



Figure S1. RNA-seq data confirming the aberrant splicing. The figure shows an IGV screenshot of the 50 bp read alignments of RNA-seq data derived from lesional skin of the affected Irish Terrier. Soft-clipped bases not aligning to the genomic reference are shown. Please note that this is a split screen view showing three different genomic locations simultaneously. The genomic coordinates are indicated at the top, the relative positions within the *ATP2A2* gene are indicated at the bottom. The position of the c.2091A>G variant is indicated in all three windows. The reference transcript NM_001003214.1 represents the A-allele, while the CanFam3.1 reference genome assembly represents the G-allele. The allelic ratio between transcripts from the A-allele and the G-allele is 89:21. Numbers from 1 to 11 indicate characteristic reads that allow to infer several splicing events.

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| 1: Read from A-allele, correct splice junction exon 14/15 as indicated by the soft-clipped bases | (n = 27) |
| 2: Read from G-allele, splicing of exon 14 to an aberrant exon | (n = 7) |
| 3: Read from A-allele, correct splice junction exon 14/15 | (n = 19) |
| 4: Read from G-allele, correct splice junction exon 14/15 | (n = 2) |
| 5: Read from G-allele, correct splice junction exon 14/15 as indicated by the soft-clipped bases | (n = 1) |
| 6: Read from G-allele, splicing of exon 14 to aberrant exon as indicated by the soft-clipped bases | (n = 2) |
| 7: Read from G-allele, splicing of exon 14 to aberrant exon as indicated by the soft-clipped bases | (n = 2) |
| 8: Read from A-allele, correct splice junction exon 14/15 as indicated by the soft-clipped bases | (n = 31) |
| 9: Read from G-allele, correct splice junction exon 14/15 as indicated by the soft-clipped bases | (n = 1) |
| 10: Read inferred to originate from G-allele, splice junction from the aberrant exon to exon 16 | (n = 2) |
| 11: Read inferred to originate from G-allele, splice junction from the aberrant exon to exon 15 | (n = 2) |

Similar results were obtained with an independent RNA sample from non-lesional skin of the affected dog (data not shown).