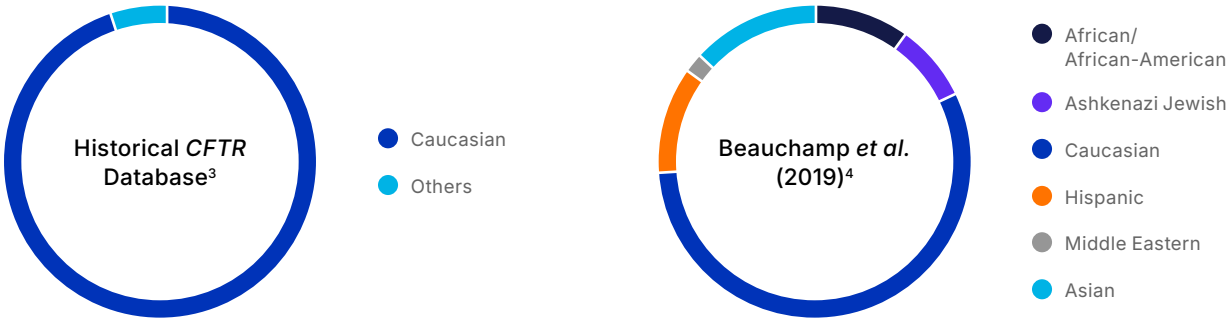
# AmplideX® PCR/CE *CFTR* Kit* Solves Cystic Fibrosis Testing Coverage Challenges

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 2,100 *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 // 01**

Most *CFTR* panels have been designed from *CFTR* Databases that were historically[3] skewed towards one ethnicity even though the U.S. population looks more like the Beauchamp *et al.* database.

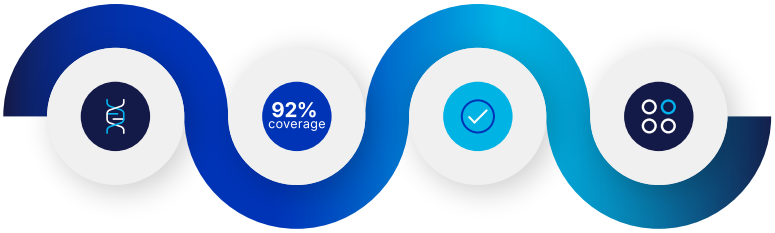


## Asuragen Addresses Critical Coverage Gaps in *CFTR* Panels

**TABLE // 01**

| 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

- Ready-to-use test kit with quality-controlled reagents reduces pipetting steps.
- Similar workflow to AmplideX PCR/CE *FMR1** and *SMN 1/2 Plus** kits eases implementation.
- Streamlined data analysis via AmplideX Reporter software.

### Optimized Workflow

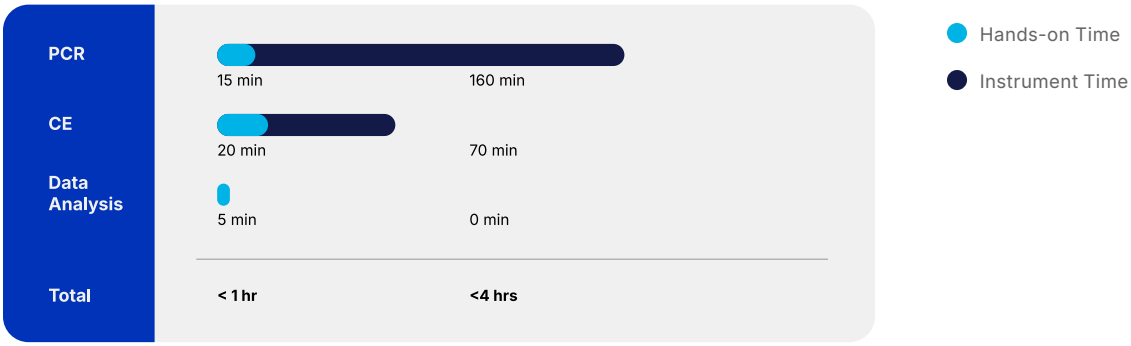
- Easy-to-use workflow designed to reduce hands-on tech time.
- Utilizes widely available laboratory PCR/CE instrumentation.
- <5 hours from DNA to data.

### Quality Results

- 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.

**FIGURE // 02**

AmplideX PCR/CE *CFTR* Kit* Workflow




Hands-on time and instrument time for the AmplideX PCR/CE *CFTR* Kit.

## Method Comparison Study

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

**TABLE // 02 A**

| | | 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%) |

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

**TABLE // 02 B**

| | | 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%) |

Sample level agreement for 47 cell lines.

## Ordering Information

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

**Contact Us:**
aus.orders@bio-techne.com

**REFERENCES**

1. Russo ML. UpToDate. 2020.
2. Opinion 691. ACOG. 2017.
3. Sosnay *et al.* Nat Genet. 2013 Oct; 45(10): 1160-1167
4. Based on data from Beauchamp KA, *et al.* Genet Med. 2019.



6394460491 2000-0193 Rev C