Figure S2: Nucleotide and amino acid sequences in wild type *dystonin* and *dt-MP* alleles

(A) The sequence shows the genomic region of the *dystonin* gene (GenBank accession number NC_000067.5) from the end of exon 39 (included only in the short *dystonin* transcript variants *dystonin-e* and *dystonin-n*) to the beginning of exon 40 (included only in the long

A wild type *dystonin* allele

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TTAGTGATCACGGGGATCAGCCACCCCGTCAGCAATAAAATGATGTCAGTGGTGGAAGCCGTGAATGCAAAT   34239563
 L  V  I  T  G  I  S  H  P  V  S  N  K  M  M  S  V  V  E  A  V  N  A  N

ATCATCAGTAAAGAAATGGGCATGCGGTGCCTGGAATTTCAGTACCTGACAGGGGGCCTGATAGAGCCGAAG   34239635
 I  I  S  K  E  M  G  M  R  C  L  E  F  Q  Y  L  T  G  G  L  I  E  P  K

GTTTTCTCGAGGCTGACCATAGAAGAGGCCCTTCACGTCGGTATCATTGATGTCCTCATCGCCACCAGACTC   34239707
 V  F  S  R  L  T  I  E  E  A  L  H  V  G  I  I  D  V  L  I  A  T  R  L

AAAGATCAAAAGTCATATGTCAGGGATATAATGTGCCCCCAGACTAAGAGAAAATTGACATATAAAGAGGCC   34239779
 K  D  Q  K  S  Y  V  R  D  I  M  C  P  Q  T  K  R  K  L  T  Y  K  E  A

CTGGAGAAAGCTGATTTTGATTTCCACACAGGACTTAAGCTGTTAGAAGTGTCTGAACCCTTGGGGACAGGG   34239851
 L  E  K  A  D  F  D  F  H  T  G  L  K  L  L  E  V  S  E  P  L  G  T  G

ATATCCAACCTCTACTATTCTTCCCAG                                                34239878
 I  S  N  L  Y  Y  S  S  Q
  stop-codon in short dystonin isoforms dystonin-e and dystonin-n
Intron 39
 ta gggcgtttga ctctgtgcag aggccgtgtc ggctgcatcc tgccatctgt tcagagca         34239938
 ga tggtgtctgc taaatgtgca gcctaggaac tgtgtcacca actcgaagcg ctgttaat         34239998
 tt cttgtaggct agaaatagct ccctgctcag aaaacacggc cattcttaag ttgaaagc         34240058
 ag aaaacgattg gctgcccctg acaaagtaac ttaaaaaaaa aaatatctta ctaaaata         34240118
 aa tgggttttac ctagaaaatg gggtgagctt ggtgaccatc cacacgttca caggcccg         34240178
 tt tcttctgacc tccttcatag tcctttcact gaatagattc aacacattaa tgatgtca         34240238
 tc cattgtacat cctgaaagag cacctcacag agcaccacag gactgggatc caaaggaa         34240298
 cg tgcaaaagtg cccccgaccc gtgaggctgg agatggctac ctgcaggcag tgatgtcc         34240358
 tc tgcatagcga agtgttcgtc gctgttaact tttactttgg ttgagacttt ggaaataa         34240418
 aa tctaacaagt gcattgtgta ttggaggagg ggtgtcatct gtgtggagtt tctctctc         34240478
 tt cttttgctct ctccttttgt ctaatagcat ctgtcaaggc atctgcgatt gtaatgct         34240538
 aa tgtaaaggca gcgattccca aatctctctc atgaataaag ttgggtttgc ttatttgt         34240598
 ca tactcctgaa gcagctgggt ggccagagca gtgatcccag actgatcata aaagcttt         34240658
 tc ccgtcctcaa agacatctgc tgtgtgatgc agtggatgtg gttaagtgaa tgaataaa         34240718
 at atcgcagtaa cctgtgtgca tgatggaaac atttgtgcct tcttctttca gcagaaag         34240778
 ga ggggtcagtt gagaagctca gtcatttatt ggaatgtttg tttgtttctt tcttcatt         34240838
 tt taaacacttg tacctgtcag                                                   34240860

Exon 40
 AAGTCACTGGATGAAGAGAAGAAACAACATATTGAAAAAGCTAAAGAATTGCAGAAATGGGTATCAAATATC   34240932
  K  S  L  D  E  E  K  K  Q  H  I  E  K  A  K  E  L  Q  K  W  V  S  N  I
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B *dt-MP* – allele

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TTAGTGATCACGGGGATCAGCCACCCCGTCAGCAATAAAATGATGTCAGTGGTGGAAGCCGTGAATGCAAAT   34239563
 L  V  I  T  G  I  S  H  P  V  S  N  K  M  M  S  V  V  E  A  V  N  A  N

ATCATCAGTAAAGAAATGGGCATGCGGTGCCTGGAATTTCAGTACCTGACAGGG                     34239617
 I  I  S  K  E  M  G  M  R  C  L  E  F  Q  Y  L  T  G
       39,553 bp deletion
Intron 61  (end of exon 39 - inbetween intron 61)
                                                ggcacaca cacacacaca         34279190

                          putative premature stop-codon in dt-MP dystonin
cacacacaca aagtgagaga cagagagaga gagacaggga gaattaaaat ccgtttttaa          34279250
                                                                  *

agtcattctg aattactaca tctcagtgaa aaggaaaaga ggaaaacctt ggaaaaagta          34279310
taaaaagagg accagatact tgatcagtgt tcagaggaat aaacttgata ttccctttgg          34279370
tagttctgaa tgcagtacaa cacaaaatca cagactttta aaacacagga gggggttggg          34279430
attttttttt tctgtgtgat ttttctttgc cacttcactg tccagttctc aagtgtgaac          34279490
tttctagaag tcagtgtccg ccacagtcac aaggctagac ttgtcctttc acagagctgc          34279550
tgagatttca tgttgtagct ttccaacagt tggttgatgc tgagtgaatt taatagaatg          34279610
cttgagggga aatattcaaa gatggaagcc atttgcctag aaatagttga ggatttttt           34279670
ttattttaat ttgaggttgt agtattgctc agtaatgaag aacatgccag atattcacaa          34279730
agccctaggt ccaatgccca gtaaatccat gtacaagtga gtgcacacgc atatggcaca          34279790
atgtattaac tatataagga cttttagaaa gagtgcaagt tgacaggaac ctgaggttca          34279850
ggtttgtttt acttatacac gagttaatac acaattacta gatccataat gctatagcga          34279910
ggaatgaaac agaggttgga ttgggttgta gagaaggaag cctaggccta tcatcatata          34279970
gaatgagcat tattaggaaa gagacctcag gaccatggag gccatcgtca ggccttgagg          34280030
cgggagggac accattagga ggaaccgtgc atcctagcag atagggaaac ccacattgca          34280090
tggctgggcc ttgagatggg agagcacggt aggcattctg agagctgctg tgcacagcat          34280150
ggcagggaag caagccttca gagacagagc cctgcagaga gggtgtattt tgactactat          34280210
agttttatgc tctggggcct gctctcccca ctcactcccc ctgcaatccc atccaatcca          34280270
accctactga ggtcaaggca caaagtatag gtcaccatgg ctgatttctg cagactgcat          34280330
ctcacagcga ccggcatctt attatggcgg gctcatgatc taacgtgtag tccatgtatc          34280390
atctcacaca agtacttctt gtaagttgga attaatagga agggggggtta ccagatgtga         34280450
acaagctaga gatgtagctt agggtcatgc ttgcctagta ccaaggccct aggtttacat          34280510
tctaacaaag ggaacaagag gaatgccatc tattatgtat cgtttgaata catagttaca          34280570
gttttctgat ggtgatatta aatgaagaaa aaaagaaaag aaaacaaata gcagcttggt          34280630
ccttttttctt gttttcag                                                       34280648

Exon 62
 ACACTAAGTGGCAAGAGCTCAACCAGTTGACCATGGACAGGCAACAAAAGCTGGAAGAGTCCTCCAATAAT   34280719
  D  T  K  W  Q  E  L  N  Q  L  T  M  D  R  Q  Q  K  L  E  E  S  S  N  N
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dystonin transcript variants). (B) In the *dt-MP* mouse mutant, the genomic region from close to the end of exon 39 up to intron 61 is deleted. Translation of putative short *dystonin* transcripts would lead to different C-termini, compared to the known *dystonin-e* and *dystonin-n* variants, disrupting the penultimate plectin-repeat and removing the last plectin-repeat of the IFBD1 domain.