Supplementary Material: Table S1

Method

In order to identify reported pathogenic variants resulting in MADD and MADD-like phenotypes, a systematic literature search was performed in PubMed (https://pubmed.ncbi.nlm.nih.gov/, June 2020) using the following terms (without quotation marks): “Multiple acyl CoA dehydrogenase deficiency”, “Glutaric aciduria type 2”, “Glutaric acidemia type 2”, “ETFDH”, “ETFA”, “ETFB”, “Flavin adenine dinucleotide synthase deficiency”, “FAD synthase deficiency”, “FLAD1”, “SLC25A32”, “Riboflavin transporter deficiency”, “Brown-Violetto-Van Laere”, “Fazio-Londe”, “SLC52A1”, “SLC52A2” and “SLC52A3”. The search yielded 872 articles (published between 1962 and July 2020), of which 179 described pathogenic variants in MADD-related genes. The remainder did not report any applicable variants (636 articles), could not be reviewed due to language restraints (51 articles), or were not available to the authors (6 articles). An additional 19 articles, which were not identified by the search, but contained MADD-related variants (cited in the identified literature), were also included. Finally, the *ETFDH* (ENST00000511912.6), *ETFA* (ENST00000557943.6), *ETFB* (ENST00000309244.9), *FLAD1* (ENST00000292180.8), *SLC25A32* (ENST00000297578.9), *SLC52A1* (ENST00000254853.10), *SCL52A2* (ENST00000643944.2) and *SLC52A3* (ENST00000645534.1) genes were searched via Ensembl (https://www.ensembl.org/, July 2020) and gnomAD v2.1.1 (Genome Aggregation Database; https://gnomad.broadinstitute.org/, July 2020) to supplement any missing information of the variants that were reported in literature, and to identify all current additional variants with a CADD (Combined Annotation Dependent Depletion) score of ≥30 (likely pathogenic/pathogenic) (Rentzsch et al., 2019). The results are displayed in Table S1.

Table S1: Reported (literature and Ensembl and gnomAD v2.1.1 databases) pathogenic variants in MADD-related genes

#	Affected gene	Exon	Intron/ non-coding	Nucleotide change (position)	Nucleotide change (allele call)	Amino acid change (predicted from cDNA)	rsID	Reference*	CADD score
1	*ETFA*	1		n.a	c.1-40G>A	n.r	n.a	van Rijt et al., 2014 van Rijt et al., 2019 van Rijt et al., 2020	n.a
2	*ETFA*	1		chr15:76311382C>T	c.7C>T	p.Arg3*	n.a	Purevjav et al., 2002 Yotsumoto et al., 2008 Yamaguchi et al., 2012 Yamada et al., 2017	0
3	*ETFA*	2		chr15:76295725G>A	c.52C>T	p.Arg18*	rs754202690	Song et al., 2009	0
4	*ETFA*	2		chr15:76295706A>T	c.71T>A	p.Val24Glu	rs979879737	-	31
5	*ETFA*	2		chr15:76295703delA	c.72delA	p.Ile25*	n.a	Schiff et al., 2006 Grünert, 2014	n.a
6	*ETFA*	2		chr15:76295702delA	c.73delA	p.Ile25*	n.a	van Rijt et al., 2019	n.a
7	*ETFA*	3		chr15:76292699A>C	c.188T>G	p.Val63Gly	rs1231262303	-	33
8	*ETFA*	3		chr15:76292686A>G	c.200T>C	p.Leu67Pro	n.a	Saral et al., 2018 van Rijt et al., 2020	n.a
9	*ETFA*	3		chr15:76292645T>G	c.242A>C	p.His81Pro	rs972293793	van Rijt et al., 2019	25
10	*ETFA*	4		chr15:76292499A>C	c.283T>G	p.Leu95Val	n.a	Yotsumoto et al., 2008 Grünert, 2014	n.a
11	*ETFA*	4		chr15:76286463A>C	c.284T>G	p.Leu95Trp	rs119458969	Ohkuma et al., 2009 Grünert, 2014	32
12	*ETFA*	4		chr15:76292436C>T	c.346G>A	p.Gly116Arg	rs119458971	[a]Freneaux et al., 1992 [a]Frerman and Goodman, 2001	27
13	*ETFA*	5		chr15:76287943G>T	c.354C>A	p.Asn118Lys	n.a	Di Giacinto et al., 2014 Chautard et al., 2020	n.a
14	*ETFA*	5		chr15:76287944ins	c.355_356insC	p.Leu119Profs*10	n.a	Schiff et al., 2006 Grünert, 2014	n.a
15	*ETFA*	5		chr15:76287932C>T	c.365G>A	p.Arg122Lys	rs773865838	Schiff et al., 2006 Grünert, 2014 van Rijt et al., 2020	27
16	*ETFA*	5		chr15:76283808C>T	c.370G>A	p.Ala124Thr	rs1390694827	van Rijt et al., 2020	24
17	*ETFA*	5		chr15:76287866A>G	c.431T>C	p.Phe144Ser	rs755055339	-	32
18	*ETFA*	6		chr15:76286480del	c.453_470del	p.152_157del	n.a	[a]Freneaux et al., 1992	n.a
19	*ETFA*	6		chr15:76286463A>C	c.470T>G	p.Val157Gly	rs119458969	[a]Indo et al., 1991 [a]Freneaux et al., 1992 [a]Frerman and Goodman, 2001	32

20	*ETFA*	6		chr15:76286455del	c.478delG	p.Asp160Metfs*4	n.a	Purevjav et al., 2002 Yotsumoto et al., 2008 Grünert, 2014	n.a
21	*ETFA*	6		chr15:76286439A>G	c.494T>C	p.Val165Ala	rs749455172	Schiff et al., 2006 Di Giacinto et al., 2014 Grünert, 2014	28
22	*ETFA*	6		chr15:76286431C>A	c.502G>T	p.Val168Phe	rs779987487	Lee et al., 2010 Grünert, 2014	24
23	*ETFA*	6		chr15:76286421G>A	c.512C>T	p.Thr171Ile	rs1801591	[a]Freneaux et al., 1992 Henriques et al., 2011 Chu and Salzman, 2019	28
24	*ETFA*		6	chr:15:76286380C>G	c.553-1G>C	exon skipping or frameshift	n.a	Yotsumoto et al., 2008 Yamada et al., 2017	n.a
25	*ETFA*	7		chr15:76285665A>G	c.635T>C	p.Leu212Pro	n.a	Schiff et al., 2006 Grünert, 2014	n.a
26	*ETFA*	7		chr15:76285649C>T	c.652G>A	p.Val218Met	rs753461278	Chautard et al., 2020	26
27	*ETFA*	7		chr15:76285645-7628653del	c.656_664del	p.Val219_Gly221del	n.a	Yamada et al., 2017	n.a
28	*ETFA*		7	chr15:76285635-76285636del	c.664+1_664+2del	n.r	rs1567215572	van Rijt et al., 2019	n.a
29	*ETFA*	8		chr15:76283774T>A	c.716A>T	p.Asp239Val	rs1260250211	-	31
30	*ETFA*	9		chr15:76274488G>A	c.740C>T	p.Ala247Val	rs1466909697	-	32
31	*ETFA*	9		chr15:76274485G>A	c.743C>T	p.Ser248Phe	rs1384518039	-	33
32	*ETFA*	9		chr15:76274483G>C	c.745C>G	p.Arg249Gly	rs1353938548	-	32
33	*ETFA*	9		chr15:76274464A>C	c.764G>T	p.Gly255Val	n.a	Purevjav et al., 2002 Yotsumoto et al., 2008 Grünert, 2014	n.a
34	*ETFA*	9		chr15:76274442T>C	c.786A>G	p.Gln262Arg	rs373871967	Lee et al., 2010 Grünert, 2014	0
35	*ETFA*	9		chr15:76274431G>A	c.797C>T	p.Thr266Met	rs119458970	[a]Freneaux et al., 1992 [a]Frerman and Goodman, 2001 Olsen et al., 2003 Schiff et al., 2006 Grünert, 2014 Pontoizeau et al., 2016 van Rijt et al., 2019 Yıldız et al., 2019 van Rijt et al., 2020	32

36	*ETFA*	9		chr15:76274428C>T	c.799G>A	p.Gly267Arg	n.a	Purevjav et al., 2002 Yotsumoto et al., 2008 Yamaguchi et al., 2012 Yamada et al., 2017	n.a
37	*ETFA*	9		chr15:76274417-76274422CTACTA>CTA	c.809_811del	p.Val270del	rs779140971	[a]Freneaux et al., 1992 van Rijt et al., 2020	0
38	*ETFA*	9		chr15:76274416G>A	c.812C>T	p.Ala271Val	rs1567211944	-	32
39	*ETFA*	10		chr15:76232280del	c.817_882del	p.Glu273_Lys294del	n.a	[a]Freneaux et al., 1992	n.a
40	*ETFA*	10		chr15:76231361A>T	c.854A>T	p.Gln285Leu	n.a	Saral et al., 2018 van Rijt et al., 2020	n.a
41	*ETFA*	10		chr15:76231336-76231344del	c.875_878del	p.Asn292Alafs*23	n.a	Schiff et al., 2006 Grünert, 2014	n.a
42	*ETFA*	10		chr15:76231335T>C	c.880A>G	p.Lys294Glu	rs774043017	-	34
43	*ETFA*	10		chr15:76231333C>G	c.882G>C	p.Lys294Asn	rs905969804	-	34
44	*ETFA*	11		chr15:76226892-76226812del	c.883_963del	p.Thr295_Lys321del	n.a	Olsen et al., 2003	n.a
45	*ETFA*	11		chr15:76225895G>A	c.917C>T	p.Pro306Leu	rs768924673	-	31
46	*ETFA*	11		chr15:76225875A>T	c.937T>A	p.Tyr313Asn	rs1482839883	Pollard et al., 2010 Grünert, 2014	27
47	*ETFA*	11		chr15:76225848-76225849delC	c.963+1delG	splice variant	rs746488455	Schiff et al., 2006 Grünert, 2014	n.a
48	*ETFB*	1		chr19:51366314G>A	c.13C>T	p.Arg5Cys	rs1422832903	-	33
49	*ETFB*	1		chr19:51366283T>G	c.44A>C	p.Asp15Ala	rs981006954	-	31
50	*ETFB*	1		chr19:51366277C>G	c.49G>C	p.Ala17Pro	n.a	Angle and Burton, 2008	n.a
51	*ETFB*	1		chr19:51366271T>A	c.55A>T	p.Lys19*	n.a	Yotsumoto et al., 2008 Sudo et al., 2015	n.a
52	*ETFB*	1		chr19:51366271T>C	c.56A>G	p.Lys19Arg	rs767751467	-	31
53	*ETFB*	2		chr19:51354289del	c.77delG	p.Gly28fs	n.a	Yotsumoto et al., 2008	n.a
54	*ETFB*	2		chr19:51354288del	c.78delG	p.Gly28fs	n.a	Sudo et al., 2015	n.a
55	*ETFB*	2		chr19:51354244A>G	c.122T>C	p.Phe41Ser	rs746082442	-	33
56	*ETFB*	2		chr19:51354242A>G	c.124T>C	p.Cys42Arg	rs774387920	Curcoy et al., 2003 Grünert, 2014	25
57	*ETFB*	2		chr19:5134223-51354225del	c.143_145delAGG	p.Glu48del	n.a	Sudo et al., 2015	n.a
58	*ETFB*	2		chr19:51354179C>T	c.187G>A	p.Ala63Thr	rs142966954	van Rijt et al., 2019	26
59	*ETFB*		2	chr19:51353294C>A	c.217-4G>T	n.r	rs149557388	van Rijt et al., 2020	0
60	*ETFB*	3		chr19:51353663-51353665del	c.217_375del	p.Glu73_Gln125del	n.a	Olsen et al., 2003	n.a

61	*ETFB*	3		chr19:51353205G>A	c.302C>T	p.Pro101Leu	rs1249159210	-	32
62	*ETFB*	3		chr19:51353191G>A	c.316C>T	p.Arg106Trp	rs748730384	-	33
63	*ETFB*	3		chr19:51353134G>C	c.373C>G	p.Gln125Glu	rs1568467526	-	32
64	*ETFB*	4		chr19:51353132C>G	c.375G>C	p.Gln125His	rs1173733121	-	31
65	*ETFB*	4		chr19:51353132-	c.375G>C + c.375_376ins(9)	p.Gln125His + p.His125_Ala126ins(3)	n.a	Olsen et al., 2003	n.a
66	*ETFB*	4		chr19:51350385C>T	c.382G>A	p.Asp128Asn	rs104894678	Olsen et al., 2003 Henriques et al., 2009 Grünert, 2014	25
67	*ETFB*		4	chr19:51350309G>A	c.438+20C>T	n.r	rs114985874	van Rijt et al., 2020	0
68	*ETFB*	5		chr19:51347045G>A	c.452C>T	p.Ser151Phe	rs74735908	-	33
69	*ETFB*	5		chr19:51347036C>T	c.461C>T	p.Thr154Met	rs1130426	[a]Colombo et al., 1994 Janeiro et al., 2019	25
70	*ETFB*	5		chr19:51347007G>A	c.490C>T	p.Arg164Trp	rs148045813	Yotsumoto et al., 2008 Sudo et al., 2015	29
71	*ETFB*	5		chr19:51347006C>T	c.491G>A	p.Arg164Gln	rs104894677	[a]Colombo et al., 1994 [a]Frerman and Goodman, 2001 Yotsumoto et al., 2008 Grünert, 2014 Sudo et al., 2015 Yamada et al., 2017	28
72	*ETFB*	5		chr19:51346926G>A	c.571C>T	p.Arg191Cys	rs772976948	Schiff et al., 2006 Grünert, 2014 Henriques et al., 2019	34
73	*ETFB*	5		chr19:51346925C>T	c.572G>A	p.Arg191His	rs769951998	-	32
74	*ETFB*	5		chr19:51346916G>A	c.581C>T	p.Thr194Met	rs369138107	-	33
75	*ETFB*	5		chr19:51346908T>C	c.589A>G	p.Asn197Asp	rs747411704	-	32
76	*ETFB*	5		chr19:51346902T>G	c.595A>C	p.Met199Leu	rs975423352	-	34
77	*ETFB*		5	chr19:51346900del	c.597+1G>C	p.Gly147_Met199del	n.a	Sudo et al., 2015	n.a
78	*ETFB*		5	chr19:51346900del	c.597+1G>C	p.Gly148_Met200del	n.a	[a]Colombo et al., 1994 Yotsumoto et al., 2008 Grünert, 2014 Yamada et al., 2017	n.a
79	*ETFB*	6		chr19:51345381T>C	c.598A>G	p.Lys200Glu	rs920672041	-	33
80	*ETFB*	6		chr19:51345375-19:51345377del	c.604_606delAAG	p.Lys202del	n.a	Curcoy et al., 2003 Grünert, 2014	n.a

81	*ETFDH*		5'UTR	chr4:158672382A>G	c.-75A>G	transcript expression decreased	rs1001221143	Wen et al., 2013 Grünert, 2014 Xi et al., 2014	0
82	*ETFDH*	1		chr4:158672458T>C	c.2T>C	p.Met1Thr	rs121964953	[a]Beard et al., 1993 [a]Frerman and Goodman, 2001 [a]Goodman et al., 2002	21
83	*ETFDH*	1		chr4:158672459G>C	c.3G>C	p.Met1Ile	rs1340862175	[a,c]Wang et al., 2011b Grünert, 2014 Xi et al., 2014 Zhu et al., 2014 [a,c]Li, 2015 Zhao et al., 2018 Chen et al., 2019b	22
84	*ETFDH*	1		chr4:158672490G>C	c.34G>C	p.Ala12Pro	rs1172887273	Hong et al., 2018 Chen et al., 2019b Hong et al., 2019 van Rijt et al., 2020	22
85	*ETFDH*	1-5		n.a	exon 1-5del	deletion of exon 1-5	n.a	Han et al., 2019	n.a
86	*ETFDH*	1-7		n.a	exon 1-7del	multiple deletions in exon 1-7	n.a	Kim et al., 2018b	n.a
87	*ETFDH*		1	chr4:158672495G>C	c.34+5G>C	splice variant	rs1373597092	Pontoizeau et al., 2016 Henriques et al., 2019 Janeiro et al., 2019	0
88	*ETFDH*		1	chr4:158680465del	c.35-2A>C	p.Ala12_59delinsGly	n.a	Hong et al., 2019	n.a
89	*ETFDH*	2		chr4:158680468del	c.36delA	p.Ala12fs*19	n.a	[a]Frerman and Goodman, 2001 Olsen et al. 2003 Trakadis et al., 2012	n.a
90	*ETFDH*	2		n.a	n.r	p.Ala12fs	n.a	Olsen et al., 2007 Grünert, 2014 Chen et al., 2019b	n.a
91	*ETFDH*	2		chr4: between 158680476 & 158680477	c.45_46insC	p.Phe16*22	n.a	Olsen et al., 2003	n.a
92	*ETFDH*	2		n.a	n.r	p.Phe16Cys	n.a	[a]Frerman and Goodman, 2001	n.a
93	*ETFDH*	2		chr4:158680483dup	c.50dup	p.His17Glnfs*6	n.a	Pontoizeau et al., 2016	n.a

94	*ETFDH*	2		chr4:158680483dup	c.51dupT	p.Ala18Cysfs*5	n.a	[a]Frerman and Goodman, 2001 [a]Goodman et al., 2002 Olsen et al., 2003 Olsen et al., 2005 Olsen et al., 2007 Trakadis et al., 2012 Fitzgerald et al., 2013 Grünert, 2014 Chen et al., 2019b van Rijt et al., 2019 van Rijt et al., 2020	n.a
95	*ETFDH*	2		chr4:158680485C>G	c.53C>G	p.Ala18Gly	n.a	Zhao et al., 2018 Chen et al., 2019b	n.a
96	*ETFDH*	2		chr4:158680493T>C	c.61T>C	p.Ser21Pro	n.a	Zhao et al., 2018 Chen et al., 2019b	n.a
97	*ETFDH*	2		chr4:158680497A>G	c.65A>G	p.Lys22Arg	n.a	Wang et al., 2016 Chen et al., 2019b	n.a
98	*ETFDH*	2		chr4:158680499G>A	c.67G>A	p.Ala23Thr	n.a	Han et al., 2019	n.a
99	*ETFDH*	2		chr4:158680511C>T	c.79C>T	p.Pro27Ser	rs537038850	Pollard et al., 2010 Grünert, 2014 Béhin et al., 2016 Chen et al., 2019b van Rijt et al., 2019 Nilipour et al., 2020	15
100	*ETFDH*	2		chr4:158680524C>T	c.92C>T	p.Thr31Ile	rs11559290	Chokchaiwong et al., 2018 Zhao et al., 2018 Chen et al., 2019b Chokchaiwong et al., 2019 Nilipour et al., 2020	10
101	*ETFDH*	2		chr4:158680524T>C	c.92T>C	p.Ile31Thr	n.a	Law et al., 2009 Er et al. 2010 Trakadis et al., 2012	n.a
102	*ETFDH*	2		chr4:158680553C>T	c.121C>T	p.Arg41*	rs773668457	[a]Frerman and Goodman, 2001 [a]Goodman et al., 2002 Peake and Kozakewich, 2017	0
103	*ETFDH*	2		chr4:158680554G>T	c.122G>T	p.Arg41Leu	n.a	Al-Jasmi et al., 2016	n.a
104	*ETFDH*	2		chr4:158680578A>G	c.146A>G	p.Tyr49Cys	n.a	[a]Goodman et al., 2002	n.a

105	*ETFDH*	2		chr4:158680584G>A	c.152G>A	p.Arg51Gln	rs534388496	Grünert, 2014 Xi et al., 2014 Angelini et al., 2018 Zhao et al., 2018 Chen et al., 2019b	26
106	*ETFDH*	2		chr4:158680589A>G	c.158A>G	p.Lys53Arg	n.a	Olsen et al., 2014	n.a
107	*ETFDH*	2		chr4:158680603G>A	c.171G>A	p.Trp57*	rs949249162	Xu et al., 2018	0
108	*ETFDH*	2		chr4:158680604G>A	c.172G>A	p.Glu58Lys	rs1473188524	Grünert, 2014 Xi et al., 2014 Chen et al., 2019b	20
109	*ETFDH*		2	chr4:158680609T>C	c.175+2T>C	truncated	n.a	Wen et al., 2013 Grünert, 2014 Xi et al., 2014	n.a
110	*ETFDH*		2	chr4:158682194G>T	c.176-1G>T	p.Gly58fs∗3	n.a	Hong et al., 2019	n.a
111	*ETFDH*	3		chr4:158682193A>G	c.176-2A>G	p.Ile72Thr	n.a	Zhao et al., 2018 Chen et al., 2019b	n.a
112	*ETFDH*	3		chr4: between 158682194 & 158682195	c.177insT	p.Asp59fs	n.a	[a,b]Cheng et al., 2017 Chen et al., 2019b	n.a
113	*ETFDH*	3		chr4:158682210G>A	c.191G>A	p.Arg64Lys	n.a	Grünert, 2014 Xi et al., 2014 Chen et al., 2019b	n.a
114	*ETFDH*	3		chr4:158682231del	c.210_212del	p.Cys71del	n.a	Wen et al., 2013 Grünert, 2014	n.a
115	*ETFDH*	3		chr4:158682229-158682234TGTTGT>TGT	c.213_215del	p.Val72del	rs796051966	Grünert, 2014 Xi et al., 2014	0
116	*ETFDH*	3		chr4:158682245G>A	c.226G>A	p.Ala76Lys	n.a	Grünert, 2014 Xi et al., 2014	n.a
117	*ETFDH*	3		chr4:158682245G>A + 4:158682247C>A	c.226G>A+ c.227C>A	p.Ala76Lys	n.a	Grünert, 2014 Xi et al., 2014 Chen et al., 2019b	n.a
118	*ETFDH*	3		chr4:158682248G>A	c.229G>A	p.Gly77Ser	n.a	Fan et al., 2018	n.a
119	*ETFDH*	3		chr4:158682255C>G	c.236C>G	p.Ala79Gly	n.a	Wen et al., 2013 Grünert, 2014 Xi et al., 2014 Xu et al., 2018	n.a

120	*ETFDH*	3		chr4:158682402T>C	c.242T>C	p.Leu81Pro	rs1554031490	Wen et al., 2010 Trakadis et al., 2012 Wen et al., 2013 Grünert, 2014 Xi et al., 2014 Wang et al., 2016 Zhao et al., 2018 Chen et al., 2019b	31
121	*ETFDH*	3		chr4:158682263T>C	c.244T>C	p.Ser82Pro	n.a	[a,b]Ramos et al., 1995 [a]Goodman et al., 2002 Olsen et al., 2007 Trakadis et al., 2012 Grünert, 2014 Chen et al., 2019b	n.a
122	*ETFDH*	3		chr4:158682264C>T	c.245C>T	p.Ser82Phe	rs887871605	[a]Goodman et al., 2002 Whitaker et al., 2015 Béhin et al., 2016 Chen et al., 2019b	27

123	*ETFDH*	3		chr4:158682269G>A	c.250G>A	p.Ala84Thr	rs121964954	Yotsumoto et al., 2008 Law et al., 2009 Liang et al., 2009 Er et al., 2010 Lan et al., 2010 [b]Maillart et al., 2010 Wasant et al., 2010 Er et al., 2011 Wang et al., 2011a [a,c]Wang et al., 2011b Trakadis et al., 2012 Chien et al., 2013 Wen et al., 2013 [a,c]Cao et al., 2014 Grünert, 2014 [b]Lu and Ji, 2014 Xi et al., 2014 [a,c]Li, 2015 Béhin et al., 2016 [b]Dai et al., 2016 Fu et al., 2016 Liu et al., 2016 [a,b]Cui et al., 2017 Xue et al., 2017 Chokchaiwong et al., 2018 Goh et al., 2018 Zhao et al., 2018 Chen et al., 2019a Chen et al., 2019b Chokchaiwong et al., 2019 Han et al., 2019 Hong et al., 2019 Lin et al., 2019 [a,c]Zhen et al., 2019 Pan et al., 2020 Santananukarn et al., 2020	28
124	*ETFDH*	3		chr4:158682269C>T	c.251C>T	p.Ala84Val	n.a	[a,c]Wang et al., 2011b Chien et al., 2013 Wen et al., 2013 Grünert, 2014 Xi et al., 2014 Chen et al., 2019b	n.a

125	*ETFDH*	3		chr4:158682275C>T	c.256C>T	p.Arg86Cys	rs1242099513	Wen et al., 2013 Grünert, 2014 Xi et al., 2014	27
126	*ETFDH*	3		chr4:158682284-4:1586822875del	c.265_266delCA	p.Gln89Valfs∗5	n.a	Hong et al., 2018 Chen et al., 2019b Hong et al., 2019	n.a
127	*ETFDH*	3		chr4:158682314C>G	c.295C>G	p.Arg99Gly	n.a	Chien et al., 2013 Grünert, 2014 Chen et al., 2019b	n.a
128	*ETFDH*	3		chr4:158682314C>T	c.295C>T	p.Arg99Cys	rs371493232	Chien et al., 2013 Zhu et al., 2014 [a,c]Li, 2015 Wang et al., 2016 Silva et al., 2018 Zhao et al., 2018 Chen et al., 2019b Hong et al., 2019	31
129	*ETFDH*	3		chr4:158682315G>A	c.296G>A	p.Arg99His	rs376630579	-	32
130	*ETFDH*	3		chr4:158682318T>A	c.299T>A	truncated	n.a	Olsen et al., 2005	n.a
131	*ETFDH*	3		chr4:158682321G>A	c.302G>A	p.Cys101Tyr	rs1436363240	-	30
132	*ETFDH*	3		chr4:158682322T>A	c.303T>A	p.Cys101*	n.a	Zhu et al., 2014 [a,c]Li, 2015 Chen et al., 2019b Hong et al., 2019	n.a
133	*ETFDH*	3		chr4:158695620A>G	c.313A>G	p.Lys105Glu	rs398124154	Pontoizeau et al., 2016	23
134	*ETFDH*	3		chr4:158684642G>A	c.315G>A	p.Met105Ile	rs1311814302	Zhao et al., 2018 Chen et al., 2019b	0
135	*ETFDH*	3		chr4:158682353C>T	c.334C>T	p.His112Tyr	n.a	Beresford et al., 2006 Olsen et al., 2007 Trakadis et al., 2012 Grünert, 2014 Chen et al., 2019b	n.a
136	*ETFDH*	3		chr4:158682365G>C	c.346G>C	p.Gly116Arg	rs1458858994	-	31
137	*ETFDH*	3		chr4:158682368G>C	c.349G>C	p.Ala117Pro	n.a	Grünert, 2014 Xi et al., 2014 Chen et al., 2019b	n.a
138	*ETFDH*	3		chr4:158682372G>T	c.353G>T	p.Cys118Phe	rs1467428857	Grünert, 2014 Xi et al., 2014 [b]Dai et al., 2016 Chen et al., 2019b	29

139	*ETFDH*	3		chr4: between 158682380 & 158682381	c.361_362insT	p.Pro121Leufs*5	n.a	Grünert, 2014 Xi et al., 2014	n.a
140	*ETFDH*	3		chr4:158682399T>A	c.380T>A	p.Leu127His	rs121964956	Liang et al., 2009 Er et al., 2010 Trakadis et al., 2012 Chien et al., 2013 Grünert, 2014 Liu et al., 2016 Chen et al., 2019b Lin et al., 2019	27
141	*ETFDH*	3		chr4:158682399T>C	c.380T>C	p.Leu127Pro	rs1244950045	[b]Lu and Ji, 2014 Chen et al., 2019b	27
142	*ETFDH*	3		chr4:158682399T>G	c.380T>G	p.Leu127Arg	n.a	Wen et al., 2010 Trakadis et al., 2012 Grünert, 2014 Xi et al., 2014	n.a
143	*ETFDH*	3		n.a	n.r	p.Leu127Cys	n.a	Xu et al., 2018	n.a
144	*ETFDH*	3		chr4:158682402T>C	c.383T>C	p.Phe128Ser	rs1554031490	Er et al., 2011 Chien et al., 2013 Grünert, 2014 Chen et al., 2019b	31
145	*ETFDH*	3		chr4:158682408A>T	c.389A>T	p.Asp130Val	rs199899494	Wen et al., 2010 [a,c]Wang et al., 2011b Trakadis et al., 2012 Wen et al., 2013 Grünert, 2014 Xi et al., 2014 Zhu et al., 2014 [a,c]Li, 2015 Zhuo et al., 2015 [a,c]Tang et al., 2017 Chen et al., 2018a Xu et al., 2018 Zhao et al., 2018 Chen et al., 2019b Han et al., 2019 Hong et al., 2019 Xu et al., 2020	29
146	*ETFDH*	3		chr4:158682410T>G	c.391T>G	p.Trp131Gly	rs758081107	-	33

147	*ETFDH*	3		chr4:158682412G>C	c.393G>C	p.Trp131Cys	rs1292908960	Wen et al., 2010 Trakadis et al., 2012 Grünert, 2014	34
148	*ETFDH*		3	chr4:158682425G>A	c.405+1G>A	truncated	n.a	Wen et al., 2010 Trakadis et al., 2012 Grünert, 2014 Xi et al., 2014	n.a
149	*ETFDH*		3	chr4:158682425G>T	c.405+1G>T	truncated	n.a	Grünert, 2014 Xi et al., 2014 Chen et al., 2019b	n.a
150	*ETFDH*		3	chr4:158682427A>T	c.405+3A>T	splice variant or truncated	rs796051965	Olsen et al., 2004 Grünert, 2014 Henriques et al., 2019 Janeiro et al., 2019	n.a
151	*ETFDH*		3	chr4:158684590A>G	c.406-2A>G	splice variant	n.a	Béhin et al., 2016 Chen et al., 2019b	n.a
152	*ETFDH*	4		chr4:158684593C>T	c.407C>T	p.Ala136Val	rs758242572	-	33
153	*ETFDH*	4		chr4:158684595C>T	c.409C>T	p.Pro137Ser	n.a	Law et al., 2009 Trakadis et al., 2012 Grünert, 2014 Xi et al., 2014 Liu et al., 2016 Chen et al., 2019b	n.a
154	*ETFDH*	4		chr4:158684599C>T	c.412C>T	p.Leu138Phe	n.a	Angelini et al., 2018 Chen et al., 2019b	n.a
155	*ETFDH*	4		chr4:158684599T>G	c.413T>G	p.Leu138Arg	rs779896449	[a]Goodman et al., 2002 Olsen et al., 2004 Cornelius et al., 2012 Grünert, 2014 Chen et al., 2019b	28
156	*ETFDH*	4		chr4:158684613-158684619del	c.427_433del	truncated	n.a	[a]Beard et al., 1995 [a]Frerman and Goodman, 2001 [a]Goodman et al., 2002	n.a
157	*ETFDH*	4		chr4:158684619G>C	c.433G>C	p.Asp145His	n.a	Grünert, 2014 Xi et al., 2014 Chen et al., 2019b	n.a
158	*ETFDH*	4		chr4:158684635-15868439	c.449_453delTAACA	p.Leu150*	n.a	Fu et al., 2016 Chen et al., 2019b	n.a
159	*ETFDH*	4		chr4:1586846375A>G	c.451A>G	p.Thr151Ala	n.a	Angelini et al., 2018 Chen et al., 2019b	n.a

160	*ETFDH*	4		chr4:158684639del	c.453delA	p.Glu152Argfs*15	n.a	Fu et al., 2016 Chen et al., 2019b	n.a
161	*ETFDH*	4		chr4:158684649A>G	c.463A>G	p.Arg155Gly	rs549150456	Henriques et al., 2019 van Rijt et al., 2020	21
162	*ETFDH*	4		chr4:158684659T>G	c.473T>G	p.Val158Gly	n.a	Tong et al., 2018	n.a
163	*ETFDH*		4	chr4:158684664T>C	c.487-9T>C	truncated	n.a	Xi et al., 2014	n.a
164	*ETFDH*		4	chr4:158685092T>C	c.488-9T>C	truncated	rs1554031705	Wen et al., 2010 Trakadis et al., 2012 Grünert, 2014	n.a
165	*ETFDH*	5		chr4:158685111G>A	c.498G>A	p.Met166Ile	n.a	Chen et al., 2019b [a,c]Zhen et al., 2019	n.a
166	*ETFDH*	5		chr4:158685116A>G	c.503A>G	p.Asn168Ser	n.a	Wen et al., 2013 Grünert, 2014 Xi et al., 2014	n.a
167	*ETFDH*	5		chr4:158685121G>T	c.508G>T	p.Gly170Cys	rs775548362	-	32
168	*ETFDH*	5		chr4:158685124A>G	c.511A>G	p.Asn171Asp	n.a	Han et al., 2019	n.a
169	*ETFDH*	5		chr4:158685131T>G	c.518T>G	p.Ile173Met	n.a	Zhu et al., 2014 [a,c]Li, 2015 Chen et al., 2019b	n.a
170	*ETFDH*	5		chr4:158685137G>A	c.524G>A	p.Arg175His	rs121964955	Olsen et al., 2007 Yotsumoto et al., 2008 Ohkuma et al., 2009 Ishii et al., 2010 Lan et al., 2010 [b]Maillart et al., 2010 Wang et al., 2011a Trakadis et al., 2012 Wen et al., 2013 [a,c]Cao et al., 2014 Grünert, 2014 Xi et al., 2014 Whitaker et al., 2015 Béhin et al., 2016 Liu et al., 2016 Yamada et al., 2017 Fan et al., 2018 Chen et al., 2019b Navarrete et al., 2019 Santananukarn et al., 2020	33
171	*ETFDH*	5		chr4:158685137G>C	c.524G>C	p.Arg175Pro	rs121964955	-	33

172	*ETFDH*	5		chr4:158685137G>T	c.524G>T	p.Arg175Leu	rs121964955	Liang et al., 2009 Er et al., 2010 Trakadis et al., 2012 Grünert, 2014 Xi et al., 2014 Zhu et al., 2014 [a,c]Li, 2015 Chen et al., 2019b Hong et al., 2019	33
173	*ETFDH*	5		chr4:158685141G>C	c.528G>C	p.Leu176Phe	n.a	Wen et al., 2013 Xi et al., 2014 Grünert, 2014 Hong et al., 2019	n.a
174	*ETFDH*	5		chr4:158685142G>A	c.529G>A	p.Gly177Arg	rs1463795907	-	32
175	*ETFDH*	5		chr4:158685148A>G	c.535A>G	p.Lys179Glu	n.a	Zhao et al., 2018 Chen et al., 2019b	n.a
176	*ETFDH*	5		chr4:158685149T>C	c.536T>C	p.Leu179Ser	n.a	Rabenstein et al., 2020	n.a
177	*ETFDH*	5		chr4:158685155G>A	c.542G>A	p.Ser181Asn	n.a	Hong et al., 2019	n.a
178	*ETFDH*	5		chr4:158685158G>A	c.545G>A	p.Trp182*	rs767249944	[a]Goodman et al., 2002	0
179	*ETFDH*	5		chr4:158685173C>T	c.560C>T	p.Ala187Val	rs369912835	Angelini et al., 2018 Chen et al., 2019b Macchione et al., 2020 Missaglia et al., 2020	26
180	*ETFDH*	5		chr4:158685184G>A	c.571G>A	p.Gly191Ser	n.a	Béhin et al., 2016 Chen et al., 2019b	n.a
181	*ETFDH*	5		chr4:158685191A>G	c.578A>G	p.Glu193Gly	rs1287661192	-	32
182	*ETFDH*	5		chr4:158685200A>G	c.587A>G	p.Tyr196Cys	n.a	Zhao et al., 2018 Chen et al., 2019b Han et al., 2019	n.a
183	*ETFDH*		5	chr4:158685224insT	c.606+5insT	n.r	n.a	Angelini et al., 2018 Chen et al., 2019b	n.a
184	*ETFDH*		5	chr4:158685220G>A	c.606+1G>A	n.r	rs1241072742	van Rijt et al., 2019	0
185	*ETFDH*		5	chr4:158685226A>G	c.606+7A>G	truncated	rs1561240026	Wen et al., 2013 Grünert, 2014 Xi et al., 2014	0
186	*ETFDH*	6		chr4:158690358A>T	c.617A>T	p.His206Leu	n.a	Zhao et al., 2018 Chen et al., 2019b	n.a
187	*ETFDH*	6		chr4:158690363G>C	c.622G>C	p.Asp208His	n.a	Béhin et al., 2016 Chen et al., 2019b Madsen et al., 2019	n.a

188	*ETFDH*	6		chr4:158690367del	c.623_626del	p.Asp208Valfs*3	n.a	Ou et al., 2020	n.a
189	*ETFDH*	6		chr4:158690384G>A	c.643G>A	p.Ala215Thr	rs755214552	Wang et al., 2011a [a,c]Wang et al., 2011b Grünert, 2014 Liu et al., 2016 Chen et al., 2019b	28
190	*ETFDH*	6		chr4:158690393G>A	c.652G>A	p.Asp218Asn	rs748289922	[a]Goodman et al., 2002 Angle and Burton, 2008 Grünert, 2014 Chen et al., 2019b	25
191	*ETFDH*	6		chr4:158690406A>C	c.665A>C	p.Gln222Pro	rs1482632936	[a]Goodman et al., 2002 van Rijt et al., 2020	23
192	*ETFDH*	6		chr4:158690420C>T	c.679C>T	p.Pro227Ser	rs141407224	Nilipour et al., 2020	25
193	*ETFDH*		6	chr4:158690426G>A	c.684+1G>A	n.r	n.a	[a,c]Wang et al., 2011b Chen et al., 2019b	n.a
194	*ETFDH*		6	chr4:158690426G>T	c.684+1G>T	splice variant	n.a	Chen et al., 2019b Xu et al., 2019	n.a
195	*ETFDH*	7		chr4:158695503T>A	c.691T>A	p.Phe231Ile	n.a	Zhao et al., 2018 Chen et al., 2019b	n.a
196	*ETFDH*	7		chr4:158695504T>C	c.692T>C	p.Phe231Ser	n.a	Grünert, 2014 Xi et al., 2014	n.a
197	*ETFDH*	7		chr4:158695512G>A	c.700G>A	p.Gly234Arg	rs1032041993	-	32
198	*ETFDH*	7		chr4:158695527G>A	c.715G>A	p.Ala239Thr	n.a	Wen et al., 2010 Trakadis et al., 2012 Grünert, 2014 Xi et al., 2014 Zhu et al., 2014 [a,c]Li, 2015 Chen et al., 2019b	n.a
199	*ETFDH*	7		chr4:158695540T>C	c.728T>C	p.Ile243Thr	rs1397900640	Köppel et al., 2006 Grünert, 2014 Béhin et al., 2016 Chen et al., 2019b	24
200	*ETFDH*	7		chr4:158697599T>C	c.731T>C	p.Phe244Ser	rs755907131	Angle and Burton, 2008 Grünert, 2014 Chen et al., 2019b	24
201	*ETFDH*	7		chr4:158695548G>A	c.736G>A	p.Glu246Lys	n.a	Zhuo et al., 2015 Hong et al., 2018 Chen et al., 2019b Hong et al., 2019	n.a

202	*ETFDH*	7		chr4:158695551G>C	c.739G>C	p.Gly247Arg	rs758218892	-	32
203	*ETFDH*	7		chr4:158695552G>T	c.740G>T	p.Gly247Val	rs1384574225	Hong et al., 2019	29
204	*ETFDH*	7		chr4:158697637T>C	c.769T>C	p.Tyr257His	rs1376613646	Béhin et al., 2016 Chen et al., 2019b	28
205	*ETFDH*	7		chr4:158697638A>G	c.770A>G	p.Tyr257Cys	rs776108871	Wen et al., 2010 Wang et al., 2011a [a,c]Wang et al., 2011b Trakadis et al., 2012 Wen et al., 2013 Grünert, 2014 Xi et al., 2014 [a,c]Zhao, 2014 Zhu et al., 2014 [a,c]Li, 2015 Peng et al., 2015 Liu et al., 2016 Wang et al., 2016 [a,c]Chen et al., 2018b Goh et al., 2018 [a,c]Hu et al., 2018 [a,c]Kong et al., 2018 Xu et al., 2018 Zhao et al., 2018 Chen et al., 2019b Han et al., 2019 Hong et al., 2019 Hu and Li, 2019 Lin et al., 2019 Nilipour et al., 2020	29
206	*ETFDH*	7		chr4:158695598G>T	c.786G>T	p.Leu262Phe	n.a	[a]Goodman et al., 2002	n.a
207	*ETFDH*	7		chr4:158695618A>T	c.806A>T	p.Gln269Leu	rs776320810	Olsen et al., 2003 Olsen et al., 2005 Olsen et al., 2007 Trakadis et al., 2012 Grünert, 2014 Chen et al., 2019b	26
208	*ETFDH*	7		chr4:158695619A>C	c.807A>C	p.Gly269His	rs1265381182	Al-Jasmi et al., 2016	23
209	*ETFDH*	7		chr4:158695624A>G	c.812A>G	p.Tyr271Cys	n.a	Tong et al., 2018	n.a

210	*ETFDH*	7		chr4:158695626G>A	c.814G>A	p.Gly272Arg	rs763541530	Angle and Burton, 2008 Grünert, 2014 Chen et al., 2018a Chen et al., 2019b Gonzalez-Perez et al., 2019 Nilipour et al., 2020	32
211	*ETFDH*	7		chr4:158695632G>T	c.820G>T	p.Gly274*	n.a	Wolfe et al., 2010 Trakadis et al., 2012 Grünert, 2014 Chen et al., 2019b van Rijt et al., 2020	n.a
212	*ETFDH*	7		chr4:158695633G>A	c.821G>A	p.Gly274Glu	n.a	[a,c]Wang et al., 2011b Zhu et al., 2014 [a,c]Li, 2015 Chen et al., 2019b	n.a
213	*ETFDH*	7		chr4:158695641G>A	c.829G>A	p.Glu277Lys	rs1554032712	-	34
214	*ETFDH*	7-8		n.a	exon 7-8del	deletion of exon 7-8	n.a	Kim et al., 2018b	n.a
215	*ETFDH*		7	chr4:158695645T>C	c.831+2T>C	n.r	rs150832878	[a,c]Wang et al., 2011b Chen et al., 2019b	n.a
216	*ETFDH*		7	chr4:158695646A>C	c.831+3A>C	n.r	n.a	Kim et al., 2018b Chen et al., 2019b	n.a
217	*ETFDH*		7	chr4:158697558G>A	c.832-1G>A	exon 8 skipping	n.a	Wasant et al., 2010 Grünert, 2014 Chen et al., 2019b	n.a
218	*ETFDH*	8		chr4:158697562T>C	c.835T>C	p.Trp279Arg	rs754082348	Wen et al., 2013 Grünert, 2014 Xi et al., 2014	33
219	*ETFDH*	8		chr4:158697563G>T	c.836G>T	p.Trp279Leu	rs1377478090	-	33
220	*ETFDH*	8		chr4:158697564G>T	c.837G>T	p.Trp279Cys	rs1230343117	-	34
221	*ETFDH*	8		chr4:158697585G>A	c.858G>A	p.Trp286*	rs1235904433	van Rijt et al., 2020	0
222	*ETFDH*	8		chr4:158697599T>G	c.872T>G	p.Val291Gly	n.a	Wen et al., 2010 Trakadis et al., 2012 Wen et al., 2013 Grünert, 2014 Xi et al., 2014 Xu et al., 2018 Zhao et al., 2018 Chen et al., 2019b Pan et al., 2020	n.a

223	*ETFDH*	8		chr4:158697604C>G	c.877C>G	p.His293Asp	n.a	[a]Goodman et al., 2002 [b]Maillart et al., 2010 Kaminsky et al., 2011 Grünert, 2014 Béhin et al., 2016 Chen et al., 2019b	n.a
224	*ETFDH*	8		chr4:158697608C>T	c.881C>T	p.Thr294Ile	n.a	Köppel et al., 2006 Grünert, 2014 Chen et al., 2019b van Rijt et al., 2019	n.a
225	*ETFDH*	8		chr4:158697617G>T	c.890G>T	p.Trp297Leu	n.a	Shioya et al., 2014 Yamada et al., 2016 Yamada et al., 2017 Chen et al., 2019b	n.a
226	*ETFDH*	8		chr4:158697619C>T	c.892C>T	p.Pro298Ser	n.a	Fu et al., 2016 Chen et al., 2019b	n.a
227	*ETFDH*	8		chr4:158697623T>C	c.896T>C	p.Leu299Ser	n.a	van Rijt et al., 2020	n.a
228	*ETFDH*	8		chr4:158697643G>A	c.916G>A	p.Gly306Arg	rs761008371	-	32
229	*ETFDH*	8		chr4:158697647C>G	c.920C>G	p.Ser307Cys	n.a	Peng et al., 2015 Wang et al., 2016 Xue et al., 2017 Wang et al., 2018 Chen et al., 2019b Hong et al., 2019	n.a
230	*ETFDH*	8		chr4:158697649T>G	c.922T>G	p.Phe308Val	rs1218671512	Yotsumoto et al., 2008 Trakadis et al., 2012 Grünert, 2014 Chen et al., 2019b	29
231	*ETFDH*	8		chr4:158697653T>C	c.926T>C	p.Leu309Pro	rs1276785426	-	32
232	*ETFDH*	8		chr4:158697653T>G	c.926T>G	p.Leu309Arg	rs1276785426	-	31
233	*ETFDH*	8		chr4:158697656A>G	c.929A>G	p.Tyr310Cys	rs796051958	-	32
234	*ETFDH*	8		chr4:158697667G>A	c.940G>A	p.Glu314Lys	n.a	van Rijt et al., 2020	n.a
235	*ETFDH*	8		chr4:158697676C>A	c.949C>A	p.Pro317Thr	n.a	Grünert, 2014 Xi et al., 2014 Chen et al., 2019b	n.a
236	*ETFDH*	8		chr4:158697677C>G	c.950C>G	p.Pro317Arg	n.a	Shioya et al., 2014 Yamada et al., 2016 Yamada et al., 2017 Chen et al., 2019b	n.a
237	*ETFDH*	8		chr4:158697680T>C	c.953T>C	p.Leu318Pro	n.a	Tong et al., 2018	n.a

238	*ETFDH*	8		chr4:158697685G>C	c.958G>C	p.Ala320Pro	rs752675985	-	33
239	*ETFDH*	8		chr4:158697686C>T	c.959C>T	p.Ala320Val	rs751725601	Chen et al., 2019a Chen et al., 2019b	25
240	*ETFDH*	8		chr4:158697691G>A	c.964G>A	p.Gly322Ser	rs867300806	-	32
241	*ETFDH*	8		chr4:158697697G>T	c.970G>T	p.Val324Leu	n.a	Xu et al., 2020	n.a
242	*ETFDH*	9		chr4:158698987G>T	c.973G>T	p.Val325Phe	rs549454407	-	34
243	*ETFDH*	9		chr4:158698675del	c.973del312	p.Val325del48	n.a	Wen et al., 2010 Trakadis et al., 2012 Grünert, 2014 Xi et al., 2014	n.a
244	*ETFDH*	9		chr4:158698990G>C	c.976G>C	p.Gly326Arg	n.a	Grünert, 2014 Xi et al., 2014 Chen et al., 2019b	n.a
245	*ETFDH*	9		chr4:158698999T>C	c.985T>C	p.Tyr329His	rs750084469	-	31
246	*ETFDH*	9		chr4:158699000A>G	c.986A>G	p.Tyr329Cys	rs1561247687	-	32
247	*ETFDH*	9		chr4:158699007T>G	c.993T>G	p.Asn331Lys	n.a	Wen et al., 2010 [a,c]Wang et al., 2011b Trakadis et al., 2012 Xi et al., 2014 Chen et al., 2019b Hu and Li, 2019	n.a
248	*ETFDH*	9		chr4:158699012A>G	c.998A>G	p.Tyr333Cys	rs781531865	Wang et al., 2011a [a,c]Wang et al., 2011b Grünert, 2014 Xi et al., 2014 Liu et al., 2016 Chen et al., 2019b Lin et al., 2019	23
249	*ETFDH*	9		chr4:158699015T>C	c.1001T>C	p.Leu334Pro	rs377686388	[a]Frerman and Goodman, 2001 [a]Goodman et al., 2002 Olsen et al., 2007 Trakadis et al., 2012 Grünert, 2014 Chen et al., 2019b van Rijt et al., 2020	30
250	*ETFDH*	9		chr4:158699040G>T	c.1026G>T	p.Arg342Ser	n.a	Grünert, 2014 Xi et al., 2014	n.a

251	*ETFDH*	9		chr4:158699041T>C	c.1027T>C	p.Trp343Arg	n.a	Wen et al., 2013 Grünert, 2014 Xi et al., 2014 Angelini et al., 2018 Chen et al., 2019b Hong et al., 2019 Missaglia et al., 2020	n.a
252	*ETFDH*	9		chr4:158699042T>C	c.1028T>C	p.Met343Thr	n.a	Zhao et al., 2018 Chen et al., 2019b	n.a
253	*ETFDH*	9		chr4:158699051A>G	c.1037A>G	p.His346Arg	rs1358691961	[a]Goodman et al., 2002	27
254	*ETFDH*	9		chr4:158699058A>C	c.1044A>C	p.Leu348Phe	n.a	Zhao et al., 2018 Chen et al., 2019b	n.a
255	*ETFDH*	9		chr4:158699063G>A	c.1049G>A	p.Arg350Gln	rs139306043	Er et al. 2010 Trakadis et al., 2012	8
256	*ETFDH*	9		chr4:158699072T>C	c.1058T>C	p.Leu353Ser	rs1286771338	-	32
257	*ETFDH*	9		chr4:158699074G>T	c.1060G>T	p.Glu354*	n.a	Olsen et al., 2007 Trakadis et al., 2012 Grünert, 2014 Chen et al., 2019b	n.a
258	*ETFDH*	9		chr4:158699077G>A	c.1063G>A	p.Gly355Ser	rs1361421769	-	31
259	*ETFDH*	9		chr4:158699081G>A	c.1067G>A	p.Gly356Glu	n.a	van der Westhuizen et al., 2018 Chen et al., 2019b Schoonen et al., 2019	n.a
260	*ETFDH*	9		chr4:158699087G>A	c.1073G>A	p.Arg358Lys	rs796051959	Creanza et al., 2018	26
261	*ETFDH*	9		chr4:158699088G>C	c.1074G>C	p.Arg358Ser	n.a	Olsen et al., 2003 Olsen et al., 2005 Trakadis et al., 2012 Grünert, 2014 Creanza et al., 2018 Chen et al., 2019b van Rijt et al., 2020	n.a
262	*ETFDH*	9		chr4:158699092G>C	c.1078G>C	p.Ala360Pro	n.a	Yamada et al., 2017	n.a

263	*ETFDH*	9		chr4:158699098G>A	c.1084G>A	p.Gly362Arg	rs369711837	Yotsumoto et al., 2008 Wen et al., 2010 Trakadis et al., 2012 Grünert, 2014 Xi et al., 2014 Zhu et al., 2014 [a,c]Li, 2015 Yamada et al., 2017 Chen et al., 2019b	32
264	*ETFDH*	9		chr4:158699110C>T	c.1096C>T	p.Leu366Phe	n.a	Yotsumoto et al., 2008 Trakadis et al., 2012 Grünert, 2014 Chen et al., 2019b	n.a
265	*ETFDH*	9		chr4:158699113A>G	c.1099A>G	p.Asn367Asp	n.a	Grünert, 2014 Xi et al., 2014 van Rijt et al., 2020	n.a
266	*ETFDH*	9		chr4:158699120G>A	c.1106G>A	p.Gly369Asp	rs754418186	Janeiro et al., 2019	32
267	*ETFDH*	9		chr4:158699120G>C	c.1106G>C	p.Gly369Ala	rs754418186	Gautschi et al., 2015 van Rijt et al., 2020	31
268	*ETFDH*		9	chr4:158703421A>G	c.1117-2A>G	splice variant	rs764060500	Hackl et al., 2017 Chen et al., 2019b	0
269	*ETFDH*	10		chr4:158703422C>T	c.1118C>T	p.Ser373Phe	n.a	van Rijt et al., 2019	n.a
270	*ETFDH*	10		chr4:158703429C>A	c.1123C>A	p.Pro375Thr	n.a	Zhao et al., 2018 Chen et al., 2019b	n.a
271	*ETFDH*	10		chr4:158703436T>C	c.1130T>C	p.Leu377Pro	rs387907170	Gempel et al., 2007 Trakadis et al., 2012 Grünert, 2014 Işıkay et al., 2017 Yiş et al., 2018 Chen et al., 2019b van Rijt et al., 2019 Yıldız et al., 2019 Nilipour et al., 2020	27

272	*ETFDH*	10		chr4:158703447G>C	c.1141G>C	p.Gly381Arg	rs1466787789	van Hove et al., 2003 Grünert, 2014 Vieira et al., 2017 Chen et al., 2019b Fischer et al., 2019a Fischer et al., 2019b Yıldız et al., 2019 van Rijt et al., 2020	33
273	*ETFDH*	10		n.a	n.r	p.Leu384Ile	n.a	Xu et al., 2018	n.a
274	*ETFDH*	10		chr4:158703465G>A	c.1157G>A	p.Gly386Asp	n.a	Fan et al., 2018 Chen et al., 2019b	n.a
275	*ETFDH*	10		chr4:158703471C>A	c.1165C>A	p.Pro389Thr	n.a	Chen et al., 2019b Yıldız et al., 2019	n.a
276	*ETFDH*	10		chr4:158703507del	c.1198_1201delACTC	p.His401Glnfs*3	n.a	Işıkay et al., 2017 Chen et al., 2019b Yıldız et al., 2019	n.a
277	*ETFDH*	10		chr4:158703510A>G	c.1204A>G	p.Thr402Ala	rs754240915	Gonzalez-Perez et al., 2019	23
278	*ETFDH*	10		chr4:158703510A>T	c.1204A>T	p.Thr402Ser	n.a	Chen et al., 2019b Xu et al., 2019	n.a
279	*ETFDH*	10		chr4:158703511C>T	c.1205C>T	p.Thr402Ile	n.a	Grünert, 2014 Xi et al., 2014	n.a
280	*ETFDH*	10		chr4:158703514C>T	c.1208C>T	p.Ala403Val	n.a	Yotsumoto et al., 2008 Ohkuma et al., 2009 Trakadis et al., 2012 Grünert, 2014 Chen et al., 2019b	n.a
281	*ETFDH*	10		chr4:158703517T>C	c.1211T>C	p.Met404Thr	rs779253471	Wen et al., 2010 Izumi et al., 2011 Trakadis et al., 2012 Grünert, 2014 Xi et al., 2014 Zhu et al., 2014 [a,c]Li, 2015 Wen et al., 2015 Hong et al., 2018 Xu et al., 2018 Zhao et al., 2018 Chen et al., 2019b Han et al., 2019 Hong et al., 2019	27

282	*ETFDH*	10		chr4:158703523G>A	c.1217G>A	p.Ser406Asn	n.a	Yamaguchi et al., 2012 Yamada et al., 2017	n.a
283	*ETFDH*	10		chr4:158703533A>C	c.1227A>C	p.Leu409Phe	rs1200031596	Wen et al., 2010 [a,c]Wang et al., 2011b Trakadis et al., 2012 Wen et al., 2013 Grünert, 2014 Xi et al., 2014 Zhu et al., 2014 [a,c]Li, 2015 Wang et al., 2016 Zhao et al., 2018 Chen et al., 2019b Han et al., 2019 Hong et al., 2019 Nilipour et al., 2020	24
284	*ETFDH*	10		chr4:158703534G>A	c.1228G>A	p.Ala410Thr	rs927133458	-	31
285	*ETFDH*	10		chr4:158703540G>T	c.1234G>T	p.Glu412*	rs398124151	van Rijt et al., 2020	0
286	*ETFDH*	10		chr4:158703552del	c.1241_1246del	p.Ile414_Phe415del	n.a	Béhin et al., 2016 Chen et al., 2019b Madsen et al., 2019	n.a
287	*ETFDH*	10		chr4:158703550T>C	c.1244T>C	p.Phe415Ser	rs1050622882	-	33
288	*ETFDH*	10		chr4:158703563del	c.1254_1257delAACT	p.Leu418Thrfs*10	n.a	Wang et al., 2011a Grünert, 2014 Chen et al., 2019b	n.a
289	*ETFDH*	10		chr4:158703579del	c.1270_1273del	n.r	n.a	[a,c]Kong et al., 2018 Chen et al., 2019b	n.a
290	*ETFDH*	10		chr4:158703577C>G	c.1271C>G	p.Thr424Ser	n.a	Zhao et al., 2018 Chen et al., 2019b	n.a
291	*ETFDH*	10		chr4:158703587-158703588AA>-	c.1281_1282del	p.Ile428Argfs*6	rs1233725939	Grünert, 2014 Xi et al., 2014 [a,c]Hu et al., 2018 Zhao et al., 2018 Chen et al., 2019b Han et al., 2019 Hu and Li, 2019	0
292	*ETFDH*	10		chr4:158703591G>C	c.1285G>C	p.Gly429Arg	rs759044284	Olsen et al., 2007 Cornelius et al., 2012 Grünert, 2014 Chen et al., 2019b	28

293	*ETFDH*		10	chr4:158703592G>A	c.1285+1G>A	p.Gly429Aspfs21*	rs767046886	Missaglia et al., 2018 Macchione et al., 2020 Missaglia et al., 2020	0
294	*ETFDH*		10	chr4:158703593T>C	c.1285+2T>C	splice variant	n.a	Han et al., 2019	n.a
295	*ETFDH*	11		chr4:158703594G>A	c.1286G>A	p.Gly429Glu	n.a	Kim et al., 2018a	n.a
296	*ETFDH*	11		chr4:158706198T>A	c.1295T>A	p.Val432Gly	n.a	Wen et al., 2013 Grünert, 2014 Xi et al., 2014	n.a
297	*ETFDH*	11		chr4:158706206T>C	c.1303T>C	p.Tyr435His	rs1057519158	-	32
298	*ETFDH*	11		chr4:158706234T>C	c.1331T>C	p.Val444Ala	rs760234838	Béhin et al., 2016 [a,b]Cheng et al., 2017 Chen et al., 2019b	23
299	*ETFDH*	11		chr4:158706236T>C	c.1333T>C	p.Trp445Arg	rs1156517377	-	32
300	*ETFDH*	11		chr4:158706254G>A	c.1351G>A	p.Gly451Arg	n.a	Han et al., 2019	n.a
301	*ETFDH*	11		chr4:158706254G>C	c.1351G>C	p.Val451Leu	rs558005496	Olsen et al., 2007 Trakadis et al., 2012 Grünert, 2014 Chen et al., 2019b van Rijt et al., 2019	24
302	*ETFDH*	11		chr4:158706257A>G	c.1354A>G	p.Arg452Gly	n.a	Kim et al., 2018b Chen et al., 2019b	n.a
303	*ETFDH*	11		chr4:158706258del	c.1355delG	p.Arg452Lysfs*3	n.a	Law et al., 2009 Trakadis et al., 2012 Grünert, 2014 Chen et al., 2019b	n.a
304	*ETFDH*	11		chr4:158706258G>A	c.1355G>A	p.Arg452Lys	n.a	[a]Goodman et al., 2002	n.a
305	*ETFDH*	11		chr4:158706269C>A	c.1366C>A	p.Pro456Thr	n.a	Beresford et al., 2006 Olsen et al., 2007 Trakadis et al., 2012 Grünert, 2014 Chen et al., 2019b	n.a
306	*ETFDH*	11		chr4:158706269C>T	c.1366C>T	p.Pro456Ser	rs751821289	Béhin et al., 2016 Touat et al., 2018 Chen et al., 2019b Rabenstein et al., 2020	25
307	*ETFDH*	11		chr4:158706270C>A	c.1367C>A	p.Pro456Gln	n.a	Nilipour et al., 2020	n.a

308	*ETFDH*	11		chr4:158706270C>T	c.1367C>T	p.Pro456Leu	rs398124152	[a]Goodman et al., 2002 [a]Henderson et al., 2002 Gempel et al., 2007 Olsen et al., 2007 Gregersen et al., 2008 Yotsumoto et al., 2008 Cornelius et al., 2012 Trakadis et al., 2012 Fitzgerald et al., 2013 Grünert, 2014 Yamada et al., 2016 Chen et al., 2019b van Rijt et al., 2020	25
309	*ETFDH*	11		chr4:158706271C>T	c.1368C>T	p.Pro456Leu	n.a	[a]Goodman et al., 2002 Yotsumoto et al., 2008	n.a
310	*ETFDH*	11		chr4:158706275-158706278del	c.1372_1375del	p.Cys458Thrfs*10	n.a	Wen et al., 2013 Grünert, 2014 Xi et al., 2014	n.a
311	*ETFDH*	11		chr4:158706281G>T	c.1378G>T	p.Gly460*	n.a	Grünert, 2014 Xi et al., 2014 Chen et al., 2019b	n.a
312	*ETFDH*	11		chr4:158706287C>G	c.1384C>G	p.Leu462Val	rs1438551187	Gorukmez et al., 2015 Chen et al., 2019b	19
313	*ETFDH*	11		chr4:158706290G>A	c.1387G>A	p.Gly463Ser	rs1561251385	-	32
314	*ETFDH*	11		chr4:158706291G>A	c.1388G>A	p.Gly463Asp	rs1561251388	-	31
315	*ETFDH*	11		chr4:158706295-158706296del	c.1392_1393del	truncated	n.a	[a]Frerman and Goodman, 2001 [a]Goodman et al., 2002	n.a
316	*ETFDH*	11		chr4:158706298T>G	c.1395T>G	p.Tyr465*	rs745714733	Wen et al., 2010 Wang et al., 2011a Trakadis et al., 2012 Wen et al., 2013 Grünert, 2014 Xi et al., 2014 Zhu et al., 2014 [a,c]Li, 2015 Liu et al., 2016 Zhao et al., 2018 Chen et al., 2019b Han et al., 2019 Hong et al., 2019	0

317	*ETFDH*	11		chr4:158706298dup	c.1395dupT	p.Gly466Trp	n.a	[a,c]Chen et al., 2018b Chen et al., 2019b	n.a
318	*ETFDH*	11		chr4:158706302G>A	c.1399G>A	p.Gly467Arg	n.a	Zhu et al., 2014 [a,c]Li, 2015 Chen et al., 2019b	n.a
319	*ETFDH*	11		chr4:158706302G>C	c.1399G>C	p.Gly467Arg	rs1174882036	Wen et al., 2010 Trakadis et al., 2012 Grünert, 2014 Xi et al., 2014 Hong et al., 2019 Lin et al., 2019 Ou et al., 2020	33
320	*ETFDH*	11		chr4:158706303G>C	c.1400G>C	p.Gly467Arg	n.a	Law et al., 2009 Trakadis et al., 2012 Grünert, 2014 Chen et al., 2019b	n.a
321	*ETFDH*	11		chr4:158706317G>A	c.1414G>A	p.Gly472Arg	rs746598421	Olsen et al. 2003 Cornelius et al., 2012 Trakadis et al., 2012 Al-Jasmi et al., 2016 van Rijt et al., 2019	33
322	*ETFDH*	11		chr4:158706318G>T	c.1415G>T	p.Gly472Val	rs768181815	-	32
323	*ETFDH*	11		chr4:158706328C>A	c.1425C>A	p.Tyr475*	n.a	Olsen et al., 2005	n.a
324	*ETFDH*	11		chr4:158706339G>C	c.1436G>C	p.Arg479Thr	n.a	Trakadis et al., 2012 Wen et al., 2010 Grünert, 2014 Xi et al., 2014	n.a
325	*ETFDH*	11		chr4:158706348A>T	c.1445A>T	p.Glu482Val	n.a	Olsen et al., 2007 Trakadis et al., 2012 Grünert, 2014 Chen et al., 2019b	n.a

326	*ETFDH*	11		chr4:158706351C>T	c.1448C>T	p.Pro483Leu	rs377656387	Gempel et al., 2007 Olsen et al., 2007 Gregersen et al., 2008 Cornelius et al., 2012 Trakadis et al., 2012 Grünert, 2014 Xi et al., 2014 van der Westhuizen et al., 2018 Chen et al., 2019b Dernoncourt et al., 2019 Schoonen et al., 2019 Yıldız et al., 2019	27
327	*ETFDH*	11		chr4:158706353T>C	c.1450T>C	p.Trp484Arg	n.a	[a,c]Wang et al., 2011b Wen et al., 2015 Wang et al., 2016 Fan et al., 2018 Xu et al., 2018 Chen et al., 2019b Hong et al., 2019	n.a
328	*ETFDH*	11		chr4:158706357C>G	c.1454C>G	p.Thr485Ser	n.a	Grünert, 2014 Xi et al., 2014 Zhao et al., 2018 Chen et al., 2019b Hong et al., 2019	n.a
329	*ETFDH*	11		chr4:158706360T>C	c.1457T>C	p.Leu486Pro	rs751839607	-	33
330	*ETFDH*		11	chr4:158706372del	c.1468+1delG	n.r	n.a	Gorukmez et al., 2015 Chen et al., 2019b	n.a
331	*ETFDH*		11	chr4:158706373T>G	c.1468+2T>G	p.Gly490fs∗15	n.a	Hong et al., 2019	n.a
332	*ETFDH*	12		chr4:158706630-158706632	c.1471_1473delins(8)	p.Ser491Glnfs*3	n.a	Béhin et al., 2016 Chen et al., 2019b	n.a
333	*ETFDH*	12		chr4:158706635A>C	c.1475A>C	p.Asp492Ala	rs763487621	-	32
334	*ETFDH*	12		chr4:158706647T>C	c.1487T>C	p.Leu496Pro	rs863224869	-	33
335	*ETFDH*	12		chr4:158706666del	c.1506delC	p.Cys502*	n.a	Kim et al., 2018a	n.a
336	*ETFDH*	12		chr4:158706679T>C	c.1519T>C	p.Tyr507His	rs779944719	-	31
337	*ETFDH*	12		chr4:158706679T>G	c.1519T>G	p.Tyr507Asp	rs779944719	Ohkuma et al., 2009 Trakadis et al., 2012 Grünert, 2014 Yamada et al., 2017 Chen et al., 2019b	32

338	*ETFDH*	12		chr4:158706682C>A	c.1522C>A	p.Pro508Thr	rs751394068	Zhao et al., 2012 Grünert, 2014 Chen et al., 2019b	26
339	*ETFDH*	12		chr4:158706684del	c.1524delA	p.Lys509Asnfs*16	n.a	Chen et al., 2019b Yıldız et al., 2019	n.a
340	*ETFDH*	12		chr4:158706688C>T	c.1528C>T	p.Pro510Ser	n.a	Wen et al., 2013 Grünert, 2014 Xi et al., 2014	n.a
341	*ETFDH*	12		chr4:158706691G>A	c.1531G>A	p.Asp511Asn	rs780768015	Wen et al., 2010 Sugai et al., 2012 Trakadis et al., 2012 Wen et al., 2013 Grünert, 2014 Xi et al., 2014 Angelini et al., 2018 Chen et al., 2019b Han et al., 2019 Macchione et al., 2020	33
342	*ETFDH*	12		chr4:158706694G>A	c.1534G>A	p.Gly512Arg	n.a	Han et al., 2019	n.a
343	*ETFDH*	12		chr4:158706704G>T	c.1544G>T	p.Ser515Ile	n.a	Grünert, 2014 Rosenbohm et al., 2014 Chen et al., 2019b	n.a
344	*ETFDH*	12		chr4:158706708T>C	c.1547T>C	p.Phe516Ser	n.a	Silva et al., 2018	n.a
345	*ETFDH*	12		chr4:158706710A>C	c.1550A>C	p.Asp517Ala	rs777302338	-	31
346	*ETFDH*	12		chr4:158706712C>G	c.1552C>G	p.Leu518Val	n.a	Han et al., 2019	n.a
347	*ETFDH*	12		chr4:158706716T>G	c.1556T>G	p.Leu519Trp	rs1270988246	-	31
348	*ETFDH*	12		chr4: between 158706743 & 158706744	c.1583_1584insA	p.Asn528Lysfs*3	n.a	Zhao et al., 2012 Grünert, 2014 Chen et al., 2019b	n.a
349	*ETFDH*	12		chr4:158706748A>G	c.1586A>G	p.His529Arg	n.a	Zhu et al., 2014 [a,c]Li, 2015 Chen et al., 2019b Hong et al., 2019	n.a
350	*ETFDH*	12		chr4:158706755A>C	c.1595A>C	p.Asp532Ala	rs766367908	-	31
351	*ETFDH*	12		chr4:158706757C>T	c.1597C>T	p.Gln533*	n.a	Zhao et al., 2018 Chen et al., 2019b	n.a

352	*ETFDH*	12		chr4:158706761C>T	c.1601C>T	p.Pro534Leu	rs200920510	Yotsumoto et al., 2008 Wolfe et al., 2010 Trakadis et al., 2012 Grünert, 2014 Liu et al., 2016 [a,c]Tong et al., 2017 Yamada et al., 2017 Tong et al., 2018 Chen et al., 2019b Dernoncourt et al., 2019 Henriques et al., 2019 Janeiro et al., 2019 van Rijt et al., 2020	27
353	*ETFDH*	12		chr4:158706783del	c.1623delT	truncated	n.a	[a]Frerman and Goodman, 2001 [a]Goodman et al., 2002	n.a
354	*ETFDH*	12		chr4:158706806del	c.1647_1648delCT	n.r	n.a	Peake and Kozakewich, 2017	n.a
355	*ETFDH*	12		chr4:158706809T>G	c.1649T>G	p.Leu550Pro	n.a	Angelini, 2015 Angelini et al., 2018 Chen et al., 2019b	n.a
356	*ETFDH*	12		chr4:158706817T>C	c.1657T>C	p.Tyr553His	rs182913453	Grünert, 2014 Xi et al., 2014 Lin et al., 2019	27
357	*ETFDH*	12		chr4:158706829G>A	c.1669G>A	p.Glu557Lys	rs1279728239	[a,c]Zhao , 2014 Chen et al., 2019b	33
358	*ETFDH*	12		chr4:158708489C>T	c.1675C>T	p.Arg559*	rs1292587000	Yotsumoto et al., 2008 [a,c]Wang et al., 2011b Trakadis et al., 2012 Yamaguchi et al., 2012 Grünert, 2014 Yamada et al., 2017 Chen et al., 2019b	26
359	*ETFDH*	12		chr4:158706836G>A	c.1676G>A	p.Arg559Gln	rs768961719	-	34
360	*ETFDH*	12		chr4:158706836G>C	c.1676G>C	p.Arg559Pro	rs768961719	-	34
361	*ETFDH*	12		chr4:158706841T>C	c.1681T>C	p.Cys561Arg	rs1561251922	-	32
362	*ETFDH*	12		chr4:158706842G>T	c.1682G>T	p.Cys561Phe	rs774278013	-	33
363	*ETFDH*	12		chr4:158706845C>T	c.1685C>T	p.Pro562Leu	rs993314323	[a]Frerman and Goodman, 2001 [a]Goodman et al., 2002	27
364	*ETFDH*		12	chr4:158706851G>T	c.1690+1G>T	p.Arg559*	rs917285990	[a]Beard et al., 1995 [a]Frerman and Goodman, 2001 [a]Goodman et al., 2002	n.a

365	*ETFDH*		12	chr4:158708361C>G	c.1691-3C>G	splice variant or absence of transcript	rs776853600	[b]Maillart et al., 2010 Wasant et al., 2010 Wen et al., 2010 Trakadis et al., 2012 Grünert, 2014 Xi et al., 2014 Béhin et al., 2016 [a,b]Cui et al., 2017 Chen et al., 2019b Hong et al., 2019	n.a
366	*ETFDH*	13		chr4:158708366G>C	c.1693G>C	p.Val565Leu	rs769893690	van Rijt et al., 2020	28
367	*ETFDH*	13		chr4:158708372G>A	c.1699G>A	p.Glu567Lys	n.a	Nilipour et al., 2020	n.a
368	*ETFDH*	13		chr4:158708376T>G	c.1703T>G	p.Phe568Ser	n.a	[a,c]Wang et al., 2011b Chen et al., 2019b	n.a
369	*ETFDH*	13		chr4:158708388A>T	c.1715A>T	p.Glu572Val	rs1169887640	-	32
370	*ETFDH*	13		chr4:158708390C>T	c.1717C>T	p.Gln573*	n.a	[a,c]Wang et al., 2011b Chen et al., 2019b	n.a
371	*ETFDH*	13		chr4:158708399G>A	c.1726G>A	p.Gly576Arg	rs760578708	-	32
372	*ETFDH*	13		chr4:158708405C>T	c.1732C>T	p.Arg578Trp	rs763912783	Béhin et al., 2016 Chen et al., 2019b	24
373	*ETFDH*	13		chr4:158708417A>T	c.1744A>T	p.Asn582Tyr	n.a	Grünert, 2014 Xi et al., 2014 Chen et al., 2019b	n.a
374	*ETFDH*	13		chr4:158708420G>A	c.1747G>A	p.Ala583Thr	rs780666554	-	32
375	*ETFDH*	13		chr4:158708436A>G	c.1763A>G	p.His588Arg	rs781498366	Grünert, 2014 Xi et al., 2014 Chen et al., 2019b	26
376	*ETFDH*	13		chr4:158708436A>T	c.1763A>T	p.His588Leu	n.a	[a,c]Wang et al., 2011b Chen et al., 2019b	n.a
377	*ETFDH*	13		chr4:158708441A>G	c.1768A>G	p.Lys590Glu	n.a	Gempel et al., 2007 Grünert, 2014 Trakadis et al., 2012 Chen et al., 2019b	n.a
378	*ETFDH*	13		chr4:158708441A>T	c.1768A>T	p.Lys590*	n.a	van Rijt et al., 2019	n.a
379	*ETFDH*	13		chr4:158708446-158708447	c.1773_1774delA	p.Cys592*	rs767795266	Grünert, 2014 Xi et al., 2014 [a,c]Tang et al., 2017 Chen et al., 2019b	n.a

380	*ETFDH*	13		chr4:158708446-158708447	c.1773-1774delAT	p.Lys590*	n.a	Zhu et al., 2014 [a,c]Li, 2015 Chen et al., 2019b	n.a
381	*ETFDH*	13		chr4:158708447T>C	c.1774T>C	p.Cys592Arg	n.a	Yotsumoto et al., 2008 Trakadis et al., 2012 Grünert, 2014 Yamada et al., 2017 Chen et al., 2019b van Rijt et al., 2020	n.a
382	*ETFDH*	13		chr4:158708447T>G	c.1774T>G	p.Cys592Arg	n.a	Ohkuma et al., 2009 Ishii et al., 2010 Chen et al., 2019b	n.a
383	*ETFDH*	13		chr4:158708456A>G	c.1783A>G	p.Lys595Glu	rs1422165935	-	31
384	*ETFDH*	13		chr4:158708459G>A	c.1786G>A	p.Asp596Asn	rs1165102742	Izumi et al., 2011 Grünert, 2014 Chen et al., 2019b	33
385	*ETFDH*	13		chr4:158708460A>G	c.1787A>G	p.Asp596Gly	n.a	[a,c]Tong et al., 2017 Yamada et al., 2017 Chen et al., 2019b	n.a
386	*ETFDH*	13		chr4:158708463C>T	c.1790C>T	p.Pro597Leu	n.a	Chen et al., 2019b Yıldız et al., 2019	n.a
387	*ETFDH*	13		chr4:158708480T>C	c.1807T>C	p.Trp603Arg	rs1561252786	-	33
388	*ETFDH*	13		chr4:158708482G>A	c.1809G>A	p.Trp603*	rs796051960	Sugai et al., 2012 Grünert, 2014 Chen et al., 2019b	0
389	*ETFDH*	13		chr4:158708483G>T	c.1810G>T	p.Val604Leu	rs1436514087	Zhu et al., 2014	28
390	*ETFDH*	13		chr4:158708485del	c.1812delG	p.Val605Tyrfs∗34	n.a	Hu et al., 2020	n.a
391	*ETFDH*	13		chr4:158708500-158709501	c.1827_1828insCAC	p.610insHis	n.a	Hong et al., 2019	n.a
392	*ETFDH*	13		chr4:158708501G>A	c.1828G>A	p.Gly610Arg	n.a	Zhu et al., 2014 [a,c]Li, 2015 Chen et al., 2019b Hong et al., 2019	n.a
393	*ETFDH*	13		chr4:158708504G>A	c.1831G>A	p.Gly611Arg	n.a	Er et al., 2010 Trakadis et al., 2012 Grünert, 2014 Chen et al., 2019b	n.a
394	*ETFDH*	13		chr4:158708505G>A	c.1832G>A	p.Gly611Glu	rs761669036	[a]Goodman et al., 2002	31
395	*ETFDH*	13		chr4:158708515C>A	c.1842C>A	p.Tyr614*	rs765049360	van Rijt et al., 2019 van Rijt et al., 2020	0

396	*ETFDH*	13		chr4:158708525T>C	c.1852T>C	p.*618Gln	rs765742496	Angle and Burton, 2008 Grünert, 2014 Chen et al., 2019b Henriques et al, 2019	0
397	*ETFDH*	13		chr4:158708525T>C	c.1852T>C	p.*618Glnext*14	rs765742496	Rocha et al., 2011 Henriques et al., 2019	n.a
398	*ETFDH*		3'UTR	chr4:158708662A>C	c.1989A>C	n.r	n.a	Grünert, 2014	n.a
399	*FLAD1*	1		chr1:154984015-154984018GGGG>GGG	c.324delG	p.Arg109Alafs*3	rs876661314	Olsen et al., 2016 Balasubramaniam et al., 2019	n.a
400	*FLAD1*	1		chr1:154984044T>A>C	c.350T>A	p.Ile117Asn	rs751960963	-	32
401	*FLAD1*	1		chr1:154984066G>T	c.372G>T	p.Lys124Asn	rs1338709810	-	35
402	*FLAD1*	1		chr1:154988105G>A	c.373G>A	p.Gly125Arg	rs747895040	-	31
403	*FLAD1*	2		chr1:154988129-154988136TTCTTTCT>TTCT	c.401_404delTTCT	p.Phe134Cysfs*8	rs876661313	Taylor et al., 2014 Olsen et al., 2016 Yıldız et al., 2018 Balasubramaniam et al., 2019	n.a
404	*FLAD1*	2		chr1:154988148T>C	c.416T>C	p.Leu139Pro	rs775833539	-	30
405	*FLAD1*	2		chr1:154988174C>T	c.442C>T	p.Arg148*	rs1426328465	Muru et al., 2019	0
406	*FLAD1*	2		chr1:154988229T>C	c.497T>C	p.Phe166Ser	rs1203866786	-	31
407	*FLAD1*	2		chr1:154988230del	c.498delC	p.Ser167Profs*20	rs876661315	Olsen et al., 2016 Balasubramaniam et al., 2019	n.a
408	*FLAD1*	2		chr1:154988240T>C	c.508T>C	p.Phe170Leu	rs761928040	Olsen et al., 2016 Balasubramaniam et al., 2019	23
409	*FLAD1*	2		chr1:154988258-154988269GCAGGGGGCATC>CA	c.526_537delinsCA	p.Ala176Glnfs*8	rs876661312	Olsen et al., 2016 Balasubramaniam et al., 2019	0
410	*FLAD1*	2		chr1:154988264G>A	c.532G>A	p.Gly178Ser	rs920886455	-	31
411	*FLAD1*	2		chr1:154988270G>A	c.538G>A	p.Gly180Ser	rs376637647	-	31
412	*FLAD1*	2		chr1:154988270G>C	c.538G>C	p.Gly180Arg	rs376637647	-	32
413	*FLAD1*	2		chr1:154988300-154988301dupGC	c.568_569dupGC	p.Val191Glnfs*10	rs876661310	Olsen et al., 2016 Auranen et al., 2017 Balasubramaniam et al., 2019	n.a
414	*FLAD1*	2		chr1:154988477C>T	c.745C>T	p.Arg249*	rs199979286	Balasubramaniam et al., 2019 Ryder et al., 2019 Yamada et al., 2019	0
415	*FLAD1*	2		chr1:154988529del	c.797_798delAGinsT	p.Glu266Valfs*3	n.a	García-Villoria et al., 2018	n.a
416	*FLAD1*	2		chr1:154988567-154988568TT>T	c.836delT	p.Phe279Serfs*45	rs876661311	Olsen et al., 2016 Balasubramaniam et al., 2019	n.a

417	*FLAD1*	2		chr1:154988663G>C	c.931G>C	p.Gly311Arg	rs764740796	-	31
418	*FLAD1*	3		chr1:154989605A>G	c.1163A>G	p.Glu388Gly	rs1043646797	-	31
419	*FLAD1*	3		chr1:154989655G>A	c.1213G>A	p.Gly405Arg	rs754788619	-	31
420	*FLAD1*	3		chr1:154989658G>T	c.1216G>T	p.Gly406Cys	rs1404487345	-	33
421	*FLAD1*	3		chr1:154989662A>T	c.1220A>T	p.Lys407Ile	rs1266041411	-	31
422	*FLAD1*	3		chr1:154989707G>C	c.1265G>C	p.Arg422Thr	rs748771067	-	33
423	*FLAD1*	5		chr1:154990341A>G	c.1367A>G	p.Tyr456Cys	rs1275925705	-	32
424	*FLAD1*	5		chr1:154990367G>A	c.1393G>A	p.Gly465Ser	rs773436746	-	32
425	*FLAD1*	5		chr1:154990382G>A	c.1408G>A	p.Ala470Thr	rs775463868	-	32
426	*FLAD1*	5		chr1:154990440G>A	c.1466G>A	p.Arg489His	rs1447858195	-	33
427	*FLAD1*	5		chr1:154990456-154990460delCCT	c.1484_1486delCCT	p.Ser495del	rs876661309	Olsen et al., 2016 Balasubramaniam et al., 2019	n.a
428	*FLAD1*	5		chr1:154990498G>T	c.1524G>T	p.Trp508Cys	rs1031039007	-	32
429	*FLAD1*		5	chr1:154992708C>G	c.1555-3C>G	splice variant	n.a	García-Villoria et al., 2018	n.a
430	*FLAD1*	6		chr1:154992746C>T	c.1588C>T	p.Arg530Cys	rs771466122	Olsen et al., 2016 Auranen et al., 2017 Balasubramaniam et al., 2019 Muru et al., 2019	26
431	*FLAD1*	6		chr1:154992747G>A	c.1589G>A	p.Arg530His	rs146007674	-	33
432	*FLAD1*	6		chr1:154992768G>C	c.1610G>C	p.Cys537Ser	rs143499209	-	32
433	*FLAD1*	6		chr1:154992777A>G	c.1619A>G	p.Tyr540Cys	rs1299471459	-	32
434	*FLAD1*	7		chr1:154993030G>A	c.1757G>A	p.Arg586His	rs766627769	-	31
435	*SLC25A32*		5' UTR	chr8:12:98593715del	c.-264_31delins(14)	deletion of Met start codon	n.a	Hellebrekers et al., 2017	n.a
436	*SLC25A32*	1		chr8:103414868C>T	c.70G>A	p.Glu24Lys	rs932761592	-	32
437	*SLC25A32*	1		chr8:103414816T>C	c.122A>G	p.His41Arg	rs1472757179	-	30
438	*SLC25A32*	1		chr8:103414810A>G	c.128T>C	p.Leu43Pro	rs149273860	-	34
439	*SLC25A32*	1		chr8:103414808C>G	c.130G>C	p.Asp44His	rs766681889	-	33
440	*SLC25A32*	1		chr8:103414795A>C	c.143T>G	p.Ile48Ser	rs567748395	-	34
441	*SLC25A32*	2		chr8:103407760G>A	c.179C>T	p.Pro60Leu	rs747038599	-	31
442	*SLC25A32*	2		chr8:103407754T>C	c.185A>G	p.Tyr62Cys	rs772386221	-	32
443	*SLC25A32*	2		chr8:103407644A>G	c.295T>C	p.Tyr99His	rs760697865	-	32
444	*SLC25A32*	3		chr8:103404859T>C	c.308A>G	p.Tyr103Cys	rs764848283	-	31
445	*SLC25A32*	3		chr8:103404778G>C	c.389C>G	p.Ala130Gly	rs973996007	-	34
446	*SLC25A32*	4		chr8:103404776C>T	c.391G>A	p.Gly131Arg	rs770178649	-	34
447	*SLC25A32*	4		chr8:103403291C>T	c.425G>A	p.Trp142*	rs147014855	Schiff et al., 2016	n.a
448	*SLC25A32*	4		chr8:103403276C>T	c.440G>A	p.Arg147His	rs142329098	Schiff et al., 2016	27
449	*SLC25A32*	4		chr8:103403195T>C	c.521A>G	p.Tyr174Cys	rs1563717284	-	30
450	*SLC25A32*	4		chr8:103403186T>C	c.530A>G	p.Glu177Gly	rs772497373	-	31

451	*SLC25A32*	4		chr8:103403178G>A	c.538C>T	p.Arg180Cys	rs374025789	-	33
452	*SLC25A32*	4		chr8:103403168T>C	c.548A>G	p.Tyr183Cys	rs864622028	-	32
453	*SLC25A32*	5		chr8:103402014A>G	c.593T>C	p.Leu198Pro	rs757326114	-	32
454	*SLC25A32*	6		chr8:103401648T>C	c.680A>G	p.Tyr227Cys	rs1202272743	-	33
455	*SLC25A32*	6		chr8:103401636G>A	c.692C>T	p.Ala231Val	rs1238398531	-	31
456	*SLC25A32*	6		chr8:103401633G>A	c.695C>T	p.Ala232Val	rs759232426	-	31
457	*SLC25A32*	6		chr8:103401630A>G	c.698T>C	p.Leu233Pro	rs145785157	-	32
458	*SLC25A32*	6		chr8:103401628A>G	c.700T>C	p.Ser234Pro	rs770819305	-	31
459	*SLC25A32*	6		chr8:103401600T>C	c.728A>G	p.Tyr243Cys	rs971518583	-	32
460	*SLC25A32*	6		chr8:103401597G>A	c.731C>T	p.Pro244Leu	rs1262662383	-	31
461	*SLC25A32*	6		chr8:103401594T>C	c.734A>G	p.Tyr245Cys	rs983458477	-	32
462	*SLC25A32*	6		chr8:103401520A>T	c.808T>A	p.Trp270Arg	rs1209946662	-	32
463	*SLC25A32*	7		chr8:103400468A>C	c.891T>G	p.Phe297Leu	rs1270709178	-	31
464	*SLC25A32*	7		chr8:103400460T>C	c.899A>G	p.Tyr300Cys	rs141856398	-	32
465	*SLC25A32*	7		chr8:103400439A>G	c.920T>C	p.Leu307Ser	rs1401357629	-	31
466	*SLC52A1*	2-3		n.r	1.9kb del	n.r	n.a	Ho et al., 2011	n.a
467	*SLC52A1*		4	chr17:5033250C>T	c.1134+11G>A	exon 4 skipping	rs141935493	Mosegaard et al., 2017	n.a
468	*SLC52A2*	1		chr8:144359384T>G	c.91T>G	p.Trp31Gly	rs1554853760	-	33
469	*SLC52A2*	1		chr8:144359385G>C	c.92G>C	p.Trp31Ser	rs797045199	Foley et al., 2014	26
470	*SLC52A2*	1		chr8:144359423G>A	c.130G>A	p.Gly44Ser	rs782089755	-	33
471	*SLC52A2*	2		chr8:144359627G>T	c.135G>T	p.Trp45Cys	rs1064795599	-	33
472	*SLC52A2*	2		chr8:144359647C>T	c.155C>T	p.Ser52Phe	rs397514657	Ciccolella et al., 2013	21
473	*SLC52A2*	2		chr8:144359674G>A	c.182G>A	p.Gly61Asp	rs1554853862	-	32
474	*SLC52A2*	2		chr8:144360215G>A	c.231G>A	p.Glu77Lys	rs1554854166	Manole et al., 2017	0
475	*SLC52A2*	2		chr8:144359737G>C	c.245G>C	p.Arg82Pro	rs377110942	Bamaga et al., 2018	12
476	*SLC52A2*	2		chr8:144359845C>A	c.353C>A	p.Ala118Asp	rs117500243	Allison et al., 2017	15
477	*SLC52A2*	3		chr8:144359860T>C	c.368T>C	p.Leu123Pro	rs397514538	Bosch et al., 2012 Haack et al., 2012	22
478	*SLC52A2*	3		chr8:144359875C>T	c.383C>T	p.Ser128Leu	rs374071862	Cosgrove et al., 2015 Manole et al., 2017	21
479	*SLC52A2*	3		chr8:144359893C>T	c.401C>T	p.Pro134Leu	rs1447838904	Guissart et al., 2016	23
480	*SLC52A2*	3		chr8:144359910C>T	c.419C>T	p.Pro140Leu	n.a	Bosch et al., 2012 Johnson et al., 2012	n.a
481	*SLC52A2*	3		chr8:144359913C>A	c.421C>A	p.Pro141Thr	rs377740960	Udhayabanu et al., 2016	22
482	*SLC52A2*	3		chr8:144359941G>A	c.449G>A	p.Gly150Asp	rs1365149837	-	32
483	*SLC52A2*	3		chr8:144359988G>A	c.496G>A	p.Gly166Ser	rs1554854006	-	32
484	*SLC52A2*	3		chr8:144359989G>A	c.497G>A	p.Gly166Asp	rs782636375	-	31

485	*SLC52A2*	3		chr8:144359997C>T	c.505C>T	p.Arg169Cys	rs782345472	Allison et al., 2017 Woodcock et al., 2018	21
486	*SLC52A2*	3		chr8:144360192C>T	c.700C>T	p.Gln234*	rs797045200	Foley et al., 2014	0
487	*SLC52A2*	3		chr8:144360300C>T	c.808C>T	p.Gln270∗	rs375088539	Petrovski et al., 2015	0
488	*SLC52A2*	3		chr8:144360343C>A	c.851C>A	p.Ala284Asp	rs398123067	Foley et al., 2014	19
489	*SLC52A2*	3		chr8:144360357C>T	c.865C>T	p.Ala288Val	rs797045201	Manole et al., 2017	0
490	*SLC52A2*	3		chr8:144360406A>G	c.914A>G	p.Tyr205Cys	rs398123068	Foley et al., 2014	23
491	*SLC52A2*	3		chr8:144360408G>A>T	c.916G>A	p.Gly306Arg	rs398124641	Bosch et al., 2012 Johnson et al., 2012 Foley et al., 2014 Srour et al., 2014 Guissart et al., 2016 Menezes et al., 2016 Allison et al., 2017 Fan and Fogel, 2018	26
492	*SLC52A2*	3		chr8:144360409G>A	c.917G>A	p.Gly306Glu	rs781923855	Nimmo et al., 2018 Shi et al., 2019	23
493	*SLC52A2*	3		chr8:144360427T>C	c.935T>C	p.Leu312Pro	rs754320812	Foley et al., 2014 Allison et al., 2017 Manole et al., 2017	24
494	*SLC52A2*	3		chr8:144360460T>C	c.968T>C	p.Leu323Pro	rs781842708	Gorcenco et al., 2019	24
495	*SLC52A2*	3		chr8:144360604T>C	c.1016T>C	p.Leu339Pro	rs148234606	Bosch et al., 2012 Haack et al., 2012 Foley et al., 2014 Gudbjartsson et al., 2015 Petrovski et al., 2015 Menezes et al., 2016 Manole et al., 2017	26
496	*SLC52A2*	3		chr8:144360610	c.1022_1023insC	p.Leu341Profs*103	n.a	Shi et al., 2019	n.a
497	*SLC52A2*	3		chr8:144360676C>T	c.1088C>T	p.Pro363Leu	rs797045202	Manole et al., 2017	28
498	*SLC52A2*	3		chr8:144360814G>C	c.1137G>C	p.Trp379Cys	rs782764685	-	34
499	*SLC52A2*	3		chr8:144360817del	c.1140delG	p.Leu381Cysfs*9	rs879254305	Bamaga et al., 2018	n.a
500	*SLC52A2*	3		chr8:144360819T>C	c.1142T>C	p.Leu381Pro	rs1554854563	-	33
501	*SLC52A2*	4		chr8:144360921G>A	c.1244G>A	p.Gly415Asp	rs782245545	-	32
502	*SLC52A2*	4		chr8:144360921G>T	c.1244G>T	p.Gly415Val	rs782245545	-	32
503	*SLC52A2*	4		chr8:144360932G>A	c.1255G>A	p.Gly419Ser	rs397514658	Ciccolella et al., 2013	26
504	*SLC52A2*	4		chr8:144360935G>A	c.1258G>A	p.Ala420Thr	rs368924997	Foley et al., 2014	23
505	*SLC52A2*	4		chr8:144361004T>C	c.1327T>C	p.Cys443Arg	n.a	Manole et al., 2017	n.a
506	*SLC52A2*	4		chr8:144361005G>A	c.1328G>A	p.Cys443Tyr	n.a	Shi et al., 2019	n.a

507	*SLC52A3*	2		chr20:765736C>T	c.39G>A	p.Gly13Arg	n.a	Manole et al., 2017	n.a
508	*SLC52A3*	2		chr20:765731C>A	c.44G>T	p.Gly15Val	n.a	Horoz et al., 2016	n.a
509	*SLC52A3*	2		chr20:765726A>G	c.49T>C	p.Trp17Arg	rs797045190	Bosch et al., 2011 Bosch et al., 2012 van Rijt et al., 2020	29
510	*SLC52A3*	2		chr20:765717T>G	c.58A>C	p.Ile20Leu	rs148387972	Manole et al., 2017	24
511	*SLC52A3*	2		chr20:765713T>C	c.62A>G	p.Asn21Ser	rs199588390	Bosch et al., 2012 Dezfouli et al., 2012 Udhayabanu et al., 2016 Gowda et al., 2018	26
512	*SLC52A3*	2		chr20:765705A>G	c.70T>C	p.Trp24Arg	rs1288873761	-	32
513	*SLC52A3*	2		chr20:765704C>T	c.71G>A	p.Trp24*	n.a	Hossain et al., 2017	n.a
514	*SLC52A3*	2		chr20:765693G>T	c.82C>A	p.Pro28Thr	rs267606688	Johnson et al., 2010 Bosch et al., 2012	27
515	*SLC52A3*	2		chr20:765669C>T	c.106G>A	p.Glu36Lys	rs267606686	Green et al., 2010 Bosch et al., 2012 Malafronte et al., 2013 Allison et al., 2017 Manole et al., 2017 Abbas et al., 2018	27
516	*SLC52A3*	2		chr20:765662C>G	c.113G>C	p.Trp38Ser	n.a	Carreau et al., 2020	n.a
517	*SLC52A3*	2		chr20:765615C>T	c.160G>A	p.Gly54Arg	rs797045191	Bosch et al., 2012 Johnson et al., 2012	25
518	*SLC52A3*	2		chr20:765602A>T	c.173T>A	p.Val58Asp	rs797045192	Bosch et al., 2012 Ciccolella et al., 2012	29
519	*SLC52A3*	2		chr20:765582G>A	c.193C>T	p.Arg65Trp	rs537244674	Davis et al., 2016	20
520	*SLC52A3*	2		chr20:765564C>T	c.211G>A	p.Glu71Lys	rs267606683	Johnson et al., 2010 Bosch et al., 2012	26
521	*SLC52A3*	2		chr20:765564C>A	c.211G>T	p.Glu71*	rs267606683	Green et al., 2010 Bosch et al., 2012	0
522	*SLC52A3*	2		chr20:765551A>G	c.224T>C	p.Ile75Thr	rs797045193	Bosch et al., 2012 Johnson et al., 2012	26
523	*SLC52A3*	2		chr20:765421C>T	c.354G>A	p.Val118Met	n.a	Manole et al., 2017	n.a
524	*SLC52A3*	2		chr20:765401G>T	c.374C>A	p.Thr125Asn	rs767263985	Manole et al., 2017 Chaya et al., 2018	27
525	*SLC52A3*	2		chr20:765392G>A	c.383C>T	p.Pro128Leu	rs890766612	-	31
526	*SLC52A3*	2		chr20:765381G>A	c.394C>T	p.Arg132Trp	rs267606684	Green et al., 2010 Bosch et al., 2012 Rabbani et al., 2020	26
527	*SLC52A3*	2		chr20:765372T>C	c.403A>G	p.Thr135Ala	rs527853872	Manole et al., 2017	5

528	*SLC52A3*	2		chr20:765320G>A	c.455C>T	p.Pro152Leu	rs1255062029	-	32
529	*SLC52A3*	2		chr20:765308G>A	c.467C>T	p.Ala156Val	rs1318600253	-	31
530	*SLC52A3*	2		chr20:765273T>G	c.502A>C	p.Asn168His	rs1337277627	Rabbani et al., 2020	23
531	*SLC52A3*	2		chr20: between 764021 & 764022	c.568-19_-18insCTGATTGAC	insertion	rs11273404	Bosch et al., 2012 Ciccolella et al., 2012	n.a
532	*SLC52A3*	3		chr20:763937G>A	c.634C>T	p.Arg212Cys	rs778479139	Manole et al., 2017	22
533	*SLC52A3*	3		chr20:763932G>C	c.639C>G	p.Tyr213*	rs778363575	Green et al., 2010 Johnson et al., 2010 Bosch et al., 2011 Bosch et al., 2012 Malafronte et al., 2013 Chaya et al., 2018 van Rijt et al., 2020	0
534	*SLC52A3*	3		chr20:763912G>T	c.659C>A	p.Pro220His	rs797045194	Bosch et al., 2012 Dezfouli et al., 2012	26
535	*SLC52A3*	3		chr20:763901A>G	c.670T>C	p.Phe224Leu	rs267606685	Green et al., 2010 Bosch et al., 2012	27
536	*SLC52A3*	3		chr20:763900A>C	c.671T>G	p.Phe224Cys	rs797045197	-	32
537	*SLC52A3*	3		chr20:763893del	c.678_680del	p.Leu227del	n.a	van Rijt et al., 2020	n.a
538	*SLC52A3*	3		chr20:763775G>A	c.796C>T	p.Arg266Trp	rs370499474	Bosch et al., 2012 Ciccolella et al., 2012	25
539	*SLC52A3*	3		chr20:763636G>A	c.935C>T	p.Ala312Val	rs752218005	Bosch et al., 2012 Dezfouli et al., 2012	24
540	*SLC52A3*	3		chr20:763616G>A	c.955C>T	p.Pro319Ser	rs797045195	Bosch et al., 2012 Ciccolella et al., 2012	29
541	*SLC52A3*	3		chr20:763615G>A	c.956C>T	p.Pro319Leu	rs760533974	-	33
542	*SLC52A3*	3		chr20:763612G>C	c.959C>G	p.Ser320Cys	rs535368648	-	33
543	*SLC52A3*	3		chr20:763612G>A	c.959C>T	p.Ser320Phe	rs535368648	-	33
544	*SLC52A3*	3		chr20:763598A>G	c.973T>C	p.Ser325Pro	rs1246925461	-	32
545	*SLC52A3*	3		chr20:763597G>A	c.974C>T	p.Ser325Phe	rs748083229	-	33
546	*SLC52A3*	3		chr20:763582C>A	c.989G>T	p.Gly330Val	rs797045196	Bosch et al., 2012 Koy et al., 2012	25
547	*SLC52A3*	3		chr20:763523A>T	c.1048T>A	p.Leu350Met	rs76947760	Green et al., 2010 Bosch et al., 2012	18
548	*SLC52A3*	3		chr20:763498C>G	c.1073G>C	p.Arg358Thr	rs892890925	-	33
549	*SLC52A3*	4		chr20:761824C>T	c.1074G>A	splice variant	rs1202138005	Manole et al., 2017	0
550	*SLC52A3*	4		chr 20:761817G>C	c.1081C>G	p.Leu361Val	n.a	Bandettini di Poggio et al., 2014	n.a
551	*SLC52A3*	4		n.a	n.r	p.Leu369Pro	n.a	Garg et al., 2018	n.a
552	*SLC52A3*	4		chr20:761774C>T	c.1124G>A	p.Gly375Asp	rs1219868273	Dezfouli et al., 2012	23

553	*SLC52A3*	4		chr20:761771T>C	c.1127A>G	p.Tyr376Cys	rs749962422	Bandettini di Poggio et al., 2014	32
554	*SLC52A3*	4		chr20:761770	c.1128_1129insT	p.Tyr376fs	n.a	Manole et al., 2017	n.a
555	*SLC52A3*	4		chr20:761742A>G	c.1156T>C	p.Cys386Arg	rs1555783543	Thulasi et al., 2017	33
556	*SLC52A3*	4		chr20:761738G>A	c.1160C>T	p.Pro387Leu	rs1164187566	-	34
557	*SLC52A3*		4	chr20:761240T>G	c.1198-2A>C	splice variant	rs754753126	Bosch et al., 2011 Bosch et al., 2012	0
558	*SLC52A3*	5		chr20:761213C>T	c.1223G>A	p.Gly408Asp	rs1057524605	Nimmo et al., 2018	25
559	*SLC52A3*	5		chr20:761204-761201	c.1232_1233ins(60)	p.Ser411_Tyr412ins(20)	n.a	Camargos et al., 2018	n.a
560	*SLC52A3*	5		chr20:761201T>C	c.1235A>G	p.Tyr412Cys	rs1359318796	-	33
561	*SLC52A3*	5		chr20:761198A>G	c.1238T>C	p.Val413Ala	rs267606687	Green et al., 2010 Bosch et al., 2012 Ciccolella et al., 2012 Davis et al., 2016 Bashford et al., 2017 Manole et al., 2017	32
562	*SLC52A3*	5		chr20:761195T>C	c.1241A>G	p.Lys414Arg	rs772753947	-	32
563	*SLC52A3*	5		chr20:761181C>T	c.1255G>A	p.Gly418Asp	rs797045198	Manole et al., 2017 Rizzo et al., 2017	20
564	*SLC52A3*	5		chr20:761171C>G	c.1265G>C	p.Arg422Pro	rs200665228	-	31
565	*SLC52A3*	5		chr20:761160G>A	c.1276C>T	p.Arg426Cys	rs780025944	-	34
566	*SLC52A3*	5		chr20:761144C>T	c.1292G>A	p.Trp431*	rs1379613754	Cosgrove et al., 2015	0
567	*SLC52A3*	5		chr20:761143C>G	c.1293G>C	p.Trp431Cys	rs1060499531	-	33
568	*SLC52A3*	5		chr20:761142C>T	c.1294G>A	p.Trp431*	n.a	Manole et al., 2017	n.a
569	*SLC52A3*	5		chr20:761140G>T	c.1296C>A	p.Cys432*	rs758570021	Bosch et al., 2012 Ciccolella et al., 2012	0
570	*SLC52A3*	5		chr20:761123A>G	c.1313T>C	p.Leu438Pro	rs1167512470	-	33
571	*SLC52A3*	5		chr20:761120C>T	c.1316G>A	p.Gly439Asp	rs1555783467	Woodcock et al., 2018	31
572	*SLC52A3*	5		chr20:761114A>G	c.1322T>C	p.Leu441Pro	rs1441733847	-	33
573	*SLC52A3*	5		chr20:761110-76111del	c.1325_1326del	p.Leu442Argfs*35	n.a	Green et al., 2010	n.a
574	*SLC52A3*	5		chr20:761110-761112GAG>G	c.1325_1326delTG	p.Leu442Argfs*64	rs794728004	Bosch et al., 2012	n.a
575	*SLC52A3*	5		chr20:761105G>A	c.1331C>T	p.Ala444Val	rs766882276	-	33
576	*SLC52A3*	5		chr20:761102A>C	c.1334T>G	p.Leu445Arg	rs761224042	-	33
577	*SLC52A3*	5		chr20:761090G>C	c.1346C>G	p.Pro449Arg	rs1322154756	-	33
578	*SLC52A3*	5		chr20:761087A>G	c.1349T>C	p.Leu450Pro	rs1237553628	-	34
579	*SLC52A3*	5		chr20:761067A>G	c.1369T>C	p.Phe457Leu	rs779750163	-	32

580	*SLC52A3*	5		chr20:761065G>C	c.1371C>G	p.Phe457Leu	rs145431028	Green et al., 2010 Bosch et al., 2012 Johnson et al., 2012 Manole et al., 2017	32
581	*SLC52A3*	5		chr20:761055A>C	c.1381T>G	p.Asp461Tyr	n.a	Bashford et al., 2017	n.a
582	*SLC52A3*		3'UTR	n.a	c.1555C>T	n.r	n.a	Rizzo et al., 2017	n.a

* No value refers to a likely pathogenic variant identified from Ensembl and gnomAD v2.1.1 databases; n.r, Not reported; n.a, Not applicable; [a]Article was not identified by the PubMed search; [b]Article is listed on PubMed, but not available in English/Dutch/German; [c]Article is not listed on PubMed and not available in English/Dutch/German. All articles not available in English/Dutch/German could not be followed-up by the authors.

Reference list:

1. Abbas, Q., Jafri, S.K., Ishaque, S., Rahman, A.J., 2018. Brown-Vialetto-Van Laere syndrome: a novel diagnosis to a common presentation. BMJ Case Rep 2018, bcr2018224958. https://doi.org/10.1136/bcr-2018-224958
2. Al-Jasmi, F.A., Al-Shamsi, A., Hertecant, J.L., Al-Hamad, S.M., Souid, A.-K., 2016. Inborn errors of metabolism in the United Arab Emirates: disorders detected by newborn screening (2011-2014). JIMD Rep. 28, 127–135. https://doi.org/10.1007/8904_2015_512
3. Allison, T., Roncero, I., Forsyth, R., Coffman, K., Pichon, J.L., 2017. Brown-Vialetto-Van Laere syndrome as a mimic of neuroimmune disorders: 3 cases from the clinic and review of the literature. J Child Neurol 32, 528–532. https://doi.org/10.1177/0883073816689517
4. Angelini, C., 2015. Spectrum of metabolic myopathies. Biochim Biophys Acta 1852, 615–621. https://doi.org/10.1016/j.bbadis.2014.06.031
5. Angelini, C., Tavian, D., Missaglia, S., 2018. Heterogeneous phenotypes in lipid storage myopathy due to ETFDH gene mutations. JIMD Rep 38, 33–40. https://doi.org/10.1007/8904_2017_27
6. Angle, B., Burton, B.K., 2008. Risk of sudden death and acute life-threatening events in patients with glutaric acidemia type II. Mol Genet Metab 93, 36–39. https://doi.org/10.1016/j.ymgme.2007.09.015
7. Auranen, M., Paetau, A., Piirilä, P., Pohju, A., Salmi, T., Lamminen, A., Löfberg, M., Mosegaard, S., Olsen, R.K., Tyni, T., 2017. Patient with multiple acyl-CoA dehydrogenation deficiency disease and FLAD1 mutations benefits from riboflavin therapy. Neuromuscul Disord 27, 581–584. https://doi.org/10.1016/j.nmd.2017.03.003
8. Balasubramaniam, S., Christodoulou, J., Rahman, S., 2019. Disorders of riboflavin metabolism. J Inherit Metab Dis 42, 608–619. https://doi.org/10.1002/jimd.12058
9. Bamaga, A.K., Maamari, R.N., Culican, S.M., Shinawi, M., Golumbek, P.T., 2018. Child neurology: Brown-Vialetto-Van Laere syndrome: dramatic visual recovery after delayed riboflavin therapy. Neurology 91, 938–941. https://doi.org/10.1212/wnl.0000000000006498
10. Bandettini di Poggio, M., Monti Bragadin, M., Reni, L., Doria-Lamba, L., Cereda, C., Pardini, M., Roccatagliata, L., Rossi, A., Schenone, A., 2014. Brown-Vialetto-Van Laere syndrome: clinical and neuroradiological findings of a genetically proven patient. Amyotroph Lateral Scler Front. Degener 15, 141–144. https://doi.org/10.3109/21678421.2013.837931

11. Bashford, J.A., Chowdhury, F.A., Shaw, C.E., 2017. Remarkable motor recovery after riboflavin therapy in adult-onset Brown-Vialetto-Van Laere syndrome. Pr. Neurol 17, 53–56. https://doi.org/10.1136/practneurol-2016-001488
12. Beard, S.E., Goodman, S.I., Bemelen, K., Frerman, F.E., 1995. Characterization of a mutation that abolishes quinone reduction by electron transfer flavoprotein-ubiquinone oxidoreductase. Hum Mol Genet 4, 157–161. https://doi.org/10.1093/hmg/4.2.157
13. Beard, S.E., Spector, E.B., Seltzer, W.K., Frerman, F.E., Goodman, S.I., 1993. Mutations in electron transfer flavoprotein:ubiquinone oxidoreductase (ETF:QO) in glutaric acidemia type II (GA2). Clin Res 41, 271A.
14. Béhin, A., Acquaviva-Bourdain, C., Souvannanorath, S., Streichenberger, N., Attarian, S., Bassez, G., Brivet, M., Fouilhoux, A., Labarre-Villa, A., Laquerrière, A., Pérard, L., Kaminsky, P., Pouget, J., Rigal, O., Vanhulle, C., Eymard, B., Vianey-Saban, C., Laforêt, P., 2016. Multiple acyl-CoA dehydrogenase deficiency (MADD) as a cause of late-onset treatable metabolic disease. Rev Neurol 172, 231–241. https://doi.org/10.1016/j.neurol.2015.11.008
15. Beresford, M.W., Pourfarzam, M., Turnbull, D.M., Davidson, J.E., 2006. So doctor, what exactly is wrong with my muscles? Glutaric aciduria type II presenting in a teenager. Neuromuscul Disord 16, 269–273. https://doi.org/10.1016/j.nmd.2006.01.001
16. Bosch, A.M., Abeling, N.G., Ijlst, L., Knoester, H., van der Pol, W.L., Stroomer, A.E., Wanders, R.J., Visser, G., Wijburg, F.A., Duran, M., Waterham, H.R., 2011. Brown-Vialetto-Van Laere and Fazio Londe syndrome is associated with a riboflavin transporter defect mimicking mild MADD: a new inborn error of metabolism with potential treatment. J Inherit Metab Dis 34, 159–164. https://doi.org/10.1007/s10545-010-9242-z
17. Bosch, A.M., Stroek, K., Abeling, N.G., Waterham, H.R., Ijlst, L., Wanders, R.J., 2012. The Brown-Vialetto-Van Laere and Fazio Londe syndrome revisited: natural history, genetics, treatment and future perspectives. Orphanet J Rare Dis 7, 83. https://doi.org/10.1186/1750-1172-7-83
18. Camargos, S., Guerreiro, R., Bras, J., Mageste, L.S., 2018. Late-onset and acute presentation of Brown-Vialetto-Van Laere syndrome in a Brazilian family. Neurol Genet 4, e215. https://doi.org/10.1212/nxg.0000000000000215
19. Cao, J., Li, Y., Yang, J., Liang, Y., Feng, S., Zhang, X., Li, J., Zhang, Y., 2014. Clinical characteristics and gene mutation analysis of riboflavin-responsive lipid storage myopathy: report of 3 cases in 2 families and review of literature. Chinese J.

Contemp. Neurol. Neurosurg. 14, 479–484. https://doi.org/10.3969/j.issn.1672-6731.2014.06.004

20. Carreau, C., Lenglet, T., Mosnier, I., Lahlou, G., Fargeot, G., Weiss, N., Demeret, S., Salachas, F., Veauville-Merllié, A., Acquaviva, C., Nadjar, Y., 2020. A juvenile ALS-like phenotype dramatically improved after high-dose riboflavin treatment. Ann Clin Transl Neurol 7, 250–253. https://doi.org/10.1002/acn3.50977
21. Chautard, R., Laroche-Raynaud, C., Lia, A.S., Chazelas, P., Derouault, P., Sturtz, F., Baaj, Y., Veauville-Merllié, A., Acquaviva, C., Favreau, F., Faye, P.A., 2020. A case report of a mild form of multiple acyl-CoA dehydrogenase deficiency due to compound heterozygous mutations in the ETFA gene. BMC Med Genomics 13, 12. https://doi.org/10.1186/s12920-020-0665-6
22. Chaya, S., Zampoli, M., Gray, D., Booth, J., Riordan, G., Ndondo, A., Fieggen, K., Rusch, J., van der Watt, G., Pillay, K., van der Westhuizen, F., Menezes, M., Wilmshurst, J., 2018. The first case of riboflavin transporter deficiency in sub-Saharan Africa. Semin Pediatr Neurol 26, 10–14. https://doi.org/10.1016/j.spen.2017.03.002
23. Chen, H.Z., Jin, M., Cai, N.Q., Lin, X.D., Liu, X.Y., Xu, L.Q., Lin, M.T., Lin, F., Wang, N., Wang, Z.Q., Xu, G.R., 2019a. Rhabdomyolysis and respiratory insufficiency due to the common ETFDH mutation of c.250G>A in two patients with late-onset multiple acyl-CoA dehydrogenase deficiency. Chin Med J 132, 1615–1618. https://doi.org/10.1097/cm9.0000000000000288
24. Chen, M., Peng, J., Wei, W., Wang, R., Xu, H., Liu, H., 2018a. A novel ETFDH mutation in an adult patient with late-onset riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency. Int J Neurosci 128, 291–294. https://doi.org/10.1080/00207454.2017.1380641
25. Chen, W., Zhang, Y., Ni, Y., Cai, S., Zheng, X., Mastaglia, F.L., Wu, J., 2019b. Late-onset riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency (MADD): case reports and epidemiology of ETFDH gene mutations. BMC Neurol 19, 330. https://doi.org/10.1186/s12883-019-1562-5
26. Chen, Y., Lui, H., Yuan, X., Wang, X., Teng, J., 2018b. Clinical analysis of riboflavin-reactive lipid storage myopathy in two families. Chinese J. Pract. Nerv. Dis. 21, 2692–2696.
27. Cheng, Y.Y., Tang, Y., Liu, A.J., Wei, L., Lin, L., Zhang, J., Zhi, L., 2017. [Clinical features and ETFDH mutations of children with late-onset glutaric aciduria type II: a report of two cases]. Zhongguo Dang Dai Er Ke Za Zhi 19, 975–978.

28. Chien, Y.H., Lee, N.C., Chao, M.C., Chen, L.C., Chen, L.H., Chien, C.C., Ho, H.C., Suen, J.H., Hwu, W.L., 2013. Fatty acid oxidation disorders in a Chinese population in Taiwan. JIMD Rep 11, 165–172. https://doi.org/10.1007/8904_2013_236
29. Chokchaiwong, S., Kuo, Y.T., Hsu, S.P., Hsu, Y.C., Lin, S.H., Zhong, W.B., Lin, Y.F., Kao, S.H., 2019. ETF-QO mutants uncoupled fatty acid β-oxidation and mitochondrial bioenergetics leading to lipid pathology. Cells 8, 106. https://doi.org/10.3390/cells8020106
30. Chokchaiwong, S., Kuo, Y.T., Lin, S.H., Hsu, Y.C., Hsu, S.P., Liu, Y.T., Chou, A.J., Kao, S.H., 2018. Coenzyme Q10 serves to couple mitochondrial oxidative phosphorylation and fatty acid β-oxidation, and attenuates NLRP3 inflammasome activation. Free Radic Res 52, 1445–1455. https://doi.org/10.1080/10715762.2018.1500695
31. Chu, G., Salzman, J., 2019. Hyperammonemia after capecitabine associated with occult impairment of the urea cycle. Cancer Med 8, 1996–2004. https://doi.org/10.1002/cam4.2036
32. Ciccolella, M., Catteruccia, M., Benedetti, S., Moroni, I., Uziel, G., Pantaleoni, C., Chiapparini, L., Bizzi, A., D'Amico, A., Fattori, F., Salsano, M.L., Pastore, A., Tozzi, G., Piemonte, F., Bertini, E., 2012. Brown-Vialetto-Van Laere and Fazio-Londe overlap syndromes: a clinical, biochemical and genetic study. Neuromuscul Disord 22, 1075–1082. https://doi.org/10.1016/j.nmd.2012.05.007
33. Ciccolella, M., Corti, S., Catteruccia, M., Petrini, S., Tozzi, G., Rizza, T., Carrozzo, R., Nizzardo, M., Bordoni, A., Ronchi, D., D'Amico, A., Rizzo, C., Comi, G.P., Bertini, E., 2013. Riboflavin transporter 3 involvement in infantile Brown-Vialetto-Van Laere disease: two novel mutations. J Med Genet 50, 104–107. https://doi.org/10.1136/jmedgenet-2012-101204
34. Colombo, I., Finocchiaro, G., Garavaglia, B., Garbuglio, N., Yamaguchi, S., Frerman, F.E., Berra, B., DiDonato, S., 1994. Mutations and polymorphisms of the gene encoding the beta-subunit of the electron transfer flavoprotein in three patients with glutaric acidemia type II. Hum Mol Genet 3, 429–435. https://doi.org/10.1093/hmg/3.3.429
35. Cornelius, N., Frerman, F.E., Corydon, T.J., Palmfeldt, J., Bross, P., Gregersen, N., Olsen, R.K., 2012. Molecular mechanisms of riboflavin responsiveness in patients with ETF-QO variations and multiple acyl-CoA dehydrogenation deficiency. Hum Mol Genet 21, 3435–3448. https://doi.org/10.1093/hmg/dds175
36. Cosgrove, J., Datta, S., Busby, M., 2015. Adult onset Brown-Vialetto-Van Laere syndrome with opsoclonus and a novel heterozygous mutation: a case report. Clin Neurol Neurosurg 128, 1–3. https://doi.org/10.1016/j.clineuro.2014.10.016

37. Creanza, A., Cotugno, M., Mazzaccara, C., Frisso, G., Parenti, G., Capaldo, B., 2018. Successful pregnancy in a young woman with multiple acyl-CoA dehydrogenase deficiency. JIMD Rep 39, 1–6. https://doi.org/10.1007/8904_2017_38
38. Cui, Y.J., Song, C.L., Cheng, Y.B., 2017. [Paroxysmal muscle weakness, liver enlargement, and hypoglycemia in a boy]. Zhongguo Dang Dai Er Ke Za Zhi 19, 1104–1108.
39. Curcoy, A., Olsen, R.K., Ribes, A., Trenchs, V., Vilaseca, M.A., Campistol, J., Osorio, J.H., Andresen, B.S., Gregersen, N., 2003. Late-onset form of beta-electron transfer flavoprotein deficiency. Mol Genet Metab 78, 247–249. https://doi.org/10.1016/s1096-7192(03)00024-6
40. Dai, D., Wen, F., Zhou, S., Chen, S., 2016. [Clinical features and gene mutations in a patient with multiple aeyl-CoA dehydrogenase deficiency with severe fatty liver]. Zhonghua Yi Xue Yi Chuan Xue Za Zhi 33, 191–194. https://doi.org/10.3760/cma.j.issn.1003-9406.2016.02.014
41. Davis, A., Josifova, D., Lloyd-Owen, S., Radunovic, A., Swash, M., 2016. Brown-Vialetto-Van Laere syndrome: a 28-year follow-up. J Neurol Neurosurg Psychiatry 87, 681–682. https://doi.org/10.1136/jnnp-2014-310088
42. Dernoncourt, A., Bouchereau, J., Acquaviva-Bourdain, C., Wicker, C., de Lonlay, P., Gourguechon, C., Sevestre, H., Merle, P.E., Maizel, J., Brault, C., 2019. Myogenic disease and metabolic acidosis: consider multiple acyl-coenzyme A dehydrogenase deficiency. Case Rep Crit Care 2019, 1598213. https://doi.org/10.1155/2019/1598213
43. Dezfouli, M.A., Yadegari, S., Nafissi, S., Elahi, E., 2012. Four novel C20orf54 mutations identified in Brown-Vialetto-Van Laere syndrome patients. J Hum Genet 57, 613–617. https://doi.org/10.1038/jhg.2012.70
44. Di Giacinto, I., Buda, S., Diamanti, M., Tonon, C., Pigna, A., Melotti, R.M., 2014. Multiple acyl-coenzyme A dehydrogenase deficiency: diagnosis in adulthood, intensive care management and sequelae. Minerva Anestesiol 80, 1145–1146.
45. Er, T.K., Chen, C.C., Liu, Y.Y., Chang, H.C., Chien, Y.H., Chang, J.G., Hwang, J.K., Jong, Y.J., 2011. Computational analysis of a novel mutation in ETFDH gene highlights its long-range effects on the FAD-binding motif. BMC Struct Biol 11, 43. https://doi.org/10.1186/1472-6807-11-43
46. Er, T.K., Liang, W.C., Chang, J.G., Jong, Y.J., 2010. High resolution melting analysis facilitates mutation screening of ETFDH gene: applications in riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency. Clin Chim Acta 411, 690–699. https://doi.org/10.1016/j.cca.2010.01.033

47. Fan, J., Fogel, B.L., 2018. Successful treatment of a genetic childhood ataxia due to riboflavin transporter deficiency. Cerebellum Ataxias 5, 12. https://doi.org/10.1186/s40673-018-0091-0

48. Fan, X., Xie, B., Zou, J., Luo, J., Qin, Z., D'Gama, A.M., Shi, J., Yi, S., Yang, Q., Wang, J., Luo, S., Chen, S., Agrawal, P.B., Li, Q., Shen, Y., 2018. Novel ETFDH mutations in four cases of riboflavin responsive multiple acyl-CoA dehydrogenase deficiency. Mol Genet Metab Rep 16, 15–19. https://doi.org/10.1016/j.ymgmr.2018.05.007

49. Fischer, T., Elpers, C., Och, U., Fobker, M., Marquardt, T., 2019a. Ketone body therapy with D/L-β-hydroxybutyric acid solution in severe MADD. Mol Genet Metab Rep 20, 100491. https://doi.org/10.1016/j.ymgmr.2019.100491

50. Fischer, T., Och, U., Marquardt, T., 2019b. Long-term ketone body therapy of severe multiple acyl-CoA dehydrogenase deficiency: a case report. Nutrition 60, 122–128. https://doi.org/10.1016/j.nut.2018.10.014

51. Fitzgerald, M., Crushell, E., Hickey, C., 2013. Cyclic vomiting syndrome masking a fatal metabolic disease. Eur J Pediatr 172, 707–710. https://doi.org/10.1007/s00431-012-1852-z

52. Foley, A.R., Menezes, M.P., Pandraud, A., Gonzalez, M.A., Al-Odaib, A., Abrams, A.J., Sugano, K., Yonezawa, A., Manzur, A.Y., Burns, J., Hughes, I., McCullagh, B.G., Jungbluth, H., Lim, M.J., Lin, J.P., Megarbane, A., Urtizberea, J.A., Shah, A.H., Antony, J., Webster, R., Broomfield, A., Ng, J., Mathew, A.A., O'Byrne, J.J., Forman, E., Scoto, M., Prasad, M., O'Brien, K., Olpin, S., Oppenheim, M., Hargreaves, I., Land, J.M., Wang, M.X., Carpenter, K., Horvath, R., Straub, V., Lek, M., Gold, W., Farrell, M.O., Brandner, S., Phadke, R., Matsubara, K., McGarvey, M.L., Scherer, S.S., Baxter, P.S., King, M.D., Clayton, P., Rahman, S., Reilly, M.M., Ouvrier, R.A., Christodoulou, J., Züchner, S., Muntoni, F., Houlden, H., 2014. Treatable childhood neuronopathy caused by mutations in riboflavin transporter RFVT2. Brain 137, 44–56. https://doi.org/10.1093/brain/awt315

53. Freneaux, E., Sheffield, V.C., Molin, L., Shires, A., Rhead, W.J., 1992. Glutaric acidemia type II. Heterogeneity in beta-oxidation flux, polypeptide synthesis, and complementary DNA mutations in the alpha subunit of electron transfer flavoprotein in eight patients. J Clin Invest 90, 1679–1686. https://doi.org/10.1172/jci116040

54. Frerman, F.E., Goodman, S.I., 2001. Defects of electron transfer flavoprotein and electron transfer flavoprotein-ubiquinone oxidoreductase: glutaric acidemia type II, in: Scriver, C.R., Sly, W.S., Childs, B., Beaudet, A.L., Valle, D., Kinzler, K.W., Vogelstein, B. (Eds.), The Metabolic and Molecular Bases of Inherited Disease. https://doi.org/10.1036/ommbid.131

55. Fu, H.X., Liu, X.Y., Wang, Z.Q., Jin, M., Wang, D.N., He, J.J., Lin, M.T., Wang, N., 2016. Significant clinical heterogeneity with similar ETFDH genotype in three Chinese patients with late-onset multiple acyl-CoA dehydrogenase deficiency. Neurol Sci 37, 1099–1105. https://doi.org/10.1007/s10072-016-2549-2
56. García-Villoria, J., de Azua, B., Tort, F., Mosegaard, S., Ugarteburu, O., Texidó, L., Morales-Romero, B., Olsen, R.K.J., Ribes, A., 2018. FLAD1, encoding FAD synthase, is mutated in a patient with myopathy, scoliosis and cataracts. Clin Genet 94, 592–593. https://doi.org/10.1111/cge.13452
57. Garg, M., Kulkarni, S.D., Hegde, A.U., Shah, K.N., 2018. Riboflavin treatment in genetically proven Brown-Vialetto-Van Laere syndrome. J Pediatr Neurosci 13, 471–473. https://doi.org/10.4103/jpn.Jpn_131_17
58. Gautschi, M., Weisstanner, C., Slotboom, J., Nava, E., Zürcher, T., Nuoffer, J.M., 2015. Highly efficient ketone body treatment in multiple acyl-CoA dehydrogenase deficiency-related leukodystrophy. Pediatr Res 77, 91–98. https://doi.org/10.1038/pr.2014.154
59. Gempel, K., Topaloglu, H., Talim, B., Schneiderat, P., Schoser, B.G., Hans, V.H., Pálmafy, B., Kale, G., Tokatli, A., Quinzii, C., Hirano, M., Naini, A., DiMauro, S., Prokisch, H., Lochmüller, H., Horvath, R., 2007. The myopathic form of coenzyme Q10 deficiency is caused by mutations in the electron-transferring-flavoprotein dehydrogenase (ETFDH) gene. Brain 130, 2037–2044. https://doi.org/10.1093/brain/awm054
60. Goh, L.L., Lee, Y., Tan, E.S., Lim, J.S.C., Lim, C.W., Dalan, R., 2018. Patient with multiple acyl-CoA dehydrogenase deficiency disease and ETFDH mutations benefits from riboflavin therapy: a case report. BMC Med Genomics 11, 37. https://doi.org/10.1186/s12920-018-0356-8
61. Gonzalez-Perez, P., Torre, M., Helgager, J., Amato, A.A., 2019. Proximal muscle weakness. Pr. Neurol 19, 321–325. https://doi.org/10.1136/practneurol-2019-002204
62. Goodman, S.I., Binard, R.J., Woontner, M.R., Frerman, F.E., 2002. Glutaric acidemia type II: gene structure and mutations of the electron transfer flavoprotein:ubiquinone oxidoreductase (ETF:QO) gene. Mol Genet Metab 77, 86–90. https://doi.org/10.1016/s1096-7192(02)00138-5
63. Gorcenco, S., Vaz, F.M., Tracewska-Siemiatkowska, A., Tranebjærg, L., Cremers, F.P.M., Ygland, E., Kicsi, J., Rendtorff, N.D., Möller, C., Kjellström, U., Andréasson, S., Puschmann, A., 2019. Oral therapy for riboflavin transporter deficiency - what is the regimen of choice? Park. Relat Disord 61, 245–247. https://doi.org/10.1016/j.parkreldis.2018.10.017

64. Gorukmez, O., Gorukmez, O., Sag, S.O., Erdol, S., Saglam, H., Yakut, T., 2015. Novel mutation of the electron transferring flavoprotein dehydrogenase (ETFDH) gene in the isolated myopathic form of coenzyme Q10 deficiency. Genet Couns 26, 259–262.
65. Gowda, V.K., Udhayabanu, T., Varalakshmi, P., Srinivasan, V.M., Ashokkumar, B., 2018. Fazio-Londe syndrome in siblings from India with different phenotypes. Brain Dev 40, 582–586. https://doi.org/10.1016/j.braindev.2018.02.010
66. Green, P., Wiseman, M., Crow, Y.J., Houlden, H., Riphagen, S., Lin, J.P., Raymond, F.L., Childs, A.M., Sheridan, E., Edwards, S., Josifova, D.J., 2010. Brown-Vialetto-Van Laere syndrome, a ponto-bulbar palsy with deafness, is caused by mutations in C20orf54. Am J Hum Genet 86, 485–489. https://doi.org/10.1016/j.ajhg.2010.02.006
67. Gregersen, N., Andresen, B.S., Pedersen, C.B., Olsen, R.K., Corydon, T.J., Bross, P., 2008. Mitochondrial fatty acid oxidation defects--remaining challenges. J Inherit Metab Dis 31, 643–657. https://doi.org/10.1007/s10545-008-0990-y
68. Grünert, S.C., 2014. Clinical and genetical heterogeneity of late-onset multiple acyl-coenzyme A dehydrogenase deficiency. Orphanet J Rare Dis 9, 117. https://doi.org/10.1186/s13023-014-0117-5
69. Gudbjartsson, D.F., Helgason, H., Gudjonsson, S.A., Zink, F., Oddson, A., Gylfason, A., Besenbacher, S., Magnusson, G., Halldorsson, B. V, Hjartarson, E., Sigurdsson, G.T., Stacey, S.N., Frigge, M.L., Holm, H., Saemundsdottir, J., Helgadottir, H.T., Johannsdottir, H., Sigfusson, G., Thorgeirsson, G., Sverrisson, J.T., Gretarsdottir, S., Walters, G.B., Rafnar, T., Thjodleifsson, B., Bjornsson, E.S., Olafsson, S., Thorarinsdottir, H., Steingrimsdottir, T., Gudmundsdottir, T.S., Theodors, A., Jonasson, J.G., Sigurdsson, A., Bjornsdottir, G., Jonsson, J.J., Thorarensen, O., Ludvigsson, P., Gudbjartsson, H., Eyjolfsson, G.I., Sigurdardottir, O., Olafsson, I., Arnar, D.O., Magnusson, O.T., Kong, A., Masson, G., Thorsteinsdottir, U., Helgason, A., Sulem, P., Stefansson, K., 2015. Large-scale whole-genome sequencing of the Icelandic population. Nat Genet 47, 435–444. https://doi.org/10.1038/ng.3247
70. Guissart, C., Drouot, N., Oncel, I., Leheup, B., Gershoni-Barush, R., Muller, J., Ferdinandusse, S., Larrieu, L., Anheim, M., Arslan, E.A., Claustres, M., Tranchant, C., Topaloglu, H., Koenig, M., 2016. Genes for spinocerebellar ataxia with blindness and deafness (SCABD/SCAR3, MIM# 271250 and SCABD2). Eur J Hum Genet 24, 1154–1159. https://doi.org/10.1038/ejhg.2015.259
71. Haack, T.B., Makowski, C., Yao, Y., Graf, E., Hempel, M., Wieland, T., Tauer, U., Ahting, U., Mayr, J.A., Freisinger, P., Yoshimatsu, H., Inui, K., Strom, T.M., Meitinger, T., Yonezawa, A., Prokisch, H., 2012. Impaired riboflavin transport due to missense

mutations in SLC52A2 causes Brown-Vialetto-Van Laere syndrome. J Inherit Metab Dis 35, 943–948. https://doi.org/10.1007/s10545-012-9513-y

72. Hackl, A., Mehler, K., Gottschalk, I., Vierzig, A., Eydam, M., Hauke, J., Beck, B.B., Liebau, M.C., Ensenauer, R., Weber, L.T., Habbig, S., 2017. Disorders of fatty acid oxidation and autosomal recessive polycystic kidney disease-different clinical entities and comparable perinatal renal abnormalities. Pediatr Nephrol 32, 791–800. https://doi.org/10.1007/s00467-016-3556-5
73. Han, J., Song, X., Lu, S., Ji, G., Xie, Y., Wu, H., 2019. Adolescent hyperuricemia with lipid storage myopathy: a clinical study. Med Sci Monit 25, 9103–9111. https://doi.org/10.12659/msm.918841
74. Hellebrekers, D., Sallevelt, S., Theunissen, T.E.J., Hendrickx, A.T.M., Gottschalk, R.W., Hoeijmakers, J.G.J., Habets, D.D., Bierau, J., Schoonderwoerd, K.G., Smeets, H.J.M., 2017. Novel SLC25A32 mutation in a patient with a severe neuromuscular phenotype. Eur J Hum Genet 25, 886–888. https://doi.org/10.1038/ejhg.2017.62
75. Henderson, M.J., Evans, C., Patterson, A., Cleary, M., Haywood, T., Thopte, I., 2002. Late presenting riboflavin responsive multiple acyl-CoA dehydrogenase deficiency with unusual features. J Inherit Metab Dis 25, 74.
76. Henriques, B.J., Fisher, M.T., Bross, P., Gomes, C.M., 2011. A polymorphic position in electron transfer flavoprotein modulates kinetic stability as evidenced by thermal stress. FEBS Lett 585, 505–510. https://doi.org/10.1016/j.febslet.2011.01.002
77. Henriques, B.J., Lucas, T.G., Martins, E., Gaspar, A., Bandeira, A., Nogueira, C., Brandão, O., Rocha, H., Vilarinho, L., Gomes, C.M., 2019. Molecular and clinical investigations on Portuguese patients with multiple acyl-CoA dehydrogenase deficiency. Curr Mol Med 19, 487–493. https://doi.org/10.2174/1566524019666190507114748
78. Henriques, B.J., Rodrigues, J. V, Olsen, R.K., Bross, P., Gomes, C.M., 2009. Role of flavinylation in a mild variant of multiple acyl-CoA dehydrogenation deficiency: a molecular rationale for the effects of riboflavin supplementation. J Biol Chem 284, 4222–4229. https://doi.org/10.1074/jbc.M805719200
79. Ho, G., Yonezawa, A., Masuda, S., Inui, K., Sim, K.G., Carpenter, K., Olsen, R.K., Mitchell, J.J., Rhead, W.J., Peters, G., Christodoulou, J., 2011. Maternal riboflavin deficiency, resulting in transient neonatal-onset glutaric aciduria type 2, is caused by a microdeletion in the riboflavin transporter gene GPR172B. Hum Mutat 32, E1976-84. https://doi.org/10.1002/humu.21399
80. Hong, D.J., Zhu, M., Zhu, Z.J., Cong, L., Zhong, S.S., Liu, L., Zhang, J., 2019. Clinical and muscle magnetic resonance image findings in patients with late-onset

multiple acyl-CoA dehydrogenase deficiency. Chin Med J 132, 275–284. https://doi.org/10.1097/cm9.0000000000000032

81. Hong, D., Yu, Y., Wang, Y., Xu, Y., Zhang, J., 2018. Acute-onset multiple acyl-CoA dehydrogenase deficiency mimicking Guillain-Barré syndrome: two cases report. BMC Neurol 18, 219. https://doi.org/10.1186/s12883-018-1221-2
82. Horoz, O.O., Mungan, N.O., Yildizdas, D., Hergüner, Ö., Ceylaner, S., Kör, D., Waterham, H., Coskun, T., 2016. Brown-Vialetto-Van Laere syndrome: two siblings with a new mutation and dramatic therapeutic effect of high-dose riboflavin. J Pediatr Endocrinol Metab 29, 227–231. https://doi.org/10.1515/jpem-2015-0198
83. Hossain, M.A., Obaid, A., Rifai, M., Alem, H., Hazwani, T., Al Shehri, A., Alfadhel, M., Eto, Y., Eyaid, W., 2017. Early onset of Fazio-Londe syndrome: the first case report from the Arabian Peninsula. Hum Genome Var 4, 17018. https://doi.org/10.1038/hgv.2017.18
84. Hu, G., Zeng, J., Wang, C., Zhou, W., Jia, Z., Yang, J., Zheng, B., 2020. A synonymous variant c.579A>G in the ETFDH gene caused exon skipping in a patient with late-onset multiple acyl-CoA dehydrogenase deficiency: a case report. Front Pediatr 8, 118. https://doi.org/10.3389/fped.2020.00118
85. Hu, X.M., Li, L., 2019. Multiple acyl-CoA dehydrogenase deficiency in an infant. Indian J Pediatr 86, 194–195. https://doi.org/10.1007/s12098-018-2768-z
86. Hu, X., Li, L., Mi, R., Zhao, Y., Li, C., 2018. A family affected by multiple acyl-CoA dehydrogenase deficiency due to compound heterozygous mutations on ETFDH and literature review. Chinese J. Neonatol. 33, 205–209. https://doi.org/10.3760/cma.j.issn.2096-2932.2018.03.012
87. Indo, Y., Glassberg, R., Yokota, I., Tanaka, K., 1991. Molecular characterization of variant alpha-subunit of electron transfer flavoprotein in three patients with glutaric acidemia type II--and identification of glycine substitution for valine-157 in the sequence of the precursor, producing an unstable mature protein in a patient. Am J Hum Genet 49, 575–580.
88. Ishii, K., Komaki, H., Ohkuma, A., Nishino, I., Nonaka, I., Sasaki, M., 2010. Central nervous system and muscle involvement in an adolescent patient with riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency. Brain Dev 32, 669–672. https://doi.org/10.1016/j.braindev.2009.08.008
89. Işıkay, S., Yaman, A., Ceylaner, S., 2017. An infant with glutaric aciduria type IIc diagnosed with a novel mutation. Turk J Pediatr 59, 315–317. https://doi.org/10.24953/turkjped.2017.03.013
90. Izumi, R., Suzuki, N., Nagata, M., Hasegawa, T., Abe, Y., Saito, Y., Mochizuki, H., Tateyama, M., Aoki, M., 2011. A case of late onset riboflavin-responsive multiple

acyl-CoA dehydrogenase deficiency manifesting as recurrent rhabdomyolysis and acute renal failure. Intern Med 50, 2663–2668. https://doi.org/10.2169/internalmedicine.50.5172

91. Janeiro, P., Jotta, R., Ramos, R., Florindo, C., Ventura, F. V, Vilarinho, L., Tavares de Almeida, I., Gaspar, A., 2019. Follow-up of fatty acid β-oxidation disorders in expanded newborn screening era. Eur J Pediatr 178, 387–394. https://doi.org/10.1007/s00431-018-03315-2

92. Johnson, J.O., Gibbs, J.R., Megarbane, A., Urtizberea, J.A., Hernandez, D.G., Foley, A.R., Arepalli, S., Pandraud, A., Simón-Sánchez, J., Clayton, P., Reilly, M.M., Muntoni, F., Abramzon, Y., Houlden, H., Singleton, A.B., 2012. Exome sequencing reveals riboflavin transporter mutations as a cause of motor neuron disease. Brain 135, 2875–2882. https://doi.org/10.1093/brain/aws161

93. Johnson, J.O., Gibbs, J.R., van Maldergem, L., Houlden, H., Singleton, A.B., 2010. Exome sequencing in Brown-Vialetto-Van Laere syndrome. Am J Hum Genet 87, 567–570. https://doi.org/10.1016/j.ajhg.2010.05.021

94. Kaminsky, P., Acquaviva-Bourdain, C., Jonas, J., Pruna, L., Chaloub, G.E., Rigal, O., Grignon, Y., Vianey-Saban, C., 2011. Subacute myopathy in a mature patient due to multiple acyl-coenzyme A dehydrogenase deficiency. Muscle Nerve 43, 444–446. https://doi.org/10.1002/mus.21881

95. Kim, S.Y., Ko, J.M., Choi, S.A., Cho, A., Lee, J.S., Lim, B.C., Kim, K.J., Chae, J.H., 2018a. A 3-month-old boy with progressive weakness. Brain Pathol 28, 773–774. https://doi.org/10.1111/bpa.12651

96. Kim, Y.J., Ko, J.M., Song, J., Lee, K.A., 2018b. Clinical features of multiple acyl-CoA dehydrogenase deficiency with ETFDH variants in the first Korean cases. Ann Lab Med 38, 616–618. https://doi.org/10.3343/alm.2018.38.6.616

97. Kong, L.E., Zhuang, S.Z., Li, C.M., Chen, L.K., Zhang, T.T., Qiu, J.H., 2018. Study of ETFDH gene mutations in a family with riboflavin-responsive lipid storage myopathy. J. Apoplexy Nerv. Dis. 35, 927–929.

98. Köppel, S., Gottschalk, J., Hoffmann, G.F., Waterham, H.R., Blobel, H., Kölker, S., 2006. Late-onset multiple acyl-CoA dehydrogenase deficiency: a frequently missed diagnosis? Neurology 67, 1519. https://doi.org/10.1212/01.wnl.0000240065.35635.a6

99. Koy, A., Pillekamp, F., Hoehn, T., Waterham, H., Klee, D., Mayatepek, E., Assmann, B., 2012. Brown-Vialetto-Van Laere syndrome: a riboflavin-unresponsive patient with a novel mutation in the C20orf54 gene. Pediatr Neurol 46, 407–409. https://doi.org/10.1016/j.pediatrneurol.2012.03.008

100. Lan, M.Y., Fu, M.H., Liu, Y.F., Huang, C.C., Chang, Y.Y., Liu, J.S., Peng, C.H., Chen, S.S., 2010. High frequency of ETFDH c.250G>A mutation in Taiwanese

patients with late-onset lipid storage myopathy. Clin Genet 78, 565–569. https://doi.org/10.1111/j.1399-0004.2010.01421.x

101. Law, L.K., Tang, N.L., Hui, J., Fung, S.L., Ruiter, J., Wanders, R.J., Fok, T.F., Lam, C.W., 2009. Novel mutations in ETFDH gene in Chinese patients with riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency. Clin Chim Acta 404, 95–99. https://doi.org/10.1016/j.cca.2009.02.015

102. Lee, H.C., Lai, C.K., Siu, T.S., Yuen, Y.P., Chan, K.Y., Chan, A.Y., Tam, S., Mak, C.M., Lam, C.W., 2010. Role of postmortem genetic testing demonstrated in a case of glutaric aciduria type II. Diagn Mol Pathol 19, 184–186. https://doi.org/10.1097/PDM.0b013e3181c9a8a8

103. Li, X.M., 2015. Clinical characteristic of lipid deposit disease in Jiangxi area and ETFA, ETFB, ETFDH gene mutation analysis. Nanchan University.

104. Liang, W.C., Ohkuma, A., Hayashi, Y.K., López, L.C., Hirano, M., Nonaka, I., Noguchi, S., Chen, L.H., Jong, Y.J., Nishino, I., 2009. ETFDH mutations, CoQ10 levels, and respiratory chain activities in patients with riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency. Neuromuscul Disord 19, 212–216. https://doi.org/10.1016/j.nmd.2009.01.008

105. Lin, Y., Zheng, Q., Zheng, T., Zheng, Z., Lin, W., Fu, Q., 2019. Expanded newborn screening for inherited metabolic disorders and genetic characteristics in a southern Chinese population. Clin Chim Acta 494, 106–111. https://doi.org/10.1016/j.cca.2019.03.1622

106. Liu, X.Y., Wang, Z.Q., Wang, D.N., Lin, M.T., Wang, N., 2016. A historical cohort study on the efficacy of glucocorticoids and riboflavin among patients with late-onset multiple acyl-CoA dehydrogenase deficiency. Chin Med J 129, 142–146. https://doi.org/10.4103/0366-6999.173438

107. Lu, J., Ji, L., 2014. [Mutation analysis for a family affected with riboflavin responsive-multiple acyl-CoA dehydrogenase deficiency]. Zhonghua Yi Xue Yi Chuan Xue Za Zhi 31, 428–432. https://doi.org/10.3760/cma.j.issn.1003-9406.2014.04.004

108. Macchione, F., Salviati, L., Bordugo, A., Vincenzi, M., Camilot, M., Teofoli, F., Pancheri, E., Zordan, R., Bertolin, C., Rossi, S., Vattemi, G., Tonin, P., 2020. Multiple acyl-CoA dehydrogenase deficiency in elderly carriers. J Neurol 267, 1414–1419. https://doi.org/10.1007/s00415-020-09729-z

109. Madsen, K.L., Preisler, N., Buch, A.E., Stemmerik, M.G., Laforêt, P., Vissing, J., 2019. Impaired fat oxidation during exercise in multiple acyl-CoA dehydrogenase deficiency. JIMD Rep 46, 79–84. https://doi.org/10.1002/jmd2.12024

110. Maillart, E., Acquaviva-Bourdain, C., Rigal, O., Brivet, M., Jardel, C., Lombès, A., Eymard, B., Vianey-Saban, C., Laforêt, P., 2010. [Multiple acyl-CoA dehydrogenase

deficiency (MADD): a curable cause of genetic muscular lipidosis]. Rev Neurol 166, 289–294. https://doi.org/10.1016/j.neurol.2009.05.009

111. Malafronte, P., Clark, H.B., Castaneda-Sanchez, I., Raisanen, J., Hatanpaa, K.J., 2013. Brown-Vialetto-Van Laere syndrome: clinical and neuropathologic findings with immunohistochemistry for C20orf54 in three affected patients. Pediatr Dev Pathol 16, 364–371. https://doi.org/10.2350/13-02-1299-cr.1

112. Manole, A., Jaunmuktane, Z., Hargreaves, I., Ludtmann, M.H.R., Salpietro, V., Bello, O.D., Pope, S., Pandraud, A., Horga, A., Scalco, R.S., Li, A., Ashokkumar, B., Lourenço, C.M., Heales, S., Horvath, R., Chinnery, P.F., Toro, C., Singleton, A.B., Jacques, T.S., Abramov, A.Y., Muntoni, F., Hanna, M.G., Reilly, M.M., Revesz, T., Kullmann, D.M., Jepson, J.E.C., Houlden, H., 2017. Clinical, pathological and functional characterization of riboflavin-responsive neuropathy. Brain 140, 2820–2837. https://doi.org/10.1093/brain/awx231

113. Menezes, M.P., O'Brien, K., Hill, M., Webster, R., Antony, J., Ouvrier, R., Birman, C., Gardner-Berry, K., 2016. Auditory neuropathy in Brown-Vialetto-Van Laere syndrome due to riboflavin transporter RFVT2 deficiency. Dev Med Child Neurol 58, 848–854. https://doi.org/10.1111/dmcn.13084

114. Missaglia, S., Pegoraro, V., Marozzo, R., Tavian, D., Angelini, C., 2020. Correlation between ETFDH mutations and dysregulation of serum myomiRs in MADD patients. Eur J Transl Myol 30, 8880. https://doi.org/10.4081/ejtm.2019.8880

115. Missaglia, S., Tavian, D., Moro, L., Angelini, C., 2018. Characterization of two ETFDH mutations in a novel case of riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency. Lipids Heal. Dis 17, 254. https://doi.org/10.1186/s12944-018-0903-5

116. Mosegaard, S., Bruun, G.H., Flyvbjerg, K.F., Bliksrud, Y.T., Gregersen, N., Dembic, M., Annexstad, E., Tangeraas, T., Olsen, R.K.J., Andresen, B.S., 2017. An intronic variation in SLC52A1 causes exon skipping and transient riboflavin-responsive multiple acyl-CoA dehydrogenation deficiency. Mol Genet Metab 122, 182–188. https://doi.org/10.1016/j.ymgme.2017.10.014

117. Muru, K., Reinson, K., Künnapas, K., Lilleväli, H., Nochi, Z., Mosegaard, S., Pajusalu, S., Olsen, R.K.J., Õunap, K., 2019. FLAD1-associated multiple acyl-CoA dehydrogenase deficiency identified by newborn screening. Mol Genet Genomic Med 7, e915. https://doi.org/10.1002/mgg3.915

118. Navarrete, R., Leal, F., Vega, A.I., Morais-López, A., Garcia-Silva, M.T., Martín-Hernández, E., Quijada-Fraile, P., Bergua, A., Vives, I., García-Jiménez, I., Yahyaoui, R., Pedrón-Giner, C., Belanger-Quintana, A., Stanescu, S., Cañedo, E., García-Campos, O., Bueno-Delgado, M., Delgado-Pecellín, C., Vitoria, I., Rausell,

M.D., Balmaseda, E., Couce, M.L., Desviat, L.R., Merinero, B., Rodríguez-Pombo, P., Ugarte, M., Pérez-Cerdá, C., Pérez, B., 2019. Value of genetic analysis for confirming inborn errors of metabolism detected through the Spanish neonatal screening program. Eur J Hum Genet 27, 556–562. https://doi.org/10.1038/s41431-018-0330-0

119. Nilipour, Y., Fatehi, F., Sanatinia, S., Bradshaw, A., Duff, J., Lochmüller, H., Horvath, R., Nafissi, S., 2020. Multiple acyl-coenzyme A dehydrogenase deficiency shows a possible founder effect and is the most frequent cause of lipid storage myopathy in Iran. J Neurol Sci 411, 116707. https://doi.org/10.1016/j.jns.2020.116707

120. Nimmo, G.A.M., Ejaz, R., Cordeiro, D., Kannu, P., Mercimek-Andrews, S., 2018. Riboflavin transporter deficiency mimicking mitochondrial myopathy caused by complex II deficiency. Am J Med Genet A 176, 399–403. https://doi.org/10.1002/ajmg.a.38530

121. Ohkuma, A., Noguchi, S., Sugie, H., Malicdan, M.C., Fukuda, T., Shimazu, K., López, L.C., Hirano, M., Hayashi, Y.K., Nonaka, I., Nishino, I., 2009. Clinical and genetic analysis of lipid storage myopathies. Muscle Nerve 39, 333–342. https://doi.org/10.1002/mus.21167

122. Olsen, R.K.J., Koňaříková, E., Giancaspero, T.A., Mosegaard, S., Boczonadi, V., Mataković, L., Veauville-Merllié, A., Terrile, C., Schwarzmayr, T., Haack, T.B., Auranen, M., Leone, P., Galluccio, M., Imbard, A., Gutierrez-Rios, P., Palmfeldt, J., Graf, E., Vianey-Saban, C., Oppenheim, M., Schiff, M., Pichard, S., Rigal, O., Pyle, A., Chinnery, P.F., Konstantopoulou, V., Möslinger, D., Feichtinger, R.G., Talim, B., Topaloglu, H., Coskun, T., Gucer, S., Botta, A., Pegoraro, E., Malena, A., Vergani, L., Mazzà, D., Zollino, M., Ghezzi, D., Acquaviva, C., Tyni, T., Boneh, A., Meitinger, T., Strom, T.M., Gregersen, N., Mayr, J.A., Horvath, R., Barile, M., Prokisch, H., 2016. Riboflavin-responsive and -non-responsive mutations in FAD synthase cause multiple acyl-CoA dehydrogenase and combined respiratory-chain deficiency. Am J Hum Genet 98, 1130–1145. https://doi.org/10.1016/j.ajhg.2016.04.006

123. Olsen, R.K., Andresen, B.S., Christensen, E., Bross, P., Skovby, F., Gregersen, N., 2003. Clear relationship between ETF/ETFDH genotype and phenotype in patients with multiple acyl-CoA dehydrogenation deficiency. Hum Mutat 22, 12–23. https://doi.org/10.1002/humu.10226

124. Olsen, R.K., Andresen, B.S., Christensen, E., Mandel, H., Skovby, F., Nielsen, J.P., Knudsen, I., Vianey-Saban, C., Simonsen, H., Gregersen, N., 2005. DNA-based prenatal diagnosis for severe and variant forms of multiple acyl-CoA dehydrogenation deficiency. Prenat Diagn 25, 60–64. https://doi.org/10.1002/pd.983

125. Olsen, R.K., Brøner, S., Sabaratnam, R., Doktor, T.K., Andersen, H.S., Bruun, G.H., Gahrn, B., Stenbroen, V., Olpin, S.E., Dobbie, A., Gregersen, N., Andresen, B.S., 2014. The ETFDH c.158A>G variation disrupts the balanced interplay of ESE- and ESS-binding proteins thereby causing missplicing and multiple acyl-CoA dehydrogenation deficiency. Hum Mutat 35, 86–95. https://doi.org/10.1002/humu.22455

126. Olsen, R.K., Olpin, S.E., Andresen, B.S., Miedzybrodzka, Z.H., Pourfarzam, M., Merinero, B., Frerman, F.E., Beresford, M.W., Dean, J.C., Cornelius, N., Andersen, O., Oldfors, A., Holme, E., Gregersen, N., Turnbull, D.M., Morris, A.A., 2007. ETFDH mutations as a major cause of riboflavin-responsive multiple acyl-CoA dehydrogenation deficiency. Brain 130, 2045–2054. https://doi.org/10.1093/brain/awm135

127. Olsen, R.K., Pourfarzam, M., Morris, A.A., Dias, R.C., Knudsen, I., Andresen, B.S., Gregersen, N., Olpin, S.E., 2004. Lipid-storage myopathy and respiratory insufficiency due to ETFQO mutations in a patient with late-onset multiple acyl-CoA dehydrogenation deficiency. J Inherit Metab Dis 27, 671–678. https://doi.org/10.1023/b:boli.0000042986.10291.e9

128. Ou, M., Zhu, L., Zhang, Y., Zhang, Y., Zhou, J., Zhang, Y., Chen, X., Yang, L., Li, T., Su, X., Hu, Q., Wang, W., 2020. A novel electron transfer flavoprotein dehydrogenase (ETFDH) gene mutation identified in a newborn with glutaric acidemia type II: a case report of a Chinese family. BMC Med Genet 21, 98. https://doi.org/10.1186/s12881-020-00995-2

129. Pan, X.Q., Chang, X.L., Zhang, W., Meng, H.X., Zhang, J., Shi, J.Y., Guo, J.H., 2020. Late-onset multiple acyl-CoA dehydrogenase deficiency with cardiac syncope: a case report. World J Clin Cases 8, 995–1001. https://doi.org/10.12998/wjcc.v8.i5.995

130. Peake, R.W., Kozakewich, H.P., 2017. A term newborn with respiratory distress, acidosis, and hypoglycemia. Clin Chem 63, 613–615. https://doi.org/10.1373/clinchem.2016.267328

131. Peng, Y., Zhu, M., Zheng, J., Zhu, Y., Li, X., Wei, C., Hong, D., 2015. Bent spine syndrome as an initial manifestation of late-onset multiple acyl-CoA dehydrogenase deficiency: a case report and literature review. BMC Neurol 15, 114. https://doi.org/10.1186/s12883-015-0380-7

132. Petrovski, S., Shashi, V., Petrou, S., Schoch, K., McSweeney, K.M., Dhindsa, R.S., Krueger, B., Crimian, R., Case, L.E., Khalid, R., El-Dairi, M.A., Jiang, Y.H., Mikati, M.A., Goldstein, D.B., 2015. Exome sequencing results in successful riboflavin treatment of a rapidly progressive neurological condition. Cold Spring Harb Mol Case Stud 1, a000257. https://doi.org/10.1101/mcs.a000257

133. Pollard, L.M., Williams, N.R., Espinoza, L., Wood, T.C., Spector, E.B., Schroer, R.J., Holden, K.R., 2010. Diagnosis, treatment, and long-term outcomes of late-onset (type III) multiple acyl-CoA dehydrogenase deficiency. J Child Neurol 25, 954–960. https://doi.org/10.1177/0883073809351984
134. Pontoizeau, C., Habarou, F., Brassier, A., Veauville-Merllié, A., Grisel, C., Arnoux, J.B., Vianey-Saban, C., Barouki, R., Chadefaux-Vekemans, B., Acquaviva, C., de Lonlay, P., Ottolenghi, C., 2016. Hyperprolinemia in type 2 glutaric aciduria and MADD-like profiles. JIMD Rep 27, 39–45. https://doi.org/10.1007/8904_2015_481
135. Purevjav, E., Kimura, M., Takusa, Y., Ohura, T., Tsuchiya, M., Hara, N., Fukao, T., Yamaguchi, S., 2002. Molecular study of electron transfer flavoprotein alpha-subunit deficiency in two Japanese children with different phenotypes of glutaric acidemia type II. Eur J Clin Invest 32, 707–712. https://doi.org/10.1046/j.1365-2362.2002.01045.x
136. Rabbani, B., Bakhshandeh, M.K., Navaeifar, M.R., Abbaskhanian, A., Soveizi, M., Geravandpoor, S., Mahdieh, N., 2020. Brown-Vialetto-Van Laere syndrome and Fazio-Londe syndrome: a novel mutation and in silico analyses. J Clin Neurosci 72, 342–349. https://doi.org/10.1016/j.jocn.2020.01.040
137. Rabenstein, M., Weis, J., Abicht, A., Fink, G.R., Lehmann, H.C., Wunderlich, G., 2020. [Multiple acyl-CoA dehydrogenase deficiency/glutaric aciduria type 2: difficult diagnosis, easy to treat]. Nervenarzt 91, 349–352. https://doi.org/10.1007/s00115-020-00886-0
138. Ramos, J.M., Martínez Pardo, M., García, M., Lorenzo, G., Merinero, B., Jiménez, A., Ugarte, M., Pallarés, E., Arenas, J., Campos, Y., Aparicio Meix, M., 1995. Miopatia lipidica por aciduria glutarica tipo II sensible a riboflavina. An Esp Pediatr 2, 207–210.
139. Rentzsch, P., Witten, D., Cooper, G.M., Shendure, J., Kircher, M., 2019. CADD: predicting the deleteriousness of variants throughout the human genome. Nucleic Acids Res. 47, D886–D894. https://doi.org/10.1093/nar/gky1016
140. Rizzo, F., Ramirez, A., Compagnucci, C., Salani, S., Melzi, V., Bordoni, A., Fortunato, F., Niceforo, A., Bresolin, N., Comi, G.P., Bertini, E., Nizzardo, M., Corti, S., 2017. Genome-wide RNA-seq of iPSC-derived motor neurons indicates selective cytoskeletal perturbation in Brown-Vialetto disease that is partially rescued by riboflavin. Sci Rep 7, 46271. https://doi.org/10.1038/srep46271
141. Rocha, H., Ferreira, R., Carvalho, J., Vitorino, R., Santa, C., Lopes, L., Gregersen, N., Vilarinho, L., Amado, F., 2011. Characterization of mitochondrial proteome in a severe case of ETF-QO deficiency. J Proteomics 75, 221–228. https://doi.org/10.1016/j.jprot.2011.04.025

142. Rosenbohm, A., Süssmuth, S.D., Kassubek, J., Müller, H.P., Pontes, C., Abicht, A., Bulst, S., Ludolph, A.C., Pinkhardt, E., 2014. Novel ETFDH mutation and imaging findings in an adult with glutaric aciduria type II. Muscle Nerve 49, 446–450. https://doi.org/10.1002/mus.23979
143. Ryder, B., Tolomeo, M., Nochi, Z., Colella, M., Barile, M., Olsen, R.K., Inbar-Feigenberg, M., 2019. A novel truncating FLAD1 variant, causing multiple acyl-CoA dehydrogenase deficiency (MADD) in an 8-year-old boy. JIMD Rep 45, 37–44. https://doi.org/10.1007/8904_2018_139
144. Santananukarn, M., Amornvit, J., Pasutharnchat, N., Jongpiputvanich, S., 2020. Needle EMG, a jigsaw to disclose lipid storage myopathy due to multiple acyl-CoA dehydrogenase deficiency. Am J Phys Med Rehabil 99, e71–e74. https://doi.org/10.1097/phm.0000000000001230
145. Saral, N.Y., Aksungar, F.B., Aktuglu-Zeybek, C., Coskun, J., Demirelce, O., Serteser, M., 2018. Glutaric acidemia type II patient with thalassemia minor and novel electron transfer flavoprotein-A gene mutations: a case report and review of literature. World J Clin Cases 6, 786–790. https://doi.org/10.12998/wjcc.v6.i14.786
146. Schiff, M., Froissart, R., Olsen, R.K., Acquaviva, C., Vianey-Saban, C., 2006. Electron transfer flavoprotein deficiency: functional and molecular aspects. Mol Genet Metab 88, 153–158. https://doi.org/10.1016/j.ymgme.2006.01.009
147. Schiff, M., Veauville-Merllié, A., Su, C.H., Tzagoloff, A., Rak, M., Ogier de Baulny, H., Boutron, A., Smedts-Walters, H., Romero, N.B., Rigal, O., Rustin, P., Vianey-Saban, C., Acquaviva-Bourdain, C., 2016. SLC25A32 mutations and riboflavin-responsive exercise intolerance. N Engl J Med 374, 795–797. https://doi.org/10.1056/NEJMc1513610
148. Schoonen, M., Smuts, I., Louw, R., Elson, J.L., van Dyk, E., Jonck, L.M., Rodenburg, R.J.T., van der Westhuizen, F.H., 2019. Panel-based nuclear and mitochondrial next-generation sequencing outcomes of an ethnically diverse pediatric patient cohort with mitochondrial disease. J Mol Diagn 21, 503–513. https://doi.org/10.1016/j.jmoldx.2019.02.002
149. Shi, K., Shi, Z., Yan, H., Wang, X., Yang, Y., Xiong, H., Gu, Q., Wu, Y., Jiang, Y., Wang, J., 2019. A Chinese pedigree with Brown-Vialetto-Van Laere syndrome due to two novel mutations of SLC52A2 gene: clinical course and response to riboflavin. BMC Med Genet 20, 76. https://doi.org/10.1186/s12881-019-0811-1
150. Shioya, A., Takuma, H., Yamaguchi, S., Ishii, A., Hiroki, M., Fukuda, T., Sugie, H., Shigematsu, Y., Tamaoka, A., 2014. Amelioration of acylcarnitine profile using bezafibrate and riboflavin in a case of adult-onset glutaric acidemia type 2 with novel

mutations of the electron transfer flavoprotein dehydrogenase (ETFDH) gene. J Neurol Sci 346, 350–352. https://doi.org/10.1016/j.jns.2014.08.040

151. Silva, A.M.S., Mendonça, R.H., Soares, D.C., Callegaro, D., Caldas, V.M., Perissinotti, I.N., Carvalho, M.S., Zanoteli, E., 2018. Pearls & oy-sters: a curable myopathy manifesting as exercise intolerance and respiratory failure. Neurology 91, 187–190. https://doi.org/10.1212/wnl.0000000000005867

152. Song, Y., Selak, M.A., Watson, C.T., Coutts, C., Scherer, P.C., Panzer, J.A., Gibbs, S., Scott, M.O., Willer, G., Gregg, R.G., Ali, D.W., Bennett, M.J., Balice-Gordon, R.J., 2009. Mechanisms underlying metabolic and neural defects in zebrafish and human multiple acyl-CoA dehydrogenase deficiency (MADD). PLoS One 4, e8329. https://doi.org/10.1371/journal.pone.0008329

153. Srour, M., Putorti, M.L., Schwartzentruber, J., Bolduc, V., Shevell, M.I., Poulin, C., O'Ferrall, E., Buhas, D., Majewski, J., Brais, B., 2014. Mutations in riboflavin transporter present with severe sensory loss and deafness in childhood. Muscle Nerve 50, 775–779. https://doi.org/10.1002/mus.24224

154. Sudo, Y., Sasaki, A., Wakabayashi, T., Numakura, C., Hayasaka, K., 2015. A novel ETFB mutation in a patient with glutaric aciduria type II. Hum Genome Var 1, 15016. https://doi.org/10.1038/hgv.2015.16

155. Sugai, F., Baba, K., Toyooka, K., Liang, W.C., Nishino, I., Yamadera, M., Sumi, H., Fujimura, H., Nishikawa, Y., 2012. Adult-onset multiple acyl CoA dehydrogenation deficiency associated with an abnormal isoenzyme pattern of serum lactate dehydrogenase. Neuromuscul Disord 22, 159–161. https://doi.org/10.1016/j.nmd.2011.08.004

156. Tang, Y., Ouyang, W., Jiang, T., Tang, L., Su, Y., Li, S., 2017. Clinical manifestation and gene analysis of multiple acyl-CoA dehydrogenase deficiency with transaminase increase. Chinese Pediatr. Integr. Tradit. West. Med. 9, 456–458. https://doi.org/10.3969/j.issn.1674-3865.2017.05.031

157. Taylor, R.W., Pyle, A., Griffin, H., Blakely, E.L., Duff, J., He, L., Smertenko, T., Alston, C.L., Neeve, V.C., Best, A., Yarham, J.W., Kirschner, J., Schara, U., Talim, B., Topaloglu, H., Baric, I., Holinski-Feder, E., Abicht, A., Czermin, B., Kleinle, S., Morris, A.A., Vassallo, G., Gorman, G.S., Ramesh, V., Turnbull, D.M., Santibanez-Koref, M., McFarland, R., Horvath, R., Chinnery, P.F., 2014. Use of whole-exome sequencing to determine the genetic basis of multiple mitochondrial respiratory chain complex deficiencies. Jama 312, 68–77. https://doi.org/10.1001/jama.2014.7184

158. Thulasi, V., Veerapandiyan, A., Pletcher, B.A., Tong, C.M., Ming, X., 2017. A case of Brown-Vialetto-Van Laere syndrome due to a novel mutation in SLC52A3 gene:

clinical course and response to riboflavin. Child Neurol Open 4, 2329048x17725610. https://doi.org/10.1177/2329048x17725610

159. Tong, F., Yan, R., Zhao, Z., 2017. One case report of late onset riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency. Chinese J. Child Heal. Care 25, 1186–1188.

160. Tong, W., Wang, Y., Lu, Y., Ye, T., Song, C., Xu, Y., Li, M., Ding, J., Duan, Y., Zhang, L., Gu, W., Zhao, X., Yang, X.A., Jin, D., 2018. Whole-exome sequencing helps the diagnosis and treatment in children with neurodevelopmental delay accompanied unexplained dyspnea. Sci Rep 8, 5214. https://doi.org/10.1038/s41598-018-23503-2

161. Touat, M., Michaud, M., Alamowitch, S., Maisonobe, T., Acquaviva-Bourdain, C., Laforêt, P., 2018. Encephalopathy associated with a reversible splenial lesion in riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency. Rev Neurol 174, 747–750. https://doi.org/10.1016/j.neurol.2017.12.004

162. Trakadis, Y., Kadlubowska, D., Barnes, R., Mitchell, J., Spector, E., Frerman, F., Melancon, S., 2012. Pregnancy of a patient with multiple acyl-CoA dehydrogenation deficiency (MADD). Mol Genet Metab 106, 491–494. https://doi.org/10.1016/j.ymgme.2012.05.001

163. Udhayabanu, T., Subramanian, V.S., Teafatiller, T., Gowda, V.K., Raghavan, V.S., Varalakshmi, P., Said, H.M., Ashokkumar, B., 2016. SLC52A2 [p.P141T] and SLC52A3 [p.N21S] causing Brown-Vialetto-Van Laere syndrome in an Indian patient: first genetically proven case with mutations in two riboflavin transporters. Clin Chim Acta 462, 210–214. https://doi.org/10.1016/j.cca.2016.09.022

164. van der Westhuizen, F.H., Smuts, I., Honey, E., Louw, R., Schoonen, M., Jonck, L.M., Dercksen, M., 2018. A novel mutation in ETFDH manifesting as severe neonatal-onset multiple acyl-CoA dehydrogenase deficiency. J Neurol Sci 384, 121–125. https://doi.org/10.1016/j.jns.2017.11.012

165. van Hove, J.L., Grünewald, S., Jaeken, J., Demaerel, P., Declercq, P.E., Bourdoux, P., Niezen-Koning, K., Deanfeld, J.E., Leonard, J. V, 2003. D,L-3-hydroxybutyrate treatment of multiple acyl-CoA dehydrogenase deficiency (MADD). Lancet 361, 1433–1435. https://doi.org/10.1016/s0140-6736(03)13105-4

166. van Rijt, W.J., Ferdinandusse, S., Giannopoulos, P., Ruiter, J.P.N., de Boer, L., Bosch, A.M., Huidekoper, H.H., Rubio-Gozalbo, M.E., Visser, G., Williams, M., Wanders, R.J.A., Derks, T.G.J., 2019. Prediction of disease severity in multiple acyl-CoA dehydrogenase deficiency: a retrospective and laboratory cohort study. J Inherit Metab Dis 42, 878–889. https://doi.org/10.1002/jimd.12147

167. van Rijt, W.J., Heiner-Fokkema, M.R., du Marchie Sarvaas, G.J., Waterham, H.R., Blokpoel, R.G., van Spronsen, F.J., Derks, T.G., 2014. Favorable outcome after physiologic dose of sodium-D,L-3-hydroxybutyrate in severe MADD. Pediatrics 134, e1224-8. https://doi.org/10.1542/peds.2013-4254

168. van Rijt, W.J., Jager, E.A., Allersma, D.P., Aktuğlu Zeybek, A.Ç., Bhattacharya, K., Debray, F.G., Ellaway, C.J., Gautschi, M., Geraghty, M.T., Gil-Ortega, D., Larson, A.A., Moore, F., Morava, E., Morris, A.A., Oishi, K., Schiff, M., Scholl-Bürgi, S., Tchan, M.C., Vockley, J., Witters, P., Wortmann, S.B., van Spronsen, F., van Hove, J.L.K., Derks, T.G.J., 2020. Efficacy and safety of D,L-3-hydroxybutyrate (D,L-3-HB) treatment in multiple acyl-CoA dehydrogenase deficiency. Genet Med 22, 908–916. https://doi.org/10.1038/s41436-019-0739-z

169. Vieira, P., Myllynen, P., Perhomaa, M., Tuominen, H., Keski-Filppula, R., Rytky, S., Risteli, L., Uusimaa, J., 2017. Riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency associated with hepatoencephalomyopathy and white matter signal abnormalities on brain MRI. Neuropediatrics 48, 194–198. https://doi.org/10.1055/s-0037-1601447

170. Wang, J., Wu, J.C., Yu, X.E., Han, Y.Z., Yang, R.M., 2018. Long-term outcomes of a patient with late-onset multiple acyl-CoA dehydrogenase deficiency caused by novel mutations in ETFDH: a case report. Med. 97, e13153. https://doi.org/10.1097/md.0000000000013153

171. Wang, Y., Zhao, D., Hong, D., Wang, Z., Yuan, Y., 2011b. Hot spot mutations in electron transfer flavoprotein dehydrogenase gene of riboflavin responsive lipid storage myopathy in 20 Chinese families. Chinese J. Neurol. 44, 309–313. https://doi.org/10.3760/cma.j.issn.1006-7876.2011.05.005

172. Wang, Z.Q., Chen, X.J., Murong, S.X., Wang, N., Wu, Z.Y., 2011a. Molecular analysis of 51 unrelated pedigrees with late-onset multiple acyl-CoA dehydrogenation deficiency (MADD) in southern China confirmed the most common ETFDH mutation and high carrier frequency of c.250G>A. J Mol Med 89, 569–576. https://doi.org/10.1007/s00109-011-0725-7

173. Wang, Z., Hong, D., Zhang, W., Li, W., Shi, X., Zhao, D., Yang, X., Lv, H., Yuan, Y., 2016. Severe sensory neuropathy in patients with adult-onset multiple acyl-CoA dehydrogenase deficiency. Neuromuscul Disord 26, 170–175. https://doi.org/10.1016/j.nmd.2015.12.002

174. Wasant, P., Kuptanon, C., Vattanavicharn, N., Liammongkolkul, S., Ratanarak, P., Sangruchi, T., Yamaguchi, S., 2010. Glutaric aciduria type 2, late onset type in Thai siblings with myopathy. Pediatr Neurol 43, 279–282. https://doi.org/10.1016/j.pediatrneurol.2010.05.018

175. Wen, B., Dai, T., Li, W., Zhao, Y., Liu, S., Zhang, C., Li, H., Wu, J., Li, D., Yan, C., 2010. Riboflavin-responsive lipid-storage myopathy caused by ETFDH gene mutations. J Neurol Neurosurg Psychiatry 81, 231–236. https://doi.org/10.1136/jnnp.2009.176404
176. Wen, B., Li, D., Li, W., Zhao, Y., Yan, C., 2015. Multiple acyl-CoA dehydrogenation deficiency as decreased acyl-carnitine profile in serum. Neurol Sci 36, 853–859. https://doi.org/10.1007/s10072-015-2197-y
177. Wen, B., Li, D., Shan, J., Liu, S., Li, W., Zhao, Y., Lin, P., Zheng, J., Li, D., Gong, Y., Yan, C., 2013. Increased muscle coenzyme Q10 in riboflavin responsive MADD with ETFDH gene mutations due to secondary mitochondrial proliferation. Mol Genet Metab 109, 154–160. https://doi.org/10.1016/j.ymgme.2013.04.007
178. Whitaker, C.H., Felice, K.J., Silvers, D., Wu, Q., 2015. Fulminant lipid storage myopathy due to multiple acyl-coenzyme a dehydrogenase deficiency. Muscle Nerve 52, 289–293. https://doi.org/10.1002/mus.24552
179. Wolfe, L.A., He, M., Vockley, J., Payne, N., Rhead, W., Hoppel, C., Spector, E., Gernert, K., Gibson, K.M., 2010. Novel ETF dehydrogenase mutations in a patient with mild glutaric aciduria type II and complex II-III deficiency in liver and muscle. J Inherit Metab Dis 33 Suppl 3, S481-7. https://doi.org/10.1007/s10545-010-9246-8
180. Woodcock, I.R., Menezes, M.P., Coleman, L., Yaplito-Lee, J., Peters, H., White, S.M., Stapleton, R., Phelan, D.G., Chong, B., Lunke, S., Stark, Z., Pitt, J., Ryan, M.M., Robertson, C., Yiu, E.M., 2018. Genetic, radiologic, and clinical variability in Brown-Vialetto-Van Laere syndrome. Semin Pediatr Neurol 26, 2–9. https://doi.org/10.1016/j.spen.2017.03.001
181. Xi, J., Wen, B., Lin, J., Zhu, W., Luo, S., Zhao, C., Li, D., Lin, P., Lu, J., Yan, C., 2014. Clinical features and ETFDH mutation spectrum in a cohort of 90 Chinese patients with late-onset multiple acyl-CoA dehydrogenase deficiency. J Inherit Metab Dis 37, 399–404. https://doi.org/10.1007/s10545-013-9671-6
182. Xu, H.L., Lian, Y.J., Chen, X., Zhang, L., Cheng, X., 2019. Two novel ETFDH mutations in a patient with lipid storage myopathy. Chin Med J 132, 1876–1878. https://doi.org/10.1097/cm9.0000000000000310
183. Xu, H., Chen, X., Lian, Y., Wang, S., Ji, T., Zhang, L., Li, S., 2020. Skin damage in a patient with lipid storage myopathy with a novel ETFDH mutation responsive to riboflavin. Int J Neurosci 1–7. https://doi.org/10.1080/00207454.2020.1730831
184. Xu, J., Li, D., Lv, J., Xu, X., Wen, B., Lin, P., Liu, F., Ji, K., Shan, J., Li, H., Li, W., Zhao, Y., Zhao, D., Pok, J.Y., Yan, C., 2018. ETFDH mutations and flavin adenine dinucleotide homeostasis disturbance are essential for developing riboflavin-

responsive multiple acyl-coenzyme A dehydrogenation deficiency. Ann Neurol 84, 659–673. https://doi.org/10.1002/ana.25338

185. Xue, Y., Zhou, Y., Zhang, K., Li, L., Kayoumu, A., Chen, L., Wang, Y., Lu, Z., 2017. Compound heterozygous mutations in electron transfer flavoprotein dehydrogenase identified in a young Chinese woman with late-onset glutaric aciduria type II. Lipids Heal. Dis 16, 185. https://doi.org/10.1186/s12944-017-0576-5

186. Yamada, K., Ito, M., Kobayashi, H., Hasegawa, Y., Fukuda, S., Yamaguchi, S., Taketani, T., 2019. Flavin adenine dinucleotide synthase deficiency due to FLAD1 mutation presenting as multiple acyl-CoA dehydrogenation deficiency-like disease: a case report. Brain Dev 41, 638–642. https://doi.org/10.1016/j.braindev.2019.04.002

187. Yamada, K., Kobayashi, H., Bo, R., Purevsuren, J., Mushimoto, Y., Takahashi, T., Hasegawa, Y., Taketani, T., Fukuda, S., Yamaguchi, S., 2017. Efficacy of bezafibrate on fibroblasts of glutaric acidemia type II patients evaluated using an in vitro probe acylcarnitine assay. Brain Dev 39, 48–57. https://doi.org/10.1016/j.braindev.2016.08.004

188. Yamada, K., Kobayashi, H., Bo, R., Takahashi, T., Purevsuren, J., Hasegawa, Y., Taketani, T., Fukuda, S., Ohkubo, T., Yokota, T., Watanabe, M., Tsunemi, T., Mizusawa, H., Takuma, H., Shioya, A., Ishii, A., Tamaoka, A., Shigematsu, Y., Sugie, H., Yamaguchi, S., 2016. Clinical, biochemical and molecular investigation of adult-onset glutaric acidemia type II: characteristics in comparison with pediatric cases. Brain Dev 38, 293–301. https://doi.org/10.1016/j.braindev.2015.08.011

189. Yamaguchi, S., Li, H., Purevsuren, J., Yamada, K., Furui, M., Takahashi, T., Mushimoto, Y., Kobayashi, H., Hasegawa, Y., Taketani, T., Fukao, T., Fukuda, S., 2012. Bezafibrate can be a new treatment option for mitochondrial fatty acid oxidation disorders: evaluation by in vitro probe acylcarnitine assay. Mol Genet Metab 107, 87–91. https://doi.org/10.1016/j.ymgme.2012.07.004

190. Yıldız, Y., Olsen, R.K.J., Sivri, H.S., Akçören, Z., Nygaard, H.H., Tokatlı, A., 2018. Post-mortem detection of FLAD1 mutations in 2 Turkish siblings with hypotonia in early infancy. Neuromuscul Disord 28, 787–790. https://doi.org/10.1016/j.nmd.2018.05.009

191. Yıldız, Y., Talim, B., Haliloglu, G., Topaloglu, H., Akçören, Z., Dursun, A., Sivri, H.S., Coşkun, T., Tokatlı, A., 2019. Determinants of riboflavin responsiveness in multiple acyl-CoA dehydrogenase deficiency. Pediatr Neurol 99, 69–75. https://doi.org/10.1016/j.pediatrneurol.2019.06.015

192. Yiş, U., Becker, K., Çırak, S., 2018. A boy with neck weakness. Neuromuscul Disord 28, 236–237. https://doi.org/10.1016/j.nmd.2017.11.016

193. Yotsumoto, Y., Hasegawa, Y., Fukuda, S., Kobayashi, H., Endo, M., Fukao, T., Yamaguchi, S., 2008. Clinical and molecular investigations of Japanese cases of glutaric acidemia type 2. Mol Genet Metab 94, 61–67. https://doi.org/10.1016/j.ymgme.2008.01.002
194. Zhao, F., 2014. Clinical, pathology, biochemical and genetic analysis of one family with lipid deposit myopathy. Jilin University.
195. Zhao, Y.W., Liu, X.J., Zhang, W., Wang, Z.X., Yuan, Y., 2018. Muscle magnetic resonance imaging for the differentiation of multiple acyl-CoA dehydrogenase deficiency and immune-mediated necrotizing myopathy. Chin Med J 131, 144–150. https://doi.org/10.4103/0366-6999.222323
196. Zhao, Z.N., Bao, M.X., Ma, G.T., Liu, X.M., Xu, W.J., Sun, Z.W., Chen, H., Zhu, M.J., 2012. A case of late-onset riboflavin responsive multiple acyl-CoA dehydrogenase deficiency with novel mutations in ETFDH gene. CNS Neurosci Ther 18, 952–954. https://doi.org/10.1111/cns.12007
197. Zhen, C., Li, Y., Tian, F., 2019. Diagnosis and management of a twin brother and sister with glutaric aciduria type II. J. Zhengzhou Univ. (Medical Sci.) 54, 309–312.
198. Zhu, M., Zhu, X., Qi, X., Weijiang, D., Yu, Y., Wan, H., Hong, D., 2014. Riboflavin-responsive multiple acyl-CoA dehydrogenation deficiency in 13 cases, and a literature review in mainland Chinese patients. J Hum Genet 59, 256–261. https://doi.org/10.1038/jhg.2014.10
199. Zhuo, Z., Jin, P., Li, F., Li, H., Chen, X., Wang, H., 2015. A case of late-onset riboflavin responsive multiple acyl-CoA dehydrogenase deficiency (MADD) with a novel mutation in ETFDH gene. J Neurol Sci 353, 84–86. https://doi.org/10.1016/j.jns.2015.04.011