

**Genetic variants in SeqDB\_v13**  
**Release date - 02/11/2015**

#	gene_name	variant_name	dbSNP_id
1	CFTR	1078delT	rs121908744
2	CFTR	1119delA	rs397508824
3	CFTR	1138insG	rs397508138
4	CFTR	1154insTC	rs387906360
5	CFTR	1161delC	rs121908774
6	CFTR	1213delT	rs387906361
7	CFTR	1215delG	rs387906375
8	CFTR	1221delCT	rs387906365
9	CFTR	1248+1G>A	rs397508158
10	CFTR	1248+1G>C	rs397508158
11	CFTR	1249-1G>A	-
12	CFTR	124del23bp	rs397508136
13	CFTR	1259insA	rs397508163
14	CFTR	1288insTA	rs121908785
15	CFTR	1341+1G>A	rs397508176
16	CFTR	1341G>A	rs397508177
17	CFTR	1342-1G>C	rs397508178
18	CFTR	1367del5	rs397508184
19	CFTR	1394delC	-
20	CFTR	1525-1G>A	rs397508200
21	CFTR	1525-2A>G	rs397508201
22	CFTR	1548delG	rs397508205
23	CFTR	1609delCA	rs121908775
24	CFTR	1660delG	-
25	CFTR	1677delTA	rs121908776
26	CFTR	1706del17	-
27	CFTR	1716+1G>A	rs397508230
28	CFTR	1716+1G>T	rs397508230
29	CFTR	1717-1G>A	rs76713772
30	CFTR	1717-8G>A	rs193922503
31	CFTR	175delC	rs397508715
32	CFTR	1782delA	rs397508251
33	CFTR	1784delG	rs397508252
34	CFTR	1811+1.6kbA>G	rs397508266
35	CFTR	1811+1G>C	rs397508263
36	CFTR	1812-1G>A	rs121908794
37	CFTR	1812-1G>C	rs121908794
38	CFTR	1824delA	rs193922505
39	CFTR	182delT	rs774973931, rs397508742
40	CFTR	185+1G>T	rs397508746
41	CFTR	1854delT	-
42	CFTR	1898+1G>A	rs121908748
43	CFTR	1898+1G>C	rs121908748
44	CFTR	1898+1G>T	rs121908748
45	CFTR	1898+3A>C	rs397508298
46	CFTR	1898+3A>G	rs397508298

47	CFTR	1898+5G>T	rs121908796
48	CFTR	1924del7	rs397508303
49	CFTR	1949del84	rs121908777
50	CFTR	2043delG	rs121908778
51	CFTR	2055del9insA	rs121908779
52	CFTR	2105-2117del13insAGAAA	rs121908780
53	CFTR	2118del4	rs397508325
54	CFTR	2143delT	rs121908812
55	CFTR	2307insA	rs121908787
56	CFTR	2347delG	rs397508353
57	CFTR	241delAT	rs397508162, rs397508164
58	CFTR	2423delG	rs387906376
59	CFTR	2522insC	rs397508372
60	CFTR	2556insAT	rs387906359
61	CFTR	2585delT	rs397515498
62	CFTR	2594delGT	-
63	CFTR	2603delT	rs397508380
64	CFTR	2622+1G>A	rs141158996
65	CFTR	2622+1G>T	rs141158996
66	CFTR	2697insT	-
67	CFTR	2711delT	rs397508399
68	CFTR	2721del11	rs397508400
69	CFTR	2732insA	rs397508405
70	CFTR	274delAATC	-
71	CFTR	2751+2T>A	rs397508407
72	CFTR	2789+2insA	rs397508414
73	CFTR	2789+5G>A	rs80224560
74	CFTR	2790-1G>C	rs397508416
75	CFTR	2790-1G>T	rs397508416
76	CFTR	2790-2A>G	-
77	CFTR	2869insG	rs121908788
78	CFTR	2896insAG	rs397508431
79	CFTR	2907delTT	rs397508433
80	CFTR	2909delT	rs397508434
81	CFTR	2942insT	rs193922510
82	CFTR	2957delT	rs397508441
83	CFTR	296+1G>A	rs397508243
84	CFTR	296+1G>C	rs397508243
85	CFTR	297-1G>A	rs397508249
86	CFTR	2991del32	rs397508445
87	CFTR	3007delG	rs397508447
88	CFTR	306insA	rs397508294
89	CFTR	3089insTC	-
90	CFTR	3120+1G>A	rs75096551
91	CFTR	3120G>A	rs121908797
92	CFTR	3121-1G>A	rs397508470
93	CFTR	3121-2A>G	rs193922515
94	CFTR	3121-2A>T	rs193922515
95	CFTR	3130delA	rs397508475
96	CFTR	3132delTG	rs397508477

97	CFTR	3171delC	rs121908781
98	CFTR	3196del54	-
99	CFTR	3199del6	rs121908767
100	CFTR	3272-11A>G	-
101	CFTR	3272-16T>A	-
102	CFTR	3272-1G>A	rs397508506
103	CFTR	3272-26A>G	rs76151804
104	CFTR	3293delA	rs387906377
105	CFTR	3320ins5	rs387906366
106	CFTR	3359delCT	
107	CFTR	3395dupA	rs397508524
108	CFTR	3500-2A>G	rs755416052
109	CFTR	3601-20T>C	rs373002889
110	CFTR	3622insT	rs397508576
111	CFTR	3659delC	rs121908747
112	CFTR	3662delA	rs397508579
113	CFTR	3667del4	rs121908782
114	CFTR	3667ins4	rs387906378
115	CFTR	3730delAAinsG	-
116	CFTR	3732delA	rs397508586
117	CFTR	3737delA	rs397508587
118	CFTR	3750delAG	rs397508589
119	CFTR	3791delC	rs121908811
120	CFTR	3821delT	rs121908783
121	CFTR	3849+10kbC>T	rs75039782
122	CFTR	3849+1G>A	-
123	CFTR	3849+4A>G	rs387906362
124	CFTR	3849G>A	rs144781064
125	CFTR	3850-1G>A	rs387906369
126	CFTR	3876delA	rs121908784
127	CFTR	3878delG	-
128	CFTR	3898insC	rs397508605, rs387906370
129	CFTR	3905insT	rs121908789
130	CFTR	3944delGT	rs397508612
131	CFTR	394delTT	rs121908769
132	CFTR	396del5	-
133	CFTR	4005+1G>A	rs143570767
134	CFTR	4005+2T>C	rs146795445
135	CFTR	4006-19del3	rs397508623
136	CFTR	4010del4	rs397508628
137	CFTR	4040delA	rs780529834, rs397508637
138	CFTR	405+1G>A	rs121908791
139	CFTR	405+3A>C	rs74467662
140	CFTR	405+4A>G	rs387906374
141	CFTR	406-1G>A	rs121908792
142	CFTR	406-6T>C	rs371315549
143	CFTR	4168delCTAAGCC	rs397508662
144	CFTR	4209TGTT>AA	rs397508668
145	CFTR	4218insT	rs397508669
146	CFTR	4279insA	rs397508685

147	CFTR	4326delTC	rs397508693
148	CFTR	4374+1G>A	rs372227120
149	CFTR	4374+1G>T	rs372227120
150	CFTR	4382delA	rs397508706
151	CFTR	440del8	-
152	CFTR	4428insGA	rs397508709
153	CFTR	444delA	rs121908801
154	CFTR	451del8	rs397508516
155	CFTR	457TAT>G	rs121908798
156	CFTR	541delC	rs397508672
157	CFTR	556delA	rs387906363
158	CFTR	557delT	rs387906364
159	CFTR	574delA	rs121908770
160	CFTR	591del18	rs387906371
161	CFTR	604insA	-
162	CFTR	621+1G>T	rs78756941
163	CFTR	621+3A>G	rs377729736
164	CFTR	663delT	rs121908771
165	CFTR	663dupT	-
166	CFTR	675del4	rs397508750
167	CFTR	711+1G>T	rs77188391
168	CFTR	711+3A>G	rs397508761
169	CFTR	711+5G>A	rs78440224
170	CFTR	712-1G>T	rs121908793
171	CFTR	852del22	rs121908804
172	CFTR	875+1G>A	rs397508791
173	CFTR	875+1G>C	rs397508791
174	CFTR	876-14del12	rs387906367
175	CFTR	935delA	rs121908772
176	CFTR	936delTA	rs121908773
177	CFTR	989-992insA	rs397508803
178	CFTR	991del5	rs397508806, rs397508805
179	CFTR	A1006E	rs397508480
180	CFTR	A1067T	rs121909020
181	CFTR	A204T	rs748026786
182	CFTR	A349V	rs121909021
183	CFTR	A46D	rs151020603
184	CFTR	A554E	rs121909022
185	CFTR	A559T	rs75549581
186	CFTR	A561E	rs121909047
187	CFTR	A96E	rs397508449
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191	CFTR	C866Y	rs193922506
192	CFTR	CFTRdele2,3(21kb)	-
193	CFTR	D110H	rs113993958
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195	CFTR	D1270N	rs11971167
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197	CFTR	D579A	rs397508288
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201	CFTR	D648V	rs121909033
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203	CFTR	E116K	rs397508571
204	CFTR	E1371X	rs397508675
205	CFTR	E193X	rs397508758
206	CFTR	E217G	rs121909046
207	CFTR	E278del	rs397508801
208	CFTR	E384X	-
209	CFTR	E504Q	rs397508223
210	CFTR	E528E	rs1800095
211	CFTR	E56K	rs397508256
212	CFTR	E585X	rs397508296
213	CFTR	E60K	rs77284892
214	CFTR	E60X	rs77284892
215	CFTR	E656X	rs397508323
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218	CFTR	E827X	rs121909018
219	CFTR	E831X	rs397508387
220	CFTR	E92K	rs121908751
221	CFTR	E92X	rs121908751
222	CFTR	F1052V	rs150212784
223	CFTR	F1074L	rs186045772
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226	CFTR	F311L	rs121909016
227	CFTR	F508del	rs199826652, rs113993960
228	CFTR	F693L(TTT>CTT)	rs397508338
229	CFTR	F693L(TTT>TTG)	rs145540754
230	CFTR	G1061R	rs142394380
231	CFTR	G1069R	rs200321110
232	CFTR	G1244E	rs267606723
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234	CFTR	G1249E	rs121909040
235	CFTR	G1349D	rs193922525
236	CFTR	G178E	rs397508748
237	CFTR	G178R	rs80282562
238	CFTR	G178X	rs80282562
239	CFTR	G27X	rs397508796
240	CFTR	G314E	rs75763344
241	CFTR	G330X	rs79031340
242	CFTR	G44D	rs1800074
243	CFTR	G480C	rs79282516
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245	CFTR	G542X	rs113993959
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248	CFTR	G551S	rs121909013
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250	CFTR	G628R(G>A)	rs397508316
251	CFTR	G628R(G>C)	rs397508316
252	CFTR	G673X	rs397508331
253	CFTR	G85E	rs75961395
254	CFTR	G91R	rs121908750
255	CFTR	G970R	rs397508453
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258	CFTR	H139L	rs76371115
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261	CFTR	H199Y	rs121908802
262	CFTR	H609R	rs397508310
263	CFTR	H949Y	rs121909035
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267	CFTR	I148N	rs35516286
268	CFTR	I148T	rs35516286
269	CFTR	I336K	rs397508139
270	CFTR	I502N	rs397508222
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273	CFTR	I507del	rs121908745
274	CFTR	I530T	-
275	CFTR	I556V	rs75789129
276	CFTR	IVS13+2T>C	
277	CFTR	IVS14a-6T>C <sup>2</sup>	rs371315682
278	CFTR	IVS19-24G>A <sup>2</sup>	rs374013084
279	CFTR	K329X	-
280	CFTR	K710X	rs75115087
281	CFTR	K716X	rs121909023
282	CFTR	L1065P	rs121909036
283	CFTR	L1077P	rs139304906
284	CFTR	L1093P	rs397508527
285	CFTR	L1254X	rs397508604
286	CFTR	L1335P	rs397508658
287	CFTR	L138ins	rs397508686
288	CFTR	L206W	rs121908752
289	CFTR	L346P	rs397508146
290	CFTR	L467F	-
291	CFTR	L467P	rs139573311
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295	CFTR	L719X	rs397508345
296	CFTR	L732X	rs397508350

297	CFTR	L863R	-
298	CFTR	L88S	rs397508412
299	CFTR	L88X (T>A)	rs397508412
300	CFTR	L88X (T>G)	rs397508412
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305	CFTR	M1137V	rs397508553
306	CFTR	M1140K	rs397508558
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308	CFTR	M348K	rs142920240
309	CFTR	M952I	rs151048781
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313	CFTR	P205S	rs121908803
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317	CFTR	P988R	-
318	CFTR	Q1038X	-
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322	CFTR	Q1291H	rs121909015
323	CFTR	Q1313K	rs121909026
324	CFTR	Q1313X	rs121909026
325	CFTR	Q1352H(G>C)	rs113857788
326	CFTR	Q1352H(G>T)	rs113857788
327	CFTR	Q1382X	rs397508684
328	CFTR	Q1411X	rs397508701
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331	CFTR	Q290X	rs397508808
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336	CFTR	Q525X	rs397508227
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370	CFTR	R709X	rs121908760
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381	CFTR	S1251N	rs74503330
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384	CFTR	S1255X	rs76649725
385	CFTR	S341P	rs397508144
386	CFTR	S42F	rs143456784
387	CFTR	S466L	rs121908805
388	CFTR	S466X (C>A)	rs121908805
389	CFTR	S466X (C>G)	rs121908805
390	CFTR	S489X	rs397508211
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395	CFTR	S549R(A>C)	rs121908757
396	CFTR	S549R(T>G)	rs121909005



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398	CFTR	S912X	rs121909034
399	CFTR	S945L	rs397508442
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409	CFTR	V520I	rs77646904
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411	CFTR	W1089X	rs78802634
412	CFTR	W1145X	rs397508561
413	CFTR	W1204X (TGG>TAG)	rs121908764
414	CFTR	W1204X (TGG>TGA)	rs121908765
415	CFTR	W1282R	rs397508616
416	CFTR	W1282X	rs77010898
417	CFTR	W1316X	rs121909010
418	CFTR	W19G	-
419	CFTR	W361R(T>A)	rs397508154
420	CFTR	W361R(T>C)	rs397508154
421	CFTR	W361X	-
422	CFTR	W401X (TGG>TAG)	rs397508174
423	CFTR	W401X (TGG>TGA)	rs397508175
424	CFTR	W496X	rs397508216
425	CFTR	W57G	rs397508272
426	CFTR	W57R	rs397508272
427	CFTR	W57X	rs121909025
428	CFTR	W846X	rs397508393
429	CFTR	W882X(TGG>TAG)	rs397508413
430	CFTR	W882X(TGG>TGA)	
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432	CFTR	Y1092X (C>A)	rs121908761
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434	CFTR	Y109C	rs121909031
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442	CFTR	Y913X	rs149790377
443	GALT	5'UTR-119delGTCA	rs111033640, rs142496102
444	GALT	A345D	rs111033815
445	GALT	A46Pfs	rs111033651
446	GALT	E203K	rs111033736

447	GALT	F171S	rs111033715
448	GALT	F194L	rs111033726
449	GALT	H184D	rs111033716
450	GALT	H184Q	rs111033717
451	GALT	H319Q	rs111033792
452	GALT	I378V	rs111033819
453	GALT	IVS3-2A>C	rs111033667
454	GALT	IVS4-27G>C	rs41274865
455	GALT	IVS5+62G>A	rs2277202
456	GALT	IVS5-24G>A	rs41274867
457	GALT	K285N	rs111033773
458	GALT	L116P	-
459	GALT	L195P	rs111033728
460	GALT	L218L	rs2070075
461	GALT	L218V	rs2070075
462	GALT	L358P	rs1133894
463	GALT	L62M	rs1800461
464	GALT	L74P	rs111033663
465	GALT	M142K	rs111033695
466	GALT	M142T	rs111033695
467	GALT	M178R	-
468	GALT	N314D	rs2070074
469	GALT	P183T	rs111033721
470	GALT	P66H	rs111033656
471	GALT	P66L	rs111033656
472	GALT	Q188R	rs75391579
473	GALT	R123G	rs111033674
474	GALT	R333G	rs111033800
475	GALT	R333W	rs111033800
476	GALT	S135L	rs111033690
477	GALT	S135W	rs111033690
478	GALT	V168L	rs367543258
479	GALT	V44L	rs111033647
480	GALT	V44M	rs111033647
481	GALT	Y209C	rs111033744
482	GALT	Y209S	rs111033744
483	PAH	1298dupA	-
484	PAH	15-BP DEL, EX11	rs62516097
485	PAH	1-BP DEL, 1129T	rs62642941
486	PAH	1-BP DEL, CODON 55	rs199475566
487	PAH	398_401del4bp	rs199475605
488	PAH	3-BP DEL, ATC	rs62514904
489	PAH	3-BP DEL, CTT	rs62516096
490	PAH	47delCT	rs62642906
491	PAH	592_613del22bp	rs199475697
492	PAH	929_939del11	rs62651568
493	PAH	A104D	rs62642929
494	PAH	A165T	rs199475626
495	PAH	A259V	rs118203921
496	PAH	A300S	rs5030853

497	PAH	A342P	rs62507282
498	PAH	A342T	rs62507282
499	PAH	A403V	rs5030857
500	PAH	D415V	-
501	PAH	E178G	rs77958223
502	PAH	E221G	rs62514934
503	PAH	E280K	rs62508698
504	PAH	E280Q	rs62508698
505	PAH	E390G	rs5030856
506	PAH	EX3DEL	rs199475570
507	PAH	F299C	rs62642933
508	PAH	F39del	rs199475565
509	PAH	F39L	rs62642926
510	PAH	G188D	rs199475689
511	PAH	G256D	-
512	PAH	G272X	rs62514952
513	PAH	G344R	rs62508679
514	PAH	G46S	rs74603784
515	PAH	I306V	rs62642934
516	PAH	I421N	rs199475696
517	PAH	I65N	rs75193786
518	PAH	I65S	rs75193786
519	PAH	I65T	rs75193786
520	PAH	IVS1+5G>T	rs62514895
521	PAH	IVS10-11G>A	rs5030855
522	PAH	IVS10-1G>A	rs62508684
523	PAH	IVS10-3C>T	rs62507344
524	PAH	IVS11+1G>C	rs62509015
525	PAH	IVS12+1G>A	rs5030861
526	PAH	IVS2+5G>A	rs18798839
527	PAH	IVS2+5G>C	rs62507288
528	PAH	IVS2-13T>G	rs62507341
529	PAH	IVS4+5G>T	rs62507321
530	PAH	IVS4-1G>A	rs62514907
531	PAH	IVS7+1G>A	rs5030852
532	PAH	IVS7+2T>A	rs62514955
533	PAH	IVS9+5G>A	rs62508637
534	PAH	K363fsdelG	rs5030654
535	PAH	L213P	rs62516109
536	PAH	L255S	rs62642930
537	PAH	L311P	rs62642936
538	PAH	L48S	rs5030841
539	PAH	M1I	rs62514893
540	PAH	M1R	rs62508575
541	PAH	M1V	rs62514891
542	PAH	P211T	rs62514931
543	PAH	P225T	rs199475589
544	PAH	P244L	rs118203923
545	PAH	P281L	rs5030851
546	PAH	P407L	rs62644473

547	PAH	R111X	rs76296470
548	PAH	R158P	rs5030843
549	PAH	R158Q	rs5030843
550	PAH	R176X	rs199475575
551	PAH	R243L	rs62508588
552	PAH	R243Q	rs62508588
553	PAH	R243X	rs5030846
554	PAH	R252G	rs5030847
555	PAH	R252W	rs5030847
556	PAH	R261G	rs5030850
557	PAH	R261P	rs5030849
558	PAH	R261Q	rs5030849
559	PAH	R261X	rs5030850
560	PAH	R297H	rs62642939
561	PAH	R408Q	rs5030859
562	PAH	R408W	rs5030858
563	PAH	R413P	rs79931499
564	PAH	S273F	rs62514953
565	PAH	S349P	rs62508646
566	PAH	S349R	rs62508646
567	PAH	S359X	rs5030854
568	PAH	T372S	rs62517163
569	PAH	T380M	rs62642937
570	PAH	V230I	rs62516152
571	PAH	V245A	rs76212747
572	PAH	V388L	rs62516101
573	PAH	V388M	rs62516101
574	PAH	V399V	rs199475584
575	PAH	W326X	rs62514959
576	PAH	Y204C	rs62514927
577	PAH	Y277D	rs78655458
578	PAH	Y356H	rs62507320
579	PAH	Y356X (G>C)	rs62516095
580	PAH	Y356X (G>T)	rs62516095
581	PAH	Y414C	rs5030860