

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	---	11delG	Spector & Sharer, unpublished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T	Spector et al, unpublished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	---	88-91del4+IVS1 ^{gtca}	Spector et al, unpublished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G	Spector et al, unpublished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A	Spector et al, unpublished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T	Goodman, unpublished
	ΔL66	196-198del3	Spector & Sharer, unpublished
	---	198delC	Spector et al, unpublished
	---	198dupC	Spector et al, unpublished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C	Lewis B, unpublished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C	Spector et al, unpublished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G	Spector & Sharer, unpublished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A	Spector et al, unpublished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T	Spector et al, unpublished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A	Spector et al, unpublished	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C	Spector et al, unpublished	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	
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	
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	
G390A		1169G>C	Goodman et al, 1998	
G390V		1169G>T	Busquets et al, 2000b	
---		1172-1173insT	Wang et al, 2014	
---		1173delG	Anikster et al, 1996	
---		1173dupG		

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Exon X	---	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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References:

- Abdul Wahab SA, Yakob Y, Abdul Azize NA et al. Clinical and mutational analysis of the GCDH gene in Malaysian patients with glutaric aciduria type 1. *Biomed Res Int* 2016;4074365. Epub 2016 Sep 8
- Anikster Y, Shaag A, Joseph A, et al. Glutaric aciduria type I in the Arab and Jewish communities in Israel. *Am J Hum Genet* 59;1012-1018, 1996
- Bahr O, Mader I, Zschocke J, et al. Adult onset glutaric aciduria type I presenting with a leukoencephalopathy. *Neurology* 59;1802-4, 2002
- Badve MS, Bhuta S, McGill J. Rare presentation of a treatable disorder: glutaric aciduria type 1. *New Zeal Med J* 128;61-64, 2015
- Biery BJ, Stein DE, Morton DH, Goodman SI. Gene structure and mutations of glutaryl-CoA dehydrogenase: Impaired association of enzyme subunits that is due to an A421V substitution causes glutaric acidemia type I in the Amish. *Am J Hum Genet* 59;1006-1011, 1996
- Bross P, Frederiksen JB, Bie AS, et al. Heterozygosity for an in-frame deletion causes glutaryl-CoA dehydrogenase deficiency in a patient detected by newborn screening: investigation of the effect of the mutant allele. *J Inherit Metab Dis* 35;787-796, 2017
- Busquets C, Begona M, Christensen E, et al. Glutaryl-CoA dehydrogenase deficiency in Spain: Evidence of two groups of patients, genetically, and biochemically distinct. *Pediatr Res* 48;315-322, 2000a
- Busquets C, Soriano M, Tavares de Alameda I, et al. Mutation analysis of the GCDH gene in Italian and Portuguese patients with glutaric aciduria type I. *Mol Genet Metab* 71;535-537, 2000b
- Busquets C, Coll MJ, Merinero B, et al. Prenatal molecular diagnosis of glutaric aciduria type 1 by direct mutation analysis. *Prenat Diagn* 20;761-764, 2000c
- Christensen E, Ribes A, Merinero B, Zschocke J. Correlation of genotype and phenotype in glutaryl-CoA dehydrogenase deficiency. *J Inherit Metab Dis* 27;861-868, 2004
- Couce ML, Lopez-Suarez O, Boveda MD et al. Glutaric aciduria type 1: Outcome of patients with early- versus late-diagnosis. *Eur J Paediatr Neurol* 17;383-389, 2013
- Goodman SI, Kratz LE, DiGiulio KA, et al. Cloning of glutaryl-CoA dehydrogenase cDNA, and expression of wild type and mutant enzymes in *Escherichia coli*. *Hum Molec Genet* 4;1493-1498, 1995
- Goodman SI, Stein DE, Schlesinger S, et al. Glutaryl-CoA dehydrogenase mutations in glutaric acidemia (type I): Review and report of thirty novel mutations. *Hum Mutat* 12;141-144, 1998
- Greenberg CR, Reimer D, Singal R, et al. A G-to-T transversion at the +5 position of intron 1 in the glutaryl-CoA dehydrogenase gene is associated with the Island Lake variant of glutaric acidemia type I. *Hum Mol Genet* 4;493-495, 1995
- Gupta N, Singh PK, Kumar M, et al. Glutaric acidemia type 1 – Clinico-molecular profile and novel mutations in GCDH gene in Indian patients. *JIMD Reports* 21;45-55, 2015
- Ikeda H, Kimura T, Ikegami T, et al. Novel mutations of the glutaryl-CoA dehydrogenase gene in two Japanese patients with glutaric aciduria type I. *Am J Med Genet* 80;327-329, 1998

Known Mutations of the Glutaryl-CoA Dehydrogenase Gene

Compiled by Stephen I. Goodman

Kalkanoglu HS, Dursun A, Tokatly A, et al. Two novel mutations in glutaric aciduria type I. *J Inher Metab Dis* 26 (Suppl 2);193, 2003 (Abstr)

Kim HS, Lu HJ, Lee J et al. A Korean patient with glutaric aciduria type 1 with a novel mutation in the glutaryl CoA dehydrogenase gene. *Ann Clin Lab Sci* 44;213-216, 2014

Kolker S, Garbade SF, Boy N, et al. Decline of acute encephalopathic crises in children with glutaryl-CoA dehydrogenase deficiency identified by newborn screening in Germany. *Pediat Res* (in press)

Korman SH, Jacobs C, Darmin PS, et al. Glutaric aciduria type 1: Clinical, biochemical and molecular findings in patients from Israel. *Eur J Paediatr Neurology* 11;81-89, 2007

Kruthika-Vinod TP, Muntaj S, Devaraju KS et al. Genetic screening of selected disease-causing mutations in glutaryl-CoA dehydrogenase gene among Indian patients with glutaric aciduria type I. *J Pediatr Genet* 6;142-148, 2017

Lin SK, Hsu SG, Ho ES, et al. Novel mutation and prenatal sonographic findings of glutaric aciduria (type I) in two Taiwanese families. *Prenat Diagn* 22;725-729, 2002

Lisyova J, Petrovic R, Jurickova K, et al. GA1 – distinct genotype and phenotype characteristics in reported Slovak patients. *Bratisl Med J* 117;631-688, 2016

Liu Q, Chen Y, Chen W. Mutation analysis of GCDH gene in four patients with glutaric acidemia type I. *Zhonghua Xi Xue Yi Za Zhi* 32;187-191, 2015 [Article in Chinese]

Mahfoud A, Dominguez CL, Rizzo C, Ribes A. In utero macrocephaly as clinical manifestation of glutaric aciduria type I. *Rev Neurol* 39;939-942, 2004

Mosaeilhy A, Mohamed MM, Priya Doss GC et al. Genotype-phenotype correlation in 18 Egyptian patients with glutaric acidemia type 1. *Metab Brain Dis* 2017 Apr 7 [Epub ahead of print]

Moseilhy A, Hassan MM, El Abd EI et al. Severe neurological manifestations in an Egyptian patient with a novel frameshift mutation in the glutaryl-CoA dehydrogenase gene. *Metab Brain Dis* 32;35-40, 2017

Muhlhausen C, Christensen E, Schwartz M, et al. Severe phenotype despite high residual glutaryl-CoA dehydrogenase activity: A novel mutation in a Turkish patient with glutaric aciduria type I. *J Inher Metab Dis* 26;713-714, 2003

Mushimoto Y, Fukuda S, Hasegawa Y, et al. Clinical and molecular investigation of 19 Japanese cases of glutaric acidemia type 1. *Molec Genet Metab* 102;343-348, 2011

Park JD, Lim B, Kim KJ, et al. Glutaric aciduria type 1 in Korea; Report of two novel mutations. *J Korean Med Sci* 25; 957-960, 2010.

Pierson TM, Nezhad M, Tremblay MA, et al. Adult-onset glutaric aciduria type I presenting with white matter abnormalities and subependymal nodules. *Neurogenetics* 16;325-328, 2015

Radha Rama Devi A, Ramesh VA, Nagarajaram HA, et al. Spectrum of mutations in glutaryl-CoA dehydrogenase gene in glutaric aciduria type I – Study from south India. *Brain Dev* 38;54-60, 2016

Schwartz M, Christensen E, Superti-Furga A, Brandt NJ. The human glutaryl-CoA dehydrogenase gene: Report of intronic sequences and of thirteen novel mutations causing glutaric aciduria type I. *Hum Genet* 102;452-458, 1998

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Tang NLS, Hui J, Law LK, et al. Recurrent and novel mutations of GCDH gene in Chinese glutaric acidemia type I families. *Human Mutation. Mutation in Brief #375* (2000) Online

Tehrani S, Zaman, Houshmand. The clinical, biochemical and molecular features of three Iranian patients with glutaryl-CoA dehydrogenase deficiency. *J Inherit Metab Dis 34 (Suppl 3); S140*, 2011

Tsai FC, Lee HJ, Wang AG, et al. Experiences during newborn screening for glutaric aciduria type 1: Diagnosis, treatment, genotype, phenotype, and outcomes. *J Chin Med Assoc 80;253-261*, 2017

Venturoni L, Woontner M, Goodman SI. Deep sequencing of GCDH in known glutaric acidemia type 1 patients. *Molec Genet Metab 111;274*, 2014 (Abstr)

Wang Q, Li X, Ding Y et al. Clinical and mutational spectra of 23 Chinese patients with glutaric aciduria type 1. *Brain and Develop 36;813-822*, 2014

Wen P-, Wang GB, Liu XH et al. Analysis of clinical features and GCDH gene mutations in four patients with glutaric acidemia type 1. *Zhonghua Yi Xue Yi Chuan Xue Za Zhi 2012 Dec;29(6):642-647*

Zhang Y, Li H, Ma R et al. Clinical and molecular investigation in Chinese patients with glutaric aciduria type 1. *Clin Chim Acta 453;75-79*, 2016

Zschocke J, Quak E, Guldberg P, Hoffmann GF. Mutation analysis in glutaric aciduria type I. *J Med Genet 37;177-181*, 2000