

Riboflavin Transporter Deficiency

Updated Dec 2020

Table 1. Report Pathogenic Variants in SLC52A2 (previously GPR172A)
Reference Sequence: NM_024531.4

DNA nucleotide changed	Frequency (n=198)	ExAC Frequency (n=~250,000)	Type	Exon	Publication	Other Source
c.92 G>C	1	0	Non-synonymous	2	Foley 2013	
c.103 C>T	2	1	Non-synonymous	2		Cure RTD Registry
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	8	2	Non-synonymous	3		Cure RTD Registry
c.231G>A	1	0	Non-synonymous	3	Manole 2017	
c.245 G>C	2	6	Non-synonymous	3		Cure RTD Registry
c.287 C>T	1	1	Non-synonymous	3		Cure RTD Registry
c.297 G>C	1	9	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	7	Non-synonymous	3	Manole 2017, SSIEM - Abstract 2016	
c.401 C>T	4	3	Non-synonymous	3	Guissart 2015	
c.421C>A	2	1	Non-synonymous	3	Udhayabanu 2016	
c.477c>G	2	0	Non-synonymous	3		Cure RTD Registry
c.505 C>T	9	5	Non-synonymous	3	Woodcock 2017, Allison 2017	
c.700 C>T	1	0	Stop-gained	3	Foley 2013	
c.751C>T	1	0		3		Cure RTD Registry
c.808 C>T	2	5	Stop-gained	3	Shashi 2017, Petrovski 2015	
c.851 C>A	2	0	Non-synonymous	3	Foley 2013	
c.861C>A	1	0		3		Cure RTD Registry
c.863 C>T	2	25	Non-synonymous	3		Cure RTD Registry
c.865 C>T	1	1	Non-synonymous	3	Manole 2017	
c.911C>T	2	0	Non-synonymous	3	SMID 2018 - Abstract	
c.914 A>G	1	2	Non-synonymous	3	Foley 2013	
c.917 G>A	2	1	Non-synonymous	3	Nimmo 2017	
c.916 G>A	63	14	Non-synonymous	3	Johnson 2012, Foley 2013, Srour 2014, Menenze 2015, Sanchez 2017	
c.935 T>C	8	7	Non-synonymous	3	Foley 2013, Allison 2017, Manole 2017,	
c.973 T>G	8	0	Non-synonymous	3	Badanejad 2018	
c.968 T>C	2	8	Non-synonymous	3	Gorcenco 2018	
c.1016 T>C	39	18	Non-synonymous	4	Haack 2012, Foley 2013, Menenze 2015, Shashi 2017, Manole 2017,	
c.1052G>A	1	0		4		Cure RTD Registry
c.1087 C>G	1	1	Non-synonymous	4		Cure RTD Registry
c.1088 C>T	1	0	Non-synonymous	4	Manole 2017	
c.1243 G>A	1	0	Non-synonymous	5		Cure RTD Registry

c.1244 G>C	4	3	Non-synonymous	5		Cure RTD Registry
c.1245C>T	2	0	Non-synonymous	5	Kranthi 2020	
c.1250 T>C	1	5	Non-synonymous	5		Cure RTD Registry
c.1255 G>A	1	0	Non-synonymous	5	Ciccolella 2012	
c.1258 G>A	7	4	Non-synonymous	5	Foley 2013, Schwartz 2015	
c.1259 C>T	4	0	Non-synonymous	5		Cure RTD Registry
c.1267 A>G	2	9	Non-synonymous	5		Cure RTD Registry
c.1268T>C	2	0		5		Cure RTD Registry
c.1289 A>G	2	1	Non-synonymous	5		Cure RTD Registry
c.1327 T>C	4	0	Non-synonymous	5	Manole 2017	
c.1328 G>A	1	0				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	0			Disease in Childhood 102(Suppl 2):A181.3-A182 · June 2017	
c.405_407delCTT	2	0	Deletion			Cure RTD Registry
1022 C insertion (p.Cys443Tyr)	1					Cure RTD Registry
c.1161_1189del21	1					Cure RTD Registry
c.131-1G>C	1					Cure RTD Registry
Phe197del	1					Cure RTD Registry
p.Pro48Leu	2					Cure RTD Registry
Phe197del	1	0	Deletion			Cure RTD Registry

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

Riboflavin Transporter Deficiency

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, Udhayabanu 2017	
c.71G4A (stop)	Hossain 2017	
c.82C>A	Johnson et al 2010	
c.106G>A	Green et al 2010, Rosser 2015, Manole 2017	
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.354 G>A	Manole 2017	
c.361C>G	Poggio 2013	
c.374C>A	Chaya 2017, Manole 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 Registry
c.436G>A	Hockaday 1981	
c.639C>G	Green et al 2010, Chaya 2017	
c.634 C>T	Manole 2017	
c.659C>A	Dezfouli et al 2012	
c.670T>C	Green et al 2010	
c.765C>T		Cure RTD Registry
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.1074 G>A	Manole 2017	
c.1121G>A		Cure RTD Registry
c.1156T>C	Thulasi 2017	
c.1198-2A>C	Bosch et al 2011	
c.1198-20_c.1237dup (insertion)		Cure RTD Registry
c.1237T>C	Green et al 2010, Bashford 2016, Rosser 2015, Manole 2017	
c.1238T>C	Ciccolella et al 2012, Davis 2015	
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 2017	
c.671T>G	Manole 2017	
c.1128-1129_insT	Manole 2017	
c.1232_1233ins	Camargos 2018	
c.1223G>A	Nimmo 2017	
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 Registry
c.1124G>A	Dezfouli 2012	WT

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