Supplementary Table 3. *NEFL* mutations

rs ID	gDNA change (chr8; hg19)	cDNA change	Amino acid change	Abbreviation	PhyloP	GERP	Grantham score	SIFT	PolyPhen2	CADD	MAF ExAC	>1 variants in amino acid	Co-segrega-tion analysis	Reported in >1 pedigree	Functional studies[b]
rs61491953	24814007G>A	c.23C>T	p.Pro8Leu	P8L	2.40	5.59	98	0.008 (D)	0.777 (P)	22.5	-	Yes	Yes[a]	Yes	Yes[1]
rs61491953	24814007G>T	c.23C>A	p.Pro8Gln	P8Q	2.40	5.59	76	0.056 (D)	0.901 (P)	22.7	-	Yes	-	-	Yes[1]
rs61491953	24814007G>C	c.23C>G	p.Pro8Arg	P8R	2.40	5.59	103	0.004 (D)	0.901 (P)	22.5	-	Yes	Yes	Yes	Yes[2-8]
rs60261494	24814007_24814008delGGinsCT	c.22_23delCCinsAG	p.Pro8Arg	P8R	2.85	5.59	103	0.004 (D)	0.901 (P)	18.8	-	Yes	Yes	-	-
rs58640772	24813970_24813982dupCTCCACGTAGCGC	c.48_60dup	p.Thr21Alafs*83	T21Afs*83	1.69	4.06	-	-	-	20.8	-	-	-	-	Yes[9]
rs28928910	24813966G>A	c.64C>T	p.Pro22Ser	P22S	1.18	5.27	74	0.095 (T)	0.137 (T)	22.3	-	Yes	Yes	Yes	Yes[3,10-12]
rs28928910	24813966G>T	c.64C>A	p.Pro22Thr	P22T	1.18	5.27	38	0.082 (T)	0.628 (P)	22.4	-	Yes	Yes	Yes	Yes[3,11]
rs267607538	24813965G>C	c.65C>G	p.Pro22Arg	P22R	2.73	5.27	103	0.001 (D)	0.912 (D)	22.5	-	Yes	Yes	-	-
rs58332872	24813762C>T	c.268G>A	p.Glu90Lys	E90K	5.91	5.27	56	0.000 (D)	0.980 (D)	23.0	-	-	Yes[a]	Yes	Yes[1]
rs62636505	24813749A>G	c.281T>C	p.Leu94Pro	L94P	4.93	5.27	98	0.000 (D)	0.999 (D)	22.8	-	-	Yes	-	-
rs58982919	24813737T>C	c.293A>G	p.Asn98Ser	N98S	4.93	5.27	46	0.000 (D)	1.000 (D)	22.7	-	Yes	Yes[a]	Yes	Yes[1,13,14]
-	24813737T>G	c.293A>C	p.Asn98Thr	N98T	4.93	5.27	65	0.000 (D)	1.000 (D)	22.8	-	Yes	-	-	-
rs121913663	24813612C>A	c.418G>T	p.Glu140*	E140*	4.36	5.54	-	-	-	23.0	-	-	-	-	-
rs59101996	24813584G>A	c.446C>T	p.Ala149Val	A149V	4.44	5.54	64	0.104 (T)	0.835 (P)	22.8	-	-	-	-	Yes[15]
-	24813474C>A	c.556G>T	p.Glu186*	E186*	4.44	4.72	-	-	-	22.8	-	-	-	-	-
rs199422214	24813402C>A	c.628G>T	p.Glu210*	E210*	6.15	5.69	-	-	-	23.0	-	-	Yes	-	Yes[16]
rs587777880	24813236T>C	c.794A>G	p.Tyr265Cys	Y265C	5.03	5.62	194	0.000 (D)	1.000 (D)	22.7	-	-	Yes	-	-
rs62636502	24813227A>C	c.803T>G	p.Leu268Arg	L268R	5.03	5.62	102	0.002 (D)	0.998 (D)	22.6	-	Yes	Yes	-	-
rs62636502	24813227A>G	c.803T>C	p.Leu268Pro	L268P	5.03	5.62	98	0.003 (D)	0.998 (D)	22.8	-	Yes	Yes	Yes	-
-	24813098A>G	c.932T>C	p.Leu311Pro	L311P	1.95	5.58	98	0.021 (D)	0.934 (D)	22.5	-	-	-	-	-
rs267607537	24813053_24813067delTTCATGCCCCGGCAT	c.963_977del	p.Cys322_Asn326del	C322_N326del	2.23	3.77	-	-	-	21.4	-	-	Yes	-	-
rs59443585	24813035T>G	c.995A>C	p.Gln332Pro	Q332P	4.85	5.10	76	0.000 (D)	0.992 (D)	22.7	-	-	Yes	Yes	Yes[2-8]
rs60930717	24813032A>G	c.998T>C	p.Leu333Pro	L333P	4.85	5.10	98	0.000 (D)	0.992 (D)	22.7	-	-	-	-	-
rs281865140	24813029T>G	c.1001A>C	p.Gln334Pro	Q334P	2.41	3.93	76	0.011 (D)	0.904 (P)	22.4	-	-	-	-	-
rs587777881	24813023A>G	c.1007T>C	p.Leu336Pro	L336P	4.85	5.10	98	0.002 (D)	0.956 (D)	22.7	-	-	-	-	-
-	24811714T>A	c.1150A>T	p.Ile384Phe	I384F	5.08	5.58	21	0.000 (D)	0.999 (D)	22.7	-	-	Yes	-	-
-	24811698T>C	c.1166A>G	p.Tyr389Cys	Y389C	5.08	5.78	194	0.024 (D)	1.000 (D)	22.3	-	-	Yes	-	-
rs62636503	24811293C>T	c.1186G>A	p.Glu396Lys	E396K	6.10	5.91	56	0.000 (D)	1.000 (D)	22.7	-	-	Yes	Yes	Yes[17,18]
-	24811218G>A	c.1261C>T	p.Arg421*	R421*	0.61	3.08	-	-	-	21.5	-	-	-	-	-
rs199775873	24811164A>T	c.1315T>A	p.Phe439Ile	F439I	2.57	5.91	21	0.138 (T)	0.666 (P)	21.0	0.011	-	-	-	-
rs587777882	24811160G>A	c.1319C>T	p.Pro440Leu	P440L	1.98	5.02	98	0.002 (D)	0.551 (P)	21.4	-	-	Yes	Yes	-

(a) Confirmed *de novo* mutations. (b) *In vitro* or *in vivo* functional studies. CADD = CADD score (http://cadd.gs.washington.edu/). D = damaging/probably damaging. ExAC = Exome Aggregation Consortium browser v0.3 (http://exac.broadinstitute.org). GERP = GERP score accessed through the UCSC Genome Browser (https://genome.ucsc.edu/cgi-bin/hgGateway). hg19 = human reference genome assembly 19 (GRCh37). MAF = minor allele frequency. P = possibly damaging. phyloP = 46 vertebrate basewise conservation phyloP score accessed through the UCSC Genome Browser (https://genome.ucsc.edu/cgi-bin/hgGateway). PolyPhen2 = PolyPhen2 score (http://genetics.bwh.harvard.edu/pph2/). rs ID = reference single nucleotide polymorphism identifier at dbSNP. SIFT = SIFT score (http://provean.jcvi.org/index.php). T = tolerated/benign.

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