

## Custom Cystic Fibrosis Analysis with the MassARRAY System

To date, independent laboratories have developed and tested over 100,000 *CFTR* samples using the MassARRAY System.

Agena Bioscience™ now offers custom services to aid in assay design for cystic fibrosis mutation analysis. Table 1 shows examples of *CFTR* mutations which have been tested against well-characterized *CFTR* HapMap samples from the Coriell Biorepository (MUTCF-2) plus additional Coriell samples known to harbor *CFTR* mutations, and showed 100% concordance with the Coriell data.<sup>6</sup>

Examples of Common <i>CFTR</i> Mutations Detectable with the MassARRAY System					
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	2055delI9->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

**Table 1**

- 23 ACMG/ACOG-recommended mutations
- 49 of the other most common variants (allele frequency >0.1%) with known *CFTR* relevance

*Additional mutations are also offered through Assays by Agena Custom Laboratory Services.*



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