

AmplideX[®]

PCR/CE *CFTR* Kit*



Offering the broadest coverage[†] of pathogenic *CFTR* variants of any commercially available, targeted testing kit

The AmplideX[®] PCR/CE *CFTR* Kit* brings the simplicity and scalability of AmplideX technology to *CFTR* variant detection. Offering the streamlined detection of 67 pathogenic variants, covering approximately 93%[†] of the US population, the assay provides the broadest coverage of any commercially available, targeted testing kit. The assay is optimized for use on widely established laboratory equipment and delivers genotype results from DNA in just under five hours.



Figure 1. AmplideX PCR/CE *CFTR* Kit* workflow.

Mix A				Mix B			
F508del	L206W	G85E	S945L	V456A	G970D	Q493X	Y122X
R117H	2789+5G>A	R347P/H	1898+1G>A	R117C	R1070W	R1158X	<i>CFTR</i> ddele20
W1282X	621+1G>T	2184insA	2184delA	2789+2insA	1154insTC	1811+1.6kbA>G	R75X
D1152H	3120+1G>A	A455E	711+1G>T	<i>CFTR</i> ddele2,3	1548delG	V520F	1898+5G>T
G542X	R553X	R347H/P	G622D	I618T	2183AA>G	1078delT	A559T
G551D	1717-1G>A	R560T	IVS8 PolyT	F311del	E60X	1677delTA	R352Q
R1070Q	I507del	3272-26A>G	IVS8 PolyTG	R1066C	P67L	M1101K	
N1303K	R334W	3659delC		3876delA	S549N	Q890X	
3849+10kbC>T	P5L	R1162X		394delTT	3905insT	Y1092X*	

RED variants indicate their inclusion in the ACMG23 listing.
*Detects both C>A and C>G

Table 1. List of variants detected by the AmplideX PCR/CE *CFTR* Kit*.

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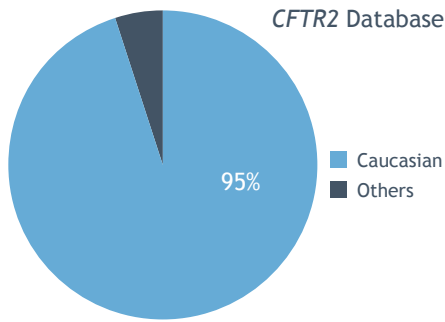


Figure 2A. Ethnic diversity of 89K patients in *CFTR2* database.

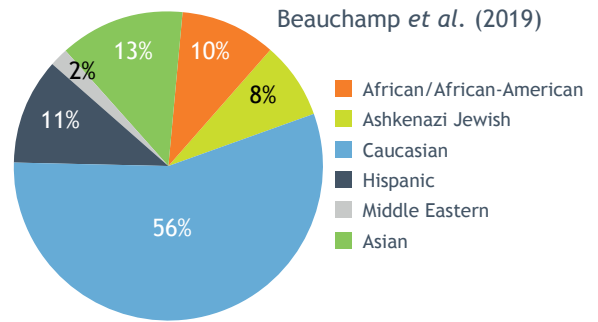


Figure 2B. Ethnic diversity of 115K subjects in US study.

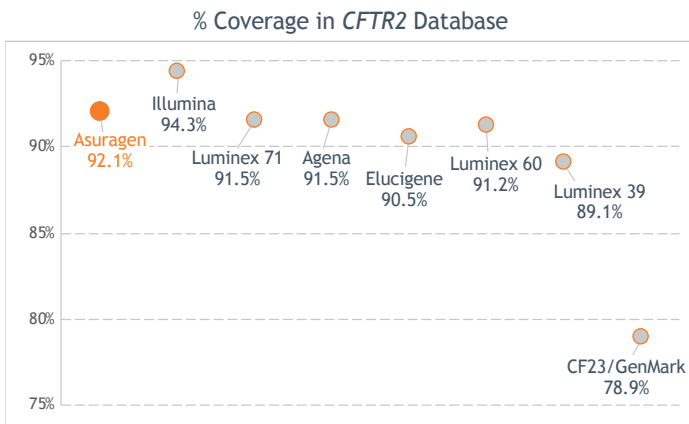


Figure 2C. Percent coverage of variants detected by commercial kits based on frequencies observed in the *CFTR2* database.

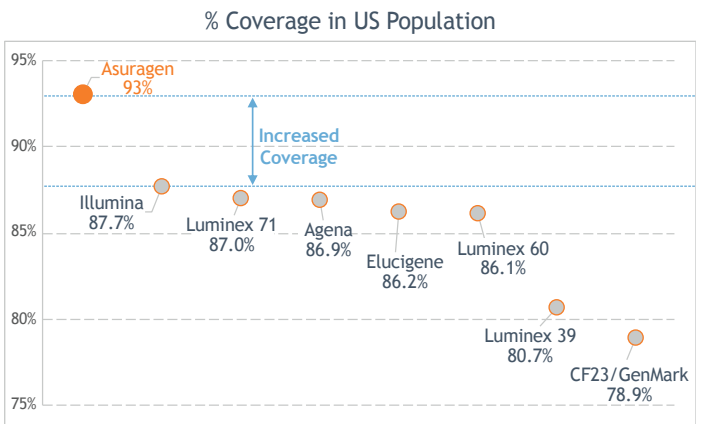


Figure 2D. Percent coverage of variants detected by commercial kits based on frequencies observed in the US population.

Assay Metric	Description
Assay Format	<ul style="list-style-type: none"> 2 PCR reactions covering 67 pathogenic variants in <i>CFTR</i> Gene Content includes variants representative of diverse ethnicities based on recent large-scale studies Similar workflow to AmplideX PCR/CE Kits (<i>FMR1</i>, <i>SMN1/2</i> Plus)
Output	<ul style="list-style-type: none"> Reports mutational status and zygosity of each variant included in the kit (SNPs, indels, repeats) PDF and CSV samples reports provides flexibility to customer Automated, push-button data reporting via AmplideX Reporter software
Sample Types	Blood
Turnaround Time	Less than 5 hours
System Compatibility	Applied Biosystems™ Genetic Analyzers
Catalog Number	50 Reaction Kit: A00076 • 100 Reaction Kit: A00077

Table 2. AmplideX PCR/CE *CFTR* Kit* Target Product Specifications.



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*Product in development. Specifications not finalized.
 †Beauchamp, et al. Genet Med. 2019 Nov;21(11):2569-2576.