Supplementary Material

Dusl *et al.*: A 3'-UTR mutation creates a microRNA target site in the *GFPT1* gene of patients with congenital myasthenic syndrome

Figure S1. Schematic representation of the sequence alignment of miR-600 with wild-type and variant (c.*22C>A) *GFPT1* mRNA. Bioinformatics tools revealed that the 3'-UTR mutation c.*22C>A may result in a gain of a putative binding site for the miR-600 [MIMAT0003268]. The mutation creates a 7mer-A1 site which is highlighted in grey. Lines indicate Watson-Crick base pairings.

Figure S2. Quantitative analysis of the microRNAs **(A)** miR-206 and **(B)** miR-206* in myoblasts of *GFPT1* patients and two unaffected controls (Control 1 and Control 2) by real-time qRT-PCR. LGM5.3 and LGM5.5: compound heterozygous for c.*22C>A and p.M492T; LGM9.3: compound heterozygous for c.*22C>A and p.V199F. Data are derived from n=3 independent experiments.

Figure S3. Renilla-to-firefly luminescence ratios observed when co-transfecting COS-7 cells with the luciferase reporter pRL-4xA (mutant) with either 0, 10, 50 or 100 nM miR-206*. Error bars indicate $\pm$SD obtained from three replicates.

Figure S4. Renilla-to-firefly luminescence ratios observed when co-transfecting COS-7 cells with the indicated luciferase reporter (pRL, pRL1xC (wild-type), pRL1xA (mutant)) and either 100nM control miRNA or miR-206* oligonucleotide. Error bars indicate $\pm$SD obtained from three replicates. p = 0.049, pRL1xC+miR-206* vs pRL1xA+miR-206*; p = 0.020, pRL1xA+controlmiRNA vs pRL1xA+miR-206*.

Figure S1.

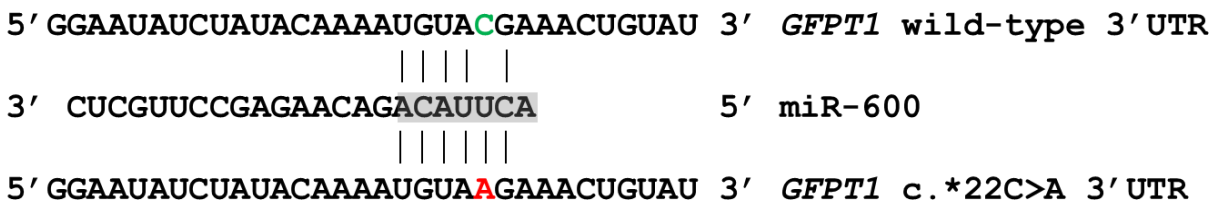
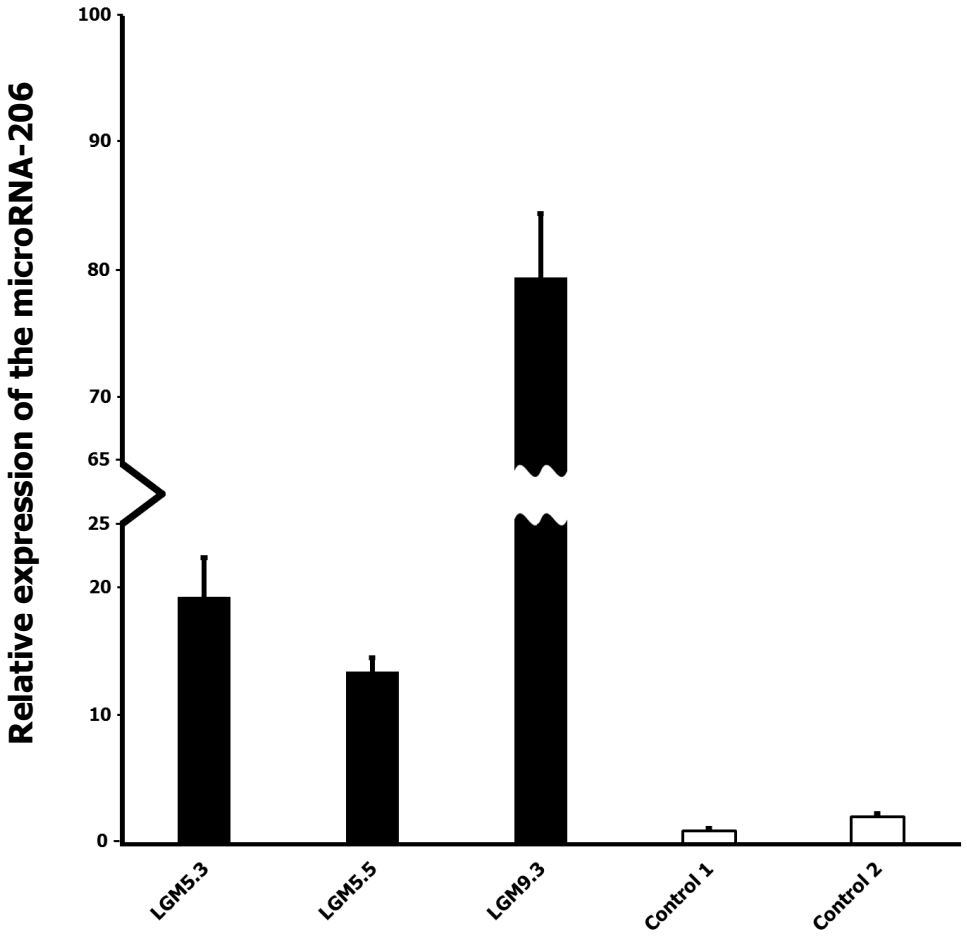

5′GGAAUAUCUAUACAAAAUGUACGAAACUGUAU 3′ GFPT1 wild-type 3′UTR
3′ CUCGUUCCGAGAACAGACAUUCA 5′ miR-600
5′GGAAUAUCUAUACAAAAUGUAAGAAACUGUAU 3′ GFPT1 c.*22C>A 3′UTR

Figure S2.

A


Relative expression of the microRNA-206
100
90
80
70
65
25
20
10
0
LGM5.3
LGM5.5
LGM9.3
Control 1
Control 2

B

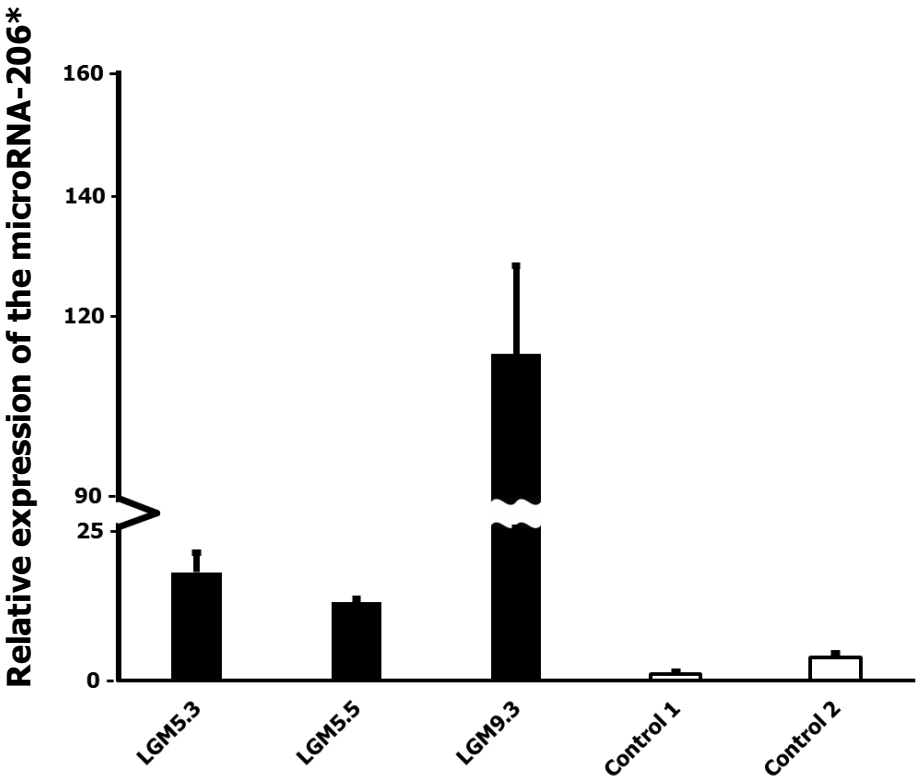

Relative expression of the microRNA-206*
160
140
120
90
25
0
LGM5.3
LGM5.5
LGM9.3
Control 1
Control 2

Figure S3.

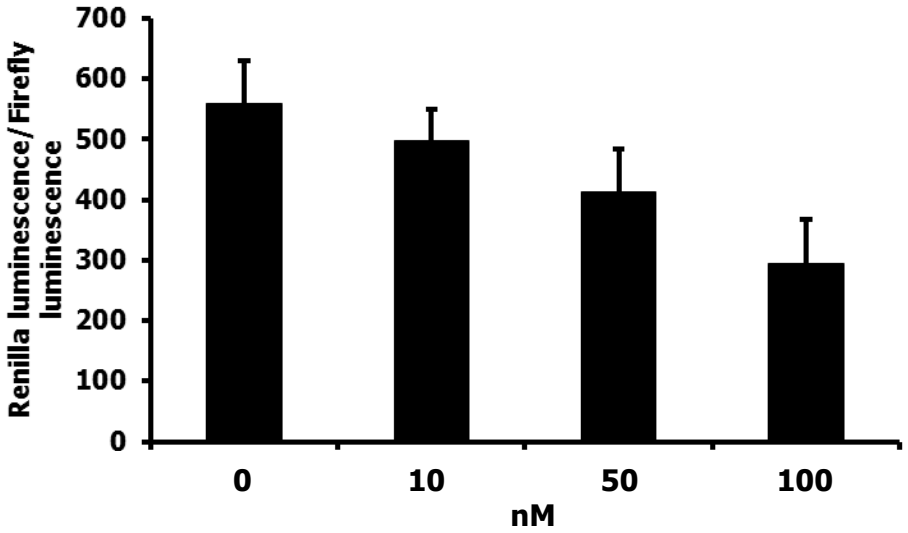


Figure S4.