



List of CFTR Gene Mutations That Are Responsive to Symdeko® (tezacaftor/ivacaftor)

546insCTA	E92K	G576A	L346P	R117G	S589N
711+3A→G	E116K	G576A; R668C [†]	L967S	R117H	S737F
2789+5G→A	E193K	G622D	L997F	R117L	S912L
3272-26A→G	E403D	G970D	L1324P	R117P	S945L
3849+10kbC→T	E588V	G1069R	L1335P	R170H	S977F
A120T	E822K	G1244E	L1480P	R258G	S1159F
A234D	E831X	G1249R	M152V	R334L	S1159P
A349V	F191V	G1349D	M265R	R334Q	S1251N
A455E	F311del	H939R	M952I	R347H	S1255P
A554E	F311L	H1054D	M952T	R347L	T338I
A1006E	F508C	H1375P	P5L	R347P	T1036N
A1067T	F508C; S1251N [†]	I148T	P67L	R352Q	T1053I
D110E	F508del [^]	I175V	P205S	R352W	V201M
D110H	F575Y	I336K	Q98R	R553Q	V232D
D192G	F1016S	I601F	Q237E	R668C	V562I
D443Y	F1052V	I618T	Q237H	R751L	V754M
D443Y; G576A; R668C [†]	F1074L	I807M	Q359R	R792G	V1153E
D579G	F1099L	I980K	Q1291R	R933G	V1240G
D614G	G126D	I1027T	R31L	R1066H	V1293G
D836Y	G178E	I1139V	R74Q	R1070Q	W1282R
D924N	G178R	I1269N	R74W	R1070W	Y109N
D979V	G194R	I1366N	R74W; D1270N [†]	R1162L	Y161S
D1152H	G194V	K1060T	R74W; V201M [†]	R1283M	Y1014C
D1270N	G314E	L15P	R74W; V201M; D1270N [†]	R1283S	Y1032C
E56K	G551D	L206W	R75Q	S549N	
E60K	G551S	L320V	R117C	S549R	

[^]A patient must have two copies of the F508del mutation or at least one copy of a responsive mutation present in the table.

[†]Complex/compound mutations where a single allele of the CFTR gene has multiple mutations; these exist independent of the presence of mutations on the other allele.

Source: U.S. Food and Drug Administration