Title: Multiminicore Disease *GeneReview* Tables 2-4

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

Table 2. Polymorphisms Identified in *SEPN1*

Exonic Variant	Amino Acid Change	Exon
c425A>G	Y142C	4
c583G>A	A195T	5
c1173T>C	P391P	9
c1329A>G	E443E	10
c1506A>C	K502N	12

Table 3. Pathologic Allelic Variants in *SEPN1* Associated with Multiminicore Disease

Nucleotide Change	Amino Acid Change	Exon
c-19/+73del	Unknown	1
c1A>G	Unknown	1
c.1AinsT	Unknown	1
c22dup10bp	Frameshift at Q8	1
c.80dup20bp	Frameshift at R27	1
c713-714insA	Frameshift at N238	5
c817G>A	G273E	6
c878A>G	H293R	7
c943G>A	G315S	7
c1019A>T	N340I	8
c1315C>T	R439X	10
c1358G>C	W453S	10
c1384T>G	U462G	10
c1385G>A	U462X	10
c1397G>A	R466Q	11
c.1446delC	Frameshift at L482	11
g.17195T>C	SECIS element	3’UTR

Table 4. Pathologic Allelic Variants in *RYR1* Associated with Multiminicore Disease

Nucleotide Change	Amino Acid Change	Exon
c.212A>C	S71Y	3
c.325C>T	R109W	4
c.1453A>G	M485V	14
c.4729G>A	A1577T	33
c.6178G>T	G2060C	38
c.6847A>C	N2283H	42
c.7268T>A	M2423K	45
c.8816G>A	R2939K	57
c.10343C>T	S3448F	68
c.10579C>T	P3527S	71
c.11315G>A	R3772Q	79
c.12986C>A	A4329D	91
c.14126C>T	T4709M	96
c.14365-2A>T	Acceptor Splice site mutation	Intron 99
c14545G>A	V4849I	101
14646+2.99 kb A>G (splice site mutation)	Frameshift additional exon of 94 AA; premature stop codon 4976X	101-102