NYS Cystic Fibrosis Newborn Screening: Custom *CFTR* Variant Panel Content

Variant: Legacy | HGVS Protein (cDNA)
124del23bp | p.? (c.-9_14del)
M1L | p.Met1? (c.1A>C)
M1V | p.Met1Val / p.Met1? (c.1A>G)
M1K | p.Met1? (c.2T>A)
M1T | p.? (c.2T>C)
M1R | p.? (c.2T>G)
M1I | p.? (c.3G>A)
c.4delC | p.Gln2Argfs*23 (c.4del)
Q2X | p.Gln2* (c.4C>T)
S4X | p.Ser4* (c.11C>A)
156insG | p.Ala9Glyfs*36 (c.25dup)
S13F | p.Ser13Phe (c.38C>T)
c.40_44delAAACT | p.Lys14Phefs*29 (c.40_44del)
K14X | p.Lys14* (c.40A>T)
174delA | p.Lys14Asnfs*11 (c.42del)
175delC | p.Leu15Phefs*10 (c.43del)
c.43dupC | p.Leu15Profs*30 (c.43dup)
181_182dup | p.Trp19Alafs*7 (c.49_50dup)
c.49_50delTT | p.Phe17Glnfs*27 (c.49_50del)
175insT | p.Ser18Glnfs*27 (c.50dup)
182delT | p.Phe17Serfs*8 (c.50del)
L15P | p.Leu15Pro (c.44T>C)
183delC | p.Phe17Leufs*8 (c.51del)
185+1G->T | p.? (c.53+1G>T)
185+2T->C | p.? (c.53+2T>C)
185+2T->G | p.? (c.53+2T>G)
186-2A->G | p.? (c.54-2A>G)
186-1G->A | p.? (c.54-1G>A)
W19X | p.Trp19* (c.56G>A)
W19X | p.Trp19* (c.57G>A)
c.71_72delTGinsA | p.Leu24* (c.71_72delinsA)
G27R | p.Gly27Arg (c.79G>A)
G27X | p.Gly27* (c.79G>T)
211delG | p.Gly27Aspfs*64 (c.80del)[#]
218insA | p.Gln30Thrfs*15 (c.87dup)
Q30X | p.Gln30* (c.88C>T)
c.88_89delCA | p.Gln30Alafs*14 (c.88_89del)
E33X | p.Glu33* (c.97G>T)
241delAT | p.Tyr38Profs*6 (c.112_113del)
Y38X | p.Tyr38* (c.114C>G)
Q39X | p.Gln39* (c.115C>T)
A46D | p.Ala46Asp (c.137C>A)
c.142_145delAATC | p.Asn48Tyrfs*42 (c.142_145del)
c.147_150delATCT | p.Ser50Lysfs*40 (c.147_150del)
284delA | p.Lys52Asnfs*39 (c.156del)
295ins8 | p.Arg55Asnfs*39 (c.156_163dup)
K52X | p.Lys52* (c.154A>T)
E54X | p.Glu54* (c.160G>T)
296+1G->A | p.? (c.164+1G>A)
296+1G->C | p.? (c.164+1G>C)
296+1G->T | p.? (c.164+1G>T)
296+2T->A | p.? (c.164+2T>A)
296+2T->C | p.? (c.164+2T>C)
296+2T->G | p.? (c.164+2T>G)
296+3insT | p.? (c.164+4dup)
297-3C->T | p.? (c.165-3C>T)
297-2A->G | p.? (c.165-2A>G)
297-1G->A | p.? (c.165-1G>A)
E56K | p.Glu56Lys (c.166G>A)
E56X | p.Glu56* (c.166G>T)
300delA | p.Glu56Aspfs*35 (c.168del)
W57G | p.Trp57Gly (c.169T>G)
W57X | p.Trp57* (c.170G>A)
W57X | p.Trp57* (c.171G>A)
305insT | p.Arg59* (c.174dup)
306delTAGA | p.Asp58Glufs*32 (c.174_177del)
306insA | p.Arg59Lysfs*10 (c.175dup)

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308insA | p.Glu60Argfs*9 (c.177dup)
E60K | p.Glu60Lys (c.178G>A)
E60X | p.Glu60* (c.178G>T)
317insC | p.Ser63Phefs*6 (c.185dup)
P67L | p.Pro67Leu (c.200C>T)
K68X | p.Lys68* (c.202A>T)
R75X | p.Arg75* (c.223C>T)
365-366insT | p.Trp79Leufs*32 (c.233dup)
c.234delC | p.Trp79Glyfs*12 (c.234del)
366insC | p.Trp79Leufs*32 (c.234dup)
W79X | p.Trp79* (c.236G>A)
c.243delT | p.Phe81Leufs*10 (c.243del)
379-381insT | p.Tyr84Leufs*27 (c.248dup)
Y84X | p.Tyr84* (c.252T>A)
G85X | p.Gly85* (c.253G>T)
G85E | p.Gly85Glu (c.254G>A)
394delTT | p.Leu88Ilefs*22 (c.262_263del)
L88X | p.Leu88* (c.263T>A)
L88X | p.Leu88* (c.263T>G)
c.264_268delATATT | p.Leu88Phefs*21 (c.264_268del)
G91R | p.Gly91Arg (c.271G>A)
405+1G->A | p.? (c.273+1G>A)
405+2T->G | p.? (c.273+2T>G)
405+3A->C | p.? (c.273+3A>C)
406-2A->C | p.? (c.274-2A>C)
406-2A->G | p.? (c.274-2A>G)
406-1G->A | p.? (c.274-1G>A)
406-1G->C | p.? (c.274-1G>C)
E92K | p.Glu92Lys (c.274G>A)
E92X | p.Glu92* (c.274G>T)
415insA | p.Ala96Serfs*15 (c.285dup)
Q98X | p.Gln98* (c.292C>T)
Q98R | p.Gln98Arg (c.293A>G)
P99L | p.Pro99Leu (c.296C>T)
L101X | p.Leu101* (c.302T>G)
435insA | p.Leu102Thrfs*9 (c.303dup)
L102R | p.Leu102Arg (c.305T>G)
G103X | p.Gly103* (c.307G>T)
442delA | p.Arg104Glufs*3 (c.310del)
R104X | p.Arg104* (c.310A>T)
444delA | p.Ile105Serfs*2 (c.313del)
c.324delC | p.Tyr109Metfs*15 (c.324del)
457TAT->G | p.Tyr109Glyfs*4 (c.325_327delinsG)
458delAT | p.Tyr109* (c.326_327del)
Y109X | p.Tyr109* (c.327T>A)
D110H | p.Asp110His (c.328G>C)
464delC | p.Pro111Argfs*13 (c.332del)
K114X | p.Lys114* (c.340A>T)
E116K | p.Glu116Lys (c.346G>A)
R117C | p.Arg117Cys (c.349C>T)
R117H | p.Arg117His (c.350G>A)[f]
R117P | p.Arg117Pro (c.350G>C)
c.353delC | p.Ser118Leufs*6 (c.353del)
489delC | p.Ile119Metfs*5 (c.357del)
Y122X | p.Tyr122* (c.366T>A)
499dupC | p.Leu123Profs*36 (c.367dup)
G126D | p.Gly126Asp (c.377G>A)
c.381_382dup | p.Cys128Tyrfs*7 (c.381_382dup)
519delT | p.Leu130Serfs*4 (c.387del)
525delT | p.Phe131Leufs*3 (c.393del)
526_527delAT | p.Ile132Glufs*25 (c.394_398del)
538insAC | p.Leu136Hisfs*18 (c.405_406dup)
541delC | p.Leu137Serfs*16 (c.409del)
541del4 | p.Leu137Tyrfs*15 (c.409_412del)
542del7 | p.Leu137Profs*14 (c.410_416del)
L138ins | p.Leu138dup (c.413_415dup)
547insTA | p.His139Leufs*15 (c.415_416insTA)
H139R | p.His139Arg (c.416A>G)

Variant: Legacy | HGVS Protein (cDNA)
556delA | p.Ile142Phefs*11 (c.424del)
557delT | p.Phe143Leufs*10 (c.429del)
565delC | p.Leu145Phefs*8 (c.433del)
574delA | p.Ile148Leufs*5 (c.442del)
G149R | p.Gly149Arg (c.445G>A)
c.450delG | p.Met150Ilefs*3 (c.450del)
582insG | p.Gln151Alafs*8 (c.450dup)
583delC | p.Gln151Argfs*2 (c.451del)
Q151X | p.Gln151* (c.451C>T)
593insT | p.Ala155Serfs*4 (c.461dup)
602del14 | p.Phe157* (c.470_483del)
604insA | p.Ser158Lysfs*5 (c.472dup)
605insT | p.Leu159Phefs*4 (c.476dup)
L159X | p.Leu159* (c.476T>A)
Y161D | p.Tyr161Asp (c.481T>G)
c.488delA | p.Lys163Argfs*3 (c.488del)
621+1G->A | p.? (c.489+1G>A)
621+1G->T | p.? (c.489+1G>T)
621+2T->C | p.? (c.489+2T>C)
621+2T->G | p.? (c.489+2T>G)
622-2A->C | p.? (c.490-2A>C)
622-1G->A | p.? (c.490-1G>A)
622-1G->C | p.? (c.490-1G>C)
622-1G->T | p.? (c.490-1G>T)
L165S | p.Leu165Ser (c.494T>C)
624delT | p.Leu165* (c.494del)
L165X | p.Leu165* (c.494T>A)
630delG | p.Lys166Asnfs*7 (c.498del)
654del5 | p.Ile175Tyrfs*6 (c.522_526del)
663delT | p.Ile177Metfs*12 (c.531del)
663insT | p.Gly178Trpfs*5 (c.531dup)
G178R | p.Gly178Arg (c.532G>A)
Q179X | p.Gln179* (c.535C>T)
675del4 | p.Leu183Phefs*5 (c.543_546del)
675del14 | p.Ser182Glnfs*6 (c.543_556del)
680delT | p.Leu183Profs*6 (c.548del)
c.551_555delTTTCC | p.Leu184Glnfs*7 (c.551_555del)
681delC | p.Leu184Phefs*5 (c.550del)
690delC | p.Asn186Lysfs*3 (c.558del)
c.560delA | p.Asn187Thrfs*2 (c.560del)
F191V | p.Phe191Val (c.571T>G)
D192G | p.Asp192Gly (c.575A>G)
708delT | p.Asp192Glufs*23 (c.576del)
E193K | p.Glu193Lys (c.577G>A)
E193X | p.Glu193* (c.577G>T)
710_711+5del7 | p.? (c.578_579+5del)
710del4 | p.? (c.579_579+3del)
711+1G->T | p.? (c.579+1G>T)
711+2T->C | p.? (c.579+2T>C)
711+3A->G | p.? (c.579+3A>G)
711+5G->A | p.? (c.579+5G>A)
712-2A->G | p.? (c.580-2A>G)
712-2A->C | p.? (c.580-2A>C)
712-1G->T | p.? (c.580-1G>T)
G194R | p.Gly194Arg (c.580G>A)
G194R | p.Gly194Arg (c.580G>C)
G194X | p.Gly194* (c.580G>T)
713delGA | p.Gly194Alafs*63 (c.581_582del)
c.583delC | p.Ala196Hisfs*19 (c.583del)
H199Y | p.His199Tyr (c.595C>T)
733delG | p.Val201Cysfs*14 (c.601del)
c.604_605delTG | p.Trp202Aspfs*55 (c.604_605del)
W202X | p.Trp202* (c.606G>A)
P205S | p.Pro205Ser (c.613C>T)
746insC | p.Leu206Phefs*52 (c.614dup)
749delT | p.Leu206Cysfs*9 (c.617del)
L206W | p.Leu206Trp (c.617T>G)
Q207X | p.Gln207* (c.619C>T)
W216X | p.Trp216* (c.647G>A)
W216X | p.Trp216* (c.648G>A)
c.650_659del | p.Glu217Glyfs*11 (c.650_659del)
L218X | p.Leu218* (c.653T>A)

Variant: Legacy | HGVS Protein (cDNA)
Q220X | p.Gln220* (c.658C>T)
C225X | p.Cys225* (c.675T>A)
L227R | p.Leu227Arg (c.680T>G)
V232D | p.Val232Asp (c.695T>A)
840insT | p.Gln237Serfs*21 (c.708dup)
840delT | p.Gln237Argfs*4 (c.708del)
Q237X | p.Gln237* (c.709C>T)
845_846insA | p.Gly239Trpfs*19 (c.713_714insA)
L240X | p.Leu240* (c.714del)
849delG | p.Leu240* (c.717del)
852del22 | p.? (c.723_743+1del)
Y247X | p.Tyr247* (c.741C>A)
Y247X | p.Tyr247* (c.741C>G)
875+1G->A | p.? (c.743+1G>A)
875+1G->C | p.? (c.743+1G>C)
875insTACA | p.? (c.743_743+1insTACA)
875+2T->A | p.? (c.743+2T>A)
875+2T->C | p.? (c.743+2T>C)
876-2A->G | p.? (c.744-2A>G)
Q250X | p.Gln250* (c.748C>T)
892delA | p.Lys254Argfs*7 (c.761del)
896delT | p.Ile255Thrfs*6 (c.764del)
905delG | p.Arg258Asnfs*3 (c.773del)
c.775delinsGCTGGGAAGAT | p.Leu259Alafs*12 (c.775delinsGCTGGGAAGAT)
909delT | p.Val260* (c.777del)
935delA | p.Asn268Ilefs*17 (c.803del)
936delTA | p.Ile269Profs*4 (c.805_806del)
Q270X | p.Gln270* (c.808C>T)
c.811delT | p.Ser271Leufs*14 (c.811del)
954delA | p.Tyr275Thrfs*10 (c.822del)
Y275X | p.Tyr275* (c.825C>G)
C276X | p.Cys276* (c.828C>A)
W277X | p.Trp277* (c.830G>A)
W277X | p.Trp277* (c.831G>A)
E279X | p.Glu279* (c.835G>T)
982delA | p.Met284* (c.850del)
977insA | p.Met284Asnfs*3 (c.850dup)
991del5 | p.Asn287Lysfs*19 (c.861_865del)
994del9 | p.? (c.863_869+2del)
R289X | p.Arg289* (c.865A>T)
c.865_869delAGACA | p.Arg289Asnfs*17 (c.865_869del)
Q290X | p.Gln290* (c.868C>T)
1001+1G->C | p.? (c.869+1G>C)
1001+2T->A | p.? (c.869+2T>A)
1001+2T->G | p.? (c.869+2T>G)
1002-2A->C | p.? (c.870-2A>C)
1002-2A->G | p.? (c.870-2A>G)
E292X | p.Glu292* (c.874G>T)
1006_1007delGA | p.Glu292Thrfs*15 (c.874_875del)
1027delG | p.Ala299Glnfs*4 (c.895del)
1037insA | p.Arg303Glufs*5 (c.905_906insA)
1040del4 | p.Arg303Thrfs*24 (c.908_911del)
Y304X | p.Tyr304* (c.912C>G)
c.912delCinsGG | p.Tyr304* (c.912delinsGG)
1058delC | p.Phe310Serfs*18 (c.927del)
F311L | p.Phe311Leu (c.933C>A)
F311L | p.Phe311Leu (c.933C>G)
F311del / F312del | p.Phe312del (c.935_937del)[j]
c.937_938delTC | p.Ser313Argfs*50 (c.937_938del)
c.942delG | p.Phe315Serfs*13 (c.942del)
1078delT | p.Phe316Leufs*12 (c.948del)
L320X | p.Leu320* (c.959T>A)
c.972delC | p.Tyr325Metfs*3 (c.972del)
1112delT | p.Leu327Glnfs*42 (c.980del)
c.982delA | p.Ile328Serfs*41 (c.982del)

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K329X | p.Lys329* (c.985A>T)
1119delA | p.Gly330Glufs*39 (c.987del)
G330X | p.Gly330* (c.988G>T)
c.991_995del | p.Ile331Profs*31 (c.991_995del)
R334W | p.Arg334Trp (c.1000C>T)
R334L | p.Arg334Leu (c.1001G>T)
1138insG | p.Ile336Serfs*28 (c.1006_1007insG)
I336K | p.Ile336Lys (c.1007T>A)
T338I | p.Thr338Ile (c.1013C>T)
1154insTC | p.Phe342Hisfs*28 (c.1021_1022dup)
S341P | p.Ser341Pro (c.1021T>C)
1157insTA | p.Cys343Thrfs*27 (c.1025_1026insTA)
1161delC | p.Cys343* (c.1029del)
L346P | p.Leu346Pro (c.1037T>C)
R347H | p.Arg347His (c.1040G>A)
R347P | p.Arg347Pro (c.1040G>C)
1185delTC | p.Arg352Alafs*11 (c.1053_1054del)
R352Q | p.Arg352Gln (c.1055G>A)
Q353X | p.Gln353* (c.1057C>T)
W356X | p.Trp356* (c.1068G>A)
1200_1219del20 | p.Trp356* (c.1068_1087del)
1199delG | p.Ala357Leufs*12 (c.1069del)
1204_1205delGT | p.Val358Thrfs*5 (c.1072_1073del)
Q359K/T360K | p.Gln359_Thr360delins LysLys (c.1075_1079delinsAAAAA)
Q359R | p.Gln359Arg (c.1076A>G)
1212delA | p.Trp361Glyfs*8 (c.1080del)
1213delT | p.Trp361Glyfs*8 (c.1081del)
W361X | p.Trp361* (c.1082G>A)
1215delG | p.Trp361Cysfs*8 (c.1083del)
W361X | p.Trp361* (c.1083G>A)
c.1084_1088dup | p.Ser364Metfs*7 (c.1084_1088dup)
Y362X | p.Tyr362* (c.1086T>A)
Y362X | p.Tyr362* (c.1086T>G)
1221delCT | p.Leu365Trpfs*16 (c.1093_1094del)
Q372X | p.Gln372* (c.1114C>T)
c.1115delA | p.Gln372Argfs*16 (c.1115del)
1248+1G->A | p.? (c.1116+1G>A)
1248+1G->C | p.? (c.1116+1G>C)
1248+1G->T | p.? (c.1116+1G>T)
1248insATCAA | p.? (c.1116_1116+1insATCAA)
1248+2T->A | p.? (c.1116+2T>A)
1248+2T->C | p.? (c.1116+2T>C)
1249-1G->A | p.? (c.1117-1G>A)
1249insA | p.Asp373Glufs*9 (c.1118dup)
Q376X | p.Gln376* (c.1126C>T)
1262delA | p.Lys377Serfs*11 (c.1130del)
1259insA | p.Gln378Alafs*4 (c.1130dup)
Q378X | p.Gln378* (c.1132C>T)
E379X | p.Glu379* (c.1135G>T)
K381X | p.Lys381* (c.1141A>T)
1288insTA | p.Asn386Ilefs*3 (c.1155_1156dup)
1288insA | p.Asn386Lysfs*25 (c.1157dup)
1289insTA | p.Leu387Thrfs*2 (c.1157_1158insTA)
1291delTT | p.Leu387Asnfs*23 (c.1159_1160del)
1294del7 | p.Thr388Glnfs*3 (c.1162_1168del)
T388X | p.Thr388* (c.1162_1163delinsTA)
1309delG | p.Val393* (c.1177del)
c.1190dupT | p.Thr398Asnfs*13 (c.1190dup)
W401X | p.Trp401* (c.1202G>A)
W401X | p.Trp401* (c.1203G>A)
E402X | p.Glu402* (c.1204G>T)

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1340delA | p.Glu403Glyfs*39 (c.1208del)
1341+1G->A | p.? (c.1209+1G>A)
1341+1G->C | p.? (c.1209+1G>C)
1341+1G->T | p.? (c.1209+1G>T)
1341+2T->A | p.? (c.1209+2T>A)
1341+2T->G | p.? (c.1209+2T>G)
1342-2A->C | p.? (c.1210-2A>C)
1342-2delAG | p.? (c.1210-2_1210-1del)
1342-1G->A | p.? (c.1210-1G>A)
1342-1G->C | p.? (c.1210-1G>C)
1342-1G->T | p.? (c.1210-1G>T)
1343delG | p.Gly404Aspfs*38 (c.1211del)
c.1219delG | p.Glu407Asnfs*35 (c.1219del)
E407X | p.Glu407* (c.1219G>T)
1353delA | p.Glu407Aspfs*35 (c.1221del)
1366delG | p.Ala412Glnfs*30 (c.1234del)
Q414X | p.Gln414* (c.1240C>T)
1367del5 | p.Asn415* (c.1243_1247del)
c.1244delA | p.Asn415Thrfs*27 (c.1244del)
1380insT | p.Asn417* (c.1248dup)
c.1262delC | p.Thr421Ilefs*21 (c.1262del)
1419delC | p.Phe430Serfs*12 (c.1287del)
S434X | p.Ser434* (c.1301C>G)
S434X | p.Ser434* (c.1301C>A)
1429del7 | p.Ser434Leufs*6 (c.1301_1307del)
1434delA | p.Leu435Phefs*7 (c.1302del)
c.1312dupA | p.Thr438Asnfs*8 (c.1312dup)
K442X | p.Lys442* (c.1324A>T)
1461ins4 | p.Ile444Argfs*3 (c.1327_1330dup)
D443fs | p.Asp443Glufs*19 (c.1329_1350del)
1460delAT | p.Ile444* (c.1330_1331del)
1465_1466insTAAT | p.Asn445Ilefs*38 (c.1333_1334insTAAT)
1471delA | p.Lys447Argfs*2 (c.1340del)
1474delA | p.Ile448* (c.1342del)
G451V | p.Gly451Val (c.1352G>T)^
Q452X | p.Gln452* (c.1354C>T)
L453S | p.Leu453Ser (c.1358T>C)
1491-1500del | p.Leu454Alafs*6 (c.1360_1387del)
A455E | p.Ala455Glu (c.1364C>A)
1497delGG | p.Val456Cysfs*25 (c.1365_1366del)
1498delG | p.Val456Leufs*13 (c.1366del)
V456F | p.Val456Phe (c.1366G>T)
V456A | p.Val456Ala (c.1367T>C)
1504delG | p.Gly458Aspfs*11 (c.1373del)
c.1375_1385del | p.Ser459Argfs*19 (c.1375_1385del)
1524+1delG | p.? (c.1392+1del)
1524+1G->A | p.? (c.1392+1G>A)
1525-2A->G | p.? (c.1393-2A>G)
1525-1G->A | p.? (c.1393-1G>A)
1525-1G->C | p.? (c.1393-1G>C)
1525-1G->T | p.? (c.1393-1G>T)
S466X | p.Ser466* (c.1397C>A)
S466X | p.Ser466* (c.1397C>G)
L467P | p.Leu467Pro (c.1400T>C)
1540del10 | p.Val470Glufs*54 (c.1409_1418del)
1548delG | p.Gly473Glufs*54 (c.1418del)
E474K | p.Glu474Lys (c.1420G>A)
1556delT | p.Leu475Argfs*52 (c.1424del)
1565delCA | p.Ser478* (c.1433_1434del)
E479X | p.Glu479* (c.1435G>T)
1571delG | p.Gly480Valfs*47 (c.1439del)
1576insT | p.Lys483* (c.1446dup)
S489X | p.Ser489* (c.1466C>A)
S489X | p.Ser489* (c.1466C>G)
1601delT | p.Phe490Serfs*37 (c.1469del)

Variant: Legacy | HGVS Protein (cDNA)
1601delTC | p.Phe490Leufs*13 (c.1470_1471del)
S492F | p.Ser492Phe (c.1475C>T)
1609delCA | p.Gln493Valfs*10 (c.1477_1478del)
Q493X | p.Gln493* (c.1477C>T)
c.1486delT | p.Trp496Glyfs*31 (c.1486del)
W496X | p.Trp496* (c.1487G>A)
I502T | p.Ile502Thr (c.1505T>C)
E504X | p.Glu504* (c.1510G>T)
I507del | p.Ile507del (c.1519_1521del)
F508del | p.Phe508del (c.1521_1523del)
1660delG | p.Val510Phefs*17 (c.1528del)
c.1530_1531delTT | p.Ser511Leufs*2 (c.1530_1531del)
D513G | p.Asp513Gly (c.1538A>G)
E514X | p.Glu514* (c.1540G>T)
1675_1687del13 | p.Tyr515Alafs*8 (c.1543_1555del)
1677delTA | p.Tyr515* (c.1545_1546del)
R516G | p.Arg516Gly (c.1546A>G)
c.1547_1548delGA | p.Arg516Ilefs*51 (c.1547_1548del)
Y517X | p.Tyr517* (c.1551C>A)
Y517X | p.Tyr517* (c.1551C>G)
V520F | p.Val520Phe (c.1558G>T)
C524X | p.Cys524* (c.1572C>A)
Q525X | p.Gln525* (c.1573C>T)
c.1573delC | p.Gln525Asnfs*2 (c.1573del)
1710del17bp | p.? (c.1579_1584+11del)
E527X | p.Glu527* (c.1579G>T)
c.1581dupA | p.Glu528Argfs*40 (c.1581dup)
E528X | p.Glu528* (c.1582G>T)
1716+2T->C | p.? (c.1584+2T>C)
1716+2delT | p.? (c.1584+2del)
1716+1G->A | p.? (c.1584+1G>A)
1716+1G->T | p.? (c.1584+1G>T)
1717-8G->A | p.? (c.1585-8G>A)
1717-2A->G | p.? (c.1585-2A>G)
1717-1G->T | p.? (c.1585-1G>T)
1717-1G->A | p.? (c.1585-1G>A)
S531X | p.Ser531* (c.1592_1593delinsG)
K536X | p.Lys536* (c.1606A>T)
c.1608delA | p.Asp537Thrfs*3 (c.1608del)
1749insTA | p.Val540* (c.1616_1617dup)
G542X | p.Gly542* (c.1624G>T)
1759delG | p.Glu543Lysfs*6 (c.1627del)
1774delCT | p.Leu548Glufs*19 (c.1642_1643del)
S549R | p.Ser549Arg (c.1645A>C)
S549N | p.Ser549Asn (c.1646G>A)
S549R | p.Ser549Arg (c.1647T>A)
S549R | p.Ser549Arg (c.1647T>G)
G550X | p.Gly550* (c.1648G>T)
1782delA | p.Gly551Valfs*8 (c.1650del)
G551S | p.Gly551Ser (c.1651G>A)
G551D | p.Gly551Asp (c.1652G>A)
1784delG | p.Gly551Valfs*8 (c.1652del)
1784insATCAT | p.Gln552Serfs*9 (c.1652_1653insATCAT)
Q552X | p.Gln552* (c.1654C>T)
1787delA | p.Gln552Hisfs*7 (c.1656del)
R553X | p.Arg553* (c.1657C>T)
R555X | p.Arg555* (c.1663A>T)
1802delC | p.Ser557Phefs*2 (c.1670del)
L558S | p.Leu558Ser (c.1673T>C)
1806delA | p.Ala559Glnfs*13 (c.1674del)
A559T | p.Ala559Thr (c.1675G>A)
1807delG | p.Ala559Glnfs*13 (c.1675del)
R560K | p.Arg560Lys (c.1679G>A)
R560T | p.Arg560Thr (c.1679G>C)
1811+1G->A | p.? (c.1679+1G>A)
1811+1G->C | p.? (c.1679+1G>C)

Variant: Legacy | HGVS Protein (cDNA)
1811+2T->C | p.? (c.1679+2T>C)
1811+1.6kbA->G | p.? (c.1680-886A>G)
1811+1643G->T | p.? (c.1680-877G>T)
1812-2A->C | p.? (c.1680-2A>C)
1812-1G->A | p.? (c.1680-1G>A)
IVS11-1G->C | p.? (c.1680-1G>C)
R560S | p.Arg560Ser (c.1680A>C)
R560S | p.Arg560Ser (c.1680A>T)
A561E | p.Ala561Glu (c.1682C>A)
1813insC | p.Val562Serfs*6 (c.1682dup)
Y563N | p.Tyr563Asn (c.1687T>A)
Y563D | p.Tyr563Asp (c.1687T>G)
Y563X | p.Tyr563* (c.1689C>A)
1824delA | p.Asp565Metfs*7 (c.1692del)
1833delT | p.Leu568Cysfs*4 (c.1703del)
L568X | p.Leu568* (c.1703T>A)
Y569D | p.Tyr569Asp (c.1705T>G)
Y569X | p.Tyr569* (c.1707T>A)
c.1708_1712delTTATT | p.Leu570Argfs*17 (c.1708_1712del)
L570X | p.Leu570* (c.1709T>A)
1845delAG/1846delGA | p.Asp572Leufs*16 (c.1714_1715del)
P574H | p.Pro574His (c.1721C>A)
1857delT | p.Phe575Leufs*4 (c.1725del)
c.1725_1727delinsAT | p.Phe575Leufs*4 (c.1725_1727delinsAT)
Y577X | p.Tyr577* (c.1731C>A)
1870delG | p.Val580Phefs*2 (c.1738del)
1874_1875insTT | p.Leu581Phefs*2 (c.1741_1742dup)
1874insT | p.Leu581Phefs*8 (c.1742dup)
L581X | p.Leu581* (c.1742T>A)
E585X | p.Glu585* (c.1753G>T)
c.1753delG | p.Glu585Lysfs*10 (c.1753del)
E588X | p.Glu588* (c.1762G>T)
1898+1G->A | p.? (c.1766+1G>A)
1898+1G->C | p.? (c.1766+1G>C)
1898+1G->T | p.? (c.1766+1G>T)
1898+2delT | p.? (c.1766+2del)
1898+2T->A | p.? (c.1766+2T>A)
1898+2T->C | p.? (c.1766+2T>C)
1898+3A->G | p.? (c.1766+3A>G)
1898+5G->T | p.? (c.1766+5G>T)
1898-2A->G | p.? (c.1767-2A>G)
1899-1G->A | p.? (c.1767-1G>A)
1918delGC | p.Ala596* (c.1786_1787del)
K598X | p.Lys598* (c.1792A>T)
1924del7 | p.Lys598Glyfs*11 (c.1792_1798del)
1932delG | p.Ile601Phefs*10 (c.1800del)
I601F | p.Ile601Phe (c.1801A>T)
c.1807delG | p.Val603Serfs*8 (c.1807del)
1942del17 | p.Thr604Phefs*5 (c.1810_1826del)
1949del84 | p.Met607_Gln634del (c.1820_1903del)
H609R | p.His609Arg (c.1826A>G)
A613T | p.Ala613Thr (c.1837G>A)
1978dupA | p.Ile616Asnfs*6 (c.1846dup)
1978delA | p.Ile616Tyrfs*2 (c.1846del)
c.1853_1863del | p.Ile618Argfs*2 (c.1853_1863del)
I618T | p.Ile618Thr (c.1853T>C)
2003del8 | p.Ser624Ilefs*15 (c.1871_1878del)
2005delTA | p.Tyr625Phefs*16 (c.1874_1875del)
G628R | p.Gly628Arg (c.1882G>A)
G628R | p.Gly628Arg (c.1882G>C)
S631X | p.Ser631* (c.1892C>G)
2025dupA | p.Glu632Argfs*10 (c.1893dup)
c.1894_1895del | p.Glu632Thrfs*9 (c.1894_1895del)
Q634X | p.Gln634* (c.1900C>T)

NYS Cystic Fibrosis Newborn Screening: Custom *CFTR* Variant Panel Content

Variant: Legacy | HGVS Protein (cDNA)
2036delA | p.Asn635Ilefs*28 (c.1904del)
2043delG | p.Gln637Hisfs*26 (c.1911del)
2053insTA | p.Ser641Ilefs*23 (c.1920_1921dup)
2055del9->A | p.Ser641Argfs*5 (c.1923_1931delinsA)
G646X | p.Gly646* (c.1936G>T)
2075delA | p.Asp648Valfs*15 (c.1943del)
Q652X | p.Gln652* (c.1954C>T)
E656X | p.Glu656* (c.1966G>T)
R657X | p.Arg657* (c.1969A>T)
c.1970delG | p.Arg657Lysfs*6 (c.1970del)
2104insA | p.Arg658Lysfs*7 (c.1972dup)
2105-2117del13insAGAAA | p.Arg658Lysfs*4 (c.1973_1985delinsAGAAA)
2108delA | p.Asn659Ilefs*4 (c.1976del)
2109-2118del10 | p.Asn659Lysfs*10 (c.1977_1986del)
S660X | p.Ser660* (c.1979C>A)
S660X | p.Ser660* (c.1979C>G)
2113delA | p.Ile661Serfs*2 (c.1981del)
2114delT | p.Ile661Thrfs*2 (c.1982del)
2118del4 | p.Thr663Argfs*8 (c.1986_1989del)
2118del14 | p.Thr663Profs*21 (c.1987_2000del)
E664X | p.Glu664* (c.1990G>T)
2132delAC | p.His667Profs*21 (c.2000_2001del)
2143delT | p.Leu671* (c.2012del)
G673X | p.Gly673* (c.2017G>T)
c.2032_2033delTC | p.Ser678Leufs*10 (c.2032_2033del)
W679X | p.Trp679* (c.2036G>A)
W679X | p.Trp679* (c.2037G>A)
2175insA | p.Thr682Asnfs*7 (c.2044dup)
2176insC | p.Gln685Thrfs*4 (c.2045dup)
c.2045delC | p.Thr682Lysfs*40 (c.2045del)
2183delAA | p.Lys684Thrfs*4 (c.2051_2052del)
2184delA | p.Lys684Asnfs*38 (c.2052del)
2184insA | p.Gln685Thrfs*4 (c.2052dup)
2183AA->G | p.Lys684Serfs*38 (c.2051_2052delinsG)
2184del4 | p.Lys684Asnfs*37 (c.2052_2055del)
c.2053delC | p.Gln685Asnfs*37 (c.2053del)
2185insC | p.Gln685Profs*4 (c.2053dup)
c.2053_2054insAA | p.Ser686Asnfs*37 (c.2053_2054insAA)
Q685X | p.Gln685* (c.2053C>T)
2189CT->A | p.Ser686Tyrfs*36 (c.2057_2058delinsA)
2193ins4 | p.Lys688Phefs*2 (c.2058_2061dup)
K688X | p.Lys688* (c.2062A>T)
E692X | p.Glu692* (c.2074G>T)
2215delG | p.Glu695Lysfs*27 (c.2083del)
2221insA | p.Arg697Lysfs*33 (c.2089dup)
c.2089delA | p.Arg697Glyfs*25 (c.2089del)
2222delG | p.Lys698Argfs*24 (c.2091del)
R709X | p.Arg709* (c.2125C>T)
K710X | p.Lys710* (c.2128A>T)
Q715X | p.Gln715* (c.2143C>T)
K716X | p.Lys716* (c.2145_2146delinsGT)
K716X | p.Lys716* (c.2146A>T)
c.2148delG | p.Thr717Leufs*5 (c.2148del)
L719X | p.Leu719* (c.2156T>A)
Q720X | p.Gln720* (c.2158C>T)
2290del16 | p.Gln720Lysfs*8 (c.2158_2173del)

Variant: Legacy | HGVS Protein (cDNA)
2307insA | p.Glu726Argfs*4 (c.2175dup)
E730X | p.Glu730* (c.2188G>T)
L732X | p.Leu732* (c.2195T>G)
2335delA | p.Arg735Glyfs*4 (c.2203del)
2347delG | p.Val739Tyrfs*16 (c.2215del)
G745X | p.Gly745* (c.2233G>T)
2372del8 | p.Ile748Serfs*28 (c.2241_2248del)
c.2250delT | p.Arg751Alafs*4 (c.2250del)
c.2261delT | p.Val754Glyfs*17 (c.2261del)
2406_2407delinsT | p.Thr760Argfs*11 (c.2274_2275delinsT)
2409delC | p.Thr760Argfs*11 (c.2277del)
R764X | p.Arg764* (c.2290C>T)
2429delG | p.Arg766Serfs*5 (c.2298del)
Q767X | p.Gln767* (c.2299C>T)
2435insC | p.Val769Cysfs*10 (c.2303dup)
2456delAC | p.His775Leufs*3 (c.2324_2325del)
2466delC | p.Gln779Lysfs*24 (c.2335del)
Q779X | p.Gln779* (c.2335C>T)
c.2337delA | p.Gly780Valfs*23 (c.2337del)
2472_2478del7 | p.Gln781Phefs*20 (c.2340_2346del)
c.2341delC | p.Gln781Argfs*22 (c.2341del)
Q781X | p.Gln781* (c.2341C>T)
2481-2482insT | p.His784Serfs*21 (c.2349dup)
R785X | p.Arg785* (c.2353C>T)
c.2357dupA | p.Thr787Aspfs*18 (c.2357dup)
R792X | p.Arg792* (c.2374C>T)
2510delAA | p.Lys793Serfs*11 (c.2378_2379del)
2512delG | p.Val794Cysfs*9 (c.2380del)
2522insC | p.Gln799Serfs*6 (c.2393dup)
Q799X | p.Gln799* (c.2395C>T)
2556insAT | p.Ser809Ilefs*13 (c.2423_2424dup)
2557delT | p.Ser809Glnfs*12 (c.2425del)
R810X | p.Arg810* (c.2428A>T)
c.2433_2437delinsATA | p.Leu812Tyrfs*10 (c.2433_2437delinsATA)
L812X | p.Leu812* (c.2435T>A)
Q814X | p.Gln814* (c.2440C>T)
E815X | p.Glu815* (c.2443G>T)
2585delT | p.Leu818Trpfs*3 (c.2453del)
2586-2687insT | p.Glu819* (c.2454_2455insT)
2594delGT | p.Ser821Argfs*4 (c.2463_2464del)
E822X | p.Glu822* (c.2464G>T)
c.2467_2470delGAAA | p.Glu823Leufs*6 (c.2467_2470del)
2603delT | p.Asn825Thrfs*5 (c.2472del)
E826X | p.Glu826* (c.2476G>T)
K830X | p.Lys830* (c.2488A>T)
c.2489_2490insA | p.Glu831Glyfs*5 (c.2489dup)
2622+1G->A | p.? (c.2490+1G>A)
2623-2A->G | p.? (c.2491-2A>G)
2623-1G->C | p.? (c.2491-1G>C)
E831X | p.Glu831* (c.2491G>T)
c.2493delG | p.Glu831Aspfs*13 (c.2493del)
2634delT | p.Phe834Leufs*10 (c.2502del)
2634insT | p.Asp835* (c.2502dup)
2640delT | p.Asp836Glufs*8 (c.2508del)
2655del26 | p.Ala842Serfs*45 (c.2523_2548del)
W846X | p.Trp846* (c.2537G>A)
W846X | p.Trp846* (c.2538G>A)
c.2538delG | p.Trp846* (c.2538del)

Variant: Legacy | HGVS Protein (cDNA)
2672delA | p.Asn847Thrfs*13 (c.2540del)
Y849X | p.Tyr849* (c.2547C>A)
R851X | p.Arg851* (c.2551C>T)
2686dupT | p.Tyr852Leufs*44 (c.2554dup)
c.2556dupT | p.Ile853Tyrfs*43 (c.2556dup)
2694delT | p.Val855Serfs*5 (c.2562del)
c.2573delG | p.Ser858Thrfs*2 (c.2573del)
2708del13 | p.Leu859* (c.2576_2588del)
2711delT | p.Phe861Leufs*3 (c.2583del)
2721del11 | p.Ile864Serfs*28 (c.2589_2599del)
2723delTT | p.Ile864Metfs*31 (c.2592_2593del)
2732insA | p.Val868Serfs*28 (c.2601dup)
2737delA | p.Ile869Phefs*37 (c.2605del)
2747delC | p.Ala872Glufs*34 (c.2615del)
2751+1G->A | p.? (c.2619+1G>A)
2751+2T->A | p.? (c.2619+2T>A)
2751+2T->C | p.? (c.2619+2T>C)
2752-2A->G | p.? (c.2620-2A>G)
2752-1G->T | p.? (c.2620-1G>T)
2777insTG | p.Trp882Cysfs*25 (c.2644_2645dup)
W882X | p.Trp882* (c.2645G>A)
W882X | p.Trp882* (c.2646G>A)
2787del16 | p.? (c.2655_2657+13del)
2789+5G->A | p.? (c.2657+5G>A)
2790-2A->G | p.? (c.2658-2A>G)
2790-1G->C | p.? (c.2658-1G>C)
2790-1G->T | p.? (c.2658-1G>T)
Q890X | p.Gln890* (c.2668C>T)
2837delG | p.Ser902Thrfs*4 (c.2705del)
c.2732_2733insA | p.Ser911Argfs*64 (c.2732_2733insA)
c.2733insA | p.Ser912Ilefs*63 (c.2733_2734insA)
S912X | p.Ser912* (c.2735C>A)
2869insG | p.Tyr913* (c.2737_2738insG)
Y913X | p.Tyr913* (c.2739T>A)
c.2745_2746delGT | p.Phe916Leufs*58 (c.2745_2746del)
2896insAG | p.Val922Glufs*2 (c.2763_2764dup)
2907delTT | p.Leu926Alafs*48 (c.2776_2777del)
2909delT | p.Leu926Cysfs*16 (c.2777del)
L927P | p.Leu927Pro (c.2780T>C)
c.2789delG | p.Gly930Aspfs*12 (c.2789del)
2935del11 | p.Leu935Alafs*36 (c.2803_2813del)
2937_2942delinsTCAGA | p.Pro936_Leu937del (c.2805_2810del)
c.2805_2810delinsTCAGA | p.Pro936Glnfs*6 (c.2805_2810delinsTCAGA)
2942insT | p.Val938Glyfs*37 (c.2810dup)
2948delA | p.His939Leufs*3 (c.2816del)
2949del5 | p.Thr940Asnfs*33 (c.2819_2823del)
c.2818_2831del | p.Thr940Valfs*30 (c.2818_2831del)
2951insA | p.Leu941Serfs*34 (c.2819_2820insA)
2954delT | p.Leu941Glnfs*27 (c.2822del)
2957delT | p.Ile942Thrfs*26 (c.2825del)
S945L | p.Ser945Leu (c.2834C>T)
2967delG | p.Ile947Phefs*21 (c.2835del)
K946X | p.Lys946* (c.2836A>T)
c.2839delA | p.Ile947Phefs*21 (c.2839del)
2975delT | p.Leu948Tyrfs*20 (c.2843del)
K951X | p.Lys951* (c.2851A>T)
2991del32 | p.Leu953Phefs*11 (c.2859_2890del)

Variant: Legacy | HGVS Protein (cDNA)
3007delG | p.Ala959Hisfs*9 (c.2875del)
3011delC | p.Pro960Leufs*8 (c.2879del)
c.2879_2882delCTAT | p.Pro960Argfs*7 (c.2879_2882del)
3012delT | p.Met961Cysfs*7 (c.2880del)
3015_3018dupGTCA | p.Thr963Valfs*13 (c.2883_2886dup)
S962X | p.Ser962* (c.2885C>A)
S962X | p.Ser962* (c.2885C>G)
3028delA | p.Thr966Argfs*2 (c.2896del)
3029delC | p.Thr966Serfs*2 (c.2897del)
3031-3032delinsA | p.Leu967Argfs*14 (c.2899_2900delinsA)
G970R | p.Gly970Arg (c.2908G>C)
3040+1G->A | p.? (c.2908+1G>A)
3040+1G->T | p.? (c.2908+1G>T)
G970D | p.Gly970Asp (c.2909G>A)
3041delG | p.Gly970Valfs*11 (c.2909del)
c.2909_2924dup16 | p.Phe976Trpfs*4 (c.2909_2924dup)
3041-1G->A | p.? (c.2909-1G>A)
3041-1G->C | p.? (c.2909-1G>C)
3056delGA | p.Arg975Ilefs*10 (c.2924_2925del)
3061insA | p.Ser977Tyrfs*9 (c.2929_2930insA)
K978X | p.Lys978* (c.2932A>T)
D979V | p.Asp979Val (c.2936A>T)
3089insTC | p.Pro988Cysfs*13 (c.2959_2960dup)
3090del4 | p.Pro988Leufs*11 (c.2958_2961del)
3095insT | p.Leu989Serfs*5 (c.2964dup)
c.2982_2988+2delCATCCAGGT | p.? (c.2982_2988+2del)
Q996X | p.Gln996* (c.2986C>T)
3120G->A | p.Gln996= / p.? (c.2988G>A)
3120+1G->A | p.? (c.2988+1G>A)
3120+1G->C | p.? (c.2988+1G>C)
3121-2A->G | p.? (c.2989-2A>G)
3121-2A->C | p.? (c.2989-2A>C)
3121-2A->T | p.? (c.2989-2A>T)
3121-1G->A | p.? (c.2989-1G>A)
3121-1G->T | p.? (c.2989-1G>T)
3126delA | p.Leu998Phefs*2 (c.2994del)
3129del4 | p.Ile1000* (c.2997_3000del)
3130delA | p.Ile1000Leufs*2 (c.2998del)
3132delTG | p.Val1001Aspfs*45 (c.3002_3003del)
G1003X | p.Gly1003* (c.3007G>T)
3139delG | p.Gly1003Glufs*3 (c.3008del)
3143delC | p.Ala1004Valfs*2 (c.3011del)
3143del9 | p.Ala1004_Ala1006del (c.3011_3019del)
A1006E | p.Ala1006Glu (c.3017C>A)
3152delT | p.Val1008Serfs*15 (c.3021del)
3154delG | p.Val1008Serfs*15 (c.3022del)
L1011X | p.Leu1011* (c.3032T>G)
c.3033dup | p.Gln1012Thrfs*35 (c.3033dup)
Q1012X | p.Gln1012* (c.3034C>T)
3171delC | p.Tyr1014Thrfs*9 (c.3039del)
3171insC | p.Tyr1014Leufs*33 (c.3039dup)
3199del6 | p.Ile1023_Val1024del (c.3067_3072del)
3213-3214insT | p.Met1028Tyrfs*19 (c.3081dup)
Q1035X | p.Gln1035* (c.3103C>T)
3238delA | p.Thr1036Profs*24 (c.3106del)
T1036N | p.Thr1036Asn (c.3107C>A)
S1037X | p.Ser1037* (c.3110C>A)
Q1038X | p.Gln1038* (c.3112C>T)

NYS Cystic Fibrosis Newborn Screening: Custom *CFTR* Variant Panel Content

Variant: Legacy | HGVS Protein (cDNA)
Q1042TfsX5 | p.Gln1042Thrfs*5 (c.3123dup)
Q1042X | p.Gln1042* (c.3124C>T)
E1044X | p.Glu1044* (c.3130G>T)
E1046X | p.Glu1046* (c.3136G>T)
3271+1delG | p.? (c.3139+1del)
3271+1G->A | p.? (c.3139+1G>A)
3271+1G->C | p.? (c.3139+1G>C)
3271+1G->T | p.? (c.3139+1G>T)
3271delGG | p.? (c.3139_3139+1del)
3271+2T->C | p.? (c.3139+2T>C)
3272-26A->G | p.? (c.3140-26A>G)
3272-1G->A | p.? (c.3140-1G>A)
H1054D | p.His1054Asp (c.3160C>G)
3293delA | p.His1054Leufs*6 (c.3161del)
L1059X | p.Leu1059* (c.3176T>G)
c.3180delA | p.Gly1061Aspfs*22 (c.3180del)
G1061R | p.Gly1061Arg (c.3181G>A)
G1061R | p.Gly1061Arg (c.3181G>C)
W1063X | p.Trp1063* (c.3189G>A)
L1065P | p.Leu1065Pro (c.3194T>C)
R1066C | p.Arg1066Cys (c.3196C>T)
R1066H | p.Arg1066His (c.3197G>A)
c.3199delG | p.Ala1067Profs*16 (c.3199del)
3349insT | p.Tyr1073Leufs*3 (c.3217dup)
3359delC | p.Thr1076Ilefs*7 (c.3227del)
3359delCTCTG | p.Thr1076Ilefs*78 (c.3227_3231del)
3359delCT | p.Leu1077Valfs*78 (c.3229_3230del)
L1077P | p.Leu1077Pro (c.3230T>C)
c.3231_3232delGT | p.Phe1078Profs*77 (c.3231_3232del)
H1085P | p.His1085Pro (c.3254A>C)
3396delC | p.Trp1089Glyfs*13 (c.3264del)
W1089X | p.Trp1089* (c.3266G>A)
Y1092X | p.Tyr1092* (c.3276C>A)
Y1092X | p.Tyr1092* (c.3276C>G)
3422del16 | p.Arg1097Glnfs*2 (c.3290_3305del)
W1098R | p.Trp1098Arg (c.3292T>C)
W1098X | p.Trp1098* (c.3293G>A)
3425delG | p.Trp1098Cysfs*4 (c.3294del)
W1098X | p.Trp1098* (c.3294G>A)
W1098C | p.Trp1098Cys (c.3294G>C)
W1098C | p.Trp1098Cys (c.3294G>T)
Q1100X | p.Gln1100* (c.3298C>T)
M1101K | p.Met1101Lys (c.3302T>A)
M1101R | p.Met1101Arg (c.3302T>G)
R1102X | p.Arg1102* (c.3304A>T)
E1104X | p.Glu1104* (c.3310G>T)
3447delG | p.Met1105Ilefs*16 (c.3315del)
c.3321dup | p.Val1108Cysfs*48 (c.3321dup)
3453delT | p.Phe1107Leufs*14 (c.3321del)
c.3322_3323delGT | p.Val1108Hisfs*47 (c.3322_3323del)
3454delG | p.Val1108Serfs*13 (c.3322del)
3456delC | p.Ile1109Serfs*12 (c.3324del)
c.3325delA | p.Ile1109Serfs*12 (c.3325del)
c.3344_3345insA | p.Phe1116Leufs*40 (c.3344_3345insA)
S1118F | p.Ser1118Phe (c.3353C>T)
3497delC | p.Thr1122Lysfs*12 (c.3365del)
3499+1G->T | p.? (c.3367+1G>T)
3499+1G->A | p.? (c.3367+1G>A)
3499+2T->C | p.? (c.3367+2T>C)
3499+2T->A | p.? (c.3367+2T>A)
3500-2A->T | p.? (c.3368-2A>T)

Variant: Legacy | HGVS Protein (cDNA)
3500-1G->A | p.? (c.3368-1G>A)
3500-2A->G | p.? (c.3368-2A>G)
c.3380_3383del | p.Gly1127Glufs*6 (c.3380_3383del)
R1128X | p.Arg1128* (c.3382A>T)
3516del5 | p.Val1129Tyrfs*25 (c.3384_3388del)
3521del14 | p.Gly1130Valfs*21 (c.3389_3402del)
3528delC | p.Leu1133* (c.3397del)
3532AC->GTA | p.Thr1134Valfs*22 (c.3400_3401delinsGTA)
3539del16 | p.Ala1136Valfs*7 (c.3407_3422del)
c.3411_3414delGAAT | p.Met1137Ilefs*3 (c.3411_3414del)
3556insAGTA | p.Thr1142Lysfs*15 (c.3421_3424dup)
c.3426_3427insGTAA | p.Leu1143Valfs*14 (c.3426_3427insGTAA)
Q1144X | p.Gln1144* (c.3430C>T)
W1145X | p.Trp1145* (c.3434G>A)
W1145X | p.Trp1145* (c.3435G>A)
3600G->A | p.Leu1156= / p.? (c.3468G>A)
3600+1G->A | p.? (c.3468+1G>A)
3600+1G->T | p.? (c.3468+1G>T)
3600+2insT | p.? (c.3468+2dup)
3600+5G->A | p.? (c.3468+5G>A)
3601-2A->C | p.? (c.3469-2A>C)
3601-2A->G | p.? (c.3469-2A>G)
3601-1G->A | p.? (c.3469-1G>A)
3601-1G->C | p.? (c.3469-1G>C)
3601-1G->T | p.? (c.3469-1G>T)
R1158X | p.Arg1158* (c.3472C>T)
S1159P | p.Ser1159Pro (c.3475T>C)
S1159F | p.Ser1159Phe (c.3476C>T)
c.3477delT | p.Val1160* (c.3477del)
3613AGCCGA->GG | p.Ser1161Glyfs*30 (c.3481_3486delinsGG)
R1162X | p.Arg1162* (c.3484C>T)
3617delGA | p.Val1163Leufs*2 (c.3486_3487del)
3622insT | p.Lys1165* (c.3492dup)
K1165X | p.Lys1165* (c.3493A>T)
c.3495delG | p.Lys1165Asnfs*27 (c.3495del)
3629delT | p.Phe1166Serfs*26 (c.3497del)
c.3524delC | p.Pro1175Leufs*17 (c.3524del)
c.3525_3537del | p.Thr1176Asnfs*12 (c.3525_3537del)
3659delC | p.Lys1177Serfs*15 (c.3528del)
3662delA | p.Lys1177Serfs*15 (c.3530del)
3667ins4 | p.Thr1179Ilefs*17 (c.3532_3535dup)
S1178X | p.Ser1178* (c.3533C>A)
S1178X | p.Ser1178* (c.3533C>G)
3667del4 | p.Thr1179Asnfs*12 (c.3536_3539del)
c.3539_3554del | p.Lys1180Thrfs*7 (c.3539_3554del)
Y1182X | p.Tyr1182* (c.3546C>G)
Q1186X | p.Gln1186* (c.3556C>T)
3695delC | p.Ser1188* (c.3563del)
c.3569_3570delTT | p.Val1190Aspfs*4 (c.3569_3570del)
S1196X | p.Ser1196* (c.3587C>A)
S1196X | p.Ser1196* (c.3587C>G)
3724delG | p.Val1198* (c.3592del)
3730A->TCT | p.Lys1200Serfs*12 (c.3598delinsTCT)
3731-3732AA->G | p.Lys1200Argfs*11 (c.3599_3600delinsG)

Variant: Legacy | HGVS Protein (cDNA)
3732delA | p.Asp1201Metfs*10 (c.3600del)
3737delA | p.Asp1202Alafs*9 (c.3605del)
W1204X | p.Trp1204* (c.3611G>A)
W1204X | p.Trp1204* (c.3612G>A)
3747delC | p.Ser1206Glnfs*5 (c.3615del)
S1206X | p.Ser1206* (c.3617C>G)
3750delA | p.Gly1208Alafs*3 (c.3618del)
3750delAG | p.Gly1208Profs*56 (c.3618_3619del)
3755delG | p.Gly1208Alafs*3 (c.3623del)
3755dupG | p.Gln1209Profs*56 (c.3623dup)
c.3635delT | p.Val1212Alafs*16 (c.3635del)
3789insA | p.Thr1220Asnfs*45 (c.3658dup)
3791delC | p.Thr1220Lysfs*8 (c.3659del)
c.3678delA | p.Leu1227* (c.3678del)
3821delT | p.Ser1231Profs*4 (c.3691del)
I1234V | p.Ile1234Val (c.3700A>G)
3840delT | p.Gly1237Alafs*22 (c.3708del)
3847delA | p.Arg1239Glyfs*20 (c.3715del)
3849G->A | p.Arg1239= / p.? (c.3717G>A)
3849+1G->A | p.? (c.3717+1G>A)
3849+4A->G | p.? (c.3717+4A>G)
3849+5G->A | p.? (c.3717+5G>A)
3849+40A->G | p.? (c.3717+40A>G)
3849+10kbC->T | p.? (c.3718-2477C>T)
3850-3T->G | p.? (c.3718-3T>G)
3850-1G->A | p.? (c.3718-1G>A)
V1240G | p.Val1240Gly (c.3719T>G)
3856delC | p.Leu1242Serfs*17 (c.3724del)
L1243X | p.Leu1243* (c.3728T>A)
G1244E | p.Gly1244Glu (c.3731G>A)
G1247X | p.Gly1247* (c.3739G>T)
S1248X | p.Ser1248* (c.3743C>A)
S1248X | p.Ser1248* (c.3743C>G)
3876delA | p.Lys1250Argfs*9 (c.3744del)
G1249R | p.Gly1249Arg (c.3745G>C)
G1249R | p.Gly1249Arg (c.3745G>A)
3878delG | p.Lys1250Argfs*9 (c.3747del)
S1251N | p.Ser1251Asn (c.3752G>A)
3886insA | p.Thr1252Asnfs*13 (c.3754dup)
3889dupT | p.Leu1253Phefs*12 (c.3758dup)
3892delTT | p.Leu1254Ilefs*10 (c.3760_3761del)
L1254X | p.Leu1254* (c.3761T>A)
L1254X | p.Leu1254* (c.3761T>G)
S1255P | p.Ser1255Pro (c.3763T>C)
S1255X | p.Ser1255* (c.3764C>A)
3898insC | p.Leu1258Phefs*7 (c.3767dup)
3905delT | p.Leu1258* (c.3773del)
3905insT | p.Leu1258Phefs*7 (c.3773dup)
L1253X | p.Leu1253* (c.3773T>A)
E1266X | p.Glu1266* (c.3796G>T)
I1269N | p.Ile1269Asn (c.3806T>A)
3940delG | p.Asp1270Metfs*8 (c.3808del)
3944delGT | p.Ser1273Leufs*28 (c.3816_3817del)
W1274X | p.Trp1274* (c.3822G>A)
3959delC | p.Ser1276* (c.3827del)
3960-3961delA | p.Ile1277* (c.3829del)
3967delTT | p.Leu1279Alafs*22 (c.3835_3836del)
Q1280X | p.Gln1280* (c.3838C>T)
Q1281X | p.Gln1281* (c.3841C>T)
W1282X | p.Trp1282* (c.3845G>A)
W1282X | p.Trp1282* (c.3846G>A)

Variant: Legacy | HGVS Protein (cDNA)
R1283M | p.Arg1283Met (c.3848G>T)
G1287X | p.Gly1287* (c.3859G>T)
Q1291X | p.Gln1291* (c.3871C>T)
4005+1G->A | p.? (c.3873+1G>A)
4005+1G->T | p.? (c.3873+1G>T)
4005+2T->C | p.? (c.3873+2T>C)
4006-2A->G | p.? (c.3874-2A>G)
4006-1G->A | p.? (c.3874-1G>A)
4006delA | p.Val1293Tyrfs*35 (c.3876del)
c.3883_3884insG | p.Ile1295Serfs*7 (c.3883_3884insG)
4010del4 | p.Ile1295Phefs*32 (c.3883_3886del)
4015delA | p.Ile1295Phefs*33 (c.3883del)
c.3889delT | p.Ser1297Leufs*31 (c.3889del)
4016insT | p.Ser1297Phefs*5 (c.3889dup)
4022insT | p.Gly1298Trpfs*4 (c.3891dup)
c.3893delG | p.Gly1298Glufs*30 (c.3893del)
K1302X | p.Lys1302* (c.3904A>T)
4040delA | p.Asn1303Thrfs*25 (c.3908del)
4040dupA | p.Asn1303Lysfs*6 (c.3908dup)
N1303K | p.Asn1303Lys (c.3909C>G)
4048insCC | p.Tyr1307Profs*22 (c.3917_3918dup)
Y1307X | p.Tyr1307* (c.3921T>A)
E1308X | p.Glu1308* (c.3922G>T)
Q1309X | p.Gln1309* (c.3925C>T)
W1310X | p.Trp1310* (c.3929G>A)
W1310X | p.Trp1310* (c.3930G>A)
4064-4065delinsAATATG | p.Ser1311Lysfs*12 (c.3932_3933delinsAATATG)
Q1313X | p.Gln1313* (c.3937C>T)
W1316X | p.Trp1316* (c.3947G>A)
4089delA | p.Asp1320Metfs*8 (c.3957del)
4089ins4 | p.Asp1320Argfs*3 (c.3957_3958insAGGG)
4095+1G->A | p.? (c.3963+1G>A)
4095+1G->C | p.? (c.3963+1G>C)
4095+1G->T | p.? (c.3963+1G>T)
4096-1G->A | p.? (c.3964-1G>A)
L1324P | p.Leu1324Pro (c.3971T>C)
4108delT | p.Ser1326Leufs*2 (c.3976del)
Q1330X | p.Gln1330* (c.3988C>T)
4120delCA | p.Gln1330Valfs*6 (c.3988_3989del)
c.3999delG | p.Lys1334Serfs*13 (c.3999del)
L1335P | p.Leu1335Pro (c.4004T>C)
c.4028delG | p.Gly1343Alafs*4 (c.4028del)
4160insG | p.Cys1344Leufs*15 (c.4028dup)
4165delGT | p.Val1345Profs*13 (c.4033_4034del)
p.S1347PfsX13 | p.Ser1347Profs*13 (c.4035_4038dup)
c.4036dupC | p.Leu1346Profs*13 (c.4036dup)
4168delCTAAGCC | p.Leu1346Metfs*6 (c.4036_4042del)
4168delCTAAG | p.Leu1346Profs*11 (c.4037_4041del)
4172delGC | p.Ser1347Thrfs*11 (c.4040_4041del)
4177delG | p.Gly1349Alafs*5 (c.4046del)
G1349D | p.Gly1349Asp (c.4046G>A)
K1351X | p.Lys1351* (c.4051A>T)
4197_4198delCT | p.Leu1356Glyfs*2 (c.4065_4066del)
4203TAG->AA | p.Arg1358Asnfs*22 (c.4071_4073delinsAA)

NYS Cystic Fibrosis Newborn Screening: Custom *CFTR* Variant Panel Content

Variant: Legacy | HGVS Protein (cDNA)
4209TGTT->AA | p.Val1360Thrfs*3 (c.4077_4080delinsAA)
c.4078delG | p.Val1360Phefs*20 (c.4078del)
4218insT | p.Lys1363* (c.4086dup)
4222delG | p.Ala1364Argfs*16 (c.4090del)
I1366N | p.Ile1366Asn (c.4097T>A)
4237-4242delinsAGAA | p.Leu1369Argfs*16 (c.4105_4110delinsAGAA)

Variant: Legacy | HGVS Protein (cDNA)
E1371X | p.Glu1371* (c.4111G>T)
H1375P | p.His1375Pro (c.4124A>C)
4259del5 | p.Leu1376Serfs*8 (c.4127_4131del)
4268+2T->G | p.? (c.4136+2T>G)
4271delC | p.Thr1380Asnfs*4 (c.4139del)
Y1381X | p.Tyr1381* (c.4143C>A)
Y1381X | p.Tyr1381* (c.4143C>G)
4279insA | p.Ile1383Asnfs*3 (c.4147dup)
Q1382X | p.Gln1382* (c.4144C>T)

Variant: Legacy | HGVS Protein (cDNA)
c.4163_4167delTAAAA | p.Leu1388Profs*5 (c.4163_4167del)
Q1390X | p.Gln1390* (c.4168C>T)
4301delA | p.Ala1391Hisfs*7 (c.4170del)
c.4187delC | p.Thr1396Lysfs*2 (c.4187del)
4326delTC | p.Cys1400* (c.4197_4198del)
4332delTG | p.Cys1400* (c.4200_4201del)
C1400X | p.Cys1400* (c.4200T>A)

Variant: Legacy | HGVS Protein (cDNA)
E1401X | p.Glu1401* (c.4201G>T)
E1409X | p.Glu1409* (c.4225G>T)
C1410X | p.Cys1410* (c.4230C>A)
Q1411X | p.Gln1411* (c.4231C>T)
Q1412X | p.Gln1412* (c.4234C>T)
4374+1G->A | p.? (c.4242+1G>A)
4374+1G->T | p.? (c.4242+1G>T)
4382delA | p.Glu1418Argfs*14 (c.4251del)
4428insGA | p.Ser1435Glyfs*14 (c.4300_4301dup)

Cystic Fibrosis (CF) is screened in New York State (NYS) infants at birth using a three-tier IRT-DNA-SEQ algorithm. ***Tier 1***: Immunoreactive trypsinogen (IRT) is tested in all infants. ***Tier 2***: A custom second-tier panel targeting 1,018 clinically-relevant *CFTR* variants (v1.2) is screened in infants with elevated IRT (top 5%). Variants included on the panel are listed in the table. The panel was updated to v1.2 on 8/15/2025. Each variant is described using legacy[1] and Human Genome Variation Society (HGVS) nomenclature,[2] with cDNA nucleotide changes with respect to NCBI transcript NM_000492.4, and amino acid changes with respect to NCBI amino acid reference sequence NP_000483.3. Of the 1,018 targeted variants, 1,014 have been classified as CF-causing by The Clinical and Functional Translation of CFTR database (CFTR2),[3,4] two variants (R117H-7T and F311del/F312del) are of varying clinical consequence,[∫] and two variants (211delG and G451V) are classified as pathogenic[#] and likely-pathogenic[^], respectively, using American College of Medical Genetics and Genomics (ACMG) standards.[5] Large deletions/duplications defined as CF-causing by CFTR2 are not included on the second-tier panel. ***Tier 3:*** Other *CFTR* variants, including pathogenic and likely pathogenic variants[5] not catalogued in CFTR2; variants of varying clinical consequence;[4] and variants of unknown[4] or uncertain significance (VUS)[5] may be detected via expanded third-tier analysis, in which the complete *CFTR* coding sequence and other relevant regions are analyzed. Third-tier analysis is only conducted for infants with one second-tier panel variant or ultra-high IRT and no panel variants. Large deletions and duplications may be detected via third-tier analysis.

Variants recommended for population-based CF carrier screening[6] are shown in bold.

Most variants with protein effects listed as p.? represent variants that alter splicing.

[∫]Defined as a variant of varying clinical consequence by CFTR2. If R117H is detected, intron 9 (HGVS; legacy intron 8) polyT/TG status at c.1210-12T/c.1210-34TG is unmasked.

References

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NYS Cystic Fibrosis Newborn Screening: Custom *CFTR* Variant Panel Content

Assay, Software and Analysis Versions

	Version	Dates in use	Additional information
Custom Archer CFTR assay	1.0.0	07/01/2019 – present	
Archer Analysis software	6.0.4	07/01/2019 – 8/14/2025	
	7.3.3	8/15/2025 – present	
Tier 2 panel variants	1.0.0	07/01/2019 – 01/31/2022	338 variants
	1.1.0	02/01/2022 – 8/14/2025	369 variants - Added 32 CF-causing variants added to CFTR2 (release dates 01/10/2020, 07/31/2020, 09/24/2021). - Removed c.165-3C>T because CFTR2 reclassified from CF-causing to unknown significance.
	1.2.0	8/15/2025 – present	1,018 variants - Added 647 CF-causing variants added to CFTR2 (release date 9/25/2024). - Added c.80del (211delG) and G451V. - c.1820_1903del (1949del84) is called with a custom script as of 8/15/2025.
Tier 3 sequence analysis	1.0.0	07/01/2019 – present	5,248 nucleotides in *CFTR* gene analyzed. Effective 02/01/2022, 5T-11TG is no longer reported. CFTR2 reclassified to non CF-causing.
Intron 9 (legacy intron 8) polyT/TG analysis	1.0.0	07/01/2019 – 01/19/2020	
	2.0.0	01/20/2020 – 8/14/2025	
	2.0.1	8/15/2025 – present	Same algorithm. Minor changes to accommodate Archer Analysis v7.3.3 changes.