Further characterization of the structural variant upstream of the KIT gene underlying head depigmentation across a diverse panel of cattle breeds

Valentin Sorin, Marie-Pierre Sanchez, Laurence Drouilhet, Gwenola Tosser-Klopp, Maulana Mughitz Naji, Didier Boichard, Hubert Pausch, Mekki Boussaha, Alexander Leonard

Session 85 Poster 32

Incorporating Breed-Specific Recombination Maps in Selection Signature Analysis of Cattle Breeds

N. Abdollahi Sisi[1], D. Wittenburg[1], H. Schwarzenbacher[2], F. Seefried[3]
[1] Forschungsinstitut für Nutztierbiologie (FBN), Wilhelm-Stahl-Allee 2, 18196 Dummerstorf, Germany, [2] Zuchtdata GmbH, Dresdner Straße, 1200 Vienna, Austria, [3] Qualitas AG, Chamerstrasse 56, 6300 Zug, Switzerland

Recombination and selection are key evolutionary processes shaping genetic variation in livestock populations. Recombination promotes genetic diversity by breaking associations between alleles, thereby reducing linkage disequilibrium (LD) across the genome. In contrast, selection can lead to extended LD regions by driving the rapid increase in frequency of beneficial alleles. To improve the accuracy of selection signature detection, we constructed breed-specific recombination maps for eight cattle breeds using LINKPHASE3, separately estimating recombination rates for males and females. We analyzed genotype data from these breeds, with sample sizes ranging from 4,181 to 367,056 individuals. Our results show that males display higher recombination rates across all cattle breeds. The resulting maps spanned 26.64–29.63 Morgan in males and 23.10–25.62 Morgan in females. These maps were included in selection signature analysis using the rehh package. Our analysis identified genomic regions under selection, revealing breed- and sex-specific differences in selection pressure. Functional annotation of these regions uncovered candidate genes associated with economically important traits, highlighting their potential role in breed-specific adaptation. Additionally, we will investigate the advantages of using breed-specific recombination maps compared to a consensus map to assess their impact on selection signature detection. These findings emphasize the importance of incorporating breed-specific recombination maps into selection signature analysis to better understand the interplay between recombination and selection in shaping genetic diversity. Keywords: Cattle breeds, recombination map, recombination rate, selection signature

Session 85 Poster 33

Further characterization of the structural variant upstream of the KIT gene underlying head depigmentation across a diverse panel of cattle breeds

V. Sorin[1], M. P. Sanchez[1], L. Drouilhet[2], G. Tosser-Klopp[2], M. M. Naji[1], D. Boichard[1], H. Pausch[3], M. Boussaha[1], A. Leonard[3]
[1] INRAE, Université Paris Saclay, INRAE, AgroParisTech, GABI, 78350 Jouy-en-Josas, France, [2] INRAE, Université de Toulouse, INRAE, GenPhySE, 31326 Castanet-Tolosan, France, [3] ETH, Animal Genomics, ETH, 8092 Zurich, Switzerland

A previous study based on a taurine pangenome uncovered a structural variant upstream of the KIT gene associated with depigmentation in white-headed cattle. In this study, we aimed to deepen our understanding of this region by analysing a collection of 80 genome assemblies from 20 different cattle breeds, including several not previously investigated. Using a combination of complementary approaches, we constructed a 2 Mb pangenome graph encompassing the KIT gene and the depigmentation-associated region to investigate the spectrum of structural variations within this region. Our analysis revealed a more fragmented structure than previously reported, mainly due to the inclusion of a larger number of assemblies (80 vs. 24) with varying sequencing technologies' quality (CLR and HiFi). Despite this increased complexity, the identified structural variant's alleles closely mirrored those reported in earlier study and depicting clear segregation of the population into two distinct groups. The first group includes breeds characterized by a white-headed phenotype and the second group contains all non-white headed breeds. We further validated this structural variant by aligning short-reads to the pangenome graph and genotyping several hundred animals for this structural variant. Our results provide new insights into the genetic mechanisms underlying head depigmentation in cattle and demonstrate the power of pangenome-based analyses for detecting trait-associated structural variations. This research was supported by funding from the Agreenium programme, with contributions from the Biosphera Graduate School, the ABIES doctoral school, the INRAE Animal Genetics division, and the INRAE GBOS team from the GABI department.