

**Known Mutations of the Glutaryl-CoA Dehydrogenase Gene:**

Compiled by Stephen I. Goodman

Updated February 08, 2005

<u>Location</u>	<u>Mutation</u>	<u>Base Change</u>	<u>Reference</u>
Exon I	---	---	---
Intron 1	---	IVS1+5G>T	Greenberg et al, 1995
Exon II	---	---	---
Intron 2	---	IVS2-2A>C	Zschocke et al, 2000
Exon III	W50C	150G>C	Zschocke et al, 2000
	---	152-153insA	Goodman et al, unpublished, 2005
	---	158delC	Goodman et al, 1998
	E64D	192G>T	Goodman et al, unpublished, 2001
	F71S	212T>C	Busquets et al, 2000a
	---	219delC	Zschocke et al, 2000
	Q76X	226C>T	Zschocke et al, 2000
	R78G	232A>G	Spector & Sharer et al, unpublished, 2005
	R88C	262C>T	Biery et al, 1996
	E90K	268G>A	Busquets et al, 2000a
Intron 3	---	IVS3+1G>A	Tang et al, 2000
	---	IVS3-2A>C	Goodman et al, unpublished, 2005
Exon IV	R94L	281G>T	Schwartz et al, 1998
	G101R	301G>A	Anikster et al, 1996
Intron 4	---	IVS4+5G>A	Goodman et al, 1998
Exon V	Y113H	337T>C	Zschocke et al, 2000
	C115Y	344G>A	Goodman et al, 1998
	S119L	356C>T	Busquets et al, 2000a
	A122V	365C>T	Goodman et al, 1998
	R128G	382C>G	Goodman et al, 1998
	R128X	382C>T	Busquets et al, 2000a
	R128Q	383G>A	Zschocke et al, 2000
	---	387-388delGC	Busquets et al, 2000a
	R132Q	395G>A	Zschocke et al, 2000
	V133M	397G>A	Zschocke et al, 2000
	R138G	412A>G	Goodman et al, 1998
	R138K	413G>A	Zschocke et al, 2000
	S139L	416C>T	Goodman et al, 1998
	---	433delT	Kalkanoglu et al, 2003
	S146F	437C>T	Spector & Sharer et al, unpublished, 2005
	S146Y	437C>A	Spector & Sharer et al, unpublished, 2005
	V148I	442G>A	Schwartz et al, 1998
	Y155H	463T>C	Busquets et al, 2000a
	Y155C	464G>A	Goodman et al, unpublished, 2001
	Q160R	479A>G	Goodman et al, unpublished, 2001
	R161W	481C>T	Busquets et al, 2000c
	R161Q	482G>A	Biery et al, 1996

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Intron 5	---	IVS5+1G>A	Zschocke et al, 2000
Exon VI	C176R	526T>C	Zschocke et al, 2000
	G178R	532G>A	Biery et al, 1996
	G178E	533G>A	Goodman et al, unpublished, 2004
	L179R	536T>G	Goodman et al, 1998
	E181Q	541G>C	Zschocke et al, 2000
	E181K	541G>A	Busquets et al, 2000a
	G185R	553G>A	Goodman et al, unpublished, 2001
	G185A	554G>C	Zschocke et al, 2000
	M191T	572T>C	Schwartz et al, 1998
	A195T	583G>A	Schwartz et al, 1998
Intron 6	---	IVS6-1G>A	Zschocke et al, 2000
Exon VII	T214M	641C>T	Goodman et al, unpublished, 2001
	S216L	647 C>T	Goodman et al, unpublished, 2003
	P217L	650C>T	Zschocke et al, 2000
	A219T	655G>A	Tang et al, 2000
	A226T	676G>A	Goodman et al, unpublished, 2001
	R227P	680G>C	Biery et al, 1996
	F236L	706T>C	Goodman et al, 1998
	L238P	713T>C	Lin et al, 2002
	G244C	730G>T	Goodman et al, unpublished, 2001
	P248L	743C>T	Zschocke et al, 2000
	Q251X	751C>T	Spector & Sharer et al, unpublished, 2005
	S255L	764C>T	Busquets et al, 2000a
	R257W	769C>T	Schwartz et al, 1998
	R257Q	770G>A	Schwartz et al, 1998
	S259P	775T>C	Zschocke et al, 2000
	M263V	787A>G	Muhlhausen et al, unpublished, 2003
	M266V	796A>G	Goodman et al, 1998
	---	802-803insG	Goodman et al, unpublished, 2002
	P278S	832C>T	Schwartz et al, 1998
	---	848delT	Busquets et al, 2000a
L283P	848T>C	Anikster et al, 1996	
Intron 7	---	IVS7+1G>A	Goodman et al, 1998
Exon VIII	A293T	877G>A	Biery et al, 1996
	R294W	880C>T	Schwartz et al, 1998
	Y295H	883T>C	Goodman et al, 1995
	A298V	893C>T	Spector & Sharer et al, unpublished, 2005
	G303R	907G>A	Goodman et al, unpublished, 2001
	A304T	910G>A	Busquets et al, 2000a
	S305L	914C>T	Anikster et al, 1996
	C308S	923G>C	Goodman et al, 1998
	L309W	926T>G	Goodman et al, 1998
	R313W	937C>T	Goodman et al, 1998
	R313Q	938G>A	Zschocke et al, 2000
	A316D	947C>A	Busquets et al, 2000a
	Intron 8	---	---

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Exon IX	Q333E	997C>G	Goodman et al, 1998	
	Q333X	997C>T	Goodman et al, unpublished, 2005	
	M339V	1015A>G	Ikeda et al, 1998	
	L340F	1018C>T	Kalkanoglu et al, 2003	
	L347F	1039C>T	Goodman et al, unpublished, 2002	
	A349T	1045G>A	Schwartz et al, 1998	
	G354R	1060G>C	Goodman et al, 1998	
	G354S	1060G>A	Schwartz et al, 1998	
	R355C	1063C>T	Goodman et al, 1998	
	R355H	1064G>A	Schwartz et al, 1998	
	Intron 9	---	---	---
	Exon X	E365K	1093G>A	Biery et al, 1996
R372K		1115G>A	Busquets et al, 2000a	
C375R		1123T>C	Goodman et al, 1998	
C375Y		1124G>A	Goodman et al, unpublished, 2002	
A382T		1144G>A	Goodman et al, 1998	
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, 2005	
---		1160-1173del	Schwartz et al, 1998	
M388T		1163T>C	Muehlhausen et al, unpublished, 2004	
G390R		1168G>C	Anikster et al, 1996	
G390A		1169G>C	Goodman et al, 1998	
G390V		1169G>T	Busquets et al, 2000b	
---		1173delG	Anikster et al, 1996	
N392D		1174A>G	Goodman et al, 1998	
V400M		1198G>A	Biery et al, 1996	
---		1198-1199insT	Goodman et al, unpublished, 2003	
R402W		1204C>T	Biery et al, 1996	
R402Q		1205G>A	Goodman et al, 1998	
H403R		1208A>G	Goodman et al, 1998	
---		1209-1210insG	Busquets et al, 2000c	
M405V		1213A>G	Goodman et al, unpublished, 2004	
N406K		1218C>G	Goodman et al, 1998	
N406S		1217A>G	Goodman et al, unpublished, 2003	
L407P		1220T>C	Goodman et al, 1998	
Y413X		1239C>A	Zschocke et al, 2000	
E414K		1240G>A	Biery et al, 1996	
Intron 10		---	IVS10+1G>C	Goodman et al, 1998
		---	IVS10-2A>C	Tang et al, 2000
	---	IVS10-2A>G	Goodman et al, unpublished, 2005	

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Exon XI	T416I	1247C>T	Anikster et al, 1996
	A421T	1261G>A	Goodman et al, 1998
	A421V (Amish)	1262C>T	Biery et al, 1996
	T429M	1286C>T	Schwartz et al, 1998
	A433E	1298C>A	Schwartz et al, 1998
	A433V	1298C>T	Busquets et al, 2000a
	X439W	1317A>G	Busquets et al, 2000b

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