MiSeqDx Cystic Fibrosis 175-Variant Assay Summary of variants:

G85E (c.254G>A), R117H (c.350G>A), 621+1G>T (c.489+1G>T), 711+1G>T (c.579+1G>T), R553* (c.1657C>T), R1162* (c.3484C>T), 3659del (c.3528del), 3849+10kbC>T (c.3717+12191C>T), W1282* (c.3846G>A), N1303K (c.3909C>G), R347P (c.1040G>C), A455E (c.1364C>A), I507del (c.1519_ 1521del), F508del (c.1521_ 1523del), 1717-1G>A (c.1585-1G>A), R560T (c.1679G>C), 1898+1G>A (c.1766+1G>A), 2184del (c.2052del), 2789+5G>A (c.2657+5G>A), G542* (c.1624G>T), R334W (c.1000C>T), G551D (c.1652G>A), 3120+1G>A (c.2988+1G>A), M1V (c.1A>G), CFTR dele2,3 (c.54-5940_273+10250dele21kb), Q39* (c.115C>T), E60* (c.178G>T), P67L (c.200C>T), R75* (c.223C>T), 394del (c.262_263del), 405+1 G>A (c.273+1G>A), 406-1G>A (c.274-1G>A), E92* (c.274G>T), E92K (c.274G>A), Q98* (c.292C>T), 457TAT>G (c.325_327delinsG), D110H (c.328G>C), R117C (c.349C>T), Y122* (c.366T>A), 574del (c.442del), 663del (c.531del), G178R (c.532G>A), 711+3A>G (c.579+3A>G), 711+5 G>A (c.579+5G>A), 712-1 G>T (c.580-1G>T), H199Y (c.595C>T), P205S (c.613C>T), L206W (c.617T>G), Q220* (c.658C>T), T338I (c.1013C>T), 3272-26A>G (c.3140-26A>G), L1065P (c.3194T>C), R1066C (c.3196C>T), R1066H (c.3197G>A), L1077P (c.3230T>C), W1089* (c.3266G>A), Y1092*(C>A) (c.3276C>A), Y1092*(C>G) (c.3276C>G), M1101K (c.3302T>A), E1104* (c.3310G>T), R1158* (c.3472C>T), S1196* (c.3587C>G), W1204* (c.3611G>A), W1204* (c.3612G>A), 3791del (c.3659del), G1244E (c.3731G>A), 3876del (c.3744del), S1251N (c.3752G>A), 3905insT (c.3773_3774insT), 4005+1G>A (c.3873+1G>A), 4016insT (c.3884_3885insT), Q1313* (c.3937C>T), 4209TGTT>AA (c.4077_ 4080delinsAA), CFTRdele22,23 (c.3964-78_ 4242+577del), 4382del (c.4251del), S341P (c.1021T>C), 1154insTC (c.1022_1023insTC), R347H (c.1040G>A), R352Q (c.1055G>A), 1213del (c.1081del), 1248+1G>A (c.1116+1G>A), 1259insA (c.1127_1128insA), W401* (c.1202G>A), W401* (c.1203G>A), 1341+1G>A (c.1209+1G>A), 1461ins4 (c.1329_ 1330insAGAT), 1525-1G>A (c.1393-1G>A), S466* (C>A) (c.1397C>A), S466* (C>G) (c.1397C>G), L467P (c.1400T>C), 1548del (c.1418del), S489* (c.1466C>A), S492F (c.1475C>T), Q493* (c.1477C>T), 1677del (c.1545_ 1546del), V520F (c.1558G>T), Q525* (c.1573C>T), 1717-8G>A (c.1585-8G>A), A559T (c.1675G>A), R560K (c.1679G>A), 1811+1.6kb A>G (c.1679+1.6kbA>G), 1812-1 G>A (c.1680-1G>A), E585* (c.1753G>T), 1898+3A>G (c.1766+3A>G), 2143del (c.2012del), 2183AA>G (c.2051_ 2052delinsG), 2184insA (c.2052_2053insA), R709* (c.2125C>T), K710* (c.2128A>T), 2307insA (c.2175_2176insA), L732* (c.2195T>G), 2347del (c.2215del), R764* (c.2290C>T), 2585del (c.2453del), E822* (c.2464G>T), 2622+1G>A (c.2490+1G>A), E831* (c.2491G>T), W846* (c.2537G>A), R851* (c.2551C>T), 2711del (c.2583del), Q890* (c.2668C>T), L927P (c.2780T>C), S945L (c.2834C>T), 52del22 (c.720_741del), 1078del (c.948del), G330* (c.988G>T), I336K (c.1007T>A), S549R (c.1645A>C), 3007del (c.2875del), S549N (c.1646G>A), G970R (c.2908G>C), S549R (c.1647T>G), 3120G>A (c.2988G>A), Q552* (c.1654C>T), 3121-1G>A (c.2989-1G>A), Q2* (c.4C>T), G91R (c.271G>A), Q98R (c.293A>G), 444del (c.313del), F191V (c.571T>G), L218* (c.653T>A), V232D (c.695T>A), 935del (c.803del), Q290* (c.868C>T), 1161del (c.1029del), 1288insTA (c.1155_1156dup), V456A (c.1367T>C), 1504del

(c.1373del), C524* (c.1572C>A), 1716+1G->A (c.1584+1G>A), G551S (c.1651G>A), L558S (c.1673T>C), 1811+1G->A (c.1679+1G>A), R560S (c.1680A>C), A561E (c.1682C>A), 1824del (c.1692del),Y569D (c.1705T>G), 1898+5G->T (c.1766+5G>T), A613T (c.1837G>A), G628R (c.1882G>A), R785* (c.2353C>T), R792* (c.2374C>T), Y913* (c.2739T>A), G970D (c.2909G>A), 3199del6 (c.3067_3072del), T1036N (c.3107C>A), W1098C (c.3294G>C), S1118F (c.3353C>T), 3849+5G->A (c.3717+5G>A), S1255* (c.3764C>A), 3940del (c.3808del)

Reflex reporting with homozygous p.F508del or p.I507del: I506V (c.1516A>G), I507V (c.1519A>G), F508C (c.1523T>G). Reflex reporting with p.R117H: PolyTG/PolyT haplotypes