Title: Riboflavin Transporter Deficiency Neuronopathy *GeneReview,* Molecular Genetics, Table 2

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Note: The following information is provided by the authors and has not been reviewed by *GeneReviews* staff.

Table 2. Pathogenic Variants in *SLC52A1, SLC52A2,* and *SLC52A3*

Pathogenic variants in *SLC52A3*			Reference sequence
DNA nucleotide changed	**Protein amino acid changed**	**Reference (first report)**	
c.49T>C	Trp17Arg	Bosch et al [2011]	NM_033409.3
c.62A>G	Asn21Ser	Dezfouli et al [2012]	NM_033409.3
c.82C>A	Pro28Thr	Johnson et al [2010]	NM_033409.3
c.106G>A	Glu36Lys	Green et al [2010]	NM_033409.3
c.160G>A	Gly54Arg	Johnson et al [2010]	NM_033409.3
c.173T>A	Val58Asp	Ciccolella et al [2012]	NM_033409.3
c.211G>A	Glu71Lys	Johnson et al [2010]	NM_033409.3
c.211G>T	Glu71Ter	Green et al [2010]	NM_033409.3
c.224T>C	Ile75Thr	Johnson et al [2010]	NM_033409.3
c.394C>T	Arg132Trp	Green et al [2010]	NM_033409.3
c.568-19_-18insCTGATTGAC	Insertion	Ciccolella et al [2012]	NM_033409.3
c.639C>G	Tyr213Ter	Green et al [2010]	NM_033409.3
c.659C>A	Pro220His	Dezfouli et al [2012]	NM_033409.3
c.670T>C	Phe224Leu	Green et al [2010]	NM_033409.3
c.796C>T	Arg266Trp	Ciccolella et al [2012]	NM_033409.3

Pathogenic variants in *SLC52A3*			Reference sequence
DNA nucleotide changed	**Protein amino acid changed**	**Reference (first report)**	
c.935C>T	Ala312Val	Dezfouli et al [2012]	NM_033409.3
c.955C>T	Pro319Ser	Ciccolella et al [2012]	NM_033409.3
c.989G>T	Gly330Val	Koy et al [2012]	NM_033409.3
c.1048T>A	Leu350Met	Green et al [2010]	NM_033409.3
c.1198-2A>C	Unknown	Bosch et al [2011]	NM_033409.3
c.1237T>C	Val413Ala	Green et al [2010]	NM_033409.3
c.1238T>C	Val413Ala	Ciccolella et al [2012]	NM_033409.3
c.1296C>A	Cys432Ter	Ciccolella et al [2012]	NM_033409.3
1325_1326delTG	Leu442ArgfsTer64	Green et al [2010]	NM_033409.3
c.1371C>G	Phe457Leu	Green et al [2010]	NM_033409.3
c.374C>A	Thr125Asn	Manole et al, unpublished	NM_033409.3
c.403A>G	Thr135Ala	Manole et al, unpublished	NM_033409.3
c.634C>T	Arg212Cys	Manole et al, unpublished	NM_033409.3
c.671T>G	Phe224Cys	Manole et al, unpublished	NM_033409.3
c.1128-1129_insT	Tyr276Ter	Manole et al, unpublished	NM_033409.3
c.1255G>A	Gly418Asp	Manole et al, unpublished	NM_033409.3
c.1294G>A	Trp431Ter	Manole et al, unpublished	NM_033409.3
Pathogenic variants in *SLC52A2*			
c.92G>C	Trp31Ser	Foley et al [2014]	NM_024531.4
c.155C>T	Ser52Phe	Ciccolella et al [2012]	NM_024531.4
c.368T>C	Leu123Pro	Haack et al [2012]	NM_024531.4

Pathogenic variants in *SLC52A3*			**Reference sequence**
DNA nucleotide changed	**Protein amino acid changed**	**Reference (first report)**	
c.383C>T	Ser128Leu	Manole et al, unpublished	NM_024531.4
c.700C>T	Gln234Ter	Foley et al [2014]	NM_024531.4
c.851C>A	Ala284Asp	Foley et al [2014]	NM_024531.4
c.865C>T	Ala288Val	Manole et al, unpublished	NM_024531.4
c.914A>G	Tyr305Cys	Foley et al [2014]	NM_024531.4
c.916G>A	Gly306Arg	Foley et al [2014]	NM_024531.4
c.935T>C	Leu312Pro	Foley et al [2014]	NM_024531.4
c.1088C>T	Pro363Leu	Manole et al, unpublished	NM_024531.4
c.1016T>C	Leu339Pro	Haack et al [2012]	NM_024531.4
c.1255G>A	Gly419Ser	Ciccolella et al [2012]	NM_024531.4
c.1258G>A	Ala420Thr	Foley et al [2014]	NM_024531.4
Pathogenic variants in *SLC52A1*			
Deletion of exons 2 and 3 using RT PCR	Deletion of exons 2 and 3, amino acids 44-379	Ho et al [2011]	NM_001104577

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