

CFTR Variant Panel

- All newborn samples with an IRT value above the daily 97th percentile were tested for the CFTR (Cystic Fibrosis Transmembrane Regulator) variants listed below, 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 CFTR variant panel.

Last updated: October 2016

9 Nov 2009 – 14 Jul 2010 (24 variants)	15 Jul 2010 – 30 Sept 2016 (38 variants)	1 Oct 2016 – Present (130 variants)
G85E	G85E	G85E
R117H	R117H	R117H
621+1G>T	621+1G>T	621+1G>T
711+1G>T	711+1G>T	711+1G>T
1078delT	1078delT	1078delT
R334W	R334W	R334W
R347P	R347P	R347P
A455E	A455E	A455E
I507del	I507del	I507del
F508del	F508del	F508del
1717-1G>A	1717-1G>A	1717-1G>A
G542X	G542X	G542X
G551D	G551D	G551D
R553X	R553X	R553X
R560T	R560T	R560T
1898+1G>A	1898+1G>A	1898+1G>A
2184delA	2184delA	2184delA
2789+5G>A	2789+5G>A	2789+5G>A
3120+1G>A	3120+1G>A	3120+1G>A
R1162X	R1162X	R1162X
3659delC	3659delC	3659delC
3849+10kbC>T	3849+10kbC>T	3849+10kbC>T
W1282X	W1282X	W1282X
N1303K	N1303K	N1303K
	Q493X	Q493X
	S549N	S549N
	3905insT	3905insT
	E60X	E60X
	Y1092X C>A	Y1092X C>A
	2183AA>G	2183AA>G
	V520F	V520F

BC Newborn Screening Program



S549R	S549R
R347H	R347H
3876delA	3876delA
394delTT	394delTT
Y122X	Y122X
D1152H	NOT IN PANEL
3849+4A>G	NOT IN PANEL
	M1V
	CFTRdele2,3
	Q39X
	P67L
	R75X
	405+1G>A
	406-1G>A
	E92X
	E92K
	Q98X
	457TAT>G
	D110H
	R117C
	663delT
	G178R
	711+3A>G
	711+5G>A
	712-1G>T
	H199Y
	P205S
	L206W
	Q220X
	852del22
	G330X
	I336K
	T338I
	S341P
	1154insTC
	R352Q
	1213delT
	1248+1G>A
	1259insA
	W401X
	1341+1G>A

BC Newborn Screening Program



1461ins4
1525-1G>A
S466X
L467P
1548delG
S489X
S492F
1677delTA
Q525X
1717-8G>A
Q552X
A559T
R560K
1181+1.6kbA>G
1812-1G>A
E585X
1898+3A>G
2143delT
2184insA
R709X
K710X
2307insA
L732X
2347delG
R764X
2585delT
E822X
2622+1G>A
E831X
W846X
R851X
2711delT
Q890X
L927P
S945L
3007delG
G970R
3120G>A
3121-1G>A
3272-26A>G
L1065P

BC Newborn Screening Program



R1066C
R1066H
L1077P
W1089X
M1101K
E1104X
R1158X
S1196X
W1204X
3791delC
G1244E
S1251N
4005+1G>A
4016insT
Q1313X
4209TGTT>AA
CFTRdele22,23
4382delA