Figure \$1 IGV screenshot of the region with the insertion. The sequence alignments of an unrelated control cow, a heterozygous carrier (Dudoc Mr Burns) and the affected calf are displayed. Read pairs mapping to BTA 11 are displayed in grey. The control animal shows normal overlapping alignments of the Illumina reads with respect to the reference sequence. Note the dashed black vertical line indicating the genome position where all read alignments are truncated and the corresponding mates of mapped reads map to other genome regions containing a ERV2-1 like transposable LTR element (each different chromosome is indicated by a unique color). In the carrier animal, about 50% of the mapped read mates map to other genome regions, for the other half of the mapped reads the corresponding mate maps in the same region of BTA 11 with insert sizes of about 400 bp, as expected. In the affected animal, no read pair bridges this region. The insertion site is within exon 5 of the APOB gene. Note the small region of overlap immediately to the right of the dashed vertical line, visible in the carrier and affected animals. This is due to the fact that the first five nucleotides of the inserted DNA element correspond perfectly to the five nucleotides of APOB exon 5 which follow the insertion site.

