

## Supplemental information

All PCR protocols had 30 cycles starting with an initial denaturation at 94°C for 30 seconds. Annealing and elongation conditions are given at each primer pair. Each PCR started with an initial denaturation at 94°C for 3 minutes and ended with a final elongation at 72°C for 6 minutes.

Name	Sequence	5'-position *	Annealing	Elongation	Reference
Fig. 3a					
Exon43-f	5'-GCC AGA GGC TTC TGC TCT ATG-3'	281064			
Exon43-r	5'-CAG GGC GGG GGA ATA GTC AAA GAC AGA-3'	281648	62°C, 45 s	72°C, 50 s	Pool et al., 2005 (IFBD2)
Deletion-f	5'-GCT GGT GGA CGA GGG CTT CG-3'	274333			
Deletion-r	5'-TGC CAT GCT GTG CAC AGC AGC-3'	315018	62°C, 45 s	72°C, 90 s	
Control-f	5'-AGA CCG CCG GTG ACG TAT CAG G-3'	272159			
Control-r	5'-GGT CGT GAC TTC TGT ATG CAT GCC-3'	272885	62°C, 45 s	72°C, 50 s	
Fig. 3b					
Wild type allele-f	5'-GCA GGC AGT GAT GTC CTC TGC-3'	275227			
Wild type allele-r	5'-CTA CCC CGT TAG CAT TAC AGC C-3'	275887	62°C, 45 s	72°C, 45 s	
dt-MP-allele-f	5'-GCT GGT GGA CGA GGG CTT CG-3'	274333			
dt-MP-allele-r	5'-TGC CAT GCT GTG CAC AGC AGC-3'	315018	62°C, 45 s	72°C, 90 s	
Fig. 5					
dystonin-f**	5'-ATT CAA GAG TTC ATG GAC CTA CGG ACA C-3'	265943			
dystonin-a-r	5'-TAA TTA GGC GGT TTT CAG TCT GGG TGA G-3'	288987	68°C, 45 s	72°C, 45 s	Leung et al., 2001
dystonin-b-r	5'-CAA TAA GGC CTC TTA AAA CTG CCT GAA A-3'	279758	68°C, 45 s	72°C, 45 s	Leung et al., 2001
dystonin-e/n-r	5'-TC ACG ATC GTC TCC AGC TCA CGG-3'	269797	68°C, 45 s	72°C, 45 s	Leung et al., 2001

\*with respect to coding strand, genomic DNA

\*\* common forward primer for all isoforms

### S1 data. Primers

Fig. 3A: Primer sequences to identify the deletion in genomic DNA. Primers binding in exon 43 (coding for the IFBD2) show a band in control, but not in affected animals. Primers binding in exon 39 and intron 61, respectively, give a band in affected animals. The internal control band spans from exon 38 via intron 38 to exon 39.

Fig. 3C: Primer sequences for genotyping. The dystonin wild type allele is detected with primers binding in introns 39 and 40. The dt-MP allele is detected with primers that bind upstream of the deletion start in exon 39 and downstream of the end in intron 61, respectively.

Fig. 4: Primer sequences to detect the different dystonin transcripts in cDNA. For all isoforms, a common intron-spanning forward primer binding in exon 36/37 was used. The backward primers bind in exon 48 (dystonin-a), exon 43 (dystonin-b) and exon 38 (dystonin-e/n), respectively.