Title: Action Myoclonus – Renal Failure Syndrome *GeneReview* – Table 4
Authors: Amrom D, Andermann F, Andermann E
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Note: The following information is provided by the authors listed above and has not been reviewed by *GeneReviews* staff.

Table 4. Pathogenic Variants in *SCARB2* Causing AMRF Syndrome

Standard Naming Convention DNA Nucleotide Change	Location	Protein Amino Acid Change	Reference Sequence	Reference
c.111delC Frameshift and premature stop	Exon 1		N/A	Hopfner et al [2011]
c.296delA Frameshift	Exon 3	p.Asn99IlefsTer34	NM_005506	Berkovic et al [2008]
c.361C>T	Exon 3	p.Arg121Ter	NM_005506	Fu et al [2014]
c.435_436insAG	Exon 4	p.Trp146SerfsTer16	NM_005506	Berkovic et al [2008]
c.533G>A Nonsense	Exon 4	p.Trp178Ter	N/A	Balreira et al [2008]
c.424-2A>C Splice-site	Intron 4	-	NM_005506	Dibbens et al [2009]
c.666delCCTTA Frameshift	Exon 5	p.Tyr222Ter	NM_005506	Dibbens et al [2009] Hopfner et al [2011]
c.704+1G>A Splice-site	Intron 5		N/A	Perandones et al [2012] Perandones et al [2014]
c.704+1G>C Splice-site	Intron 5		NM_005506	Dibbens et al [2009]
c.862C>T Nonsense	Exon 7	p.Gln288Ter	NM_005506	Berkovic et al [2008] Dibbens et al [2011]
c.1015_1016insT	Exon 8	p.His341ThrfsTer7	NM_005506	Guerrero-Lopez [2012]
c.1087C>A Missense	Exon 8	p.His363Asn	N/A	Dardis et al [2009]
c.1116-2A>C Splice-site	Intron 8	-	NM_005506	Dibbens et al [2009]
c.1187+3insT Splice-site	Intron 9		NM_005506	Dibbens et al [2011]
c.1239+1G>T Splice-site	Intron 10	-	NM_005506	Berkovic et al [2008]
c.1258delG Frameshift	Exon 11	p.Glu420ArgfsTer5	NM_005506	Dibbens et al [2009]
c.1270C>T	Exon 11	p.Arg42Ter	NM_005506	Zeigler et al [2014] He et al [2014]
c.1385_1390del6insATGCATGCACC	Exon 11	p.Gly462AspfsTer34	NM_005506	Higashiyama et al [2013] Fu et al [2013]
c.1412A>G	Exon 12	p.Gln471Gly	NM_005506	Velayati et al [2011]

N/A = not available

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