Evidence-Based CFTR Variant Coverage

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Optimized Advanta™CFTR NGS Library Prep Assay design supports target enrichment of all 27 CFTR exons and select intronic regions, which cover >250 variants derived from the CFTR2 mutation list*. Sequence the spectrum of variant types, from simple SNPs to large deletions, in a single NGS-based workflow.

Variant legacy name

1078delT (d)	2185insC	3850-1G->A	C276X	**I507del** (a) (d)	**R553X** (a) (d)
1119delA	2307insA	3850-3T->G	C524X	**K710X**	**R560K**
1138insG	**2347delG**	**3876delA**	CFTR50kbdel	**L1065P**	R560S
1154insTC	2372del8	3878delG	CFTRdele1	**L1077P**	**R560T** (a)
1161delC	2556insAT	3905insT (d)	CFTRdele14b-17b	L1254X	**R709X**
1213delT	**2585delT**	**394delTT**	CFTRdele17a-18	**L206W**	**R75X**
1248+1G->A	2594delGT	**4005+1G->A**	CFTRdele17a,17b	**L227R**	**R764X**
1249-1G->A	**2622+1G->A**	4010del4	CFTRdele2	**L467P**	R785X
124del23bp	**2711delT**	4015delA	CFTRdele2-4	**L732X**	R792X
1259insA	2721del11	4016insT	CFTRdele2,3 (d)	L88X	**R851X**
1288insTA	2732insA	**4021dupT**	CFTRdele22-24	**L927P**	**S1196X**
1341+1G->A	**2789+5G->A** (a) (d)	4022insT	CFTRdele22,23	**M1101K** (d)	**S1251N**
1461ins4	2790-1G->C	4040delA	CFTRdele4-7	**M1V**	S1255P
1471delA	**2869insG**	405+1G->A	CFTRdup6b-10	**N1303K** (a) (d)	**S1255X**
1497delGG	2896insAG	**405+3A->C**	**D110H**	**P205S**	**S341P**
1525-1G->A	2942insT	**406-1G->A**	E1104X	**P67L**	**S466X**
1525-2A->G	2957delT	**4209TGTT->AA**	E1371X	Q1042X	**S489X**
1548delG	296+1G->A	4218insT	E193X	**Q1313X**	**S492F**
1609delCA	296+1G->T	4279insA	E56K	Q1382X	S4X
1677delTA (d)	297-1G->A	4326delTC	**E585X**	Q1411X	**S549N**
1717-1G->A (a) (d)	2991del32	4374+1G->A	**E60X** (d)	Q1412X	**S549R**
1717-8G->A	**3007delG**	4374+1G->T	**E822X**	**Q220X**	S912X
1782delA	3028delA	**4382delA**	**E831X**	**Q2X**	**S945L**
1811+1.6kbA->G	306delTAGA	4428insGA	**E92K**	**Q39X**	**T338I**
1812+1G->C	306 insA	442delA	**E92X** (d)	Q414X	**V520F**
1812-1G->A	**3120+1G->A**	**444delA**	**F508del** (a) (d)	**Q493X**	**W1089X**
1824delA	**3120G->A** (a)	**457TAT->G**	**G1061R**	Q525X	W1098X
182delT	**3121-1G->A**	541delC	**G1244E**	**Q552X**	W1145X
1833delT	3121-2A->G	**574delA**	**G1349D**	Q685X	W1204X
185+1G->T	3132delTG	**621+1G->T** (a) (d)	**G178R**	Q715X	**W1282X** (a) (d)
1898+1G->A (a)	**3171delC**	**663delT**	G27X	**Q890X**	W19X
1898+1G->C	3171insC	675del4	**G330X**	**Q98X**	W216X
1898+3A->G	3271delGG	**711+1G->T** (a)	**G542X** (a) (d)	**R1066C**	**W401X**
1924del7	**3272-26A->G** (d)	**711+3A->G**	G550X	**R1066H**	W496X
2055del9->A	3500-2A->G	**711+5G->A**	**G551D** (a) (d)	R1102X	W57X
2105-2117del13in-sAGAAA	365-366insT	**712-1G->T**	G551S	**R1158X**	**W846X**
2118del4	**3659delC** (a) (d)	**852del22**	G673X	**R1162X** (a) (d)	W882X
2143delT (d)	3667ins4	**935delA**	**G85E** (a) (d)	**R117C**	**Y1092X** (d)
2183AA->G or **2183delAA->G** (d)	3737delA	991del5	**G970R**	**R117H;5T** (a)	**Y122X**
2184delA (a) (d)	**3791delC**	**A455E** (a) (d)	H1054D	**R334W** (a) (d)	Y275X
2184insA (d)	3821delT	A46D	**H199Y**	**R347H**	**Y569D**
	3849+10kbC->T (a) (d)	**A559T**	**I1234V**	**R347P** (a) (d)	Y849X
	3849+4A->G	A561E	**I336K** (d)	**R352Q**	Y913X

Table 1. CFTR variants targeted by the Advanta CFTR NGS Library Prep Assay. Ordered by variant legacy name (alphanumerically). (a) indicates variants (23) within the ACMG/ACOG recommendations. (d) indicates the minimum variants (31) required by German guidelines. Bold indicates the inclusion of at least one variant-bearing sample in the analytical validation.

*The Clinical and Functional Translation of CFTR (CFTR2); available at cftr2.org (CF-causing variants from CFTR2_8August2016.xlsx)

Optimized Advanta CFTR NGS Library Prep Assay design supports target enrichment of all 27 CFTR exons and select intronic regions, which cover >250 variants derived from the CFTR2 mutation list*. Sequence the spectrum of variant types, from simple SNPs to large deletions, in a single NGS-based workflow.

Variant cDNA name

c.-9_14del23
c.(?_1)_(53+1_54-1)del
c.(273+1_274-1)_(1116+1_1117-1)del
c.(273+1_274-1)_(1116+1_1117-1)del(1584+1_1585-1)_(3468+1_3469-1)del
c.(53+1_54-1)_(164+1_165-1)del
c.(53+1_54-1)_(489+1_490-1)del
c.(743+1_744-1)_(1584+1_1585-1)dup
c.[350G>A;1210–12[5]] (a)
c.1000C>T (a) (d)
c.1006_1007insG
c.1007T>A (d)
c.1013C>T
c.1021T>C
c.1022_1023insTC
c.1029delC
c.1040G>A
c.1040G>C (a) (d)
c.1055G>A
c.1081delT
c.1116+1G>A
c.1117-1G>A
c.1127_1128insA
c.1153_1154insAT
c.115C>T
c.11C>A
c.1202G>A or **c.1203G>A**
c.1209+1G>A
c.1240C>T
c.137C>A
c.164+1G>A
c.164+1G>T
c.165-1G>A
c.166G>A
c.170G>A or c.171G>A
c.174_175insA
c.174_177delTAGA
c.178G>T (d)
c.1A>G
c.200C>T
c.223C>T

c.233dupT
c.254G>A (a) (d)
c.262_263delTT
c.263T>A or c.263T>G
c.273+1G>A
c.273+3A>C
c.274-1G>A
c.274G>A
c.274G>T (d)
c.292C>T
c.310delA
c.313delA
c.325_327delTATinsG
c.328G>C
c.349C>T
c.409delC
c.442delA
c.489+1G>T (a) (d)
c.4C>T
c.50delT
c.53+1G>T
c.531delT
c.532G>A
c.54-5940_273+10250del21kb (d)
c.543_546delTAGT
c.577G>T
c.579+1G>T (a)
c.579+3A>G
c.579+5G>A
c.57G>A
c.580-1G>T
c.595C>T
c.613C>T
c.617T>G
c.647G>A
c.658C>T
c.680T>G
c.720_741delAGGGAGAATGATGATGAAGTAC
c.79G>T
c.803delA
c.825C>G
c.828C>A
c.859_863delAACTT
c.948delT (d)

c.987delA
c.988G>T
c.(2619+1_2620-1)_(3367+1_3368-1)del
c.1329_1330insAGAT
c.1340delA
c.1364C>A (a) (d)
c.1365_1366delGG
c.1393-1G>A
c.1393-2A>G
c.1397C>A or **c.1397C>G**
c.1400T>C
c.1418delG
c.1466C>A
c.1475C>T
c.1477_1478delCA
c.1477C>T
c.1487G>A
c.1519_1521delATC (a) (d)
c.1521_1523delCTT (a) (d)
c.1545_1546delTA (d)
c.1558G>T
c.1572C>A
c.1573C>T
c.1585-1G>A (a) (d)
c.1585-8G>A
c.1624G>T (a) (d)
c.1645A>C or **c.1647T>G**
c.1646G>A
c.1648G>T
c.1650delA
c.1651G>A
c.1652G>A (a) (d)
c.1654C>T
c.1657C>T (a) (d)
c.1675G>A
c.1679+1.6kbA>G
c.1679+1G>A
c.1679G>A
c.1679G>C (a)
c.1680-1G>A
c.1680A>C
c.1682C>A
c.1692delA
c.1703delT
c.1705T>G

c.1753G>T
c.1766+1G->C
c.1766+1G>A (a)
c.1766+3A>G
c.1792_1798delAAAACTA
c.1923_1931del9insA
c.1973_1985del13insAGAAA
c.1986_1989delAACT
c.2012delT (d)
c.2017G>T
c.2051_2052delAAinsG (d)
c.2052_2053insA (d)
c.2052delA (a) (d)
c.2053_2054insC
c.2053C>T
c.2125C>T
c.2128A>T
c.2143C>T
c.2175_2176insA
c.2195T>G
c.2215delG
c.2240_2247delCGATACTG
c.2290C>T
c.2353C>T
c.2374C>T
c.2424_2425insAT
c.2453delT
c.2462_2463delGT
c.2464G>T
c.2490+1G>A
c.2491G>T
c.2537G>A
c.2547C>A
c.2551C>T
c.2583delT
c.2589_2599delAATTTGGTGCT
c.2600_2601insA
c.2645G>A
c.2657+5G>A (a) (d)
c.2658-1G>C
c.2668C>T
c.2735C>A

c.2737_2738insG
c.2739T>A
c.2764_2765insAG
c.(2988+1_2989-1)_(3367+1_3368-1)del
c.(2988+1_2989-1)_(3468+1_3469-1)del
c.(3963+1_3964-1)_(*1_?)del
c.2780T>C
c.2810_2811insT
c.2825delT
c.2834C>T
c.2859_2890delACATTCTGTTCTTCAAGCACCTATGTCAACCC
c.2875delG
c.2896delA
c.2908G>C
c.2988+1G>A
c.2988G>A (a)
c.2989-1G>A
c.2989-2A>G
c.3002_3003delTG
c.3039_3040insC
c.3039delC
c.3124C>T
c.3139_3139+1delGG
c.3140-26A>G (d)
c.3160C>G
c.3181G>C
c.3194T>C
c.3196C>T
c.3197G>A
c.3230T>C
c.3266G>A
c.3276C>A or **c.3276C>G** (d)
c.3293G>A or c.3294G>A
c.3302T>A (d)
c.3304A>T
c.3310G>T
c.3368-2A>G
c.3435G>A
c.3472C>T
c.3484C>T (a) (d)
c.3528delC (a) (d)

c.3535_3536insTCAA
c.3587C>G
c.3605delA
c.3611G>A or c.3612G>A
c.3659delC
c.366T>A
c.3691delT
c.3700A>G
c.3717+12191C>T (a) (d)
c.3717+4A>G
c.3718-1G>A
c.3718-3T>G
c.3731G>A
c.3744delA
c.3747delG
c.3752G>A
c.3761T>G
c.3763T>C
c.3764C>A
c.3773_3774insT (d)
c.3846G>A (a) (d)
c.3873+1G>A
c.3882_3885delTATT
c.3883delA
c.3884_3885insT
c.3890_3891insT
c.3899dupT
c.3908delA
c.3909C>G (a) (d)
c.3937C>T
c.3964-78_4242+577del
c.4046G>A
c.4077_4080delTGTTinsAA
c.4086_4087insT
c.4111G>T
c.4144C>T
c.4147_4148insA
c.4196_4197delTC
c.4231C>T
c.4234C>T
c.4242+1G>A
c.4242+1G>T
c.4251delA
c.4296_4297insGA

Table 2. CFTR variants targeted by the Advanta CFTR NGS Library Prep Assay. Ordered by variant cDNA name (5' to 3'). (a) indicates variants (23) within the ACMG/ACOG recommendations. (d) indicates the minimum variants (31) required by German guidelines. Bold indicates the inclusion of at least one variant-bearing sample in the analytical validation.

*The Clinical and Functional Translation of CFTR (CFTR2); available at cftr2.org (CF-causing variants from CFTR2_8August2016.xlsx)

Panel availability subject to confirmation.

For Research Use Only. Not for use in diagnostic procedures.

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