### appliedbiosystems

# CFTR gene testing using real-time PCR

A flexible, scalable, and low-cost real-time PCR solution to suit your *CFTR* gene testing needs

Combining Applied Biosystems<sup>™</sup> TaqMan<sup>®</sup> Genotyping Assays with the Applied Biosystems<sup>™</sup> QuantStudio<sup>™</sup> 12K Flex Real-Time PCR System delivers a sample-to-answer workflow for cystic fibrosis transmembrane conductance regulator (*CFTR*) mutation research. Features include:

- Fully qualified content—all TaqMan Assays demonstrate accurate and reproducible performance
- Multiple sample types—optimized for whole blood and buccal swabs
- Flexible formats—QuantStudio 12K Flex Real-Time PCR System provides breadth in content and the choice of four assay formats, including prespotted 384-well arrays and Applied Biosytems<sup>™</sup> OpenArray<sup>™</sup> plates

- Extensive menu of targets—design custom research panels from a menu of 199 *CFTR* gene targets, including full coverage of the ACMG-23 panel and majority coverage of the *CFTR2* mutation list
- Low cost per sample—significantly lower reagent costs with OpenArray nanofluidic technology



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#### Focusing on gene-related disease states

Inherited diseases like cystic fibrosis are genetically determined conditions stemming from a single gene mutation, a multifactorial inheritance, or a chromosomal abnormality. Researchers have identified over 1,000 disease-related mutations in the *CFTR* gene.

## Fully qualified *CFTR* assays to meet call rate and accuracy requirements

We continue to build new and valuable content to use on your Applied Biosystems<sup>™</sup> real-time PCR system, helping you to maximize the use of your instrument. All qualified assays are tested against extracted nucleic acids (Table 1), cell-line DNA, and synthetic controls.

#### Results you can trust

For our *CFTR* gene mutation detection assays, we've made it easy for you to implement this area of testing into your laboratory quicker and with confidence. These assays come complete with:

- Positive controls
- User guide detailing sample-to-answer workflow from whole blood and buccal swabs
- Analytical validation services

To view a full list of our *CFTR* gene mutation assays, download the menu from **thermofisher.com/cftr** 



## Table 1. High-performance results withOpenArray plates.

Performance metrics						
	Rep 1	Rep 2	Rep 3	Overall		
Call rate	99.6%	99.5%	99.7%	99.6%		
Accuracy	100%	99.99%	100%	99.99%		
Data points	8,100	8,100	8,100	24,300		

Study included 64 whole blood samples, including 28 CF-positive samples and 36 wild-type samples.

#### **Ordering information**

Product	Number of assays per plate	Number of samples per plate	Cat. No.
TaqMan OpenArray Genotyping Plate, Custom Format 32	26	96	4471114
TaqMan OpenArray Genotyping Plate, Custom Format 64	60	48	4471115
TaqMan OpenArray Genotyping Plate, Custom Format 128	120	24	4471116
TaqMan OpenArray Genotyping Plate, Custom Format 192	180	16	4471117
CFTR TaqMan Assay Controls	_	_	Contact sales representative

Design your own custom panel from a menu of 199 *CFTR* TaqMan gene mutation assays at **thermofisher.com/cftr** or contact your sales representative.



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