

FDA Approved 139 Variant Panel

| Legacy Name | Standard Nomenclature | |
|---------------|-----------------------|----------------------|
| | (protein) | (nucleotide) |
| 1078delT | p.Phe316LeufsX12 | c.948delT |
| 1154insTC | p.Phe342HisfsX28 | c.1022_1023insTC |
| 1213delT | p.Trp361GlyfsX8 | c.1081delT |
| 1248+1G>A | | c.1116+1G>A |
| 1259insA | p.Gln378AlafsX4 | c.1127_1128insA |
| 1341+1G>A | | c.1209+1G>A |
| 1461ins4 | p.Ile444ArgfsX3 | c.1329_1330insAGAT |
| 1525-1G>A | | c.1393-1G>A |
| 1548delG | p.Gly473GlufsX54 | c.1418delG |
| 1677delTA | p.Tyr515X | c.1545_1546delTA |
| 1717-1G>A | | c.1585-1G>A |
| 1717-8G>A | | c.1585-8G>A |
| 1811+1.6kbA>G | | c.1679+1.6kbA>G |
| 1812-1G>A | | c.1680-1G>A |
| 1898+1G>A | | c.1766+1G>A |
| 1898+3A>G | | c.1766+3A>G |
| 2143delT | p.Leu671X | c.2012delT |
| 2183AA>G | p.Lys684SerfsX38 | c.2051_2052delAAinsG |
| 2184delA | p.Lys684AsnfsX38 | c.2052delA |
| 2184insA | p.Gln685ThrfsX4 | c.2052_2053insA |
| 2307insA | p.Glu726ArgfsX4 | c.2175_2176insA |
| 2347delG | p.Val739TyrfsX16 | c.2215delG |
| 2585delT | p.Leu818TrpfsX3 | c.2453delT |
| 2622+1G>A | | c.2490+1G>A |
| 2711delT | p.Phe861LeufsX3 | c.2583delT |
| 2789+5G>A | | c.2657+5G>A |
| 3007delG | p.Ala959HisfsX9 | c.2875delG |
| 3120+1G>A | | c.2988+1G>A |
| 3120G>A | | c.2988G>A |
| 3121-1G>A | | c.2989-1G>A |
| 3272-26A>G | | c.3140-26A>G |
| 3659delC | p.Lys1177SerfsX15 | c.3528delC |
| 3791delC | p.Thr1220LysfsX8 | c.3659delC |
| 3849+10kbC>T | | c.3717+12191C>T |
| 3876delA | p.Lys1250ArgfsX9 | c.3744delA |
| 3905insT | p.Leu1258PhefsX7 | c.3773_3774insT |
| 394delTT | p.Leu881IlefsX22 | c.262_263delTT |
| 4005+1G>A | | c.3873+1G>A |
| 4016insT | p.Ser1297PhefsX5 | c.3884_3885insT |
| 405+1G>A | | c.273+1G>A |
| 406-1G>A | | c.274-1G>A |

| | | |
|---------------|-------------------|------------------------------------|
| 4209TGTT>AA | | c.4077_4080delTGTTinsAA |
| 4382delA | p.Glu1418ArgfsX14 | c.4251delA |
| 457TAT>G | p.Tyr109GlyfsX4 | c.325_327delTATinsG |
| 574delA | p.Ile148LeufsX5 | c.442delA |
| 621+1G>T | | c.489+1G>T |
| 663delT | p.Ile177MetfsX12 | c.531delT |
| 711+1G>T | | c.579+1G>T |
| 711+3A>G | | c.579+3A>G |
| 711+5G>A | | c.579+5G>A |
| 712-1G>T | | c.580-1G>T |
| 852del22 | p.Gly241GlufsX13 | c.720_741delAGGGAGAATGATGATGAAGTAC |
| A455E | p.Ala455Glu | c.1364C>A |
| A559T | p.Ala559Thr | c.1675G>A |
| CFTRdele2,3 | p.Ser18ArgfsX16 | c.54-5940_273+10250del21kb |
| CFTRdele22,23 | | c.3964-78_4242+577del |
| D110H | p.Asp110His | c.328G>C |
| E1104X | p.Glu1104X | c.3310G>T |
| E585X | p.Glu585X | c.1753G>T |
| E60X | p.Glu60X | c.178G>T |
| E822X | p.Glu822X | c.2464G>T |
| E831X | p.Glu831X | c.2491G>T |
| E92K | p.Glu92Lys | c.274G>A |
| E92X | p.Glu92X | c.274G>T |
| F508C | | |
| F508del | p.Phe508del | c.1521_1523delCTT |
| G1244E | p.Gly1244Glu | c.3731G>A |
| G178R | p.Gly178Arg | c.532G>A |
| G330X | p.Gly330X | c.988G>T |
| G542X | p.Gly542X | c.1624G>T |
| G551D | p.Gly551Asp | c.1652G>A |
| G85E | p.Gly85Glu | c.254G>A |
| G970R | p.Gly970Arg | c.2908G>C |
| H199Y | p.His199Tyr | c.595C>T |
| I336K | p.Ile336Lys | c.1007T>A |
| I506V | | |
| I507del | p.Ile507del | c.1519_1521delATC |
| I507V | | |
| K710X | p.Lys710X | c.2128A>T |
| L1065P | p.Leu1065Pro | c.3194T>C |
| L1077P | p.Leu1077Pro | c.3230T>C |
| L206W | p.Leu206Trp | c.617T>G |
| L467P | p.Leu467Pro | c.1400T>C |
| L732X | p.Leu732X | c.2195T>G |
| L927P | p.Leu927Pro | c.2780T>C |
| M1101K | p.Met1101Lys | c.3302T>A |
| M1V | p.Met1Val | c.1A>G |

| | | |
|-----------------------|--------------|-----------|
| N1303K | p.Asn1303Lys | c.3909C>G |
| P205S | p.Pro205Ser | c.613C>T |
| P67L | p.Pro67Leu | c.200C>T |
| PolyTG/PolyT | | |
| Q1313X | p.Gln1313X | c.3937C>T |
| Q220X | p.Gln220X | c.658C>T |
| Q39X | p.Gln39X | c.115C>T |
| Q493X | p.Gln493X | c.1477C>T |
| Q525X | p.Gln525X | c.1573C>T |
| Q552X | p.Gln552X | c.1654C>T |
| Q890X | p.Gln890X | c.2668C>T |
| Q98X | p.Gln98X | c.292C>T |
| R1066C | p.Arg1066Cys | c.3196C>T |
| R1066H | p.Arg1066His | c.3197G>A |
| R1158X | p.Arg1158X | c.3472C>T |
| R1162X | p.Arg1162X | c.3484C>T |
| R117C | p.Arg117Cys | c.349C>T |
| R117H | p.Arg117His | c.350G>A |
| R334W | p.Arg334Trp | c.1000C>T |
| R347H | p.Arg347His | c.1040G>A |
| R347P | p.Arg347Pro | c.1040G>C |
| R352Q | p.Arg352Gln | c.1055G>A |
| R553X | p.Arg553X | c.1657C>T |
| R560K | p.Arg560Lys | c.1679G>A |
| R560T | p.Arg560Thr | c.1679G>C |
| R709X | p.Arg709X | c.2125C>T |
| R75X | p.Arg75X | c.223C>T |
| R764X | p.Arg764X | c.2290C>T |
| R851X | p.Arg851X | c.2551C>T |
| S1196X | p.Ser1196X | c.3587C>G |
| S1251N | p.Ser1251Asn | c.3752G>A |
| S341P | p.Ser341Pro | c.1021T>C |
| S466X(C>A) | p.Ser466X | c.1397C>A |
| S466X(C>G) | p.Ser466X | c.1397C>G |
| S489X | p.Ser489X | c.1466C>A |
| S492F | p.Ser492Phe | c.1475C>T |
| S549N | p.Ser549Asn | c.1646G>A |
| S549R (c.1645A>C) | p.Ser549Arg | c.1645A>C |
| S549R (c.1647T>G) | p.Ser549Arg | c.1647T>G |
| S945L | p.Ser945Leu | c.2834C>T |
| T338I | p.Thr338Ile | c.1013C>T |
| V520F | p.Val520Phe | c.1558G>T |
| W1089X | p.Trp1089X | c.3266G>A |
| W1204X (c.3611G>A) | p.Trp1204X | c.3611G>A |
| W1204X | p.Trp1204X | c.3612G>A |

| | | |
|-------------------|------------|-----------|
| (c.3612G>A) | | |
| W1282X | p.Trp1282X | c.3846G>A |
| W401X (c.1202G>A) | p.Trp401X | c.1202G>A |
| W401X (c.1203G>A) | p.Trp401X | c.1203G>A |
| W846X | p.Trp846X | c.2537G>A |
| Y1092X (C>A) | p.Tyr1092X | c.3276C>A |
| Y1092X (C>G) | p.Tyr1092X | c.3276C>G |
| Y122X | p.Tyr122X | c.366T>A |