Table S1. SMA disease modifiers*.

Disease modifier	References*
Established / identified in human studies	
Level of SMN protein determined by	
SMN2 copy number / expression level	[7,8,87]
SMN2 SVs and their combinations with SVs in other genes	[78–83]
SMN2 promoter CpG methylation	[84]
NAIP copy number	[88–90]
SERF1A copy number	[87]
Plastin 3 expression level	[91,92,100–103]
Coronin C expression level	[96]
Neuritin 1 expression level	[102]
Neurocalcin delta expression level	[106,107]
Zinc finger protein ZPR1 expression level	[72,108]
TLL2 SVs	[112]
Proposed / identified *in vitro* or in animal studies	
Level of SMN protein determined by	
Proteasomal degradation	[113]
mRNA transport	[113]
Splicing**	[22,115]
Non-coding RNAs	[70,71]
Hspa8 SVs impacting formation of SNARE complex***	[63]

* selected; **modulated by factors other than SVs in *SMN2;* *** demonstrated in animal model only.