

# VCGS variant list for cystic fibrosis

## screening & diagnostic testing

Exon/intron number	Nucleotide change (NM_000492.3)	Protein change (NP_000483.3)	Diagnostic/screening
4	c.350G>A	p.Arg117His	Diagnostic assay only
Intron 9	c.1210-34TG[11_13]	polyTG tract	Diagnostic assay only
Intron 9	c.1210-12T[5_9]	polyT tract	Diagnostic assay only
Intron 2	c.165-3C>T	No protein name	Diagnostic and prepari <sup>®</sup> screening
Intron 2	c.165-1G>A	No protein name	Diagnostic and prepari <sup>®</sup> screening
3	c.166G>A	p.Glu56Lys	Diagnostic and prepari <sup>®</sup> screening
3	c.169T>G	p.Trp57Gly	Diagnostic and prepari <sup>®</sup> screening
3	c.170G>A	p.Trp57*	Diagnostic and prepari <sup>®</sup> screening
3	c.171G>A	p.Trp57*	Diagnostic and prepari <sup>®</sup> screening
3	c.175dupA	p.Arg59Lysfs*10	Diagnostic and prepari <sup>®</sup> screening
3	c.174_177delTAGA	p.Asp58Glufs*32	Diagnostic and prepari <sup>®</sup> screening
3	c.178G>T	p.Glu60*	Diagnostic and prepari <sup>®</sup> screening
3	c.200C>T	p.Pro67Leu	Diagnostic and prepari <sup>®</sup> screening
3	c.223C>T	p.Arg75*	Diagnostic and prepari <sup>®</sup> screening
3	c.233dupT	p.Trp79Leufs*32	Diagnostic and prepari <sup>®</sup> screening
3	c.254G>A	p.Gly85Glu	Diagnostic and prepari <sup>®</sup> screening
3	c.262_263delTT	p.Leu88Ilefs*22	Diagnostic and prepari <sup>®</sup> screening
3	c.263T>A	p.Leu88*	Diagnostic and prepari <sup>®</sup> screening
3	c.263T>G	p.Leu88*	Diagnostic and prepari <sup>®</sup> screening
Intron 3	c.273+1G>A	No protein name	Diagnostic and prepari <sup>®</sup> screening
Intron 3	c.273+3A>C	No protein name	Diagnostic and prepari <sup>®</sup> screening
Intron 3	c.274-2A>G	No protein name	Diagnostic and prepari <sup>®</sup> screening
Intron 3	c.274-1G>A	No protein name	Diagnostic and prepari <sup>®</sup> screening
4	c.274G>A	p.Glu92Lys	Diagnostic and prepari <sup>®</sup> screening
4	c.274G>T	p.Glu92*	Diagnostic and prepari <sup>®</sup> screening
4	c.292C>T	p.Gln98*	Diagnostic and prepari <sup>®</sup> screening
4	c.293A>G	p.Gln98Arg	Diagnostic and prepari <sup>®</sup> screening
4	c.296C>T	p.Pro99Leu	Diagnostic and prepari <sup>®</sup> screening
4	c.310delA	p.Arg104Glufs*3	Diagnostic and prepari <sup>®</sup> screening
4	c.313delA	p.Ile105Serfs*2	Diagnostic and prepari <sup>®</sup> screening
4	c.325_327delinsG	p.Tyr109Glyfs*4	Diagnostic and prepari <sup>®</sup> screening
4	c.328G>C	p.Asp110His	Diagnostic and prepari <sup>®</sup> screening
4	c.349C>T	p.Arg117Cys	Diagnostic and prepari <sup>®</sup> screening
4	c.366T>A	p.Tyr122*	Diagnostic and prepari <sup>®</sup> screening
4	c.409delC	p.Leu137Serfs*16	Diagnostic and prepari <sup>®</sup> screening
4	c.442delA	p.Ile148Leufs*5	Diagnostic and prepari <sup>®</sup> screening
Intron 4	c.489+1G>T	No protein name	Diagnostic and prepari <sup>®</sup> screening

5	c.494T>C	p.Leu165Ser	Diagnostic and prepar <sup>®</sup> screening
5	c.531delT	p.Ile177Metfs*12	Diagnostic and prepar <sup>®</sup> screening
5	c.532G>A	p.Gly178Arg	Diagnostic and prepar <sup>®</sup> screening
5	c.543_546delTAGT	p.Leu183Phefs*5	Diagnostic and prepar <sup>®</sup> screening
5	c.577G>T	p.Glu193*	Diagnostic and prepar <sup>®</sup> screening
Intron 5	c.579+1G>T	No protein name	Diagnostic and prepar <sup>®</sup> screening
Intron 5	c.579+3A>G	No protein name	Diagnostic and prepar <sup>®</sup> screening
Intron 5	c.579+5G>A	No protein name	Diagnostic and prepar <sup>®</sup> screening
8	c.933C>G	p.Phe311Leu	Diagnostic and prepar <sup>®</sup> screening
8	c.948delT	p.Phe316Leufs*12	Diagnostic and prepar <sup>®</sup> screening
8	c.987delA	p.Gly330Glufs*39	Diagnostic and prepar <sup>®</sup> screening
8	c.988G>T	p.Gly330*	Diagnostic and prepar <sup>®</sup> screening
8	c.1000C>T	p.Arg334Trp	Diagnostic and prepar <sup>®</sup> screening
8	c.1001G>T	p.Arg334Leu	Diagnostic and prepar <sup>®</sup> screening
8	c.1006_1007insG	p.Ile336Serfs*28	Diagnostic and prepar <sup>®</sup> screening
8	c.1007T>A	p.Ile336Lys	Diagnostic and prepar <sup>®</sup> screening
8	c.1013C>T	p.Thr338Ile	Diagnostic and prepar <sup>®</sup> screening
8	c.1021T>C	p.Ser341Pro	Diagnostic and prepar <sup>®</sup> screening
8	c.1021_1022dupTC	p.Phe342Hisfs*28	Diagnostic and prepar <sup>®</sup> screening
8	c.1029delC	p.Cys343*	Diagnostic and prepar <sup>®</sup> screening
8	c.1040G>A	p.Arg347His	Diagnostic and prepar <sup>®</sup> screening
8	c.1040G>C	p.Arg347Pro	Diagnostic and prepar <sup>®</sup> screening
8	c.1055G>A	p.Arg352Gln	Diagnostic and prepar <sup>®</sup> screening
8	c.1081delT	p.Trp361Glyfs*8	Diagnostic and prepar <sup>®</sup> screening
10	c.1240C>T	p.Gln414*	Diagnostic and prepar <sup>®</sup> screening
10	c.1301_1307delCACTTCT	p.Ser434Leufs*6	Diagnostic and prepar <sup>®</sup> screening
10	c.1327_1330dupGATA	p.Ile444Argfs*3	Diagnostic and prepar <sup>®</sup> screening
10	c.1340delA	p.Lys447Argfs*2	Diagnostic and prepar <sup>®</sup> screening
10	c.1364C>A	p.Ala455Glu	Diagnostic and prepar <sup>®</sup> screening
10	c.1365_1366delGG	p.Val456Cysfs*25	Diagnostic and prepar <sup>®</sup> screening
10	c.1367T>C	p.Val456Ala	Diagnostic and prepar <sup>®</sup> screening
10	c.1373delG	p.Gly458Aspfs*11	Diagnostic and prepar <sup>®</sup> screening
11	c.1397C>A	p.Ser466*	Diagnostic and prepar <sup>®</sup> screening
11	c.1397C>G	p.Ser466*	Diagnostic and prepar <sup>®</sup> screening
11	c.1400T>C	p.Leu467Pro	Diagnostic and prepar <sup>®</sup> screening
11	c.1418delG	p.Gly473Glufs*54	Diagnostic and prepar <sup>®</sup> screening
11	c.1438G>T	p.Gly480Cys	Diagnostic and prepar <sup>®</sup> screening
11	c.1466C>A	p.Ser489*	Diagnostic and prepar <sup>®</sup> screening
11	c.1475C>T	p.Ser492Phe	Diagnostic and prepar <sup>®</sup> screening
11	c.1477C>T	p.Gln493*	Diagnostic and prepar <sup>®</sup> screening
11	c.1477_1478delCA	p.Gln493Valfs*10	Diagnostic and prepar <sup>®</sup> screening
11	c.1487G>A	p.Trp496*	Diagnostic and prepar <sup>®</sup> screening
11	c.1519_1521delATC	p.Ile507del	Diagnostic and prepar <sup>®</sup> screening
11	c.1521_1523delCTT	p.Phe508del	Diagnostic and prepar <sup>®</sup> screening
11	c.1538A>G	p.Asp513Gly	Diagnostic and prepar <sup>®</sup> screening
11	c.1545_1546delTA	p.Tyr515*	Diagnostic and prepar <sup>®</sup> screening
11	c.1558G>T	p.Val520Phe	Diagnostic and prepar <sup>®</sup> screening
11	c.1572C>A	p.Cys524*	Diagnostic and prepar <sup>®</sup> screening
11	c.1573C>T	p.Gln525*	Diagnostic and prepar <sup>®</sup> screening

Intron 11	c.1584+1G>A	No protein name	Diagnostic and prepar <sup>®</sup> screening
Intron 11	c.1585-8G>A	No protein name	Diagnostic and prepar <sup>®</sup> screening
Intron 11	c.1585-1G>A	No protein name	Diagnostic and prepar <sup>®</sup> screening
12	c.1624G>T	p.Gly542*	Diagnostic and prepar <sup>®</sup> screening
12	c.1645A>C	p.Ser549Arg	Diagnostic and prepar <sup>®</sup> screening
12	c.1646G>A	p.Ser549Asn	Diagnostic and prepar <sup>®</sup> screening
12	c.1647T>G	p.Ser549Arg	Diagnostic and prepar <sup>®</sup> screening
12	c.1648G>T	p.Gly550*	Diagnostic and prepar <sup>®</sup> screening
12	c.1650delA	p.Gly551Valfs*8	Diagnostic and prepar <sup>®</sup> screening
12	c.1651G>A	p.Gly551Ser	Diagnostic and prepar <sup>®</sup> screening
12	c.1652G>A	p.Gly551Asp	Diagnostic and prepar <sup>®</sup> screening
12	c.1654C>T	p.Gln552*	Diagnostic and prepar <sup>®</sup> screening
12	c.1657C>T	p.Arg553*	Diagnostic and prepar <sup>®</sup> screening
12	c.1670delC	p.Ser557Phefs*2	Diagnostic and prepar <sup>®</sup> screening
12	c.1675G>A	p.Ala559Thr	Diagnostic and prepar <sup>®</sup> screening
12	c.1679G>C	p.Arg560Thr	Diagnostic and prepar <sup>®</sup> screening
Intron 12	c.1679+1G>C	No protein name	Diagnostic and prepar <sup>®</sup> screening
Intron 12	c.1680-1G>A	No protein name	Diagnostic and prepar <sup>®</sup> screening
13	c.1680A>C	p.Arg560Ser	Diagnostic and prepar <sup>®</sup> screening
13	c.1682C>A	p.Ala561Glu	Diagnostic and prepar <sup>®</sup> screening
13	c.1687T>A	p.Tyr563Asn	Diagnostic and prepar <sup>®</sup> screening
13	c.1687T>G	p.Tyr563Asp	Diagnostic and prepar <sup>®</sup> screening
13	c.1692delA	p.Asp565Metfs*7	Diagnostic and prepar <sup>®</sup> screening
13	c.1703delT	p.Leu568Cysfs*4	Diagnostic and prepar <sup>®</sup> screening
13	c.1705T>G	p.Tyr569Asp	Diagnostic and prepar <sup>®</sup> screening
13	c.1721C>A	p.Pro574His	Diagnostic and prepar <sup>®</sup> screening
13	c.1753G>T	p.Glu585*	Diagnostic and prepar <sup>®</sup> screening
Intron 13	c.1766+1G>A	No protein name	Diagnostic and prepar <sup>®</sup> screening
Intron 13	c.1766+1G>C	No protein name	Diagnostic and prepar <sup>®</sup> screening
Intron 13	c.1766+3A>G	No protein name	Diagnostic and prepar <sup>®</sup> screening
Intron 13	c.1766+5G>T	No protein name	Diagnostic and prepar <sup>®</sup> screening
14	c.1986_1989delAACT	p.Thr663Argfs*8	Diagnostic and prepar <sup>®</sup> screening
14	c.2012delT	p.Leu671*	Diagnostic and prepar <sup>®</sup> screening
14	c.2017G>T	p.Gly673*	Diagnostic and prepar <sup>®</sup> screening
14	c.2051_2052delinsG	p.Lys684Serfs*38	Diagnostic and prepar <sup>®</sup> screening
14	c.2052delA	p.Lys684Asnfs*38	Diagnostic and prepar <sup>®</sup> screening
14	c.2052dupA	p.Gln685Thrfs*4	Diagnostic and prepar <sup>®</sup> screening
14	c.2053C>T	p.Gln685*	Diagnostic and prepar <sup>®</sup> screening
14	c.2053dupC	p.Gln685Profs*4	Diagnostic and prepar <sup>®</sup> screening
14	c.2125C>T	p.Arg709*	Diagnostic and prepar <sup>®</sup> screening
14	c.2128A>T	p.Lys710*	Diagnostic and prepar <sup>®</sup> screening
14	c.2143C>T	p.Gln715*	Diagnostic and prepar <sup>®</sup> screening
14	c.2158C>T	p.Gln720*	Diagnostic and prepar <sup>®</sup> screening
14	c.2175dupA	p.Glu726Argfs*4	Diagnostic and prepar <sup>®</sup> screening
14	c.2195T>G	p.Leu732*	Diagnostic and prepar <sup>®</sup> screening
16	c.2645G>A	p.Trp882*	Diagnostic and prepar <sup>®</sup> screening
Intron 16	c.2657+5G>A	No protein name	Diagnostic and prepar <sup>®</sup> screening
18	c.2936A>T	p.Asp979Val	Diagnostic and prepar <sup>®</sup> screening
18	c.2988G>A	No protein name	Diagnostic and prepar <sup>®</sup> screening

Intron 18	c.2988+1G>A	No protein name	Diagnostic and prepar <sup>®</sup> screening
Intron 18	c.2989-2A>G	No protein name	Diagnostic and prepar <sup>®</sup> screening
Intron 18	c.2989-1G>A	No protein name	Diagnostic and prepar <sup>®</sup> screening
19	c.3002_3003delTG	p.Val1001Aspfs*45	Diagnostic and prepar <sup>®</sup> screening
19	c.3039delC	p.Tyr1014Thrfs*9	Diagnostic and prepar <sup>®</sup> screening
19	c.3039dupC	p.Tyr1014Leufs*33	Diagnostic and prepar <sup>®</sup> screening
19	c.3067_3072del	p.Ile1023_Val1024del	Diagnostic and prepar <sup>®</sup> screening
19	c.3124C>T	p.Gln1042*	Diagnostic and prepar <sup>®</sup> screening
22	c.3472C>T	p.Arg1158*	Diagnostic and prepar <sup>®</sup> screening
22	c.3476C>T	p.Ser1159Phe	Diagnostic and prepar <sup>®</sup> screening
22	c.3484C>T	p.Arg1162*	Diagnostic and prepar <sup>®</sup> screening
22	c.3528delC	p.Lys1177Serfs*15	Diagnostic and prepar <sup>®</sup> screening
22	c.3532_3535dupTCAA	p.Thr1179Ilefs*17	Diagnostic and prepar <sup>®</sup> screening
22	c.3587C>G	p.Ser1196*	Diagnostic and prepar <sup>®</sup> screening
22	c.3605delA	p.Asp1202Alafs*9	Diagnostic and prepar <sup>®</sup> screening
22	c.3611G>A	p.Trp1204*	Diagnostic and prepar <sup>®</sup> screening
22	c.3612G>A	p.Trp1204*	Diagnostic and prepar <sup>®</sup> screening
22	c.3659delC	p.Thr1220Lysfs*8	Diagnostic and prepar <sup>®</sup> screening
22	c.3691delT	p.Ser1231Profs*4	Diagnostic and prepar <sup>®</sup> screening
22	c.3700A>G	p.Ile1234Val	Diagnostic and prepar <sup>®</sup> screening
Intron 22	c.3718-2477C>T	No protein name	Diagnostic and prepar <sup>®</sup> screening
Intron 22	c.3718-3T>G	No protein name	Diagnostic and prepar <sup>®</sup> screening
Intron 22	c.3718-1G>A	No protein name	Diagnostic and prepar <sup>®</sup> screening
23	c.3731G>A	p.Gly1244Glu	Diagnostic and prepar <sup>®</sup> screening
23	c.3744delA	p.Lys1250Argfs*9	Diagnostic and prepar <sup>®</sup> screening
23	c.3747delG	p.Lys1250Argfs*9	Diagnostic and prepar <sup>®</sup> screening
23	c.3752G>A	p.Ser1251Asn	Diagnostic and prepar <sup>®</sup> screening
23	c.3761T>G	p.Leu1254*	Diagnostic and prepar <sup>®</sup> screening
23	c.3763T>C	p.Ser1255Pro	Diagnostic and prepar <sup>®</sup> screening
23	c.3764C>A	p.Ser1255*	Diagnostic and prepar <sup>®</sup> screening
23	c.3773dupT	p.Leu1258Phefs*7	Diagnostic and prepar <sup>®</sup> screening
23	c.3846G>A	p.Trp1282*	Diagnostic and prepar <sup>®</sup> screening
23	c.3848G>T	p.Arg1283Met	Diagnostic and prepar <sup>®</sup> screening
Intron 23	c.3873+1G>A	No protein name	Diagnostic and prepar <sup>®</sup> screening
Intron 23	c.3873+2T>C	No protein name	Diagnostic and prepar <sup>®</sup> screening
24	c.3883_3886delATTT	p.Ile1295Phefs*32	Diagnostic and prepar <sup>®</sup> screening
24	c.3883delA	p.Ile1295Phefs*33	Diagnostic and prepar <sup>®</sup> screening
24	c.3889dupT	p.Ser1297Phefs*5	Diagnostic and prepar <sup>®</sup> screening
24	c.3891dupT	p.Gly1298Trpfs*4	Diagnostic and prepar <sup>®</sup> screening
24	c.3900dupT	p.Arg1301*	Diagnostic and prepar <sup>®</sup> screening
24	c.3908delA	p.Asn1303Thrfs*25	Diagnostic and prepar <sup>®</sup> screening
24	c.3909C>G	p.Asn1303Lys	Diagnostic and prepar <sup>®</sup> screening
24	c.3937C>T	p.Gln1313*	Diagnostic and prepar <sup>®</sup> screening