

Riboflavin Transporter Deficiency

Updated April 2018

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

| DNA nucleotide changed | Frequency (n=177) | ExAC Frequency | Type | Exon | Publication | Other Source |
|------------------------|-------------------|----------------|----------------|------|--|-------------------|
| c.92 G>C | 1 | 0 | Non-synonymous | 2 | Foley 2013 | |
| c.103 C>T | 2 | 0 | 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 | 6 | 1 | Non-synonymous | 3 | | Cure RTD Registry |
| c.231G>A | 1 | 0 | Non-synonymous | 3 | Manole 2017 | |
| 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 | Manole 2017, SSIEM - Abstract 2016 | |
| c.401 C>T | 4 | 0 | Non-synonymous | 3 | Guissart 2015 | |
| c.421C>A | 2 | 1 | Non-synonymous | 3 | Udhayabanu 2016 | |
| 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 2017 | |
| c.911C>T | 2 | 0 | Non-synonymous | 3 | SMID 2018 - Abstract | |
| c.914 A>G | 1 | 0 | Non-synonymous | 3 | Foley 2013 | |
| c.917 G>A | 2 | 1 | Non-synonymous | 3 | Nimmo 2017 | |
| 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, Manole 2017, | |
| c.973 T>G | 8 | 0 | Non-synonymous | 3 | Badanejad 2018 | |
| c.1016 T>C | 30 | 8 | Non-synonymous | 4 | Haack 2012, Foley 2013, Menenze 2015, Shashi 2017, Manole 2017, | |
| 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 | 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.1267 A>G | 2 | 1 | Non-synonymous | 5 | | Cure RTD Registry |
| c.1289 A>G | 1 | 1 | Non-synonymous | 5 | | Cure RTD Registry |

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|---------------|---|---|----------------|---|---|-------------------|
| c.1327 T>C | 4 | 0 | Non-synonymous | 5 | Manole 2017 | |
| 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, 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 |

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|---------------------------------|---|---------------------------|
| 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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