HEREDITARY GENETICS

CFTR Mutation Analysis

All 100 ACMG *CFTR* Variants. Comprehensive Coverage. One Panel.

Cystic fibrosis is one of the most common life-threatening genetic conditions, with as many as 1 in 25 being a carrier for the autosomal recessive condition[1]. Many assays designed to detect variants in the cystic fibrosis transmembrane conductance regulator (*CFTR*) gene lack the minimum recommended content coverage outlined by leading societies and are restricted to a limited number of targets that are only applicable to a single population.

The CFTR 100+ Panel, run on the MassARRAY® System, provides a fast, cost-effective, high-throughput option for detecting American College of Medical Genetics and Genomics (ACMG)-recommended *CFTR* variants in a single panel.

- Includes all 100 variants recommended by ACMG[2], as well as 21 variants with known *CFTR* relevance, to ensure diverse population coverage.
- 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.

Agena® BIOSCIENCE

CFTR 100+ Panel

Variants (Legacy nomenclature)			
1078delT	3876delA	G628R	R347P
1154insTC	3905insT	G628R_G>C	R352Q
1161delC	3940delG	G85E	R553X
1288insTA	394delTT	G91R	R560T
1461ins4	4016insT	G970D	R560S
1504delG	406-1G>A	I336K	R709X
1525-1G>A	444delA	I506L	R75X
1677delTA	621+1G>T	I506V	R764X
1716+1G>A	711+1G>T	I507del	R785X
1717-1G>A	711+3A>G	I507V	R792X
1811+1634A>G	935delA	K710X	S1118F
1811+1G>A	A455E	L206W	S1255X
1811+1G>C	A559T	L218X	S466X
1824delA	A561E	L467P	S549N
1898+1G>A	A613T	L558S	S549R
1898+5G>T	C524X	M1101K	S945L
2055del9>A	CFTRdele2_3	N1303K	T1036N
2143delT	D110H	P67L	T338I
2183AA>G	D1152H	Q290X	V232D
2184delA	E585X	Q2X	V456A
2184insA	E60X	Q493X	V520F
2307insA	E725K	Q890X	W1089X
2622+1G>A	E92K	Q98R	W1098C
2789+5G>A	E92X	Q98X	W1204X
3120+1G>A	F191V	R1066C	W1282X
3120G>A	F508del	R1066H	Y569D
3199del6	F508C	R1158X	Y913X
3272-26A>G	G178R	R1162X	Y1092X_C>A
3659delC	G330X	R117C	Y1092X_C>G
3791delC	G542X	R117H	PolyT (5T)
3849+10kbC>T	G551D	R334W	PolyT (9T)
3849+5G>A	G551S	R347H	

Legend

ACMG-recommended *Supplemental* *Not reported; used for discrimination of other variants.*

ORDERING INFORMATION

Catalog No.	Item	# Samples	Chip Format
13402F	CFTR 100+ Panel Set - CPM (2x96)	32	96 CPM
13404F	CFTR 100+ Panel Set - CPM (10x96)	160	96 CPM
13403D	CFTR 100+ Panel Set - CPM (2x384)	128	384 CPM
13405D	CFTR 100+ Panel Set - CPM (10x384)	640	384 CPM

References

1. 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.
2. Deignan, J.L., et al. CFTR variant testing: a technical standard of the American College of Medical Genetics and Genomics (ACMG). *Genet Med* 22, 1285–1295 (2020).

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