

The estimation of additive and dominance effects underlying lameness in Fleckvieh and Braunvieh cows

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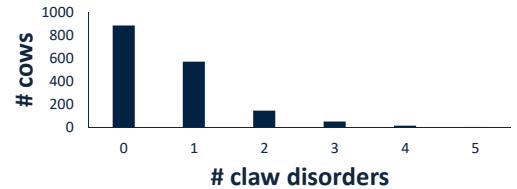
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CONCLUSIONS

- Claw diseases → polygenic determination → 301 SNPs with significant additive effects (15 dominance, 3 097 pairwise additive:additive epistases)
- Claw diseases → related to rump and legs conformation → most significant SNPs in QTL regions
- Claw diseases → biochemically related to inositol metabolisms in cell membranes

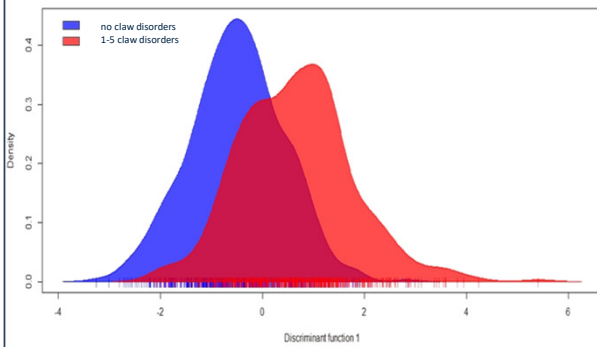
MATERIAL

- Animals → 1 998 Fleckvieh, 979 Braunvieh cows
- SNPs → GeneSeek® Genomic Profiler™ HD, 70 026 SNPs
- Trait → number of claw disorders (0-5)



METHODS & RESULTS

GWAS model → identification of significant SNPs



$$y = \mu + l + b + s + a + p + e$$

y number of claw disorders (0-5)
 l parity (1, 2, ≥ 3)
 b breed (Fleckvieh, Braunvieh)
 s additive effect of a SNP (-1, 0, 1)
 a additive polygenic $a \sim N(0, A\sigma_a^2)$
 p permanent environmental
 $p \sim N(0, I\sigma_p^2)$
 e residual $e \sim N(0, I\sigma_e^2)$

- SNP effect test – Wald test
- multiple testing correction accounting for LD
- 301 SNPs with significant additive effects
- good discrimination between healthy and sick cows

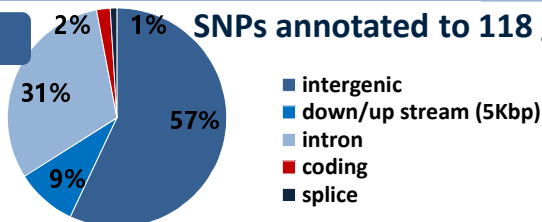
Beyond GWAS → Bioinformatics of 301 significant SNPs

Genomic annotation



Two-scale approach to SNP selection SNPassoc

additive SNPs annotated to 118 genes



dominance

- 15 significant SNP effects
- Most significant SNP on bta16
- Intergenic region

epistasis

- 3 097 significant effects

Functional annotation

KOBAS custom

- GO – plasma membrane and small molecules
- KEGG – inositol phosphate metabolism
- SNP – most significant SNPs located on bta29

