



University of
Natural Resources and
Life Sciences, Vienna



Department for
Sustainable Agriculture Systems

Division of Livestock Sciences
WG Animal Breeding

High overlap of CNVs and selection signatures by varLD analyses of taurine and zebu cattle

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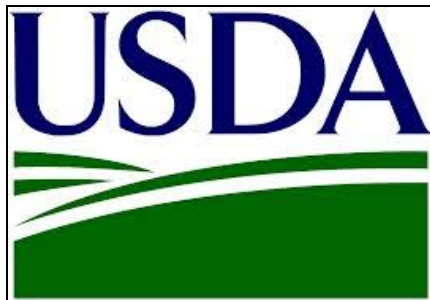
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Acknowledgements

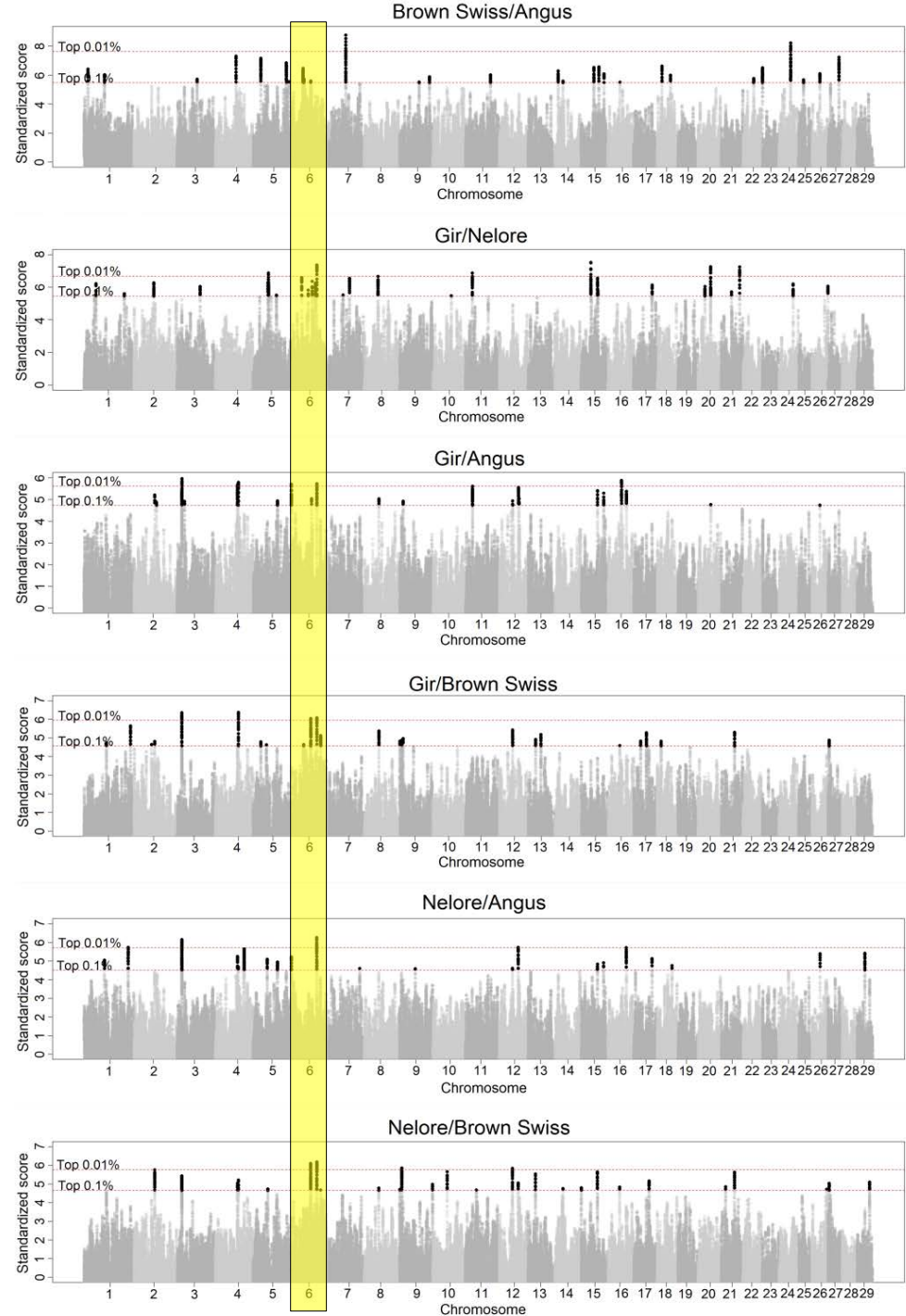
- George Liu and Derek Bickhart, USA
- Data Providers:



- ISAG 2012 –
VarLD results

- ✓ BTA6 -
TECRL gene

- Liu and
Bickhart



Background

- Selection Signature (SS): Selection pressure push towards fixation
- Genomic region with low variation
- Hitch-hiking effect on surrounding regions
- High homozygosity and LD
- Length of sweep related to age and strength of selective pressure
- Different methodologies

- CNV: segment present at a variable copy number in comparison with a reference genome
- Forms of genome structural variation (indels)
 - Additional copies (duplications), losses (deletions), inversions and translocations
- Change physical arrangement of genes:
 - Bridges, 1936: **Bar** duplication in *Drosophila*
 - Eye field narrower than in wild-type eyes

- VarLD: LCT gene
 - Lactose tolerance
 - European vs. East Asian/African
- VarLD-CNV in Human HapMap populations
 - European/East Asian: 8/20
 - European/African: 7/20
 - East Asian/African: 11/20

Teo et. al 2009, Ong and Teo
2010, Hee 2012

- Effects of CNVs on gene expression (Drosophila)
 - Increase, decrease or stable
 - Dosage dependent interactions
- SNPs vs. other kinds of genetic variations:
 - Small indels
 - Copy Number variants
- Potential selective effect

Materials and methods

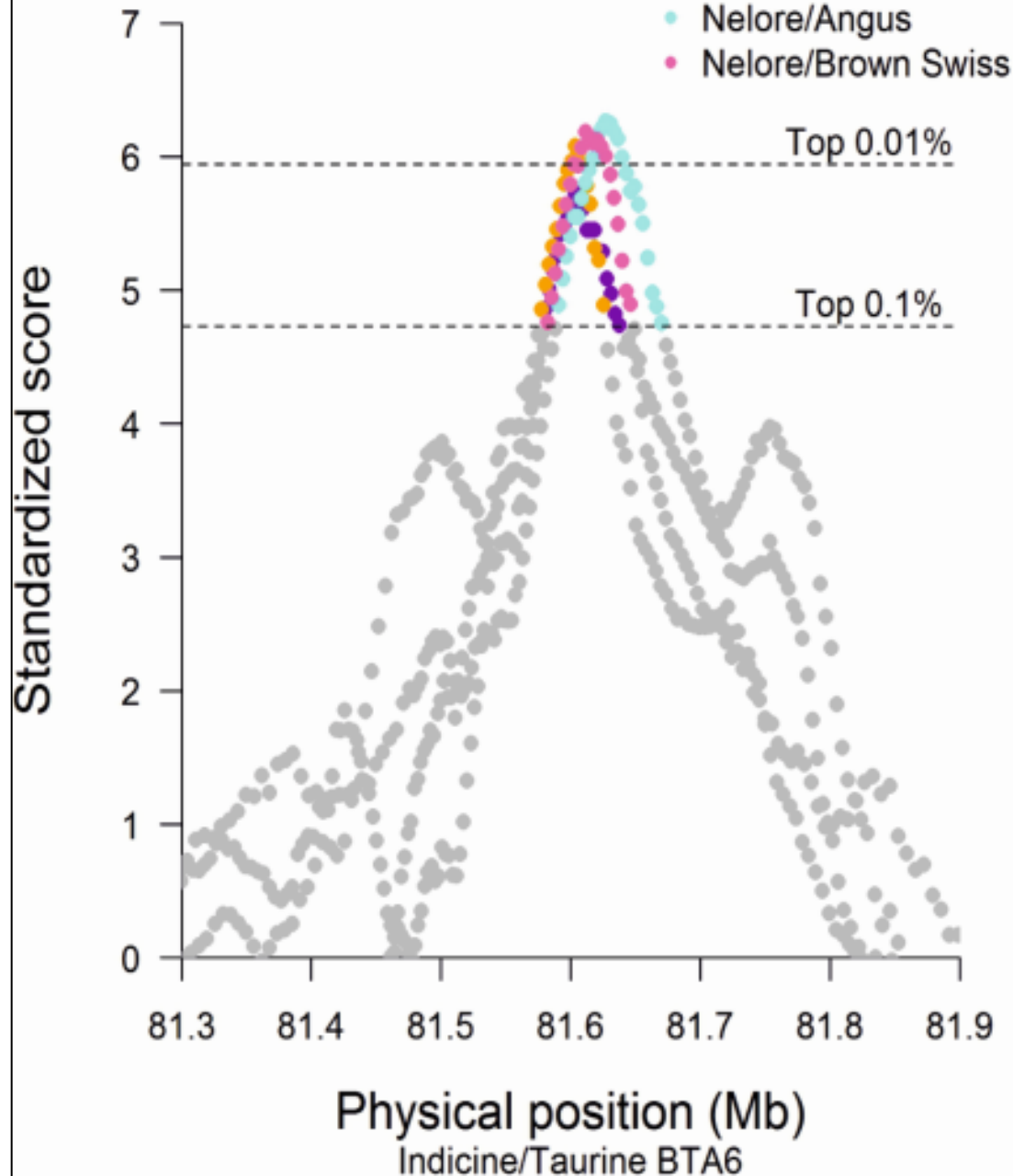
- varLD analysis (Teo et al. 2009):
 - Breeds: Nelore, Gir, Brown Swiss, Angus (30-100)
 - Illumina Bovine HD Beadchip (777K)
 - Variation in regional LD compared to background
 - Breed pairwise comparison of regional scores
 - Regions with difference in the variation highlighted

Materials and methods

- Comparison of VarLD signals with reported CNVs
 - Breeds: Nelore, Gir, Brown Swiss, Angus
 - Illumina Bovine HD Beadchip (777K)
 - Top 0.1% of VarLD Signals: 165 signals
 - 0.43% of autosomal genome
- Bovine CNVs:
 - NGS: 1265 = 2.1% (Bickhart et al. 2012)
 - HD SNP chip, 3,4384 = 5.6% (Hou et al. 2012)

Results

- 34 of our signals overlapped with reported CNV
- Common regions cover 1.84Mb, or 0.07% of the autosomal genome
- Many CNV positions coincided in both studies
- 17 Genes found inside the signals and containing the CNVs



TECRL

- BTA6
- 81.51 to 81.65
- Trans-2,3-enoyl-CoA reductase-like gene

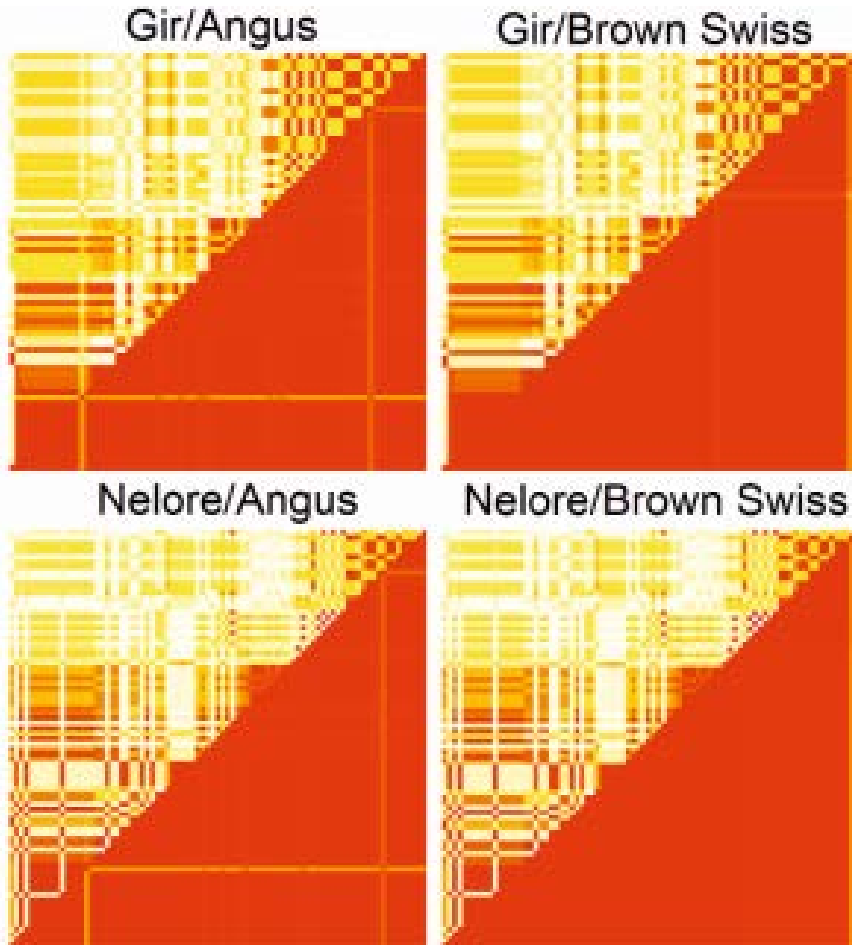
TECRL



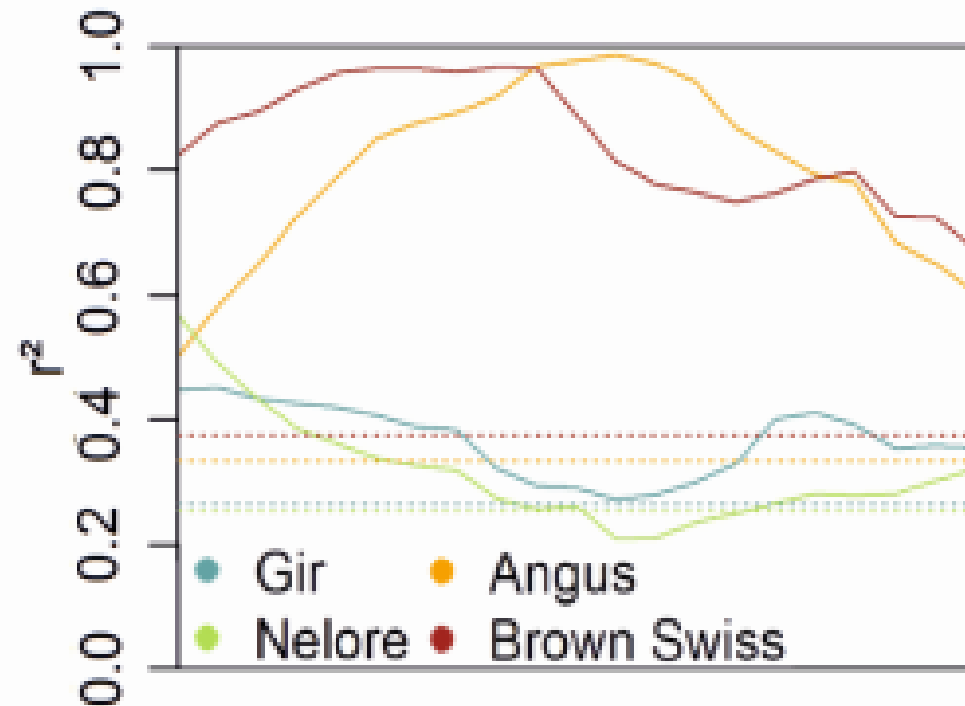
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r^2 color key



- Ubiquitin-like domain
 - Enoyl reductase: elongation of long chain fatty acids
- Steroid 5-alpha-reductase
 - dihydrotestosterone
 - Male genitalia during fetal development.
 - Mutations in humans '
Pseudohermaphroditism

Other related genes

- Cell cycle related: phosphorylation and ubiquitin-directed proteolysis
 - LMLN
 - ZNF804B
 - Protocadherin family
 - ADAMTS12
 - ASCC3

CONCLUSIONS

- 20.6% of the signals overlap with CNV
 - CNVs covering less than 7.7% of the genome
- CNVs are a likely mechanism for selection processes
 - Regions that differ in copy number between subspecies ' ancient adaptations, species-specific phenotypes
 - Recent copy number changes ' human selection = differences between breeds

Thank you for your attention



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