GeneReview Title: Mitochondrial Neurogastrointestinal Encephalopathy Disease Tables 3-7
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Table 3. Missense and Nonsense Variants Identified in MNGIE Disease

Exon	Pathogenic Variant	Codon	Amino Acid Change	Reference
Exon 2	c.112G>T	Codon 38	Gln>Ter	Garone et al [2011]
Exon 2	c.128A>C	Codon 43	Lys>Thr	Hirano et al [2004]
Exon 2	c.131G>A	Codon 44	Arg>Gln	Gamez et al [2002]
Exon 2	c.146T>G	Codon 49	Leu>Arg	Garone et al [2011]
Exon 2	c.162C>G	Codon 54	Ile>Met	Kocaefe et al [2003]
Exon 3	c.228G>A	Codon 76	Met>Ile	Martin et al [2004]
Exon 3	c.261G>C	Codon 87	Glu>Asp	Labauge et al [2002]
Exon 3	c.261G>T	Codon 87	Glu>Asp	Slama et al [2005]
Exon 3	c.275C>A	Codon 92	Thr>Asn	Schüpbach et al [2007]
Exon 3	c.328C>T	Codon 110	Gln>Ter	Garone et al [2011]
Exon 3	c.340G>A	Codon 114	Asp>Asn	Slama et al [2005]
Exon 3	c.398T>C	Codon 133	Leu>Pro	Monoy et al [2008]
Exon 3	c.401C>A	Codon 134	Ala>Gln	Garone et al [2011]
Exon 4	c.433G>A	Codon 145	Gly>Arg	Nishino et al [1999]
Exon 4	c.457G>A	Codon 153	Gly>Ser	Nishino et al [1999]
Exon 4	c.467A>G	Codon 156	Asp>Gly	Hirano et al [2004]
Exon 4	c.478T>C	Codon 160	Ser>Pro	Nishino et al [2000]
Exon 5	c.518T>G	Codon 173	Met>Arg	Nishino et al [2000]
Exon 5	c.530T>C	Codon 177	Leu>Pro	Hirano et al [2004]
Exon 5	c.605G>A	Codon 202	Arg>Lys	Poulton et al 2009
Exon 5	c.605G>C	Codon 202	Arg>Thr	Martí et al [2005]
Exon 5	c.622G>A	Codon 208	Val>Met	Martí et al [2005]
Exon 5	c.623T>G	Codon 208	Val>Gly	Garone et al [2011]
Exon 6	c.665A>G	Codon 222	Lys>Arg	Nishino et al [1999]
Exon 6	c.707T>C	Codon 236	Phe>Ser	Said et al [2005]
Exon 6	c.715G>A	Codon 239	Ala>Thr	Garone et al [2011]
Exon 6	c.760A>C	Codon 254	Thr>Pro	Hirano et al [2004]
Exon 7	c.847C>G	Codon 283	His>Asp	Martin et al [2004]
Exon 7	c.854T>C	Codon 285	Leu>Pro	Martí et al [2005]

Exon	Pathogenic Variant	Codon	Amino Acid Change	Reference
Exon 7	c.856G>A	Codon 286	Glu>Lys	Slama et al [2005]
Exon 7	c.865G>A	Codon 289	Glu>Lys	Nishino et al [2000]
Exon 7	c.866A>C	Codon 289	Glu>Ala	Nishino et al [1999]
Exon 7	c.893G>A	Codon 298	Gly>Asp	Garone et al [2011]
Exon 8	c.931G>C	Codon 311	Gly>Arg	Martí et al [2005]
Exon 8	c.931G>A	Codon 311	Gly>Ala	Garone et al [2011]
Exon 8	c.931G>T	Codon 311	Gly>Cys	Garone et al [2011]
Exon 8	c.938T>C	Codon 313	Leu>Pro	Hirano et al [2004]
Exon 8	c.1067T>C	Codon 356	Leu>Pro	Garone et al [2011]
Exon 8	c.1112T>C	Codon 371	Leu>Pro	Kocaefe et al [2003]
Exon 8	c.1159G>A	Codon 387	Gly>Ser	Garone et al [2011]
Exon 8	c.1160G>A	Codon 387	Gly>Asp	Slama et al [2005]
Exon 9	c.1282G>A	Codon 428	Gly>Ser	Hirano et al [2004]
Exon 10	c.1311G>A	Codon 437	Trp>Ter	Weiss et al [2004]
Exon 10	c.1360G>C	Codon 454	Ala>Pro	Garone et al [2011]
Exon 10	c.1412C>A	Codon 471	Ser>Ter	Carod-Artal et al [2007]

Table 4. Pathogenic Variants Affecting Exon Splicing in MNGIE Disease

Intron	Pathogenic Variant	Splice Site	Reference
Intron 1	c.1-11G>C	Acceptor site	Szigeti et al [2004]
Intron 2	c.215-1G>C	Acceptor site	Hirano et al [2004]
Intron 4	c.516+2T>C	Donor site	Nishino et al [1999]
Intron 7	c.928+1G>A	Donor site	Slama et al [2005]
Intron 7	c.929-3G>A	Acceptor site	Kocaefe et al [2003]
Intron 8	c.1160-1G>A	Acceptor site	Nishino et al [2000]
Intron 8	c.1160-1G>C	Acceptor site	Nishino et al [1999]
Intron 8	c.1159+2T>A	Donor site	Kocaefe et al [2003]
Intron 8	c.1160-1G>C	Acceptor site	Nishino et al [1999]
Intron 8	c.1160-1G>A	Acceptor site	Nishino et al [2000]
Intron 8	c.1160-2A>C	Acceptor site	Garone et al [2011]
Intron 8	c.1160-2A>G	Acceptor site	Garone et al [2011]
Intron 9	c.1300+1G>A	Donor site	Taanman et al [2009]
Intron 9	c.1300+2T>A	Donor site	Kocaefe et al [2003]
Intron 9	c.1301-1G>A	Acceptor site	Nishino et al [1999]

Table 5. Small Exon Insertions in MNGIE Disease

Exon	Pathogenic Variant	Reference
Exon 2	c.99C insertion	Nishino et al [2000]
Exon 8	c.994_1011 duplication	Gamez et al [2005]
Exon 9	c.1211T insertion	Hirano et al [2004]
Exon 10	c.1319G insertion	Weiss et al [2004]
Exon 10	c.1351C insertion	Nishino et al [1999]
Exon 10	c.1431T insertion	Poulton et al 2009

Table 6. Small Deletions in MNGIE Disease

Exon/Intron	Nucleotide Positions of Variant	Deletion	Reference
Exon 2	c.52_53del	2-base pair deletion (CT)	Nishino et al [2000]
Exon 3	c.263_264del	2-base pair deletion (CC)	Garone et al [2011]
Exon 6	c.720del	1-base pair deletion (C)	Labauge et al [2002]
Exon 7	c.784del	1-base pair deletion (C)	Slama et al [2005]
Intron 7	c.929-3_929-6del	4-base pair deletion (CCGC)	Nishino et al [2000]
Exon 8	c.1088del	1-base pair deletion (G)	Poulton et al [2009]
Exon 9	c.1193_1198del	6-base pair deletion (CGCTGG)	Nishino et al [1999]
Exon 10	c.1311del	1-base pair deletion (G)	Garone et al [2011]
Exon 10	c.1327_1346del	20-base pair deletion (GACGCCCCCGCGCTCAGCGG)	Blazquez et al [2005]
Exon 10	c.1394_1400del	6-base pair deletion (GCCATT)	Garone et al [2011]

Table 7. Small Deletion and Insertion in MNGIE Disease

Exon/Intron	Nucleotide Positions of Variant	Deletion	Reference
Exon 8	c.1010_1019del_insAA	10-base pair deletion (GCTCGGCCCT) 2-base pair insertion (AA)	Garone et al [2011]

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