

Known Mutations of the Glutaryl-CoA Dehydrogenase Gene
 Compiled by Stephen I. Goodman

<u>Location</u>	<u>Mutation</u>	<u>Base Change</u>	<u>Reference</u>
Exon I	M1V	1A>G	Kolker et al, 2007
	---	11delG	Spector & Sharer, unpublished
	S25L	74C>T	Spector et al, unpublished
	---	89/90 delC	Mushimoto et al, 2011
	---	88-91del4+IVS1 ^{gtca}	Spector et al, unpublished
Intron 1	IVS1+5G>T	---	Greenberg et al, 1995
Exon II	---	109-110del2	Tsai et al, 2017
	K42R	125A>G	Spector et al, unpublished
Intron 2	IVS2+1G>A	---	Venturoni et al, 2014
	IVS2-1G>C	---	Venturoni et al, 2014
	IVS2-2A>C	---	Zschocke et al, 2000
	IVS2-2A>T	---	Christensen et al, 2004
Exon III	---	146-149del4	Wen et al, 2012
	W50R	148T>C	Wang et al, 2014
	W50R	148T>A	Mosaeilhy et al, 2017
	W50G	148T>G	
	W50X	149G>A	Spector et al, unpublished
	W50C	150G>C	Zschocke et al, 2000
	---	152dupA	Goodman et al, 2005
	P53S	157C>T	Christensen et al, 2004
	P53Q	157C>A	Mosaeilhy et al, 2017
	---	158delC	Goodman et al, 1998
	---	174dupG	Christensen et al, 2004
	E64D	192G>T	Goodman, unpublished
	ΔL66	196-198del3	Spector & Sharer, unpublished
	---	198delC	Spector et al, unpublished
	---	198dupC	Spector et al, unpublished
	F71S	212T>C	Busquets et al, 2000a
	R72G	214G>C	Lewis B, unpublished
	R72L	215G>T	Mushimoto et al, 2011
	---	219delC	Zschocke et al, 2000
	C75G	223T>G	Christensen et al, 2004
	C75S	224G>C	Spector et al, unpublished
	Q76X	226C>T	Zschocke et al, 2000
	Q76P	227A>C	Abdul Wahab et al, 2016
	R78G	232A>G	Spector & Sharer, unpublished
	R78K	233G>A	Spector et al, unpublished
	R82P	245G>C	Liu et al, 2015
	A86G	257C>G	Christensen et al, 2004
	R88S	262C>A	Venturoni et al, 2014
	R88C	262C>T	Biery et al, 1996
	R88H	263G>A	Couce et al, 2013
	E90K	268G>A	Busquets et al, 2000a
Intron 3	IVS3+1G>A	---	Tang et al, 2000
	IVS3-2A>C	---	Goodman et al, 2005
Exon IV	H93L	278A>T	Spector et al, unpublished
	R94L	281G>T	Schwartz et al, 1998

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Exon IV	R94Q	281G>A	Gupta et al, 2015	
	M100I	300G>A	Spector et al, unpublished	
	G101R	301G>A	Anikster et al, 1996	
	G112X	334G>T	Zhang et al, 2016	
Intron 4	IVS4+2T>C	---	Mushimoto et al, 2011	
	IVS4+5G>A	---	Goodman et al, 1998	
Exon V	Y113H	337T>C	Zschocke et al, 2000	
	C115Y	344G>A	Goodman et al, 1998	
	G117R	349G>C	Christensen et al, 2004	
	S119L	356C>T	Busquets et al, 2000a	
	A122V	365C>T	Goodman et al, 1998	
	Y123H	367T>C	Spector et al, unpublished	
	G124E	371G>A	Wang et al, 2014	
	R128G	382C>G	Goodman et al, 1998	
	R128X	382C>T	Busquets et al, 2000a	
	R128Q	383G>A	Zschocke et al, 2000	
	---	387-388del2	Busquets et al, 2000a	
	E131V	392A>T	Abdul Wahab et al, 2016	
	R132G	394C>G	Bahr et al, 2002	
	R132Q	395G>A	Zschocke et al, 2000	
	V133M	397G>A	Zschocke et al, 2000	
	V133L	397G>T	Venturoni et al, 2014	
	D134G	401A>G	Gupta et al, 2015	
	S135G	403A>G	Spector et al, unpublished	
	G136C	406G>T	Wang et al, 2014	
	Y137X	411C>G	Wang et al, 2014	
	R138G	412A>G	Goodman et al, 1998	
	R138K	413G>A	Zschocke et al, 2000	
	S139L	416C>T	Goodman et al, 1998	
	S139W	416C>G	Wang et al, 2014	
	---	420del10	Wen et al, 2012	
	---	423-424del2	Christensen et al, 2004	
	Q144P	431A>C	Kruthika-Vinod et al, 2017	
	---	433delT	Kalkanoglu et al, 2003	
	S145Y	434C>A	Spector et al, unpublished	
	S146F	437C>T	Spector & Sharer, unpublished	
	S146Y	437C>A	Spector & Sharer, unpublished	
	V148I	442G>A	Schwartz et al, 1998	
	I152M	456C>G	Kruthika-Vinod et al, 2017	
	Y155H	463T>C	Busquets et al, 2000a	
	Y155C	464G>A	Goodman, unpublished	
	E158K	472G>A	Spector et al, unpublished	
	Q160X	478C>T	Park et al, 2010	
	Q160R	479A>G	Goodman, unpublished	
	R161W	481C>T	Busquets et al, 2000c	
	R161Q	482G>A	Biery et al, 1996	
	Q162R	485A>G	Radha Rama Devi et al, 2015	
	Intron 5	IVS5+1G>A	---	Zschocke et al, 2000
		IVS5-2A>G	---	Mahfoud et al, 2004

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Exon VI	K170E	508A>G	Wen et al, 2012	
	K170N	510G>C	Spector et al, unpublished	
	G171W	511G>T	Kolker et al, 2007	
	L174P	521T>C	Spector et al, unpublished	
	L174P	523T>C	Goodman et al, unpublished	
	C176R	526T>C	Zschocke et al, 2000	
	G178R	532G>A	Biery et al, 1996	
	G178E	533G>A	Goodman et al, unpublished	
	L179P	536T>C	Tehrani S et al, 2011	
	L179R	536T>G	Goodman et al, 1998	
	E181Q	541G>C	Zschocke et al, 2000	
	E181K	541G>A	Busquets et al, 2000a	
	E181G	542A>G	Spector & Sharer, unpublished	
	S184G	550A>G	Spector et al, unpublished	
	G185R	553G>A	Goodman, unpublished	
	G185A	554G>C	Zschocke et al, 2000	
	dG185-S190	---	Bross et al, 2012	
	S186C	556A>T	Mushimoto et al, 2011	
	S186I	557G>T	Spector et al, unpublished	
	M191T	572T>C	Schwartz et al, 1998	
	M191R	572T>G	Spector et al, unpublished	
	T193_R194insH	---	Korman et al, 2007	
	A195T	583G>A	Schwartz et al, 1998	
	K210N	630G>T	Spector et al, unpublished	
	Intron 6	IVS6-1G>A	---	Zschocke et al, 2000
	Exon VII	---	636-10_642dup	Badve et al, 2015
T214A		640A>G	Wang et al, 2014	
T214M		641C>T	Goodman, unpublished	
N215S		644A>G	Spector et al, unpublished	
---		644dupCTCG	Moseilhy et al, 2016	
S216L		647C>T	Goodman, unpublished	
P217L		650C>T	Zschocke et al, 2000	
A219T		655G>A	Tang et al, 2000	
D220N		658G>A	Spector et al, 2008	
D220Y		658G>T	Park et al, 2010	
L221P		662T>C	Gupta et al, 2015	
W225X		675G>A	Radha Rama Devi et al, 2016	
A226T		676G>A	Goodman, unpublished	
R227P		680G>C	Biery et al, 1996	
C228R		682T>C	Kolker et al, 2007	
C228F		683G>T	Spector & Sharer, unpublished	
R234W		700C>T	Spector & Sharer, unpublished	
R234Q		701G>A	Spector & Sharer, unpublished	
F236L		706T>C	Goodman et al, 1998	
L238P		713T>C	Lin et al, 2002	
G244C		730G>T	Goodman, unpublished	
G244S		730G>A	Mushimoto et al, 2011	
P248L		743C>T	Zschocke et al, 2000	
Q251X		751C>T	Spector & Sharer, unpublished	
S255L		764C>T	Busquets et al, 2000a	

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Exon VII	S255W	764C>G	Kolker et al, 2007	
	L256P	767T>C	Spector et al, unpublished	
	R257W	769C>T	Schwartz et al, 1998	
	R257Q	770G>A	Schwartz et al, 1998	
	S259P	775T>C	Zschocke et al, 2000	
	S259L	776C>T	Christensen et al, 2004	
	M263V	787A>G	Muhlhausen et al, 2003	
	M266V	796A>G	Goodman et al, 1998	
	M266T	797T>C	Wen et al, 2012	
	---	802dupG	Goodman, unpublished	
	P278S	832C>T	Schwartz et al, 1998	
	P278R	833C>T	Spector & Sharer, 2006	
	---	848delT	Busquets et al, 2000a	
	L283P	848T>C	Anikster et al, 1996	
	L283R	848T>G	Spector et al, unpublished	
	Intron 7	IVS7+1G>A	---	Goodman et al, 1998
		IVS7+1delG	---	Spector et al, unpublished
Exon VIII	P286S	856C>T	Radha Rama Devi et al, 2016	
	P286L	857C>T	Spector et al, unpublished	
	N291K	873C>A	Tsai et al, 2017	
	---	873delC	Wang et al, 2014	
	A293T	877G>A	Biery et al, 1996	
	R294W	880C>T	Schwartz et al, 1998	
	R294P	881G>C	Gupta et al, 2015	
	R294Q	881G>A	Christensen et al, 2004	
	Y295H	883T>C	Goodman et al, 1995	
	G296S	886G>A	Spector, unpublished	
	A298T	892G>A	Spector et al, unpublished	
	A298V	893C>T	Spector & Sharer, unpublished	
	A298T	894G>A	Christensen et al, 2004	
	V301M	901G>A	Wang et al, 2014	
	V301A	902T>C	Spector & Sharer, unpublished	
	G303R	907G>A	Goodman, unpublished	
	A304T	910G>A	Busquets et al, 2000a	
	A304P	910G>C	Spector et al, unpublished	
	S305L	914C>T	Anikster et al, 1996	
	E306G	917A>G	Spector et al, unpublished	
	F307S	920T>C	Spector et al, unpublished	
	C308R	922T>C	Kim et al, 2014	
	C308S	923G>C	Goodman et al, 1998	
	L309W	926T>G	Goodman et al, 1998	
	---	932dupC	Spector et al, unpublished	
	R313W	937C>T	Goodman et al, 1998	
	R313Q	938G>A	Zschocke et al, 2000	
	A316D	947C>A	Busquets et al, 2000a	
D318N	952G>A	Spector et al, unpublished		
Intron 8	IVS8+1G>A	---	Spector et al, unpublished	
Exon IX	Q321E	961C>G	Spector et al, unpublished	
	A327T	979G>A	Wang et al, 2014	

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Exon IX	N329S	986A>G	Korman et al, 2007	
	Q333E	997C>G	Goodman et al, 1998	
	Q333X	997C>T	Goodman et al, 2005	
	K335Q	1003A>C	Christensen, unpublished	
	D338Y	1012G>T	Spector et al, unpublished	
	M339V	1015A>G	Ikeda et al, 1998	
	L340F	1018C>T	Kalkanoglu et al, 2003	
	T341P	1021A>C	Korman et al, 2007	
	T341I	1022C>T	Spector & Sharer, 2006	
	L347F	1039C>T	Goodman, unpublished	
	A349T	1045G>A	Schwartz et al, 1998	
	Q352X	1054C>T	Mushimoto et al, 2011	
	G354R	1060G>C	Goodman et al, 1998	
	G354S	1060G>A	Schwartz et al, 1998	
		1060-1065del6	Spector et al, unpublished	
	G354A	1061G>C	Mushimoto et al, 2011	
	R355C	1063C>T	Goodman et al, 1998	
	R355H	1064G>A	Schwartz et al, 1998	
	L356P	???	Spector, 2008	
	D360Y	1078G>T	Spector et al, unpublished	
	K361E	1081A>G	Mushimoto et al, 2011	
	---	1084del161	Christensen et al, 2004	
	Intron 9	---	---	---
	Exon X	A362D	1085C>A	Goodman et al, unpublished
		E365K	1093G>A	Biery et al, 1996
		M366I	1098G>A	Spector et al, unpublished
L369P		1106T>C	Spector & Sharer, unpublished	
K371X		1111A>T	Spector et al, unpublished	
R372K		1115G>A	Busquets et al, 2000a	
N373S		1118A>G	Pirzadeh et al, 2017	
C375R		1123T>C	Goodman et al, 1998	
C375Y		1124G>A	Goodman et al, 2002	
I381T		1142T>C	Spector et al, unpublished	
A382T		1144G>A	Goodman et al, 1998	
---		1144-1145del2	Mushimoto et al, 2011	
R383S		1147C>A	Shadmehri et al, 2019	
R383C		1147C>T	Goodman et al, 1998	
R383H		1148G>A	Goodman et al, 1998	
A385V		1154C>T	Zschocke et al, 2000	
R386X		1156C>T	Biery et al, 1996	
R386G		1156C>G	Tang et al, 2000	
R386Q		1157G>A	Goodman et al, 1998	
R386P		1157G>C	Goodman et al, unpublished	
---		1160-1173del14	Schwartz et al, 1998	
M388T		1163T>C	Muhlhausen et al, unpublished	
G390R		1168G>C	Anikster et al, 1996	
G390W		1168G>T	Abdul Wahab et al, 2016	
G390E		1169G>A	Chen et al, 2018	
G390A		1169G>C	Goodman et al, 1998	
G390V	1169G>T	Busquets et al, 2000b		

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Exon X	---	1172-1173insT	Wang et al, 2014	
	---	1173delG	Anikster et al, 1996	
	---	1173dupG	Spector & Scharer, unpublished	
	N392D	1174A>G	Goodman et al, 1998	
	D396N	1186G>A	Spector et al, unpublished	
	E397X	1189G>T	Mosaeilhy et al, 2017	
	E397K	1189G>A	Spector et al, unpublished	
	Y398C	1193A>G	Couce et al, 2013	
	V400M	1198G>A	Biery et al, 1996	
	---	1199dupT	Goodman, unpublished	
	R402W	1204C>T	Biery et al, 1996	
	R402Q	1205G>A	Goodman et al, 1998	
	---	1207delG	Christensen et al, 2004	
	H403Y	1207C>T	Wang et al, 2014	
	H403R	1208A>G	Goodman et al, 1998	
	---	1210delG	Christensen et al, 2004	
	---	1210dupG	Busquets et al, 2000c	
	M405V	1213A>G	Goodman, unpublished	
	---	1213dupA	Lisyova et al, 2016	
	N406S	1217A>G	Goodman, unpublished	
	N406K	1218C>G	Goodman et al, 1998	
	---	1219-1233del15	Christensen et al, 2004	
	L407V	1219C>G	Pierson et al, 2015	
	L407P	1220T>C	Goodman et al, 1998	
	A409T	1225G>A	Lisyova et al, 2016	
	V410M	1228G>A	Kolker et al, 2007	
	Y413D	1237T>G	Mushimoto et al, 2011	
	Y413C	1238A>G	Gupta et al, 2015	
	Y413X	1239C>A	Zschocke et al, 2000	
	E414K	1240G>A	Biery et al, 1996	
	E414X	1240G>T	Kruthika-Vinod et al, 2017	
	E414A	1241A>C	Gupta et al, 2015	
	Intron 10	IVS10+1G>C	---	Goodman et al, 1998
		IVS10-2A>C	---	Tang et al, 2000
		IVS10-2A>G	---	Goodman et al, unpublished
	Exon XI	T416I	1247C>T	Anikster et al, 1996
		A421T	1261G>A	Goodman et al, 1998
		A421P	1261G>C	Spector et al, unpublished
		A421V (Amish)	1262C>T	Biery et al, 1996
		---	1276dupA	Spector et al, unpublished
---		1282-1285ins71	Wang et al, 2014	
I428M		1284C>G	Mosaeilhy et al, 2017	
T429M		1286C>T	Schwartz et al, 1998	
A433E		1298C>A	Schwartz et al, 1998	
A433V		1298C>T	Busquets et al, 2000a	
F434L		1300T>C/1302C>A	Christensen, 2005	
X439W		1317A>G	Busquets et al, 2000b	

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