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## Whole-genome sequencing identifies a homozygous deletion encompassing exons 17 to 22 of the integrin beta 4 gene in a Charolais calf with junctional epidermolysis bullosa

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### **P0304. Whole-Genome Sequencing Identifies a Homozygous Deletion encompassing Exons 17 to 22 of the *Integrin Beta 4* Gene in a Charolais Calf with Junctional Epidermolysis Bullosa**

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Since 2010, four Charolais calves with a congenital mechanobullous skin disorder that were born in the same herd from consanguineous matings were reported to us. Clinical and histopathological examination revealed lesions that are compatible with junctional epidermolysis bullosa (JEB). Fifty-four extended regions of homozygosity (>1 Mb) were identified after analysing the whole genome sequencing (WGS) data from the only case available for DNA sampling at the beginning of the study. Filtering of variants located in these regions for (i) homozygous polymorphisms observed in the WGS data from eight healthy Charolais animals and (ii) homozygous or heterozygous polymorphisms found in the genomes of 234 animals from different breeds did not reveal any deleterious candidate SNPs (single nucleotide polymorphisms) or small indels. Subsequent screening for structural variants in candidate genes located in the same regions identified a homozygous deletion that includes exons 17 to 22 of the *integrin beta 4* (*ITGB4*), a gene which has been previously associated with the same defect in humans. Genotyping of a second case and of two sires and four dams revealed a perfect association between this mutation and the assumed genotypes of the individuals. Mining of Illumina BovineSNP50 Beadchip genotyping data from 6870 Charolais cattle detected only 44 animals heterozygous for a 5.6-Mb haplotype around *ITGB4* that was shared with the carriers of the mutation. Interestingly, none of the 16 animals genotyped for the deletion carried the mutation, which suggests a rather recent origin for the mutation. In conclusion, we successfully identified the causative mutation for a very rare autosomal recessive mutation with only one case by exploiting the most recent DNA sequencing technologies.