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VEREIN ZUR FÖRDERUNG DER FORSCHUNG IM GESUNDHEITSSEKTOR VON LAMAS UND ALPAKAS e.V.

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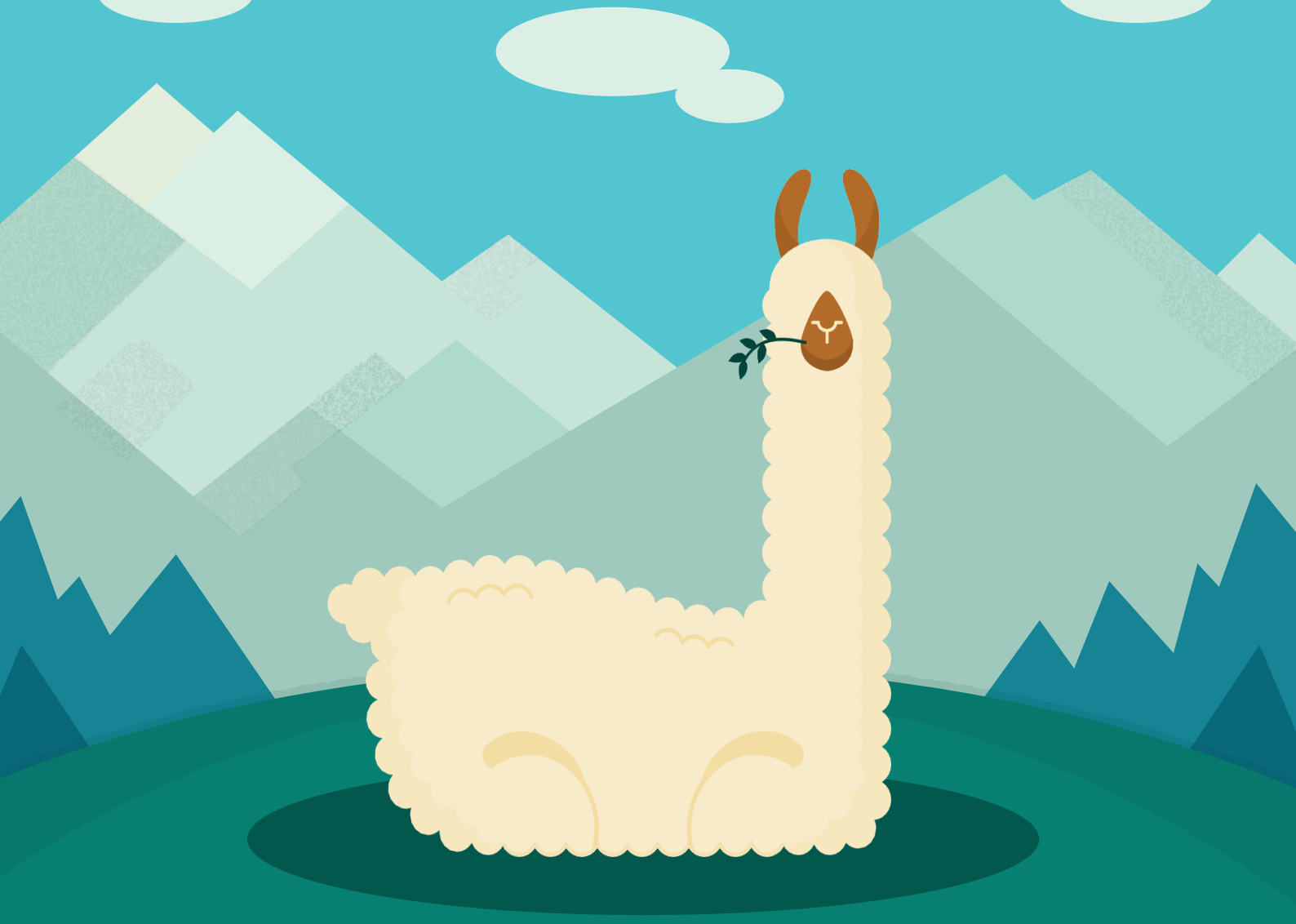
BOOK OF ABSTRACTS



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Polymorphisms in *MC1R* and *ASIP* genes associated with color phenotypes in alpaca huacaya

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The wide phenotypic diversity in alpacas results from human selection by color phenotypes that favored the fixation of main alleles in *MC1R* and *ASIP* genes. In this context, the objectives of this study were: to characterize the fiber color by colorimetry and identify the main polymorphisms in *MC1R* and *ASIP* genes associated with black and brown alpacas. Fiber and blood samples of 98 alpacas were obtained from Pacomarca Research Station; and 9 vicuñas from Abancay province were considered as reference for this study (Peru). Fleece color phenotypes were determined by colorimetry using Chroma Meter CR-210. DNA was extracted from 200 µL of EDTA anticoagulated blood using the commercial kit (Quick-DNA™ Miniprep Plus Kit). Polymerase chain reaction (PCR) primers were designed to amplify *MC1R* and *ASIP* genes following Daverio et al. (2016). For all PCR products purification we used exo-sap method, with enzyme ExoASP-IT® (usb). The purified amplicons were sequenced by the Sanger method at Macrogen Inc using the original PCR forward and reverse primers. Complete *MC1R* and *ASIP* coding sequences for each animal were obtained and analyzed using Geneious Software Version 11.1.5. The *CIE L*a*b** system, L* = lightness showed low values in eumelanic alpacas (black and black-brown) and high values in white, pheomelanic brown alpacas and vicuñas. Coding sequence of *MC1R* (CDS) consisted of 954bp and encoded a 317 amino acid protein. Inside of CDS a heterozygous deletion *c.224_227del* and nine SNPs were observed; 5 non synonymous SNPs: c.82A>G, c.259G>A, c.376G>A, c.587T>C, c.901C>T (p.T28A, p.M87V, p.G126S, p.F196S, p.R301C, respectively) and 4 synonymous SNPs: c.126C>T, c.354C>T, c.618G>A, c.933A>G. Two non-synonymous polymorphisms (c.292C>T and c.353G>A) and a 57bp deletion (c.325_381del) were identified within exon 4 of *ASIP* gene. The five non synonymous SNPs at *MC1R* and the mutations at *ASIP* define the recessive genotypes (*ASIP*) together with the dominant genotypes at *MC1R* (*EEaa*) in black and black-brown alpacas, heterozygote genotypes for both genes (*EeAa*) was observed in brown, dark brown and black alpacas. The wild genotype ($E^+E^+A^+A^+$) was observed in white, brown alpaca and vicuña. The nine vicuñas have the wild allele without deletion in *MC1R* and *ASIP* genes. In sum, there is more than one genotype for black and brown phenotypes. Likewise, for each genotype described we also observed black and brown phenotypes; then it would be explained by the epistatic interaction between *MC1R* and *ASIP* genes.