

BC Newborn Screening Program

CFTR Mutation Panel

9 Nov 2009 – 14 Jul 2010 (24 mutations)	15 Jul 2010 – Current (38 mutations)
1078delT	1078delT
1717-1G>A	1717-1G>A
1898+1G>A	1898+1G>A
2184delA	2184delA
2789+5G>A	2789+5G>A
3120+1G>A	3120+1G>A
3659delC	3659delC
3849+10kbC>T	3849+10kbC>T
621+1G>T	621+1G>T
711+1G>T	711+1G>T
A455E	A455E
dF508	deltaF508
dI507	deltaI507
G542X	G542X
G551D	G551D
G85E	G85E
N1303K	N1303K
R1162X	R1162X
R117H	R117H
R334W	R334W
R347P	R347H
R553X	R553X
R560T	R560T
W1282X	W1282X
	2183AA>G
	3849+4A>G
	3876delA
	3905insT
	394delTT
	D1152H
	E60X
	Q493X
	R347P
	S549N
	S549R
	V520F
	Y1092X
	Y122X

- All newborn samples with an IRT value above the daily 97th percentile were tested for the CFTR (Cystic Fibrosis Transmembrane Regulator) mutations listed above as part of the Cystic Fibrosis screening program.
- The sample receipt date listed on the original newborn screening report can be used to identify the appropriate mutation panel.