The MiSeqDx Cystic Fibrosis 139-Variant Assay Offers the Largest Panel of Clinically Relevant *CFTR* Variants

General Population[1]				
M1V	1213delT	1898+3A>G	2347delG	M1101K
CFTRdele2,3	1248+1G>A	1717-8G>A	R764X	E1104X
Q39X	1259insA	**1717-1G>A**	2585delT	**3659delC**
G85E	W401X(c.1202G>A)	**G542X**	2622+1G>A	**3849+10kbC>T**
E92X	W401X(c.1203G>A)	S549R(c.1645A>C)	E831X	**W1282X**
Q98X	1341+1G>A	S549R(c.1647T>G)	R851X	Q1313X
R117H	1461ins4	S549N	**2789+5G>A**	4209TGTT>AA
621+1G>T	**A455E**	**G551D**	L927P	CFTRdele22,23
711+3A>G	L467P	**R553X**	3007delG	4382delA
R334W	S489X	R560K	G970R	*I506V*
S341P	**I507del**	R709X	3120G>A	*I507V*
R347H	**F508del**	**2184delA**	**3120+1G>A**	*F508C*
R347P	Q525X[2]	L732X	3121-1G>A	

Regional European[1]				
E60X	G178R	1525-1G>A	2184insA	W1089X
P67L	**711+1G>T**	Q493X	E822X	Y1092X(C>A)
R75X	712-1G>T	1677delTA	W846X	Y1092X(C>G)
394delTT	Q220X	V520F	2711delT	R1158X
405+1G>A	852del22	Q552X	Q890X	S1196X
E92K	1078delT	**R560T**	S945L	G1244E
457TAT>G	I336K	E585X	3272-26A>G	S1251N
D110H	T338I	**1898+1G>A**	L1065P	3905insT
R117C	1154insTC	2143delT	R1066C	4005+1G>A
Y122X	R352Q	K710X	R1066H	**N1303K**
574delA	*PolyTG/PolyT*	2183AA>G	L1077P	4016insT

Middle Eastern[1]	US Hispanic[1]	Hispanic[1]	African American[1]	Native American[1]
S466X(C>A)	406-1G>A	663delT	G330X	**R1162X**
S466X(C>G)	711+5G>A	H199Y	A559T	
1548delG[2]	1812-1G>A	P205S	2307insA	
	S492F	L206W	3791delC	
	W1204X (c.3611G>A)	1811+1.6kb A>G		
	W1204X (c.3612G>A)	3876delA		

Listed within each demographic by genomic coordinate order. **Bold** indicates that these mutations are part of the ACMG-23 list recommended for CF screening. *Italics* indicates that these mutations are conditionally reported.

References:

1. Demographic data source: Castellani C, Cuppens H, Macek Jr M, Cassiman JJ, Kerem E, et al. (2008) Consensus on the use and interpretation of cystic fibrosis mutation analysis in clinical practice. J Cyst Fibros 7: 179–196.
2. Mutation is classified in the CFTR2 database (www.cftr2.org) as a CF-causing variant while Sosnay et al. (Sosnay PR, Siklosi KR, Van Goor F, Kaniecki K,Yu H, et al. (2013 Defining the disease liability of variants in the cystic fibrosis transmembrane conductance regulator gene. Nat Genet. 45: 1160–1167.) classifies the variant as a mutation of unknown significance. The database classification is more current and reflects the completed functional testing which was not available at the time of the Sosnay publication.