Elenco mutazioni analizzate mediante SmartSeq (Amplicon Suite) versione 3.3.0 (*analisi "screening"*)

Variant	Legacy	c.HGVS	p.HGVS
SNP	1078delT (CF-causing)	c.948delT	p.Phe316Leufs*12
SNP	1119delA (CF-causing)	c.987delA	p.Gly330Glufs*39
SNP	1138insG (CF-causing)	c.1006_1007insG	p.Ile336Serfs*28
SNP	1154insTC (CF-causing)	c.1021_1022dupTC	p.Phe342Hisfs*28
SNP	1161delC (CF-causing)	c.1029delC	p.Cys343Ter
SNP	1213delT (CF-causing)	c.1081delT	p.Trp361Glyfs*8
SNP	1248+1G->A (CF-causing)	c.1116+1G>A	-
SNP	1249-1G->A (CF-causing)	c.1117-1G>A	-
SNP	124del23bp (CF-causing)	c.-9_14del23	-
SNP	1259insA (CF-causing)	c.1130dupA	p.Gln378Alafs*4
SNP	1288insTA (CF-causing)	c.1155_1156dupTA	p.Asn386Ilefs*3
SNP	1341+1G->A (CF-causing)	c.1209+1G>A	-
SNP	1343delG (CF-causing)	c.1211delG	p.Gly404Aspfs*38
SNP	1429del7 (CF-causing)	c.1301_1307delCACTTCT	p.Ser434LeufsX6
SNP	1461ins4 (CF-causing)	c.1327_1330dupGATA	p.Ile444Argfs*3
SNP	1471delA (CF-causing)	c.1340delA	p.Lys447Argfs*2
SNP	1497delGG (CF-causing)	c.1365_1366delGG	p.Val456Cysfs*25
SNP	1504delG (CF-causing)	c.1373delG	p.Gly458AspfsX11
SNP	1525-1G->A (CF-causing)	c.1393-1G>A	-
SNP	1525-2A->G (CF-causing)	c.1393-2A>G	-
SNP	1548delG (CF-causing)	c.1418delG	p.Gly473Glufs*54
SNP	1609delCA (CF-causing)	c.1477_1478delCA	p.Gln493Valfs*10
SNP	1677delTA (CF-causing)	c.1545_1546delTA	p.Tyr515Ter
SNP	1716+1G->A (CF-causing)	c.1584+1G>A	-
SNP	1717-1G->A (CF-causing)	c.1585-1G>A	-
SNP	1717-8G->A (CF-causing)	c.1585-8G>A	-
SNP	1782delA (CF-causing)	c.1650delA	p.Gly551Valfs*8
SNP	1802delC (CF-causing)	c.1670delC	p.Ser557PhefsX2
SNP	1811+1.6kbA->G (CF-causing)	c.1680-886A>G	-
SNP	1811+1643G->T (CF-causing)	c.1680-877G>T	-
SNP	1811+1G->A (CF-causing)	c.1679+1G>A	-
SNP	1811+1G->C (CF-causing)	c.1679+1G>C	-
SNP	1812-1G->A (CF-causing)	c.1680-1G>A	-
SNP	1824delA (CF-causing)	c.1692delA	p.Asp565Metfs*7
SNP	182delT (CF-causing)	c.50delT	p.Phe17Serfs*8
SNP	1833delT (CF-causing)	c.1703delT	p.Leu568Cysfs*4
SNP	185+1G->T (CF-causing)	c.53+1G>T	-
SNP	1898+1G->A (CF-causing)	c.1766+1G>A	-
SNP	1898+1G->C (CF-causing)	c.1766+1G>C	-

Screening database version: CFTR2_Agosto 2018 (mutazioni causanti malattia: 336; varianti con conseguenze cliniche variabili:35)

Elenco mutazioni analizzate mediante SmartSeq (Amplicon Suite) versione 3.3.0 (*analisi "screening"*)

SNP	1898+1G->T (CF-causing)	c.1766+1G>T	-
SNP	1898+3A->G (CF-causing)	c.1766+3A>G	-
SNP	1898+5G->T (CF-causing)	c.1766+5G>T	-
SNP	1924del7 (CF-causing)	c.1792_1798delAAAACTA	p.Lys598Glyfs*11
SNP	2055del9->A (CF-causing)	c.1923_1931delCTCAAAACTinsA	p.Ser641Argfs*5
SNP	2075delA (CF-causing)	c.1943delA	p.Asp648ValfsX15
SNP	2105-2117del13insAGAAA (CF-causing)	c.1973_1985delGAAATTCAATCCTinsAGAAA	p.Arg658Lysfs*4
SNP	2118del4 (CF-causing)	c.1986_1989delAACT	p.Thr663Argfs*8
SNP	2143delT (CF-causing)	c.2012delT	p.Leu671Ter
SNP	2183AA->G or 2183delAA->G (CF-causing)	c.2051_2052delAAinsG	p.Lys684Serfs*38
SNP	2184delA (CF-causing)	c.2052delA	p.Lys684Asnfs*38
SNP	2184insA (CF-causing)	c.2052dupA	p.Gln685Thrfs*4
SNP	2185insC (CF-causing)	c.2053dupC	p.Gln685Profs*4
SNP	2307insA (CF-causing)	c.2175dupA	p.Glu726Argfs*4
SNP	2347delG (CF-causing)	c.2215delG	p.Val739Tyrfs*16
SNP	2372del8 (CF-causing)	c.2241_2248delGATACTGC	p.Ile748Serfs*28
SNP	2556insAT (CF-causing)	c.2423_2424dupAT	p.Ser809Ilefs*13
SNP	2585delT (CF-causing)	c.2453delT	p.Leu818Trpfs*3
SNP	2594delGT (CF-causing)	c.2463_2464delTG	p.Ser821Argfs*4
SNP	2622+1G->A (CF-causing)	c.2490+1G>A	-
SNP	2711delT (CF-causing)	c.2583delT	p.Phe861Leufs*3
SNP	2721del11 (CF-causing)	c.2589_2599delAATTTGGTGCT	p.Ile864Serfs*28
SNP	2732insA (CF-causing)	c.2601dupA	p.Val868Serfs*28
SNP	2789+5G->A (CF-causing)	c.2657+5G>A	-
SNP	2790-1G->C (CF-causing)	c.2658-1G>C	-
SNP	2869insG (CF-causing)	c.2737_2738insG	p.Tyr913Ter
SNP	2896insAG (CF-causing)	c.2763_2764dupAG	p.Val922Glufs*2
SNP	2942insT (CF-causing)	c.2810dupT	p.Val938Glyfs*37
SNP	2957delT (CF-causing)	c.2825delT	p.Ile942Thrfs*26
SNP	296+1G->A (CF-causing)	c.164+1G>A	-
SNP	296+1G->T (CF-causing)	c.164+1G>T	-
SNP	296+2T->C (CF-causing)	c.164+2T>C	-
SNP	296+3insT (CF-causing)	c.164+3_164+4insT	-
SNP	297-1G->A (CF-causing)	c.165-1G>A	-
SNP	297-3C->T (CF-causing)	c.165-3C>T	-
SNP	2991del32 (CF-causing)	c.2859_2890del32	p.Leu953Phefs*11
SNP	3007delG (CF-causing)	c.2875delG	p.Ala959Hisfs*9
SNP	3028delA (CF-causing)	c.2896delA	p.Thr966Argfs*2

Screening database version: CFTR2_Agosto 2018 (mutazioni causanti malattia: 336; varianti con conseguenze cliniche variabili:35)

Elenco mutazioni analizzate mediante SmartSeq (Amplicon Suite) versione 3.3.0 (*analisi "screening"*)

SNP	306delTAGA (CF-causing)	c.174_177delTAGA	p.Asp58Glufs*32
SNP	306insA (CF-causing)	c.175dupA	p.Arg59Lysfs*10
SNP	3120+1G->A (CF-causing)	c.2988+1G>A	-
SNP	3120G->A (CF-causing)	c.2988G>A	p.=
SNP	3121-1G->A (CF-causing)	c.2989-1G>A	-
SNP	3121-2A->G (CF-causing)	c.2989-2A>G	-
SNP	3132delTG (CF-causing)	c.3002_3003delTG	p.Val1001Aspfs*45
SNP	3143del9 (CF-causing)	c.3011_3019delCTATAGCAG	p.Ala1004_Ala1006del
SNP	3143del9 (CF-causing)	c.3009_3017delAGCTATAGC	p.Ala1004_Ala1006del
SNP	3171delC (CF-causing)	c.3039delC	p.Tyr1014Thrfs*9
SNP	3171insC (CF-causing)	c.3039dupC	p.Tyr1014Leufs*33
SNP	3271delGG (CF-causing)	c.3139_3139+1delGG	-
SNP	3272-26A->G (CF-causing)	c.3140-26A>G	-
SNP	3349insT (CF-causing)	c.3217dupT	p.Tyr1073Leufs*3
SNP	3500-2A->G (CF-causing)	c.3368-2A>G	-
SNP	3600+2insT (CF-causing)	c.3468+2dupT	-
SNP	3600+5G->A (CF-causing)	c.3468+5G>A	-
SNP	3600G->A (CF-causing)	c.3468G>A	p.=
SNP	365-366insT (CF-causing)	c.233dupT	p.Trp79Leufs*32
SNP	3659delC (CF-causing)	c.3528delC	p.Lys1177Serfs*15
SNP	3667ins4 (CF-causing)	c.3532_3535dupTCAA	p.Thr1179Ilefs*17
SNP	3737delA (CF-causing)	c.3605delA	p.Asp1202Alafs*9
SNP	3791delC (CF-causing)	c.3659delC	p.Thr1220Lysfs*8
SNP	3821delT (CF-causing)	c.3691delT	p.Ser1231Profs*4
SNP	3849+10kbC->T (CF-causing)	c.3718-2477C>T	-
SNP	3849+40A->G (CF-causing)	c.3717+40A>G	-
SNP	3849+4A->G (CF-causing)	c.3717+4A>G	-
SNP	3849+5G->A (CF-causing)	c.3717+5G>A	-
SNP	3849G->A (CF-causing)	c.3717G>A	p.=
SNP	3850-1G->A (CF-causing)	c.3718-1G>A	-
SNP	3850-3T->G (CF-causing)	c.3718-3T>G	-
SNP	3876delA (CF-causing)	c.3744delA	p.Lys1250Argfs*9
SNP	3878delG (CF-causing)	c.3747delG	p.Lys1250Argfs*9
SNP	3905insT (CF-causing)	c.3773dupT	p.Leu1258Phefs*7
SNP	394delTT (CF-causing)	c.262_263delTT	p.Leu88Ilefs*22
SNP	4005+1G->A (CF-causing)	c.3873+1G>A	-
SNP	4005+2T->C (CF-causing)	c.3873+2T>C	-
SNP	4010del4 (CF-causing)	c.3883_3886delATTT	p.Ile1295Phefs*32
SNP	4015delA (CF-causing)	c.3883delA	p.Ile1295Phefs*33
SNP	4016insT (CF-causing)	c.3889dupT	p.Ser1297Phefs*5

Screening database version: CFTR2_Agosto 2018 (mutazioni causanti malattia: 336; varianti con conseguenze cliniche variabili:35)

Elenco mutazioni analizzate mediante SmartSeq (Amplicon Suite) versione 3.3.0 (*analisi "screening"*)

SNP	4022insT (CF-causing)	c.3891dupT	p.Gly1298Trpfs*4
SNP	4040delA (CF-causing)	c.3908delA	p.Asn1303Thrfs*25
SNP	405+1G->A (CF-causing)	c.273+1G>A	-
SNP	405+3A->C (CF-causing)	c.273+3A>C	-
SNP	406-1G->A (CF-causing)	c.274-1G>A	-
SNP	406-2A->G (CF-causing)	c.274-2A>G	-
SNP	4168delCTAAGCC (CF-causing)	c.4036_4042delCTAAGCC	p.Leu1346MetfsX6
SNP	4209TGTT->AA (CF-causing)	c.4077_4080delTGTTinsAA	p.Val1360Thrfs*3
SNP	4218insT (CF-causing)	c.4086dupT	p.Lys1363Ter
SNP	4259del5 (CF-causing)	c.4127_4131delTGGAT	p.Leu1376Serfs*8
SNP	4279insA (CF-causing)	c.4147dupA	p.Ile1383Asnfs*3
SNP	4326delTC (CF-causing)	c.4197_4198delCT	p.Cys1400Ter
SNP	4374+1G->A (CF-causing)	c.4242+1G>A	-
SNP	4374+1G->T (CF-causing)	c.4242+1G>T	-
SNP	4382delA (CF-causing)	c.4251delA	p.Glu1418Argfs*14
SNP	4428insGA (CF-causing)	c.4300_4301dupAG	p.Ser1435Glyfs*14
SNP	442delA (CF-causing)	c.310delA	p.Arg104Glufs*3
SNP	444delA (CF-causing)	c.313delA	p.Ile105Serfs*2
SNP	457TAT->G (CF-causing)	c.325_327delTATinsG	p.Tyr109Glyfs*4
SNP	541delC (CF-causing)	c.409delC	p.Leu137Serfs*16
SNP	574delA (CF-causing)	c.442delA	p.Ile148Leufs*5
SNP	602del14 (CF-causing)	c.470_483del14	p.Phe157Ter
SNP	621+1G->T (CF-causing)	c.489+1G>T	-
SNP	621+3A->G (V.c.c.)	c.489+3A>G	-
SNP	663delT (CF-causing)	c.531delT	p.Ile177Metfs*12
SNP	675del4 (CF-causing)	c.543_546delTAGT	p.Leu183Phefs*5
SNP	711+1G->T (CF-causing)	c.579+1G>T	-
SNP	711+3A->G (CF-causing)	c.579+3A>G	-
SNP	711+5G->A (CF-causing)	c.579+5G>A	-
SNP	712-1G->T (CF-causing)	c.580-1G>T	-
SNP	849delG (CF-causing)	c.717delG	p.Leu240Ter
SNP	852del22 (CF-causing)	c.723_743+1delGAGAATGATGA TGAAGTACAGG	-
SNP	935delA (CF-causing)	c.803delA	p.Asn268Ilefs*17
SNP	977insA (CF-causing)	c.850dupA	p.Met284AsnfsX3
SNP	991del5 (CF-causing)	c.861_865delCTTAA	p.Asn287Lysfs*19
SNP	A1006E (CF-causing)	c.3017C>A	p.Ala1006Glu
SNP	A455E (CF-causing)	c.1364C>A	p.Ala455Glu
SNP	A46D (CF-causing)	c.137C>A	p.Ala46Asp
SNP	A559T (CF-causing)	c.1675G>A	p.Ala559Thr

Screening database version: CFTR2_Agosto 2018 (mutazioni causanti malattia: 336; varianti con conseguenze cliniche variabili:35)

Elenco mutazioni analizzate mediante SmartSeq (Amplicon Suite) versione 3.3.0 (*analisi "screening"*)

SNP	A561E (CF-causing)	c.1682C>A	p.Ala561Glu
SNP	A613T (CF-causing)	c.1837G>A	p.Ala613Thr
SNP	C276X (CF-causing)	c.828C>A	p.Cys276Ter
SNP	C524X (CF-causing)	c.1572C>A	p.Cys524Ter
SNP	D110E (V.c.c.)	c.330C>A	p.Asp110Glu
SNP	D110H (CF-causing)	c.328G>C	p.Asp110His
SNP	D1152H (V.c.c.)	c.3454G>C	p.Asp1152His
SNP	D1270N (V.c.c.)	c.3808G>A	p.Asp1270Asn
SNP	D443Y (V.c.c.)	c.1327G>T	p.Asp443Tyr
SNP	D513G (CF-causing)	c.1538A>G	p.Asp513Gly
SNP	D579G (V.c.c.)	c.1736A>G	p.Asp579Gly
SNP	D614G (V.c.c.)	c.1841A>G	p.Asp614Gly
SNP	D979V (CF-causing)	c.2936A>T	p.Asp979Val
SNP	E1104X (CF-causing)	c.3310G>T	p.Glu1104Ter
SNP	E1371X (CF-causing)	c.4111G>T	p.Glu1371Ter
SNP	E193X (CF-causing)	c.577G>T	p.Glu193Ter
SNP	E474K (CF-causing)	c.1420G>A	p.Glu474Lys
SNP	E56K (CF-causing)	c.166G>A	p.Glu56Lys
SNP	E585X (CF-causing)	c.1753G>T	p.Glu585Ter
SNP	E588V (V.c.c.)	c.1763A>T	p.Glu588Val
SNP	E60X (CF-causing)	c.178G>T	p.Glu60Ter
SNP	E822X (CF-causing)	c.2464G>T	p.Glu822Ter
SNP	E831X (CF-causing)	c.2491G>T	p.Glu831Ter
SNP	E92K (CF-causing)	c.274G>A	p.Glu92Lys
SNP	E92X (CF-causing)	c.274G>T	p.Glu92Ter
SNP	F1016S (V.c.c.)	c.3047T>C	p.Phe1016Ser
SNP	F1052V (V.c.c.)	c.3154T>G	p.Phe1052Val
SNP	F1074L (V.c.c.)	c.3222T>A	p.Phe1074Leu
SNP	F1099L (V.c.c.)	c.3297C>A	p.Phe1099Leu
SNP	F311L (CF-causing)	c.933C>G	p.Phe311Leu
SNP	F508del (CF-causing)	c.1521_1523delCTT	p.Phe508del
SNP	F575Y (V.c.c.)	c.1724T>A	p.Phe575Tyr
SNP	G1061R (CF-causing)	c.3181G>C	p.Gly1061Arg
SNP	G1069R (V.c.c.)	c.3205G>A	p.Gly1069Arg
SNP	G1244E (CF-causing)	c.3731G>A	p.Gly1244Glu
SNP	G1349D (CF-causing)	c.4046G>A	p.Gly1349Asp
SNP	G178R (CF-causing)	c.532G>A	p.Gly178Arg
SNP	G27R (CF-causing)	c.79G>A	p.Gly27Arg
SNP	G27X (CF-causing)	c.79G>T	p.Gly27Ter
SNP	G330X (CF-causing)	c.988G>T	p.Gly330Ter

Screening database version: CFTR2_Agosto 2018 (mutazioni causanti malattia: 336; varianti con conseguenze cliniche variabili:35)

Elenco mutazioni analizzate mediante SmartSeq (Amplicon Suite) versione 3.3.0 (*analisi "screening"*)

SNP	G542X (CF-causing)	c.1624G>T	p.Gly542Ter
SNP	G550X (CF-causing)	c.1648G>T	p.Gly550Ter
SNP	G551D (CF-causing)	c.1652G>A	p.Gly551Asp
SNP	G551S (CF-causing)	c.1651G>A	p.Gly551Ser
SNP	G622D (V.c.c.)	c.1865G>A	p.Gly622Asp
SNP	G673X (CF-causing)	c.2017G>T	p.Gly673Ter
SNP	G85E (CF-causing)	c.254G>A	p.Gly85Glu
SNP	G91R (CF-causing)	c.271G>A	p.Gly91Arg
SNP	G970D (CF-causing)	c.2909G>A	p.Gly970Asp
SNP	G970R (CF-causing)	c.2908G>C	p.Gly970Arg
SNP	H1054D (CF-causing)	c.3160C>G	p.His1054Asp
SNP	H1375P (CF-causing)	c.4124A>C	p.His1375Pro
SNP	H139R (CF-causing)	c.416A>G	p.His139Arg
SNP	H199Y (CF-causing)	c.595C>T	p.His199Tyr
SNP	H609R (CF-causing)	c.1826A>G	p.His609Arg
SNP	I1234V (CF-causing)	c.3700A>G	p.Ile1234Val
SNP	I336K (CF-causing)	c.1007T>A	p.Ile336Lys
SNP	I502T (CF-causing)	c.1505T>C	p.Ile502Thr
SNP	I507del (CF-causing)	c.1519_1521delATC	p.Ile507del
SNP	K710X (CF-causing)	c.2128A>T	p.Lys710Ter
SNP	L102R (CF-causing)	c.305T>G	p.Leu102Arg
SNP	L1065P (CF-causing)	c.3194T>C	p.Leu1065Pro
SNP	L1077P (CF-causing)	c.3230T>C	p.Leu1077Pro
SNP	L1254X (CF-causing)	c.3761T>G	p.Leu1254Ter
SNP	L1324P (CF-causing)	c.3971T>C	p.Leu1324Pro
SNP	L1335P (CF-causing)	c.4004T>C	p.Leu1335Pro
SNP	L138ins (CF-causing)	c.413_415dupTAC	p.Leu138dup
SNP	L15P (CF-causing)	c.44T>C	p.Leu15Pro
SNP	L165S (CF-causing)	c.494T>C	p.Leu165Ser
SNP	L206W (CF-causing)	c.617T>G	p.Leu206Trp
SNP	L227R (CF-causing)	c.680T>G	p.Leu227Arg
SNP	L453S (CF-causing)	c.1358T>C	p.Leu453Ser
SNP	L467P (CF-causing)	c.1400T>C	p.Leu467Pro
SNP	L558S (CF-causing)	c.1673T>C	p.Leu558Ser
SNP	L732X (CF-causing)	c.2195T>G	p.Leu732Ter
SNP	L88X (CF-causing)	c.263T>A	p.Leu88Ter
SNP	L88X (CF-causing)	c.263T>G	p.Leu88Ter
SNP	L927P (CF-causing)	c.2780T>C	p.Leu927Pro
SNP	L967S (V.c.c.)	c.2900T>C	p.Leu967Ser
SNP	M1101K (CF-causing)	c.3302T>A	p.Met1101Lys

Screening database version: CFTR2_Agosto 2018 (mutazioni causanti malattia: 336; varianti con conseguenze cliniche variabili:35)

Elenco mutazioni analizzate mediante SmartSeq (Amplicon Suite) versione 3.3.0 (*analisi "screening"*)

SNP	M1V (CF-causing)	c.1A>G	p.Met1Val
SNP	M265R (V.c.c.)	c.794T>G	p.Met265Arg
SNP	N1303K (CF-causing)	c.3909C>G	p.Asn1303Lys
SNP	P205S (CF-causing)	c.613C>T	p.Pro205Ser
SNP	P574H (CF-causing)	c.1721C>A	p.Pro574His
SNP	P5L (V.c.c.)	c.14C>T	p.Pro5Leu
SNP	P67L (CF-causing)	c.200C>T	p.Pro67Leu
SNP	P750L (V.c.c.)	c.2249C>T	p.Pro750Leu
SNP	P99L (CF-causing)	c.296C>T	p.Pro99Leu
SNP	Q1042X (CF-causing)	c.3124C>T	p.Gln1042Ter
SNP	Q1291R (V.c.c.)	c.3872A>G	p.Gln1291Arg
SNP	Q1313X (CF-causing)	c.3937C>T	p.Gln1313Ter
SNP	Q1330X (CF-causing)	c.3988C>T	p.Gln1330Ter
SNP	Q1382X (CF-causing)	c.4144C>T	p.Gln1382Ter
SNP	Q1411X (CF-causing)	c.4231C>T	p.Gln1411Ter
SNP	Q1412X (CF-causing)	c.4234C>T	p.Gln1412Ter
SNP	Q220X (CF-causing)	c.658C>T	p.Gln220Ter
SNP	Q2X (CF-causing)	c.4C>T	p.Gln2Ter
SNP	Q30X (CF-causing)	c.88C>T	p.Gln30X
SNP	Q359K/T360K (CF-causing)	c.[1075C>A	1079C>A]
SNP	Q39X (CF-causing)	c.115C>T	p.Gln39Ter
SNP	Q414X (CF-causing)	c.1240C>T	p.Gln414Ter
SNP	Q493X (CF-causing)	c.1477C>T	p.Gln493Ter
SNP	Q525X (CF-causing)	c.1573C>T	p.Gln525Ter
SNP	Q552X (CF-causing)	c.1654C>T	p.Gln552Ter
SNP	Q685X (CF-causing)	c.2053C>T	p.Gln685Ter
SNP	Q715X (CF-causing)	c.2143C>T	p.Gln715Ter
SNP	Q720X (CF-causing)	c.2158C>T	p.Gln720X
SNP	Q890X (CF-causing)	c.2668C>T	p.Gln890Ter
SNP	Q98R (CF-causing)	c.293A>G	p.Gln98Arg
SNP	Q98X (CF-causing)	c.292C>T	p.Gln98Ter
SNP	R1066C (CF-causing)	c.3196C>T	p.Arg1066Cys
SNP	R1066H (CF-causing)	c.3197G>A	p.Arg1066His
SNP	R1070Q (V.c.c.)	c.3209G>A	p.Arg1070Gln
SNP	R1070W (V.c.c.)	c.3208C>T	p.Arg1070Trp
SNP	R1102X (CF-causing)	c.3304A>T	p.Arg1102Ter
SNP	R1158X (CF-causing)	c.3472C>T	p.Arg1158Ter
SNP	R1162X (CF-causing)	c.3484C>T	p.Arg1162Ter
SNP	R117C (CF-causing)	c.349C>T	p.Arg117Cys
SNP	R117G (V.c.c.)	c.349C>G	p.Arg117Gly

Screening database version: CFTR2_Agosto 2018 (mutazioni causanti malattia: 336; varianti con conseguenze cliniche variabili:35)

Elenco mutazioni analizzate mediante SmartSeq (Amplicon Suite) versione 3.3.0 (*analisi "screening"*)

SNP	R117H (please check polyT) (V.c.c.)	c.350G>A	p.Arg117His
SNP	R1283M (CF-causing)	c.3848G>T	p.Arg1283Met
SNP	R334L (CF-causing)	c.1001G>T	p.Arg334Leu
SNP	R334Q (V.c.c.)	c.1001G>A	p.Arg334Gln
SNP	R334W (CF-causing)	c.1000C>T	p.Arg334Trp
SNP	R347H (CF-causing)	c.1040G>A	p.Arg347His
SNP	R347P (CF-causing)	c.1040G>C	p.Arg347Pro
SNP	R352Q (CF-causing)	c.1055G>A	p.Arg352Gln
SNP	R352W (V.c.c.)	c.1054C>T	p.Arg352Trp
SNP	R553X (CF-causing)	c.1657C>T	p.Arg553Ter
SNP	R560K (CF-causing)	c.1679G>A	p.Arg560Lys
SNP	R560S (CF-causing)	c.1680A>C	p.Arg560Ser
SNP	R560T (CF-causing)	c.1679G>C	p.Arg560Thr
SNP	R709X (CF-causing)	c.2125C>T	p.Arg709Ter
SNP	R74W (V.c.c.)	c.220C>T	p.Arg74Trp
SNP	R75X (CF-causing)	c.223C>T	p.Arg75Ter
SNP	R764X (CF-causing)	c.2290C>T	p.Arg764Ter
SNP	R785X (CF-causing)	c.2353C>T	p.Arg785Ter
SNP	R792X (CF-causing)	c.2374C>T	p.Arg792Ter
SNP	R851X (CF-causing)	c.2551C>T	p.Arg851Ter
SNP	S1118F (CF-causing)	c.3353C>T	p.Ser1118Phe
SNP	S1159F (CF-causing)	c.3476C>T	p.Ser1159Phe
SNP	S1159P (CF-causing)	c.3475T>C	p.Ser1159Pro
SNP	S1196X (CF-causing)	c.3587C>G	p.Ser1196Ter
SNP	S1251N (CF-causing)	c.3752G>A	p.Ser1251Asn
SNP	S1255P (CF-causing)	c.3763T>C	p.Ser1255Pro
SNP	S1255X (CF-causing)	c.3764C>A	p.Ser1255Ter
SNP	S13F (CF-causing)	c.38C>T	p.Ser13Phe
SNP	S341P (CF-causing)	c.1021T>C	p.Ser341Pro
SNP	S466X (CF-causing)	c.1397C>A	p.Ser466Ter
SNP	S466X (CF-causing)	c.1397C>G	p.Ser466Ter
SNP	S489X (CF-causing)	c.1466C>A	p.Ser489Ter
SNP	S492F (CF-causing)	c.1475C>T	p.Ser492Phe
SNP	S4X (CF-causing)	c.11C>A	p.Ser4Ter
SNP	S549N (CF-causing)	c.1646G>A	p.Ser549Asn
SNP	S549R (CF-causing)	c.1645A>C	p.Ser549Arg
SNP	S549R (CF-causing)	c.1647T>G	p.Ser549Arg
SNP	S912X (CF-causing)	c.2735C>A	p.Ser912Ter
SNP	S945L (CF-causing)	c.2834C>T	p.Ser945Leu
SNP	S977F (V.c.c.)	c.2930C>T	p.Ser977Phe

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Elenco mutazioni analizzate mediante SmartSeq (Amplicon Suite) versione 3.3.0 (*analisi "screening"*)

SNP	T1246I (V.c.c.)	c.3737C>T	p.Thr1246Ile
SNP	T338I (CF-causing)	c.1013C>T	p.Thr338Ile
SNP	V1153E (V.c.c.)	c.3458T>A	p.Val1153Glu
SNP	V1240G (CF-causing)	c.3719T>G	p.Val1240Gly
SNP	V232D (CF-causing)	c.695T>A	p.Val232Asp
SNP	V456A (CF-causing)	c.1367T>C	p.Val456Ala
SNP	V520F (CF-causing)	c.1558G>T	p.Val520Phe
SNP	W1089X (CF-causing)	c.3266G>A	p.Trp1089Ter
SNP	W1098C (CF-causing)	c.3294G>C	p.Trp1098Cys
SNP	W1098C (CF-causing)	c.3294G>T	p.Trp1098Cys
SNP	W1098X (CF-causing)	c.3293G>A	p.Trp1098Ter
SNP	W1098X (CF-causing)	c.3294G>A	p.Trp1098Ter
SNP	W1145X (CF-causing)	c.3435G>A	p.Trp1145Ter
SNP	W1204X (CF-causing)	c.3611G>A	p.Trp1204Ter
SNP	W1204X (CF-causing)	c.3612G>A	p.Trp1204Ter
SNP	W1282X (CF-causing)	c.3846G>A	p.Trp1282Ter
SNP	W19X (CF-causing)	c.57G>A	p.Trp19Ter
SNP	W216X (CF-causing)	c.647G>A	p.Trp216Ter
SNP	W401X (CF-causing)	c.1202G>A	p.Trp401Ter
SNP	W401X (CF-causing)	c.1203G>A	p.Trp401Ter
SNP	W496X (CF-causing)	c.1487G>A	p.Trp496Ter
SNP	W57G (CF-causing)	c.169T>G	p.Trp57Gly
SNP	W57X (CF-causing)	c.170G>A	p.Trp57Ter
SNP	W57X (CF-causing)	c.171G>A	p.Trp57Ter
SNP	W846X (CF-causing)	c.2537G>A	p.Trp846Ter
SNP	W882X (CF-causing)	c.2645G>A	p.Trp882Ter
SNP	Y1032C (V.c.c.)	c.3095A>G	p.Tyr1032Cys
SNP	Y1092X (CF-causing)	c.3276C>A	p.Tyr1092Ter
SNP	Y1092X (CF-causing)	c.3276C>G	p.Tyr1092Ter
SNP	Y122X (CF-causing)	c.366T>A	p.Tyr122Ter
SNP	Y161D (CF-causing)	c.481T>G	p.Tyr161Asp
SNP	Y275X (CF-causing)	c.825C>G	p.Tyr275Ter
SNP	Y563D (CF-causing)	c.1687T>G	p.Tyr563Asp
SNP	Y563N (CF-causing)	c.1687T>A	p.Tyr563Asn
SNP	Y569D (CF-causing)	c.1705T>G	p.Tyr569Asp
SNP	Y849X (CF-causing)	c.2547C>A	p.Tyr849Ter
SNP	Y913X (CF-causing)	c.2739T>A	p.Tyr913Ter
CNV	CFTRdele1	c.4_53+69delins299	-
CNV	CFTRdele2,3	c.54-5940_273+10250del21kb	-
CNV	CFTRdele2ins182	c.54-	-

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Elenco mutazioni analizzate mediante SmartSeq (Amplicon Suite) versione 3.3.0 (*analisi "screening"*)

		5811_164+2186del8108ins182	
CNV	CFTRdele2	c.54-1161_164+1603del2875	-
CNV	CFTRdele14b-17b	c.2620-674_3367+198del	-
CNV	CFTRdele17a-18	c.2988+1173_3468+2111del	-
CNV	CFTRdele22-24	c.3964-3890_4443+3143del9454ins5	-
CNV	CFTRdele22,23	c.3964-78_4242+577del	-

Screening database version: CFTR2_Agosto 2018 (mutazioni causanti malattia: 336; varianti con conseguenze cliniche variabili:35)