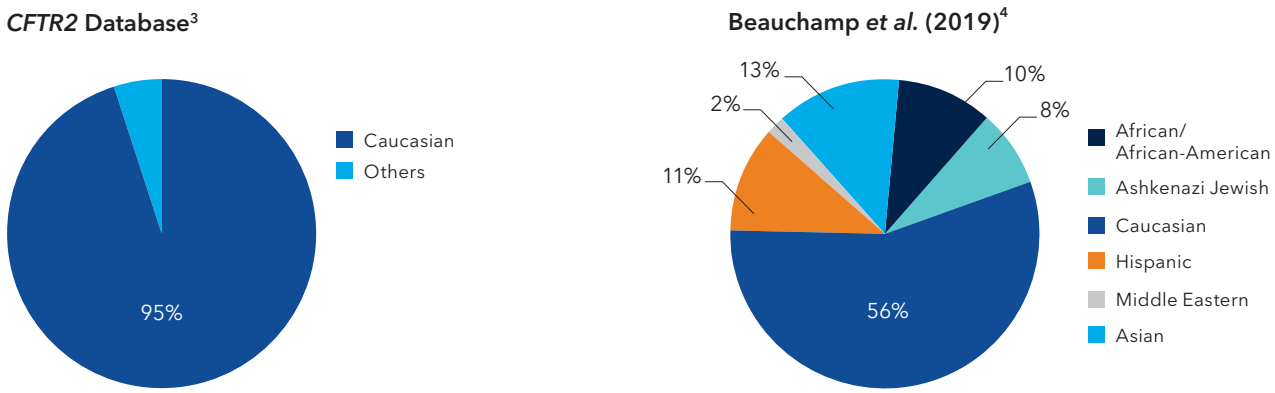
AmplideX® PCR/CE *CFTR* Kit* Solves Cystic Fibrosis Testing Coverage Challenges

Many commercially available *CFTR* panels were designed using variant information from disease databases that are heavily skewed toward individuals with European ancestry, making equitable coverage for ethnically diverse populations challenging. To address these coverage gaps, Asuragen, a Bio-Techne brand's AmplideX PCR/CE *CFTR* Kit was designed with diversity in mind and based on recent, large-scale population studies that include data from a range of ethnic groups more representative of the U.S. population.

Although more than 2100 *CFTR* variants have been documented,these variants differ in levels of pathogenicity and prevalence between ethnicities.[1] ACOG guidelines state full gene sequencing is inappropriate for screening and should be reserved for specific clinical cases, so it is paramount to have a commercially available, targeted panel with a selection of variants that provides equitable coverage and is less likely to miss carriers in diverse populations.[2]

Figure 1.

Most *CFTR* panels have been designed from this type of *CFTR*2 Database even though the U.S. population looks more like the Beauhchamp *et al.* database.

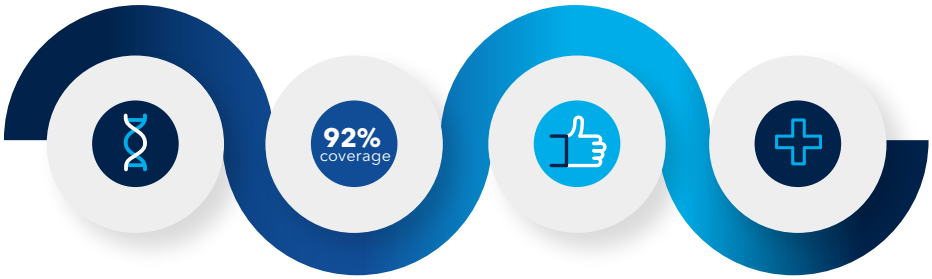


Asuragen Addresses Critical Coverage Gaps in *CFTR* Panels

Table 2.

Manufacturer	Product Name	Variants	Coverage, U.S. Population†	Coverage Difference, U.S. Population†
Asuragen	**AmplideX PCR/CE *CFTR* Kit**	**65**	**92.2%**	**-**
Illumina	MiSeqDx Cystic Fibrosis 139 Variant Assay	139	87.7%	~4% less
Agena	iPLEXPro *CFTR* Panel	74	86.9%	~5% less
Devyser	*CFTR* 68	68	87.1%	~5% less
ACMG 2023 Variant Set	-	100	86.4%	~6%
Elucigene	CF-EU2v1	50	86.2%	~6%
Luminex 60	xTAG Cystic Fibrosis (CFTR) 60 kit v2	60	86.1%	~6%

Designed to solve complex *CFTR* coverage challenges to provide the broadest coverage† of the U.S. population of any available targeted kit with an easy-to-use AmplideX® workflow and a complementary assay portfolio.



Reduced Complexity	Optimized Workflow	Quality Results
• Ready-to-use test kit with quality-controlled reagents reduces pipetting steps. • Similar workflow to AmplideX PCR/CE *FMR1** and *SMN1/2* Plus* kits eases implementation. • Streamlined data analysis via AmplideX Reporter software.	• Easy-to-use workflow designed to reduce hands-on tech time. • Utilizes widely available laboratory PCR/CE instrumentation. • <5 hrs from DNA to data.	• Built on the latest prevalence data to provide the best coverage† for all U.S. ethnicities.[4] • Detects complex yet key *CFTR* variants (STRs, SNPs, INDELs) and resolves zygosity. • Excellent concordance with other methods.

AmplideX PCR/CE *CFTR* Kit* Workflow

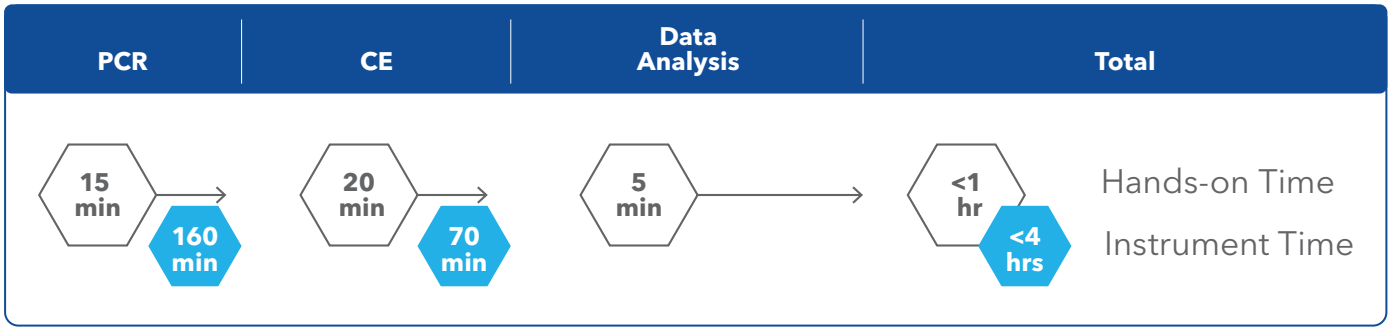


Figure 2. Hands-on time and instrument time for the AmplideX PCR/CE Kit.

Method Comparison Study

Sample level genotype agreement between AmplideX PCR/CE *CFTR* Kit and Reference Method.

Table 3A.

		Reference Genotype		
Sample Genotype Agreement		Homozygous WT	Heterozygous MUT	Homozygous MUT/ Compound HET/Multiple
AmplideX PCR/CE *CFTR* Kit	Homozygous WT	139	0	0
	Heterozygous MUT	0	466	0
	Homozygous MUT/Compound HET/Multiple	0	3	370
	Overall Sample Agreement	139/139 (100%)	466/469 (99.36%)	370/370 (100%)

Table 3A. Sample level agreement for 146 total samples (51 DBS, 91 whole blood and 4 cell lines) run on 7 CE configurations.

Table 3B.

		Reference Genotype		
Sample Genotype Agreement		Homozygous WT	Heterozygous MUT	Homozygous MUT/ Compound HET/Multiple
AmplideX PCR/CE *CFTR* Kit	Homozygous WT	1	0	0
	Heterozygous MUT	0	23	0
	Homozygous MUT/Compound HET/Multiple	0	0	23
	Overall Sample Agreement	1/1 (100%)	23/23 (100%)	23/23 (100%)

Table 3B. Sample level agreement for 47 cell line samples.

Ordering Information

Part Number	Product	Number of Reactions
A00519	AmplideX PCR/CE *CFTR* Kit	50
A00520	AmplideX PCR/CE *CFTR* Kit	100

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References

1. Russo ML. UpToDate. 2020.
2. Opinion 691. ACOG. 2017.
3. Clinical and Functional Translation of *CFTR* (*CFTR2*). www.CFTR2.org.
4. Based on data from Beauchamp KA, *et al.* Genet Med. 2019.

Bio-Techne® | R&D Systems™ Novus Biologicals™ Tocris Bioscience™ ProteinSimple™ ACD™ ExosomeDx™ Asuragen®

*For Research Use Only. Not for use in diagnostic procedures.†Based on pathogenic alleles in the U.S. demographic from Beauchamp KA, et al. Genet Med. 2019.

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