



## Cystic Fibrosis Mutation Analysis



### Flexible Solutions for Cystic Fibrosis Mutation Analysis

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

#### AGENA'S SOLUTION

➤ **iPLEX® Pro Cystic Fibrosis Panel**

The iPLEX Pro Cystic Fibrosis panel is a cost-effective way to detect 74 of the most common disease-causing mutations of the *CFTR* gene. It includes the 23 mutations recommended by the ACMG and ACOG and 49 variants with known *CFTR* relevance and allele frequency >0.1%.

➤ **Custom Panel Design**

Agena Bioscience offers two options for users interested in creating custom panels. You can independently design content using our online assay design software, or partner with our scientists for assay development and verification services.

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

## iPLEX Pro Cystic Fibrosis 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

23 ACMG/ACOG-recommended variants

49 of the other most common variants (allele frequency >0.1%) with known CFTR relevance

## ASSAY WORKFLOW

DNA to data in as little as 8 hours, with minimal manual processing time enables greater lab efficiency. Simplified reporting with automated software generates clear results.

## ORDERING INFORMATION

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

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

## 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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