



# CFTR Mutation Analysis



## Flexible Solutions for CFTR Mutation Analysis

Cystic Fibrosis is one of the most common life-threatening genetic conditions, with as many as 1 in 25 people serving as a carrier for the autosomal recessive condition<sup>1,2</sup>. Cystic fibrosis transmembrane conductance regulator (CFTR) genotyping assays can be expensive and restrict assay content to a limited number of targets that are only applicable to a single population.

The MassARRAY® System with iPLEX® Pro CFTR Panel provides a fast and cost-effective way to detect 74 of the most common mutations of the CFTR gene.

- Includes mutations recommended by ACMG and ACOG as well as 51 variants with known CFTR relevance and allele frequency >0.1% (Table 1).
- Enables greater workflow efficiency with minimal manual processing - DNA to data in as little as 8 hours.
- Delivers simplified reporting and clear results with automated software provided at no additional cost.

For Research Use Only.  
Not for use in diagnostic procedures.

## iPLEX Pro CFTR Panel Targets

### 74 Total Targets

F508del	I507del	G542X	G85E	R560T	621+1G->T
711+1G->T	R334W	R347P	A455E	1717-1G->A	R553X
G551D	1898+1G->A	2184delA	2789+5G->A	3120+1G->A	3659delC
3849+10kbC->T	W1282X	N1303K	R117H	R1162X	1078delT
394delTT	Y122X	R347H	M1101K	S1255P/X	1898+5G->T
2183AA->G	2307insA	Y1092X	3876delA	3905insT	S549N
S549R_1645A->C	S549R-1647T->G	V520F	A559T	1677delTA	2055del9->A
2143delT	3199del6	3791delC	406-1G->A	935delA	D1152H
CFTRdele2,3	E60X	G178R	G330X	K710X	L206W
Q493X	Q890X	R1066C	R1158X	R75X	S1196X
W1089X	G1244E	G1349D	G551S	R1162L/Q	S1251N
R117C	F508C	I507V	I506V	T5/T7/T9	R560K

Society-recommended variants

Common variants (allele frequency >0.1%) with known CFTR relevance

## ORDERING INFORMATION

Catalog No.	Item	# Samples	Chip Format
13162	Complete CFTR Panel Set	320	96
13162F	Complete CFTR Panel Set CPM	320	96 CPM
13163	Complete CFTR Panel Set	1280	384
13163D	Complete CFTR Panel Set CPM	1280	384 CPM

*These panel sets contain assay specific primers and all the required reagents to process DNA samples on the MassARRAY system.*

## CUSTOM PANEL DESIGN

Don't see what you need? Agena Bioscience offers two options to create your CFTR custom panel. You can independently design content using our online assay design software, or partner with our scientists for assay development services.

## References

1. Grody WW, Cutting GR, Klinger KW, Richards CS, Watson MS, et al. (2001) Laboratory standards and guidelines for population-based cystic fibrosis carrier screening. *Genet Med* 3(2): 149-54
2. Langfelder-Schwind E, Kloza E, Sugarman E, Pettersen B, Brown T, et al. (2005) Cystic fibrosis prenatal screening in genetic counseling practice: recommendations of the National Society of Genetic Counselors. *JGenet Couns* 14(1):1-15

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