

Cystic fibrosis (CFTR) – first line screening

Panel information

Gene panel	Cystic fibrosis (CFTR) - first line screening
Version	1
Total variants	50
Activation data	15/04/2016
Publisher	Center for Medical Genetics, Ghent

Variants

c.HGVS	p.HGVS	Legacy
c.1521_1523del	p.Phe508del	F508del
c.350G>A	p.Arg117His	R117H
c.1624G>T	p.Gly542Ter	G542X
c.3909C>G	p.Asn1303Lys	N1303K
c.3752G>A	p.Ser1251Asn	S1251N
c.1657C>T	p.Arg553Ter	R553X
c.3140-26A>G	p.?	3272-26A->G
c.3454G>C	p.Asp1152His	D1152H
c.1364C>A	p.Ala455Glu	A455E
c.1585-1G>A	p.?	1717-1G->A
c.3718-2477C>T	p.?	3849+10kbC->T
c.2657+5G>A	p.?	2789+5G->A
c.349C>T	p.Arg117Cys	R117C
c.2052del	p.Lys684AsnfsTer38	2184delA
c.1519_1521del	p.Ile507del	I507del
c.2051_2052delinsG	p.Lys684SerfsTer38	2183AA->G
c.3276C>A	p.Tyr1092Ter	Y1092X
c.2780T>C	p.Leu927Pro	L927P
c.3846G>A	p.Trp1282Ter	W1282X
c.579+1G>T	p.?	711+1G->T
c.3484C>T	p.Arg1162Ter	R1162X
c.2988+1G>A	p.?	3120+1G->A
c.178G>T	p.Glu60Ter	E60X
c.948del	p.Phe316LeufsTer12	1078delT
c.1040G>C	p.Arg347Pro	R347P
c.1000C>T	p.Arg334Trp	R334W
c.3528del	p.Lys1177SerfsTer15	3659delC
c.1652G>A	p.Gly551Asp	G551D
c.489+1G>T	p.?	621+1G->T
c.262_263del	p.Leu88IlefsTer22	394delTT
c.254G>A	p.Gly85Glu	G85E
c.1040G>A	p.Arg347His	R347H

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c.3196C>T	p.Arg1066Cys	R1066C
c.1646G>A	p.Ser549Asn	S549N
c.3773dup	p.Leu1258PhefsTer7	3905insT
c.1766+1G>A	p.?	1898+1G->A
c.2012del	p.Leu671Ter	2143delT
c.1647T>G	p.Ser549Arg	S549R
c.532G>A	p.Gly178Arg	G178R
c.579+5G>A	p.?	711+5G->A
c.1679G>C	p.Arg560Thr	R560T
c.175dup	p.Arg59LysfsTer10	306insA
c.1654C>T	p.Gln552Ter	Q552X
c.2908G>C	p.Gly970Arg	G970R
c.3276C>G	p.Tyr1092Ter	Y1092X
c.1477C>T	p.Gln493Ter	Q493X
c.1651G>A	p.Gly551Ser	G551S
c.3731G>A	p.Gly1244Glu	G1244E
c.3763T>C	p.Ser1255Pro	S1255P
c.4046G>A	p.Gly1349Asp	G1349D