

Supplementary Table S4. Characteristics of the *SOD1* variants relevant to the PVS1 criterion.

_Coding change (NM_000454.5)	Protein change (NP_000445.1)	Consequence Type	Predict/Not predict to undergo NMD	Location. Protein Position. Splice distance.
c.301G>T	p.Glu101Ter	stop gained	Not predict to undergo NMD	Nonsense mutation in exon 4, position 2 of 118. Protein position 101 of 155. Splice distance -57.
c.380T>A	p.Leu127Ter	stop gained	Not predict to undergo NMD	Nonsense in exon 5, position 23 of 461. Protein position 127 of 155. Splice distance 23
c.441T>A	p.Cys147Ter	nonsense variant	Not predict to undergo NMD	Nonsense in exon 5, position 84 of 461. Protein position 147 of 155. Splice distance 84.
c.424G>T	p.Gly142Ter	stop gained	Not predict to undergo NMD	Nonsense in exon 5, position 67 of 461. Protein position 142 of 155. Splice distance 67.
c.383_384insACCC	p.Lys129ProfsTer6	frameshift variant	Not predict to undergo NMD	Frameshift variants in exon 5 before poistion 27 of 461. Protein position 128 of 155. Splice distance 27.
c.384_385insTGGG	p.Lys129TrpfsTer6	frameshift variant	Not predict to undergo NMD	Frameshift variants in exon 5 before poistion 28 of 461. Protein position 129 of 155. Splice distance 28.
c.409A>T	p.Lys137Ter	nonsense variant	Not predict to undergo NMD	Nonsense variant in exon 4, position 52 of 461. Protein position 137 of 155. Splice distance 52.
c.275_276delAA	p.Lys92ArgfsTer9	frameshift variant	Predict to undergo NMD	Frameshift variant in exon 4 position 36-37 of 118. Protein position 92 of 155. Splice distance 36
c.320_321insT	p.Ser108LeufsTer15	frameshift variant	Not predict to undergo NMD	Frameshift variant in exon 4 before position 82 of 118. Protein position 107 of 155. Splice distance -38
c.383_392dup	p.Asn132GlnfsTer5	frameshift variant	Not predict to undergo NMD	Frameshift variant in exon 5 position 36 of 461. Protein position 131 of 155. Splice distance 36
c.335dup	p.Cys112TrpfsTer11	frameshift variant	Not predict to undergo NMD	Frameshit variant in exon 4 before position 97 of 118 Protein position 112 of 155. Splice distance -23,