, c.316-3C>A, c.316-2A>C*, c.316-2A>G, c.316-1G>T, c.364G>A, c.383_385del, c.*113A>G* Deletion analysis of exon 1–2, exon 3
Hemoglobinopathy, Hb C		c.19G>A
Hemoglobinopathy, Hb D		c.364G>C
Hemoglobinopathy, Hb E		c.79G>A*
Hemoglobinopathy, Hb S		c.20A>T*
Spinal muscular atrophy	*SMN1*[1,2]	Deletion analysis of exon 7–8
	SMN1 (NM_000344)[1,2]	c.*3+80T>G*
	SMN2[1,2]	Deletion analysis of exon 7–8 in cases with 0 copies of *SMN1*

[1] CFSMN / Cystic Fibrosis and Spinal Muscular Atrophy Carrier Screen Panel, Varies

[2] CSFP / Carrier Screen, Focused Panel, Varies

* Previously detected in a known positive sample