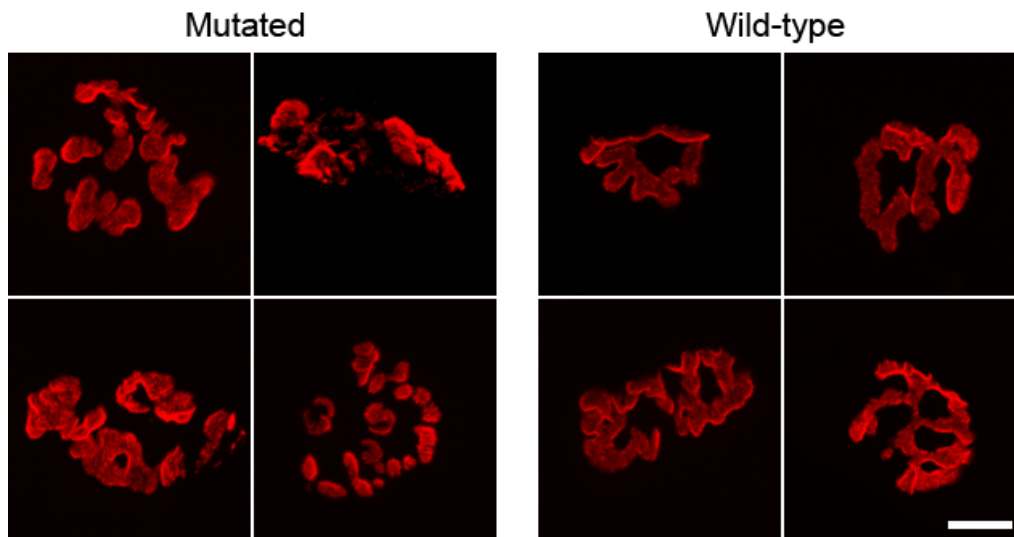**Supplemental Data**

Identification of an Agrin Mutation

that Causes Congenital Myasthenia
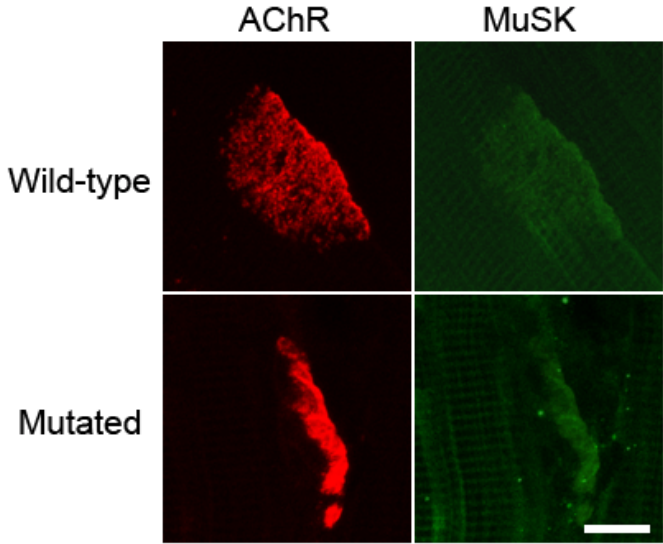
and Affects Synapse Function

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Figure S1. Fragmentation of the NMJs

Fragmentation was determined using "en face" NMJs stained with α-bungarotoxin. The number of fragments which forms one given NMJ was counted and the mean value obtained in each experimental condition (after wild-type or mutated agrin injection) was calculated. The same measurements were made for the human muscle biopsies. Calibration bar represents 10 µm and applies to the eight prints.

Figure S2. MuSK localization on aneural clusters

Colocalization of MuSK (green) with the AChR (red) can be observed. Calibration bar represents 4 µm and applies to the four prints.

Table S1. List of primers for amplification

Fragments	Primers (5'→ 3')	Fragments	Primers (5'→ 3')
Agrin-1F Agrin-1R	CGTCCCGTCCTGTCCAGTCC AGCGACCGGGGCTGGTTC	Agrin-22F Agrin-22R	CTTCCTGGGAGGCAATGG AGAGGAGGGGGCAGGTAAG
Agrin-2F Agrin-2R	CCTTCCCCAGGAGAGGACTA GACCCCTGCAACCTGTCC	Agrin-23F Agrin-23R	CCCTGCTCCCAGGAAACC CCAACTCAAAGCCTGATGGT
Agrin-1SNF Agrin-1SNR	TTCTTCTCGCTCCCGACG GGCCTCCAGCCTCTATGTG	Agrin-24F Agrin-24R	ATCCTCGGAGCTTTTCCAG AGGTGCTCAGGACCATCG
Agrin-3F Agrin-3R	GCCTGCTCAGAGGAGCCTA CCAGACACACTCCACACGTC	Agrin-25F Agrin-25R	CGATGGTCCTGAGCACCTG GCAAAGCCACCTCGTACAC
Agrin-4F Agrin-4R	CTGAGCTTTCTCCCCTACCC AGCCTCGCTCCATCTCATAG	Agrin-26F Agrin-26R	GAGGTGGCTTTGCCTGTG GGTCCTGGATGGAGGAAGAC
Agrin-5+6F Agrin-5+6R	CCCGTCTGACCGGCAAAG CACCTCGACCCTGGCACTG	Agrin-27F Agrin-27R	CTGTGGGCGGTACCCAAC ACTGAGCCGCATCCTAACTG
Agrin-7F Agrin-7R	AGGGTGCTCCAGCCTCTC AGATGGCATGTGATGTCCAG	Agrin-28F Agrin-28R	TGGGTTTTGAGTTAGGATCCAC CCACAGGAGAGACGGAGGT
Agrin-8F Agrin-8R	TCTCTCTTCCTCCACCATCC GCGACCAGGAACCTCTCTCT	Agrin-29F Agrin-29R	CCACCTCCGTCTCTCCTGT GTGGGCTCTGTCCACCAC
Agrin-9F Agrin-9R	GGGAGAGAGAGGTTCCTGGT CTGAGGGAGGCACAGGAC	Agrin-30F Agrin-30R	CACCAGCAGGTCGCTCAG GAGGAAGAGCAGGCAGCA
Agrin-10F Agrin-10R	GTCCTGTGCCTCCCTCAG CCAGAGCCCAGCAGTTTTC	Agrin-YF Agrin-YR	CGCTCACGGAGCTGTTTTTC ACCCGCCCATTGCCTATCTA
Agrin-11+12F Agrin-11+12R	GCTCTGGCTTTGGACAAGAA CACAATACACACGTCCACAGG	Agrin-31+32F Agrin-31+32R	GCTCTGCACAGCCACTTACC GTGAGGGTGGGGCCTCGT
Agrin-13+14F Agrin-13+14R	GTCCAGCACTGCATGAAATC GTGATGGGGGTGAACAGGTA	Agrin-33F Agrin-33R	GGATTCTCAGGACCGCACT CGAGGGCACAGATGGAAC
Agrin-15F Agrin-15R	ACCAGGCTCTGGAGGAGGT CTCTGTGGCTCGGGTGAC	Agrin-Z1F Agrin-Z1R	TTCCATCTGTGCCCTCGG GGATGGAGGAGTGGGAGTGA
Agrin-16+17F Agrin-16+17R	GTCACCCGAGCCACAGAG AGGCCTTCCTGGTCAGTTCT	Agrin-Z2F Agrin-Z2R	TGTTGAGATGGGTTTGCATTGG CCGACAGTGGGAGGGAGAA
Agrin-18F Agrin-18R	CTGACGCTGCCCTAAATCC CTCTGGTGGAGCCTCTCG	Agrin-34F Agrin-34R	GTCCTGTTGCCACCTTCCTA TCACAAGTGACTGGACACAGC
Agrin-19F Agrin-19R	CGAGAGGCTCCACCAGAG CCAGGCTATCCTCACACTCC	Agrin-35F Agrin-35R	AGAGGAGGCAGAGGGAACTC TGGAGTCTGTCCCTCGGATA
Agrin-20+21F Agrin-20+21R	CTGGATGCCAGGCAGATG GAGGGAGCTCTGAGCATGG	Agrin-36F Agrin-36R	GCCCCTGCAGTTCCCAGTGC AGCAGTCCCTCCGGCCAACA

Grey background: fragments to be amplified with CG rich mix
Blue background: neural Y and Z specific fragments

Table S2. *AGRN* **polymorphisms**

Missense polymorphisms	Mutated allele frequency	Indicated when already known
p.Val23Leu	0.016	
p.Asp58Asn	0.01	
p.Asp105Ile	0.01	
p.Thr267Met	0.02	
p.Ala375Ser	0.01	
p.Glu728Val	0.021	
p.Gln852Arg	0.005	rs9697293
p.Val984Met	0.01	
p.Leu1088Phe	0.01	
p.Thr1118Lys	0.01	
p.Gln1135Arg	0.016	
p.Pro1240Leu	0.01	
p.Gly1341Arg	0.01	
p.Pro1451Leu	0.01	
p.Ala1513Thr	0.016	
p.Gln1565His	0.01	
p.Val1666Ile	0.02	rs17160775
p.Arg1671Gln	0.01	
p.Arg1698Pro	0.01	
p.Gly1709Arg	0.02	
p.Arg1734His	0.01	
p.Asp1785Asn	0.01	
p.Gly2023Val	0.01	

AGRN missense polymorphisms only are presented in this Table. Intronic polymorphisms as well as synonymous polymorphisms are not. They can be obtained on demand.