

Cystic fibrosis comprehensive gene sequencing

Division of Genome Diagnostics
at BC Children's and BC Women's Hospitals

CFTR regions sequenced: CFTR_ROIv1.1

Region	Target	Location	hg19 coordinates	Size (bp)
CFTR-E01	Exon 1	E1[122..10]	chr7:117120139-117120211	73
CFTR-E02	Exon 2	E2[-10..10]	chr7:117144297-117144427	131
CFTR-E03	Exon 3	E3[-10..10]	chr7:117149078-117149206	129
CFTR-E04	Exon 4	E4[-10..10]	chr7:117170943-117171178	236
CFTR-E05	Exon 5	E5[-10..10]	chr7:117174320-117174429	110
CFTR-E06	Exon 6	E6[-10..10]	chr7:117175292-117175475	184
CFTR-E07	Exon 7	E7[-10..10]	chr7:117176592-117176737	146
CFTR-E08	Exon 8	E8[-10..10]	chr7:117180144-117180410	267
CFTR-E09	Exon 9	E9[-10..10]	chr7:117182060-117182172	113
CFTR-E10	Exon 10	E10[-5..10]	chr7:117188690-117188887	198
CFTR-E11	Exon 11	E11[-10..10]	chr7:117199508-117199719	212
CFTR-E12	Exon 12	E12[-10..10]	chr7:117227783-117227897	115
CFTR-I12a	Intron 12	E13[-886..-972]	chr7:117229521-117229521	1
CFTR-I12b	Intron 12	E13[-877..-963]	chr7:117229530-117229530	1
CFTR-E13	Exon 13	E13[-10..10]	chr7:117230397-117230503	107
CFTR-E14	Exon 14	E14[-10..10]	chr7:117231978-117232721	744
CFTR-E15	Exon 15	E15[-10..10]	chr7:117234974-117235122	149
CFTR-E16	Exon 16	E16[-10..10]	chr7:117242870-117242927	58
CFTR-E17	Exon 17	E17[-10..10]	chr7:117243576-117243846	271
CFTR-E18	Exon 18	E18[-10..10]	chr7:117246718-117246817	100
CFTR-E19	Exon 19	E19[-10..10]	chr7:117250563-117250733	171
CFTR-I19	Intron 19	E20[-26..-253]	chr7:117251609-11725109	1
CFTR-E20	Exon 20	E20[-10..10]	chr7:117251625-117251872	248
CFTR-E21	Exon 21	E21[-10..10]	chr7:117254657-117254777	121
CFTR-E22	Exon 22	E22[-10..10]	chr7:117267566-117267834	269
CFTR-I22a	Intron 22	E22[288..40]	chr7:117267864-117267864	1
CFTR-I22b	Intron 22	E23[-2477..-2632]	chr7:117280015-117280015	1
CFTR-E23	Exon 23	E23[-10..10]	chr7:117282482-117282657	176
CFTR-E24	Exon 24	E24[-10..10]	chr7:117292886-117292995	110
CFTR-E25	Exon 25	E25[-10..10]	chr7:117304732-117304924	193
CFTR-E26	Exon 26	E26[-10..10]	chr7:117305503-117305628	126
CFTR-E27	Exon 27	E27[-10..-1546]	chr7:117306952-117307172	221

Reference sequence: NM_000492.3; Total size: 4983bp

This assay may not detect all insertions or deletions greater than 5 base pairs. Rare genetic abnormalities in these regions may be undetectable due to the technical limitations outlined on the genebc.ca website under "Cautions"

Assay QC metrics:

Analytical sensitivity for single nucleotide variants (95% CI): 100% (95.0-100%)

Analytical sensitivity for insertions and deletions (95% CI): 100% (95.6-100%)

Analytical specificity (95% CI): 100% (99.99-100%)

Minimum read depth: >100