

## Mutazioni approvate FDA

1	3141del9	26	E116K	51	G480C
2	546insCTA	27	E60K	52	G27R
3	A1067T	28	E588V	53	G463V
4	A455E	29	E92K	54	G628R
5	A46D	30	E403D	55	G622D
6	A349V	31	E822K	56	G126D
7	A1006E	32	F508del	57	G970D
8	A120T	33	F1052V	58	G194R
9	A234D	34	F1074L	59	G314E
10	A554E	35	F311L	60	G194V
11	D110E	36	F575Y	61	G576A
12	D110H	37	F1099L	62	G1249R
13	D1152H	38	F508C	63	G178E
14	D1270N	39	F311del	64	G576A;R668C
15	D579G	40	F191V	65	H139R
16	D443Y	41	F1016S	66	H1085R
17	D924N	42	F508C;S1251N	67	H1085P
18	D192G	43	G1069R	68	H939R
19	D979V	44	G1244E	69	H1375P
20	D614G	45	G1349D	70	H1054D
21	D836Y	46	G178R	71	H199Y
22	D443Y;G576A;R668C	47	G551D	72	I502T
23	E193K	48	G551S	73	I618T
24	E56K	49	G85E	74	I175V
25	E474K	50	G1061R	75	I1366N

76	I601F	102	P67L	128	R751L
77	I980K	103	P5L	129	R347L
78	I1269N	104	P205S	130	R1283S
79	I1027T	105	P574H	131	R1162L
80	I148T	106	Q98R	132	R553Q
81	I336K	107	Q1291R	133	R792G
82	I807M	108	Q237E	134	R1066H
83	I1139V	109	Q237H	135	R668C
84	K1060T	110	Q359R	136	R334Q
85	L206W	111	R1070Q	137	R1283M
86	L1077P	112	R1070W	138	R75Q
87	L165S	113	R117C	139	R74Q
88	L1335P	114	R117H	140	R74W;V201M;D127N
89	L967S	115	R347H	141	E74W;D1270N
90	L320V	116	R352Q	142	R74W;V201M
91	L346P	117	R74W	143	S1251N
92	L1324P	118	R347P	144	S1255P
93	L997F	119	R117G	145	S549R
94	L453S	120	R170H	146	S945R
95	L453S	121	R334L	147	S945L
96	L1480P	122	R117L	148	S977F
97	M1101K	123	R31L	149	S13F
98	M265R	124	R933G	150	S1159F
99	M152V	125	R352W	151	S912L
100	M952I	126	R117P	152	S1159P
101	M952T	127	R258G	153	S580N

154	S341P	163	V201M	172	W1282R
155	S492F	164	V562I	173	Y563N
156	S737F	165	V1153E	174	Y161D
157	S264P	166	V1240G	175	Y109N
158	T1053I	167	V1293G	176	Y1014C
159	T1036N	168	V754M	177	Y161S
160	T338I	169	V232D	178	Y1032C
161	V456A	170	W361R		
162	V456F	171	W1098C		

#### Mutazioni testate nello studio clinico da Vertex

1. 2789+5G→A
2. 3272-26A→G
3. 3849+10kbC→T
4. P5L
5. R117C
6. L206W
7. V232D
8. T338I
9. R347H
10. A455E
11. S945L
12. L997F
13. R1066H
14. D1152H

15. G85E
16. R347P
17. L1077P
18. M1101K

#### **5 mutazioni splicing specifiche non canoniche**

1. 711+3A→G
2. 2789+5G→A
3. 3272-26A→G
4. 3849+10kbC→T
5. E831X

*Fonti: comunicato stampa Vertex, sito FDA, sito CF Europe*