Riboflavin Transporter Deficiency

Updated September 2017

Table 1. Report Pathogenic Variants in SLC52A2 (previously GPR172A) Reference Sequence: NM_ 024531.4

DNA nucleotide changed	Frequency (n=148)	ExAC Frequency	Type	Exon	Publication	Other Source
c.92 G>C	1	0	Non-synonymous	2	Foley 2013	
c.135 G>T	1	0	Non-synonymous	3		Cure RTD Registry
c.155 C>T	5	3	Non-synonymous	3	Ciccolella 2012	
c.185 T>G	6	1	Non-synonymous	3		Cure RTD Registry
c.245 G>C	2	0	Non-synonymous	3		Cure RTD Registry
c.287 C>T	1	0	Non-synonymous	3		Cure RTD Registry
c.297G>C	1	4	Non-synonymous	3	Disease in Childhood 102(Suppl 2):A181.3-A182 · June 2017	
c.368 T>C	2	1	Non-synonymous	3	Haack 2012	
c.377 G>A	1	0	Non-synonymous	3		Cure RTD Registry
c.383 C>T	3	3	Non-synonymous	3	SSIEM - Abstract 2016	
c.401 C>T	4	0	Non-synonymous	3	Guissart 2015	
c.505 C>T	9	1	Non-synonymous	3	Woodcock 2017, Allison 2017	
c.700 C>T	1	0	Stop-gained	3	Foley 2013	
c.808 C>T	2	3	Stop-gained	3	Shashi 2017, Petrovski 2015	
c.851 C>A	2	0	Non-synonymous	3	Foley 2013	
c.863 C>T	2	9	Non-synonymous	3		Cure RTD Registry
c.865 C>T	1	0	Non-synonymous	3		Manole et al, unpublished
c.914 A>G	1	0	Non-synonymous	3	Foley 2013	
c.916 G>A	54	4	Non-synonymous	3	Johnson 2012, Foley 2013, Srour 2014, Menenze 2015, Sanchez 2017	
c.935 T>C	8	4	Non-synonymous	3	Foley 2013, Allison 2017	
c.1016 T>C	26	8	Non-synonymous	4	Haack 2012, Foley 2013, Menenze 2015, Shashi 2017	
c.1088 C>T	1	0	Non-synonymous	4		Pandraud PhD Thesis
c.1243 G>A	1	0	Non-synonymous	5		Cure RTD Registry
c.1244 G>C	2	2	Non-synonymous	5		Cure RTD Registry
c.1255 G>A	1	0	Non-synonymous	5	Ciccolella 2012	
c.1258 G>A	2	2	Non-synonymous	5	Foley 2013, Schwartz 2015	
c.1259 C>T	2	0	Non-synonymous	5		Cure RTD Registry
c.1289 A>G	1	1	Non-synonymous	5		Cure RTD Registry
DEL_1030_1031	2	0	Deletion	4		Cure RTD Registry
DEL_750_751	1	0	Deletion	3		Cure RTD Registry
DEL_292_293	1	0	Deletion	3		Cure RTD Registry
DEL_1140	2	0	Deletion	5		Cure RTD Registry
c.-110–1G> A	1				Disease in Childhood 102(Suppl 2):A181.3-A182 · June 2017	

ExAC = Exome Aggregation Consortium (http://exac.broadinstitute.org/)

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Table 2. Report Pathogenic Variants in SLC52A3 (previously C20orf54)
Reference Sequence: NM_ 033409.3

DNA nucleotide changed	Publication	Other Source
c.44G>T	Horoz 2015	
c.49T>C	Bosch et al 2011	
c.62A>G	Dezfouli et al 2012	
c.71G4A (stop)	Hossain 2017	
c.82C>A	Johnson et al 2010	
c.106G>A	Green et al 2010,Rosser 2015	
c.173T>A	Ciccolella et al 2012	
c.193C>T	Davis 2015	
c.211G>A	Green et al 2010	
c.211G>T	Green et al 2010	
c.224T>C	Johnson et al 2010	
c.361C>G	Poggio 2013	
c.374C>A	Chaya 2017	
c.376A>G	Poggio 2013	
c.383C>T	Cosgrove 2015	
c.394C>T	Green et al 2010	
c.403A>G		Manole et al, unpublished
c.446G>A		Cure RTD Registery
c.436G>A	Hockaday 1981	
c.639C>G	Green et al 2010, Chaya 2017	
c.659C>A	Dezfouli et al 2012	
c.670T>C	Green et al 2010	
c.765C>T		Cure RTD Registery
c.796C>T	Ciccolella et al 2012	
c.935C>T	Dezfouli et al 2012, Khadilkar 2017	
c.955C>T	Ciccolella et al 2012	
c.989G>T	Koy et al 2012	
c.1048T>A	Green et al 2010	
c.1121G>A		Cure RTD Registery
c.1156T>C		Cure RTD Registery
c.1198-2A>C	Bosch et al 2011	
c.1198-20_c.1237dup (insertion)		Cure RTD Registery
c.1237T>C	Green et al 2010, Bashford 2016, Rosser 2015	

c.1238T>C	Ciccolella et al 2012, Davis 2015	

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Table 2. Report Pathogenic Variants in SLC52A3 (previously C20orf54)
Reference Sequence: NM_ 033409.3

Continued……

DNA nucleotide changed	Publication	Other Source
c.1296C>A	Ciccolella et al 2012	
c.1325_1326delTG	Green et al 2010	
c.1371C>G	Green et al 2010	
c.634C>T		Manole et al, unpublished
c.671T>G		Manole et al, unpublished
c.1128-1129_insT		Manole et al, unpublished
c.1255G>A		Manole et al, unpublished
c.1238T>C	Davis 2015	
c.1292G>A	Cosgrove 2015	
c.1294G>A		Manole et al, unpublished
c.1381T>G	Bashford 2016	
c.1316G>A	Woodcock 2017	
c.1313T>C		Cure RTD Registery
c.1124G>A	Dezfouli 2012	WT
c.1296C>A	Ciccolella 2012	WT

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