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## **Fine mapping of birthcoat type in the Romanov breed sheep**

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**The agouti gene in black and brown alpaca**

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The Agouti gene encodes agouti signaling protein which regulates pheomelanin and eumelanin synthesis in mammals. To investigate the role of Agouti in coat color variation of alpaca, we characterize the agouti gene on 27 black and 12 brown alpaca. The exon-4 hosts three loss of function recessive mutations, g.3836C>T, g.3896G>A and g.3866\_3923del57, involved in eumelanin synthesis. The deletion at the position p.C109\_Rdel19 eliminates the two beta sheets and the R-F-F- motif from the agouti functional domain, which are essential against alfa-MSH. Therefore, the deleted allele appears to lose function. The other SNPs observed at the amino acid position 98 and 118 change the conserved R to C and the R-F-F- motif into H-F-F-. The R-F-F- motif is important for functioning at MCRs; the disruption in this motif may result in a non functional agouti protein since the alteration of residues in and around R-F-F- causes a decrease in agouti protein inhibition of alfa-MSH binding to MCRs during signal trasduction. The three mutations are randomly distributed among the black alpaca. In our sample, we observed two genotypes: g/3836C>T / g.3896G>A (10 animals) and g.3836C>T / g.3866\_3923del57 (17 animals). Among the brown alpaca, 2 are homozygous for the wild allele, twelve are heterozygous for g.3896G>A mutation, carriers for black phenotype.

**Fine mapping of birthcoat type in the Romane breed sheep**

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Birthcoat type is an important component of lamb survival for sheep raised under harsh environment. At birth two types of coat were observed: a long hairy coat or a short woolly one. It was shown that hairy coat lambs are more adapted to survive around lambing time due to a better coat protection with less heat losses at coat surface and show better growth performances up to the age of 10 days than woolly coat lambs. Birthcoat type was estimated to be a highly heritable trait and it was reported that its determinism seems to be under the control a few major genes. A QTL detection design was initiated in a Romane breed population to search for loci influencing adaptive traits including birthcoat type through a whole genome scan with the OvineSNP50 beadchip on 824 lambs issued from 8 sire families. A highly significant ( $P < 0.1\%$  Genome Wide (GW)) and a putative QTL affecting birthcoat type were found on chromosomes 25 and 13 respectively. Fine mapping with additional markers, comparative mapping and sequencing of the QTL segment on OAR25 revealed the presence of a 2 kb DNA deletion segment. All animals from the experimental design were genotyped for the presence or absence (ins/del) of this segment on OAR25. Homozygous del/del animals were all bearing a hairy coat at birth but not all the homozygous ins/ins animals were bearing a woolly coat. When including the ins/del genotype on OAR25 as a fixed effect within the linkage analysis model, a highly significant ( $P < 0.1\%$  GW) QTL was found on OAR13 with a significant interaction ( $P < 0.001$ ) between QTL on OAR13 and the ins/del genotype fixed effect of OAR25. It was suggested that both QTL on OAR13 and OAR25 are involved as major genes in the determinism of birthcoat type.

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