

Title: Marinesco-Sjögren Syndrome *GeneReview*, Table 2

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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. Pathogenic Variants in *SIL1*

Exon	Nucleotide Change	Predicted Protein Change	Reference	Comment
Exon 3	c.178C>T	p.Glu60Ter	Senderek et al [2005]	
Exon 3	c.212dupA	p.His71GlnfsTer5	Anttonen et al [2005]	
Exon 4	c.274C>T	p.Arg92Trp	Riazuddin et al [2009]	
Exon 4	c.331C>T	p.Arg111Ter	Anttonen et al [2005], Senderek et al [2005], Annesi et al [2007]	
Exon 4	c.346delG	p.Gly116fs	Senderek et al [2005]	
Exon 6-7	c.454-?_767+?del	58-kb deletion	Takahata et al [2010]	
Exon 6	c.506_509dupAAGA	p.Asp170GlufsTer4	Anttonen et al [2005]	
Exon 6	c.603_607delGAAGA	p.Glu200AspfsTer6	Takahata et al [2010]	originally c.598_602delGAAGA
Intron 6	c.645+1G>A	Skipping of exon 6	Senderek et al [2005]	
Intron 6	c.645+2T>C	p.Val186_Gln215del p.Ala152_Gln215del	Anttonen et al [2005]	
Exon 9	c.936dupG	p.Leu313AlafsTer39	Anttonen et al [2008], Eriguchi et al [2008]	
Exon 9	c.947_948insT	p.Leu316fs	Senderek et al [2005]	
Intron 9	c.1029+1G>A	Skipping of exon 9	Senderek et al [2005]	
Intron 9	c.1030-18G>A	p.Leu344fs	Senderek et al [2005]	
Intron 9	c.1030-9G>A	p.Phe345AlafsTer9	Anttonen et al [2008]	
Exon 10	c.1240C>T	p.Gln414Ter	Riazuddin et al [2009]	

Exon	Nucleotide Change	Predicted Protein Change	Reference	Comment
Exon 10	c.1249C>T	p.Gln417Ter	Senderek et al [2005]	
Exon 10	c.1312C>T	p.Gln438Ter	Karim et al [2006]	
Exon 10	c.1366delT	p.Leu456fs	Senderek et al [2005]	
Exon 10	c.1367T>A	p.Leu456Ter	Anttonen et al [2008]	
Exon 10	c.1370T>C	p.Leu457Pro	Anttonen et al [2008]	

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