BC Newborn Screening Program

Cystic fibrosis targeted 130 variant assay

Active: 1 Oct 2016 – Present

Last Updated: August 11, 2020

Legacy Name	cDNA	Predicted Protein
M1V	c.1A>G	p.? [1]
Q39X	c.115C>T	p.Gln39Ter
E60X	c.178G>T	p.Glu60Ter
P67L	c.200C>T	p.Pro67Leu
R75X	c.223C>T	p.Arg75Ter
G85E	**c.254G>A**	**p.Gly85Glu**
394delTT	c.262_263delTT	p.Leu88IlefsTer22
405+1G>A	c.273+1G>A	p.?
406-1G>A	c.274-1G>A	p.?
E92K	c.274G>A	p.Glu92Lys
E92X	c.274G>T	p.Glu92Ter
Q98X	c.292C>T	p.Gln98Ter
457TAT>G	c.325_327delinsG	p.Tyr109GlyfsTer4
D110H	c.328G>C	p.Asp110His
R117C	c.349C>T	p.Arg117Cys
R117H	**c.350G>A**	**p.Arg117His**
Y122X	c.366T>A	p.Tyr122Ter
574delA	c.442delA	p.Ile148LeufsTer5
621+1G>T	**c.489+1G>T**	**p.?**
663delT	c.531delT	p.Ile177MetfsTer12
G178R	c.532G>A	p.Gly178Arg
711+1G>T	**c.579+1G>T**	**p.?**
711+3A>G	c.579+3A>G	p.?
711+5G>A	c.579+5G>A	p.?
712-1G>T	c.580-1G>T	p.?
H199Y	c.595C>T	p.His199Tyr
P205S	c.613C>T	p.Pro205Ser
L206W	c.617T>G	p.Leu206Trp
Q220X	c.658C>T	p.Gln220Ter
852del22	c.723_743+1del [2]	p.? [3]

[1] Alternative historical name for this variant is p.Met1Val

[2] Full variant name is c.723_743+1delGAGAATGATGATGAAGTACAGG

[3] Alternative historical name for this variant is p.Gly241GlufsTer13

Legacy Name	cDNA	Predicted Protein
1078delT	c.948delT	p.Phe316LeufsTer12
G330X	c.988G>T	p.Gly330Ter
R334W	**c.1000C>T**	**p.Arg334Trp**
I336K	c.1007T>A	p.Ile336Lys
T338I	c.1013C>T	p.Thr338Ile
S341P	c.1021T>C	p.Ser341Pro
1154insTC	c.1021_1022dupTC	p.Phe342HisfsTer28
R347H	c.1040G>A	p.Arg347His
R347P	**c.1040G>C**	**p.Arg347Pro**
R352Q	c.1055G>A	p.Arg352Gln
1213delT	c.1081delT	p.Trp361GlyfsTer8
1248+1G>A	c.1116+1G>A	p.?
1259insA	c.1130dupA	p.Gln378AlafsTer4
W401X	c.1202G>A	p.Trp401Ter
W401X	c.1203G>A	p.Trp401Ter
1341+1G>A	c.1209+1G>A	p.?
1461ins4	c.1327_1330dupGATA	p.Ile444ArgfsTer3
A455E	**c.1364C>A**	**p.Ala455Glu**
1525-1G>A	c.1393-1G>A	p.?
S466X	c.1397C>A	p.Ser466Ter
S466X	c.1397C>G	p.Ser466Ter
L467P	c.1400T>C	p.Leu467Pro
1548delG	c.1418delG	p.Gly473GlufsTer54
S489X	c.1466C>A	p.Ser489Ter
S492F	c.1475C>T	p.Ser492Phe
Q493X	c.1477C>T	p.Gln493Ter
I507del	**c.1519_1521delATC**	**p.Ile507del**
F508del	**c.1521_1523delCTT**	**p.Phe508del**
1677delTA	c.1545_1546delTA	p.Tyr515Ter
V520F	c.1558G>T	p.Val520Phe
Q525X	c.1573C>T	p.Gln525Ter
1717-1G>A	**c.1585-1G>A**	**p.?**
1717-8G>A	c.1585-8G>A	p.?
G542X	**c.1624G>T**	**p.Gly542Ter**
S549R	c.1645A>C	p.Ser549Arg
S549N	c.1646G>A	p.Ser549Asn
S549R	c.1647T>G	p.Ser549Arg
G551D	**c.1652G>A**	**p.Gly551Asp**
Q552X	c.1654C>T	p.Gln552Ter
R553X	**c.1657C>T**	**p.Arg553Ter**

Legacy Name	cDNA	Predicted Protein
A559T	c.1675G>A	p.Ala559Thr
1811+1.6kbA>G	c.1680-886A>G	p.?
R560K	c.1679G>A	p.Arg560Lys
R560T	**c.1679G>C**	**p.Arg560Thr**
1812-1G>A	c.1680-1G>A	p.?
E585X	c.1753G>T	p.Glu585Ter
1898+1G>A	**c.1766+1G>A**	**p.?**
1898+3A>G	c.1766+3A>G	p.?
2143delT	c.2012delT	p.Leu671Ter
2183AA>G	c.2051_2052delinsG	p.Lys684SerfsTer38
2184insA	c.2052dupA	p.Gln685ThrfsTer4
2184delA	**c.2052delA**	**p.Lys684AsnfsTer38**
R709X	c.2125C>T	p.Arg709Ter
K710X	c.2128A>T	p.Lys710Ter
2307insA	c.2175dupA	p.Glu726ArgfsTer4
L732X	c.2195T>G	p.Leu732Ter
2347delG	c.2215delG	p.Val739TyrfsTer16
R764X	c.2290C>T	p.Arg764Ter
2585delT	c.2453delT	p.Leu818TrpfsTer3
E822X	c.2464G>T	p.Glu822Ter
2622+1G>A	c.2490+1G>A	p.?
E831X	c.2491G>T	p.Glu831Ter
W846X	c.2537G>A	p.Trp846Ter
R851X	c.2551C>T	p.Arg851Ter
2711delT	c.2583delT	p.Phe861LeufsTer3
2789+5G>A	**c.2657+5G>A**	**p.?**
Q890X	c.2668C>T	p.Gln890Ter
L927P	c.2780T>C	p.Leu927Pro
S945L	c.2834C>T	p.Ser945Leu
3007delG	c.2875delG	p.Ala959HisfsTer9
G970R	c.2908G>C	p.Gly970Arg
3120G>A	c.2988G>A	p.?
3120+1G>A	**c.2988+1G>A**	**p.?**
3121-1G>A	c.2989-1G>A	p.?
3272-26A>G	c.3140-26A>G	p.?
L1065P	c.3194T>C	p.Leu1065Pro
R1066C	c.3196C>T	p.Arg1066Cys
R1066H	c.3197G>A	p.Arg1066His
L1077P	c.3230T>C	p.Leu1077Pro
W1089X	c.3266G>A	p.Trp1089Ter

Legacy Name	cDNA	Predicted Protein
Y1092X	c.3276C>A	p.Tyr1092Ter
Y1092X	c.3276C>G	p.Tyr1092Ter
M1101K	c.3302T>A	p.Met1101Lys
E1104X	c.3310G>T	p.Glu1104Ter
R1158X	c.3472C>T	p.Arg1158Ter
R1162X	**c.3484C>T**	**p.Arg1162Ter**
3659delC	**c.3528delC**	**p.Lys1177SerfsTer15**
S1196X	c.3587C>G	p.Ser1196Ter
W1204X	c.3611G>A	p.Trp1204Ter
W1204X	c.3612G>A	p.Trp1204Ter
3791delC	c.3659delC	p.Thr1220LysfsTer8
3849+10kbC>T	**c.3718-2477C>T**	**p.?**
G1244E	c.3731G>A	p.Gly1244Glu
3876delA	c.3744delA	p.Lys1250ArgfsTer9
S1251N	c.3752G>A	p.Ser1251Asn
3905insT	c.3773dupT	p.Leu1258PhefsTer7
W1282X	**c.3846G>A**	**p.Trp1282Ter**
4005+1G>A	c.3873+1G>A	p.?
4016insT (4021dupT)	c.3889dupT	p.Ser1297PhefsTer5
N1303K	**c.3909C>G**	**p.Asn1303Lys**
Q1313X	c.3937C>T	p.Gln1313Ter
4209TGTT>AA	c.4077_4080delinsAA	p.Val1360ThrfsTer3
4382delA	c.4251delA	p.Glu1418ArgfsTer14
CFTRdele2,3	c.54-5940_273+10250del	p.? [4]
CFTRdele22,23	c.3964-78_4242+577del	p.?

HGVS nomenclature version 20.05

- Reference sequences: coding variants NM_000492.3, NP_000483.3
- Reference sequences: non-coding variants NC_000007.13 (NM_000492.3)

Bold = ACMG (2004) 23 mutation panel

The length of the poly-T tract of intron 8 is reported according to published guidelines.

[4] Alternative historical name for this variant is p.Ser18ArgfsX16