

Cystic fibrosis (CFTR) – first line screening

Panel information

Gene panel	Cystic fibrosis (CFTR) - first line screening
Version	2
Total variants	159
Activation date	31/12/2025
Publisher	Center for Medical Genetics, Ghent

Variants

c.HGVS	p.HGVS	Legacy
c.948del	p.Phe316LeufsTer12	1078delT
c.1021_1022dup	p.Phe342HisfsTer28	1154insTC
c.1029del	p.Cys343Ter	1161delC
c.1130dup	p.Gln378AlafsTer4	1259insA
c.1155_1156dup	p.Asn386IlefsTer3	1288insTA
c.1327_1330dup	p.Ile444ArgfsTer3	1461ins4
c.1373del	p.Gly458AspfsTer11	1504delG
c.1393-1G>A	-	1525-1G->A
c.1418del	p.Gly473GlufsTer54	1548delG
c.1477_1478del	p.Gln493ValfsTer10	1609delCA
c.1545_1546del	p.Tyr515Ter	1677delTA
c.1584+1G>A	-	1716+1G->A
c.1585-1G>A	-	1717-1G->A
c.1680-886A>G	-	1811+1634A->G 1811+1.6kbA->G
c.1679+1G>A	-	1811+1G->A
c.1680-1G>A	-	1812-1G->A
c.1692del	p.Asp565MetfsTer7	1824delA
c.1766+1G>A	-	1898+1G->A
c.1766+3A>G	-	1898+3A->G
c.1766+5G>T	-	1898+5G->T
c.2012del	p.Leu671Ter	2143delT
c.2051_2052delinsG	p.Lys684SerfsTer38	2183AA->G
c.2052del	p.Lys684AsnfsTer38	2184delA
c.2052dup	p.Gln685ThrfsTer4	2184insA
c.2175dup	p.Glu726ArgfsTer4	2307insA
c.2215del	p.Val739TyrfsTer16	2347delG
c.2490+1G>A	-	2622+1G->A
c.2583delT	p.Phe861Leufs*3	2711delT
c.2657+5G>A	-	2789+5G->A
c.2737_2738insG	p.Tyr913Ter	2869insG
c.175dup	p.Arg59LysfsTer10	306insA
c.2988+1G>A	-	3120+1G->A
c.2988G>A	p.Gln996=	3120G->A

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c.3067_3072del	p.Ile1023_Val1024del	3199del6
c.3140-26A>G	-	3272-26A->G
c.3468G>A	p.Leu1156=	3600G->A
c.3528del	p.Lys1177SerfsTer15	3659delC
c.3659del	p.Thr1220LysfsTer8	3791delC
c.3718-2477C>T	-	3849+10kbC->T
c.3717+5G>A	-	3849+5G->A
c.3744del	p.Lys1250ArgfsTer9	3876delA
c.3773dup	p.Leu1258PhefsTer7	3905insT
c.3808del	p.Asp1270MetfsTer8	3940delG
c.262_263del	p.Leu88IlefsTer22	394delTT
c.3889dup	p.Ser1297PhefsTer5	4016insT
c.274-1G>A	-	406-1G->A
c.4251del	p.Glu1418ArgfsTer14	4382delA
c.313del	p.Ile105SerfsTer2	444delA
c.489+1G>T	-	621+1G->T
c.489+2T>C	-	621+2T->C
c.579+1G>T	-	711+1G->T
c.579+3A>G	-	711+3A->G
c.579+5G>A	-	711+5G->A
c.580-1G>T	-	712-1G->T
c.723_743+1del	-	852del22
c.803del	p.Asn268IlefsTer17	935delA
c.3017C>A	p.Ala1006Glu	A1006E
c.1364C>A	p.Ala455Glu	A455E
c.137C>A	p.Ala46Asp	A46D
c.1675G>A	p.Ala559Thr	A559T
c.1682C>A	p.Ala561Glu	A561E
c.1837G>A	p.Ala613Thr	A613T
c.870-1113_870-1110del	-	c.870-1113_870-1110del
c.1572C>A	p.Cys524Ter	C524X
c.328G>C	p.Asp110His	D110H
c.3454G>C	p.Asp1152His	D1152H
c.2936A>T	p.Asp979Val	D979V
c.1753G>T	p.Glu585Ter	E585X
c.178G>T	p.Glu60Ter	E60X
c.274G>A	p.Glu92Lys	E92K
c.274G>T	p.Glu92Ter	E92X
c.571T>G	p.Phe191Val	F191V
c.1521_1523del	p.Phe508del	F508del
c.3731G>A	p.Gly1244Glu	G1244E
c.4046G>A	p.Gly1349Asp	G1349D
c.532G>A	p.Gly178Arg	G178R
c.988G>T	p.Gly330Ter	G330X

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c.1624G>T	p.Gly542Ter	G542X
c.1652G>A	p.Gly551Asp	G551D
c.1651G>A	p.Gly551Ser	G551S
c.1882G>A	p.Gly628Arg	G628R
c.254G>A	p.Gly85Glu	G85E
c.271G>A	p.Gly91Arg	G91R
c.2909G>A	p.Gly970Asp	G970D
c.2908G>C	p.Gly970Arg	G970R
c.595C>T	p.His199Tyr	H199Y
c.1007T>A	p.Ile336Lys	I336K
c.1519_1521del	p.Ile507del	I507del
c.2128A>T	p.Lys710Ter	K710X
c.3194T>C	p.Leu1065Pro	L1065P
c.3230T>C	p.Leu1077Pro	L1077P
c.617T>G	p.Leu206Trp	L206W
c.653T>A	p.Leu218Ter	L218X
c.1400T>C	p.Leu467Pro	L467P
c.1673T>C	p.Leu558Ser	L558S
c.2780T>C	p.Leu927Pro	L927P
c.3302T>A	p.Met1101Lys	M1101K
c.1A>G	p.Met1?	M1V
c.3909C>G	p.Asn1303Lys	N1303K
c.613C>T	p.Pro205Ser	P205S
c.200C>T	p.Pro67Leu	P67L
c.868C>T	p.Gln290Ter	Q290X
c.4C>T	p.Gln2Ter	Q2X
c.115C>T	p.Gln39Ter	Q39X
c.1477C>T	p.Gln493Ter	Q493X
c.1654C>T	p.Gln552Ter	Q552X
c.2668C>T	p.Gln890Ter	Q890X
c.293A>G	p.Gln98Arg	Q98R
c.292C>T	p.Gln98Ter	Q98X
c.3196C>T	p.Arg1066Cys	R1066C
c.3197G>A	p.Arg1066His	R1066H
c.3472C>T	p.Arg1158Ter	R1158X
c.3484C>T	p.Arg1162Ter	R1162X
c.349C>T	p.Arg117Cys	R117C
c.350G>A	p.Arg117His	R117H
c.3848G>T	p.Arg1283Met	R1283M
c.1000C>T	p.Arg334Trp	R334W
c.1040G>A	p.Arg347His	R347H
c.1040G>C	p.Arg347Pro	R347P
c.1055G>A	p.Arg352Gln	R352Q
c.1657C>T	p.Arg553Ter	R553X

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c.1680A>C	p.Arg560Ser	R560S
c.1679G>C	p.Arg560Thr	R560T
c.2125C>T	p.Arg709Ter	R709X
c.223C>T	p.Arg75Ter	R75X
c.2290C>T	p.Arg764Ter	R764X
c.2353C>T	p.Arg785Ter	R785X
c.2374C>T	p.Arg792Ter	R792X
c.3353C>T	p.Ser1118Phe	S1118F
c.3752G>A	p.Ser1251Asn	S1251N
c.3763T>C	p.Ser1255Pro	S1255P
c.3764C>A	p.Ser1255Ter	S1255X
c.1397C>G	p.Ser466Ter	S466X
c.1646G>A	p.Ser549Asn	S549N
c.1647T>G	p.Ser549Arg	S549R
c.2834C>T	p.Ser945Leu	S945L
c.3107C>A	p.Thr1036Asn	T1036N
c.1013C>T	p.Thr338Ile	T338I
c.695T>A	p.Val232Asp	V232D
c.1367T>C	p.Val456Ala	V456A
c.1558G>T	p.Val520Phe	V520F
c.3266G>A	p.Trp1089Ter	W1089X
c.3294G>C	p.Trp1098Cys	W1098C
c.3612G>A	p.Trp1204Ter	W1204X
c.3846G>A	p.Trp1282Ter	W1282X
c.2538G>A	p.Trp846Ter	W846X
c.3276C>G	p.Tyr1092Ter	Y1092X
c.3276C>A	p.Tyr1092Ter	Y1092X
c.366T>A	p.Tyr122Ter	Y122X
c.1705T>G	p.Tyr569Asp	Y569D
c.2739T>A	p.Tyr913Ter	Y913X
c.4_53+69delins299	-	CFTRdele1
c.2620-674_3367+198del	-	CFTRdele14b-17b
c.2988+1173_3468+2111del	-	CFTRdele17a-18
c.54-1161_164+1603del2875	-	CFTRdele2
c.54-5940_273+10250del21kb	-	CFTRdele2,3
c.3964-78_4242+577del	-	CFTRdele22,23
c.3964-3890_4443+3143del9454ins5	-	CFTRdele22-24
c.54-5811_164+2186del8108ins182	-	CFTRdele2ins182