



Elenco varianti patogenetiche analizzate mediante SmartSeq (Amplicon Suite) versione 3.5.1 (analisi "screening") aggiornato al 19 luglio 2022

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

1833delT	c.1703delT	p.Leu568Cysfs*4
185+1G->T	c.53+1G>T	-
1898+1G->A	c.1766+1G>A	-
1898+1G->C	c.1766+1G>C	-
1898+1G->T	c.1766+1G>T	-
1898+3A->G	c.1766+3A>G	-
1898+5G->T	c.1766+5G>T	-
1924del7	c.1792_1798delAAAACTA	p.Lys598Glyfs*11
2055del9->A	c.1923_1931delCTCAAAACTinsA	p.Ser641Argfs*5
2075delA	c.1943delA	p.Asp648ValfsX15
2105-2117del13insAGAAA	c.1973_1985delGAAATTCAATCCTinsAGAAA	p.Arg658Lysfs*4
2118del4	c.1986_1989delAACT	p.Thr663Argfs*8
2143delT	c.2012delT	p.Leu671Ter
2183AA->G or 2183delAA->G	c.2051_2052delAAinsG	p.Lys684Serfs*38
2184delA	c.2052delA	p.Lys684Asnfs*38
2184insA	c.2052dupA	p.Gln685Thrfs*4
2185insC	c.2053dupC	p.Gln685Profs*4
2307insA	c.2175dupA	p.Glu726Argfs*4
2347delG	c.2215delG	p.Val739Tyrfs*16
2372del8	c.2241_2248delGATACTGC	p.Ile748Serfs*28
2556insAT	c.2423_2424dupAT	p.Ser809Ilefs*13
2585delT	c.2453delT	p.Leu818Trpfs*3
2594delGT	c.2463_2464delTG	p.Ser821Argfs*4
2622+1G->A	c.2490+1G>A	-
2711delT	c.2583delT	p.Phe861Leufs*3
2721del11	c.2589_2599delAATTTGGTGCT	p.Ile864Serfs*28
2732insA	c.2601dupA	p.Val868Serfs*28
2789+5G->A	c.2657+5G>A	-
2790-1G->C	c.2658-1G>C	-
2869insG	c.2737_2738insG	p.Tyr913Ter
2896insAG	c.2763_2764dupAG	p.Val922Glufs*2
2942insT	c.2810dupT	p.Val938Glyfs*37
2957delT	c.2825delT	p.Ile942Thrfs*26
296+1G->A	c.164+1G>A	-
296+1G->T	c.164+1G>T	-
296+2T->C	c.164+2T>C	-
296+3insT	c.164+3_164+4insT	-
297-1G->A	c.165-1G>A	-
2991del32	c.2859_2890del32	p.Leu953Phefs*11
3007delG	c.2875delG	p.Ala959Hisfs*9
3028delA	c.2896delA	p.Thr966Argfs*2
306delTAGA	c.174_177delTAGA	p.Asp58Glufs*32
306insA	c.175dupA	p.Arg59Lysfs*10
3120+1G->A	c.2988+1G>A	-
3120G->A	c.2988G>A	p.=

3121-1G->A	c.2989-1G>A	-
3121-2A->G	c.2989-2A>G	-
3132delTG	c.3002_3003delTG	p.Val1001Aspfs*45
3143del9	c.3011_3019delCTATAGCAG	p.Ala1004_Ala1006del
3171delC	c.3039delC	p.Tyr1014Thrfs*9
3171insC	c.3039dupC	p.Tyr1014Leufs*33
3271delGG	c.3139_3139+1delGG	-
3272-26A->G	c.3140-26A>G	-
3349insT	c.3217dupT	p.Tyr1073Leufs*3
3500-2A->G	c.3368-2A>G	-
3600+2insT	c.3468+2dupT	-
3600+5G->A	c.3468+5G>A	-
3600G->A	c.3468G>A	p.=
365-366insT	c.233dupT	p.Trp79Leufs*32
3659delC	c.3528delC	p.Lys1177Serfs*15
3667ins4	c.3532_3535dupTCAA	p.Thr1179Ilefs*17
3737delA	c.3605delA	p.Asp1202Alafs*9
3791delC	c.3659delC	p.Thr1220Lysfs*8
3821delT	c.3691delT	p.Ser1231Profs*4
3849+10kbC->T	c.3718-2477C>T	-
3849+40A->G	c.3717+40A>G	-
3849+4A->G	c.3717+4A>G	-
3849+5G->A	c.3717+5G>A	-
3849G->A	c.3717G>A	p.=
3850-1G->A	c.3718-1G>A	-
3850-3T->G	c.3718-3T>G	-
3876delA	c.3744delA	p.Lys1250Argfs*9
3878delG	c.3747delG	p.Lys1250Argfs*9
3905insT	c.3773dupT	p.Leu1258Phefs*7
394delTT	c.262_263delTT	p.Leu88Ilefs*22
4005+1G->A	c.3873+1G>A	-
4005+2T->C	c.3873+2T>C	-
4010del4	c.3883_3886delATTT	p.Ile1295Phefs*32
4015delA	c.3883delA	p.Ile1295Phefs*33
4016insT	c.3889dupT	p.Ser1297Phefs*5
4022insT	c.3891dupT	p.Gly1298Trpfs*4
4040delA	c.3908delA	p.Asn1303Thrfs*25
405+1G->A	c.273+1G>A	-
405+3A->C	c.273+3A>C	-
406-1G->A	c.274-1G>A	-
406-2A->G	c.274-2A>G	-
4168delCTAAGCC	c.4036_4042delCTAAGCC	p.Leu1346MetfsX6
4209TGTT->AA	c.4077_4080delTGTTinsAA	p.Val1360Thrfs*3
4218insT	c.4086dupT	p.Lys1363Ter
4259del5	c.4127_4131delTGGAT	p.Leu1376Serfs*8
4279insA	c.4147dupA	p.Ile1383Asnfs*3

4326delTC	c.4197_4198delCT	p.Cys1400Ter
4374+1G->A	c.4242+1G>A	-
4374+1G->T	c.4242+1G>T	-
4382delA	c.4251delA	p.Glu1418Argfs*14
4428insGA	c.4300_4301dupAG	p.Ser1435Glyfs*14
442delA	c.310delA	p.Arg104Glufs*3
444delA	c.313delA	p.Ile105Serfs*2
457TAT->G	c.325_327delTATinsG	p.Tyr109Glyfs*4
541delC	c.409delC	p.Leu137Serfs*16
557delT	c.429del	p.Phe143LeufsTer10
574delA	c.442delA	p.Ile148Leufs*5
602del14	c.470_483del14	p.Phe157Ter
621+1G->T	c.489+1G>T	-
663delT	c.531delT	p.Ile177Metfs*12
675del4	c.543_546delTAGT	p.Leu183Phefs*5
711+1G->T	c.579+1G>T	-
711+3A->G	c.579+3A>G	-
711+5G->A	c.579+5G>A	-
712-1G->T	c.580-1G>T	-
849delG	c.717delG	p.Leu240Ter
852del22	c.723_743+1delGAGAATGATGATGAAGTACAGG	-
935delA	c.803delA	p.Asn268Ilefs*17
977insA	c.850dupA	p.Met284AsnfsX3
991del5	c.861_865delCTTAA	p.Asn287Lysfs*19
A1006E	c.3017C>A	p.Ala1006Glu
A455E	c.1364C>A	p.Ala455Glu
A46D	c.137C>A	p.Ala46Asp
A559T	c.1675G>A	p.Ala559Thr
A561E	c.1682C>A	p.Ala561Glu
A613T	c.1837G>A	p.Ala613Thr
C276X	c.828C>A	p.Cys276Ter
C524X	c.1572C>A	p.Cys524Ter
CFTRdele1	c.4_53+69delins299	-
CFTRdele14b-17b	c.2620-674_3367+198del	-
CFTRdele17a-18	c.2988+1173_3468+2111del	-
CFTRdele2	c.54-1161_164+1603del2875	-
CFTRdele2,3	c.54-5940_273+10250del21kb	-
CFTRdele22,23	c.3964-78_4242+577del	-
CFTRdele22-24	c.3964-3890_4443+3143del9454ins5	-
CFTRdele2ins182	c.54-5811_164+2186del8108ins182	-
D110H	c.328G>C	p.Asp110His
D192G	c.575A>G	p.Asp192Gly
D513G	c.1538A>G	p.Asp513Gly
D979V	c.2936A>T	p.Asp979Val
E1104X	c.3310G>T	p.Glu1104Ter
E1371X	c.4111G>T	p.Glu1371Ter

E193K	c.577G>A	p.Glu193Lys
E193X	c.577G>T	p.Glu193Ter
E379X	c.1135G>T	p.Glu379Ter
E474K	c.1420G>A	p.Glu474Lys
E56K	c.166G>A	p.Glu56Lys
E585X	c.1753G>T	p.Glu585Ter
E60K	c.178G>A	p.Glu60Lys
E60X	c.178G>T	p.Glu60Ter
E656X	c.1966G>T	p.Glu656Ter
E664X	c.1990G>T	p.Glu664Ter
E822X	c.2464G>T	p.Glu822Ter
E831X	c.2491G>T	p.Glu831Ter
E92K	c.274G>A	p.Glu92Lys
E92X	c.274G>T	p.Glu92Ter
F191V	c.571T>G	p.Phe191Val
F311L	c.933C>G	p.Phe311Leu
F508del	c.1521_1523delCTT	p.Phe508del
G1061R	c.3181G>C	p.Gly1061Arg
G1244E	c.3731G>A	p.Gly1244Glu
G1249R	c.3745G>A	p.Gly1249Arg
G126D	c.377G>A	p.Gly126Asp
G1349D	c.4046G>A	p.Gly1349Asp
G178R	c.532G>A	p.Gly178Arg
G194R	c.580G>A	p.Gly194Arg
G27R	c.79G>A	p.Gly27Arg
G27X	c.79G>T	p.Gly27Ter
G330X	c.988G>T	p.Gly330Ter
G542X	c.1624G>T	p.Gly542Ter
G550X	c.1648G>T	p.Gly550Ter
G551D	c.1652G>A	p.Gly551Asp
G551S	c.1651G>A	p.Gly551Ser
G628R	c.1882G>C	p.Gly628Arg
G628R	c.1882G>A	p.Gly628Arg
G673X	c.2017G>T	p.Gly673Ter
G745X	c.2233G>T	p.Gly745Ter
G85E	c.254G>A	p.Gly85Glu
G91R	c.271G>A	p.Gly91Arg
G970D	c.2909G>A	p.Gly970Asp
G970R	c.2908G>C	p.Gly970Arg
H1054D	c.3160C>G	p.His1054Asp
H1375P	c.4124A>C	p.His1375Pro
H139R	c.416A>G	p.His139Arg
H199Y	c.595C>T	p.His199Tyr
H609R	c.1826A>G	p.His609Arg
I1234V	c.3700A>G	p.Ile1234Val
I1269N	c.3806T>A	p.Ile1269Asn

I1366N	c.4097T>A	p.Ile1366Asn
I336K	c.1007T>A	p.Ile336Lys
I502T	c.1505T>C	p.Ile502Thr
I507del	c.1519_1521delATC	p.Ile507del
I601F	c.1801A>T	p.Ile601Phe
K710X	c.2128A>T	p.Lys710Ter
L102R	c.305T>G	p.Leu102Arg
L1065P	c.3194T>C	p.Leu1065Pro
L1077P	c.3230T>C	p.Leu1077Pro
L1254X	c.3761T>G	p.Leu1254Ter
L1324P	c.3971T>C	p.Leu1324Pro
L1335P	c.4004T>C	p.Leu1335Pro
L138ins	c.413_415dupTAC	p.Leu138dup
L15P	c.44T>C	p.Leu15Pro
L165S	c.494T>C	p.Leu165Ser
L206W	c.617T>G	p.Leu206Trp
L227R	c.680T>G	p.Leu227Arg
L346P	c.1037T>C	p.Leu346Pro
L453S	c.1358T>C	p.Leu453Ser
L467P	c.1400T>C	p.Leu467Pro
L558S	c.1673T>C	p.Leu558Ser
L732X	c.2195T>G	p.Leu732Ter
L88X	c.263T>A	p.Leu88Ter
L88X	c.263T>G	p.Leu88Ter
L927P	c.2780T>C	p.Leu927Pro
M1101K	c.3302T>A	p.Met1101Lys
M1101R	c.3302T>G	p.Met1101Arg
M1V	c.1A>G	p.Met1Val
N1303K	c.3909C>G	p.Asn1303Lys
P205S	c.613C>T	p.Pro205Ser
P574H	c.1721C>A	p.Pro574His
P67L	c.200C>T	p.Pro67Leu
P99L	c.296C>T	p.Pro99Leu
Q1042X	c.3124C>T	p.Gln1042Ter
Q1313X	c.3937C>T	p.Gln1313Ter
Q1330X	c.3988C>T	p.Gln1330Ter
Q1382X	c.4144C>T	p.Gln1382Ter
Q1411X	c.4231C>T	p.Gln1411Ter
Q1412X	c.4234C>T	p.Gln1412Ter
Q220X	c.658C>T	p.Gln220Ter
Q2X	c.4C>T	p.Gln2Ter
Q30X	c.88C>T	p.Gln30X
Q359K/T360K	c.[1075C>A;1079C>A]	p.[Gln359Lys;Thr360Lys]
Q39X	c.115C>T	p.Gln39Ter
Q414X	c.1240C>T	p.Gln414Ter
Q493X	c.1477C>T	p.Gln493Ter

Q525X	c.1573C>T	p.Gln525Ter
Q552X	c.1654C>T	p.Gln552Ter
Q685X	c.2053C>T	p.Gln685Ter
Q715X	c.2143C>T	p.Gln715Ter
Q720X	c.2158C>T	p.Gln720X
Q890X	c.2668C>T	p.Gln890Ter
Q98R	c.293A>G	p.Gln98Arg
Q98X	c.292C>T	p.Gln98Ter
R1066C	c.3196C>T	p.Arg1066Cys
R1066H	c.3197G>A	p.Arg1066His
R1102X	c.3304A>T	p.Arg1102Ter
R1158X	c.3472C>T	p.Arg1158Ter
R1162X	c.3484C>T	p.Arg1162Ter
R117C	c.349C>T	p.Arg117Cys
R117P	c.350G>C	p.Arg117Pro
R1283M	c.3848G>T	p.Arg1283Met
R334L	c.1001G>T	p.Arg334Leu
R334W	c.1000C>T	p.Arg334Trp
R347H	c.1040G>A	p.Arg347His
R347P	c.1040G>C	p.Arg347Pro
R352Q	c.1055G>A	p.Arg352Gln
R553X	c.1657C>T	p.Arg553Ter
R560K	c.1679G>A	p.Arg560Lys
R560S	c.1680A>C	p.Arg560Ser
R560T	c.1679G>C	p.Arg560Thr
R709X	c.2125C>T	p.Arg709Ter
R75X	c.223C>T	p.Arg75Ter
R764X	c.2290C>T	p.Arg764Ter
R785X	c.2353C>T	p.Arg785Ter
R792X	c.2374C>T	p.Arg792Ter
R851X	c.2551C>T	p.Arg851Ter
S1118F	c.3353C>T	p.Ser1118Phe
S1159F	c.3476C>T	p.Ser1159Phe
S1159P	c.3475T>C	p.Ser1159Pro
S1196X	c.3587C>G	p.Ser1196Ter
S1251N	c.3752G>A	p.Ser1251Asn
S1255P	c.3763T>C	p.Ser1255Pro
S1255X	c.3764C>A	p.Ser1255Ter
S13F	c.38C>T	p.Ser13Phe
S341P	c.1021T>C	p.Ser341Pro
S434X	c.1301C>A	p.Ser434Ter
S434X	c.1301C>G	p.Ser434Ter
S466X	c.1397C>A	p.Ser466Ter
S466X	c.1397C>G	p.Ser466Ter
S489X	c.1466C>A	p.Ser489Ter
S492F	c.1475C>T	p.Ser492Phe

S4X	c.11C>A	p.Ser4Ter
S549N	c.1646G>A	p.Ser549Asn
S549R	c.1645A>C	p.Ser549Arg
S549R	c.1647T>G	p.Ser549Arg
S549R	c.1647T>A	p.Ser549Arg
S912X	c.2735C>A	p.Ser912Ter
S945L	c.2834C>T	p.Ser945Leu
T1036N	c.3107C>A	p.Thr1036Asn
T338I	c.1013C>T	p.Thr338Ile
V1240G	c.3719T>G	p.Val1240Gly
V232D	c.695T>A	p.Val232Asp
V456A	c.1367T>C	p.Val456Ala
V520F	c.1558G>T	p.Val520Phe
W1089X	c.3266G>A	p.Trp1089Ter
W1098C	c.3294G>C	p.Trp1098Cys
W1098C	c.3294G>T	p.Trp1098Cys
W1098R	c.3292T>C	p.Trp1098Arg
W1098X	c.3293G>A	p.Trp1098Ter
W1098X	c.3294G>A	p.Trp1098Ter
W1145X	c.3435G>A	p.Trp1145Ter
W1204X	c.3611G>A	p.Trp1204Ter
W1204X	c.3612G>A	p.Trp1204Ter
W1282X	c.3846G>A	p.Trp1282X
W19X	c.57G>A	p.Trp19Ter
W216X	c.647G>A	p.Trp216Ter
W401X	c.1202G>A	p.Trp401Ter
W401X	c.1203G>A	p.Trp401Ter
W496X	c.1487G>A	p.Trp496Ter
W57G	c.169T>G	p.Trp57Gly
W57X	c.170G>A	p.Trp57Ter
W57X	c.171G>A	p.Trp57Ter
W846X	c.2537G>A	p.Trp846Ter
W846X	c.2538G>A	p.Trp846Ter
W882X	c.2645G>A	p.Trp882Ter
Y1092X	c.3276C>A	p.Tyr1092Ter
Y1092X	c.3276C>G	p.Tyr1092Ter
Y122X	c.366T>A	p.Tyr122Ter
Y161D	c.481T>G	p.Tyr161Asp
Y275X	c.825C>G	p.Tyr275Ter
Y563D	c.1687T>G	p.Tyr563Asp
Y563N	c.1687T>A	p.Tyr563Asn
Y569D	c.1705T>G	p.Tyr569Asp
Y849X	c.2547C>A	p.Tyr849Ter
Y913X	c.2739T>A	p.Tyr913Ter

Le 354 varianti analizzate sono quelle inserite nel database di riferimento per la Fibrosi Cistica come “causanti malattia” (CFTR2: <http://www.cftr2.org/index.php>) alla data del 31/07/2020. Sono escluse le varianti con conseguenze cliniche variabili e le varianti benigne