ETH zürich

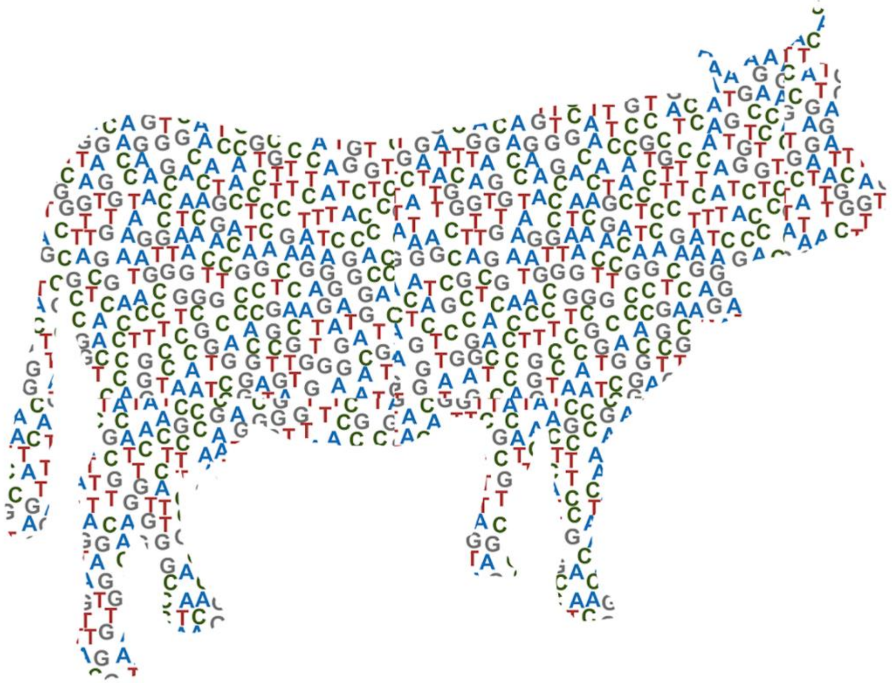
D-USYS
Institute of Agricultural Sciences

Current projects at the ETH Animal Genomics group

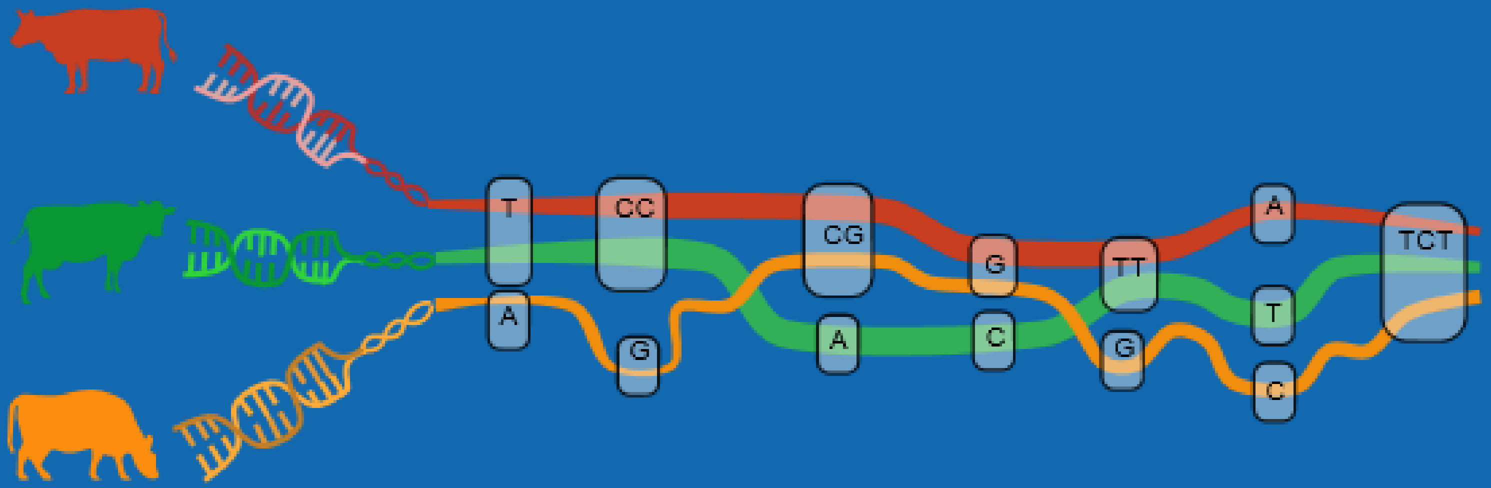
Hubert Pausch
Animal Genomics
04 December 2025

Topics covered today

- Bovine Pangenomics - Genome assembly and pangenome integration
- Structural variant analyses



Bovine pangenomes*.



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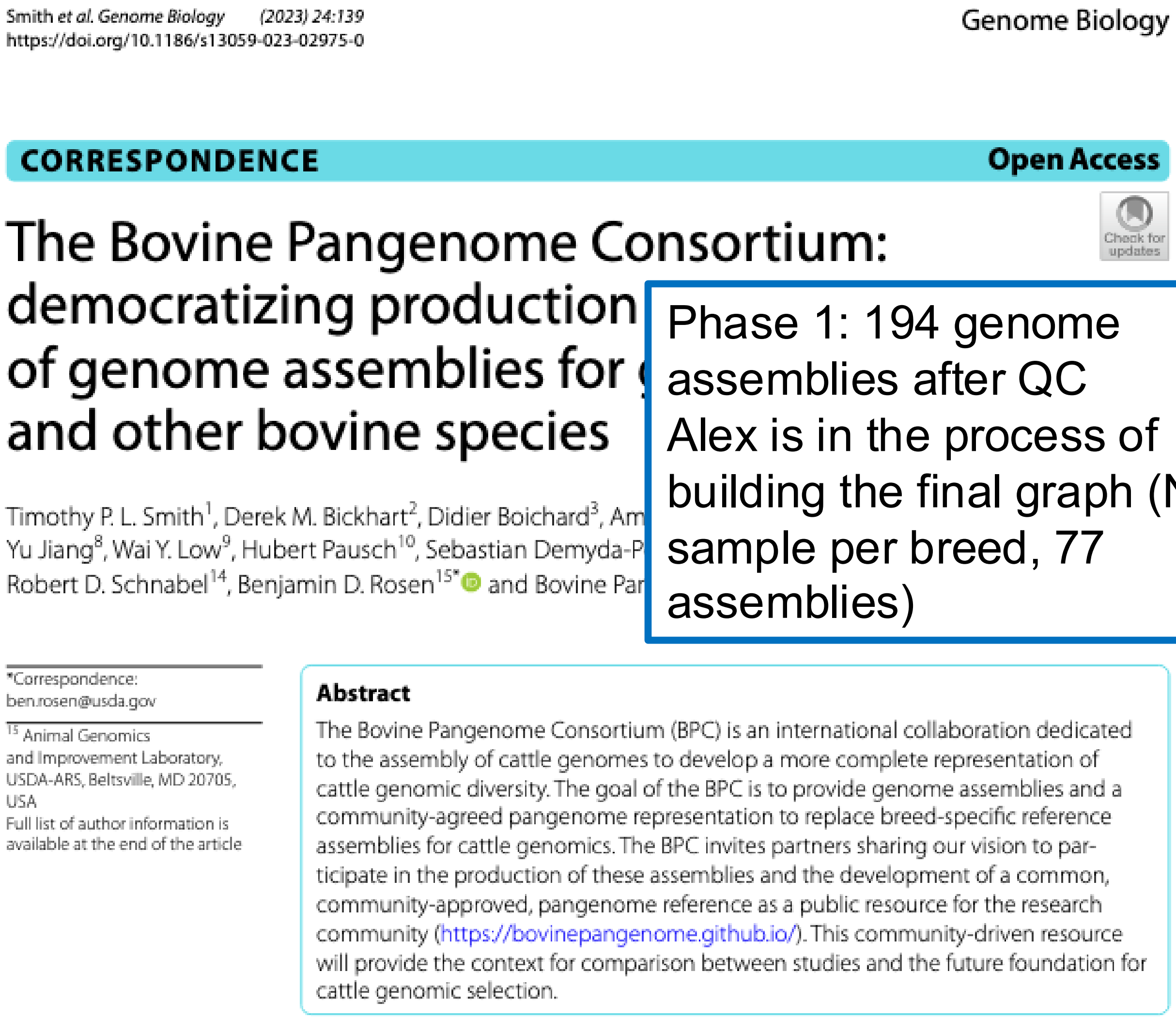
*full set of genomic elements from all species within a genus

The Bovine Pangenome Consortium



Smith *et al. Genome Biology* (2023) 24:139
https://doi.org/10.1186/s13059-023-02975-0

Genome Biology

CORRESPONDENCE **Open Access**

The Bovine Pangenome Consortium:
democratizing production
of genome assemblies for
and other bovine species

Timothy P. L. Smith[1], Derek M. Bickhart[2], Didier Boichard[3], Am
Yu Jiang[8], Wai Y. Low[9], Hubert Pausch[10], Sebastian Demyda-P
Robert D. Schnabel[14], Benjamin D. Rosen[15*] and Bovine Par

*Correspondence:
ben.rosen@usda.gov

[15] Animal Genomics and Improvement Laboratory, USDA-ARS, Beltsville, MD 20705, USA
Full list of author information is available at the end of the article

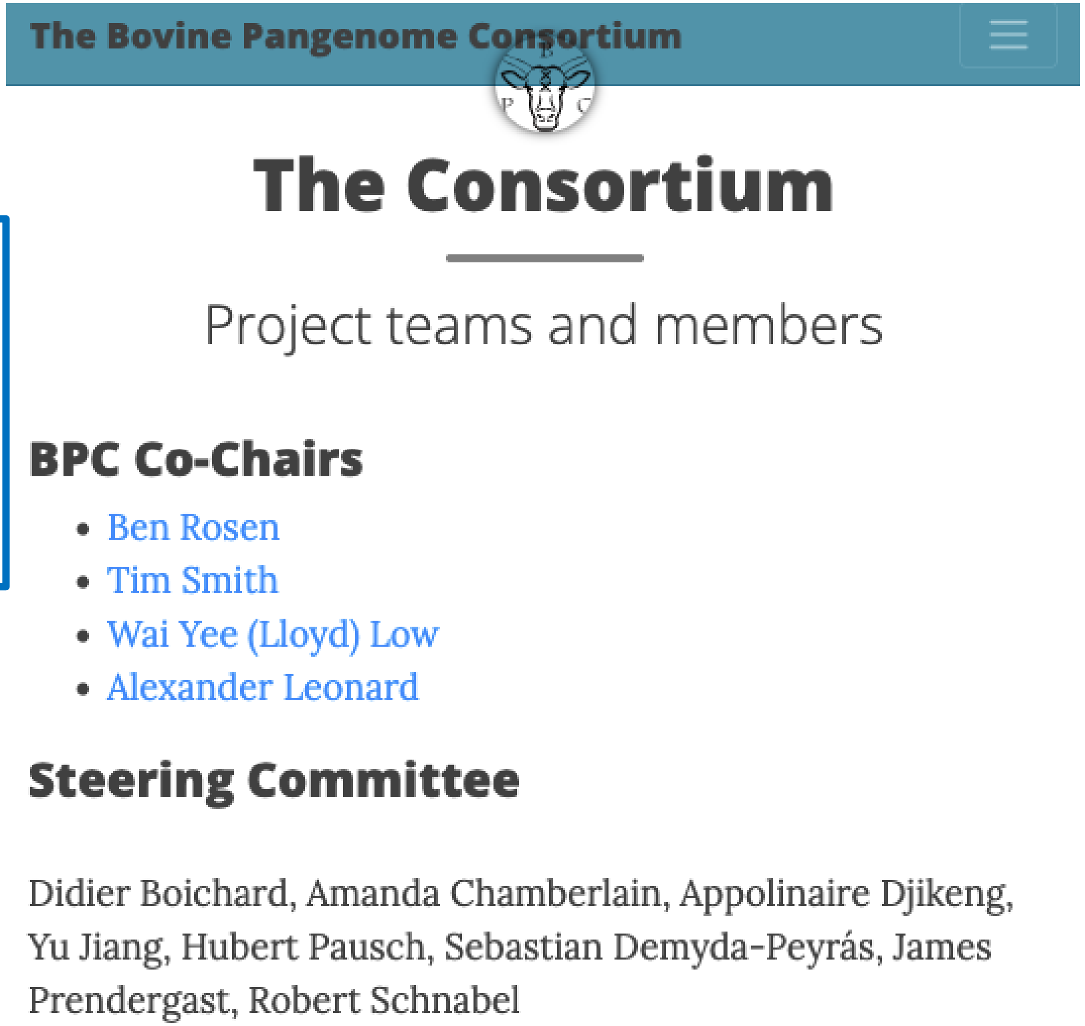
Abstract

The Bovine Pangenome Consortium (BPC) is an international collaboration dedicated to the assembly of cattle genomes to develop a more complete representation of cattle genomic diversity. The goal of the BPC is to provide genome assemblies and a community-agreed pangenome representation to replace breed-specific reference assemblies for cattle genomics. The BPC invites partners sharing our vision to participate in the production of these assemblies and the development of a common, community-approved, pangenome reference as a public resource for the research community (https://bovinepangenome.github.io/). This community-driven resource will provide the context for comparison between studies and the future foundation for cattle genomic selection.

Phase 1: 194 genome assemblies after QC
Alex is in the process of building the final graph (N=1 sample per breed, 77 assemblies)

Dr. Alexander Leonard

Dr. Tobias van Elst



The Bovine Pangenome Consortium

The Consortium

Project teams and members

BPC Co-Chairs

- Ben Rosen
- Tim Smith
- Wai Yee (Lloyd) Low
- Alexander Leonard

Steering Committee

Didier Boichard, Amanda Chamberlain, Appolinaire Djikeng, Yu Jiang, Hubert Pausch, Sebastian Demyda-Peyrás, James Prendergast, Robert Schnabel

Bovine pangenomics projects

- How does an optimal pangenome look like? (Sotiria Milia, Tristan Cumer, Alexander Leonard)
- Can we determine ancestral and derived alleles from pangenomes and use this information to detect signatures of selection? (Guoli Li)
- Can we detect introgression and hybridization events from pangenome variation (Tobias van Elst)
- Can we use pangenomes to reliably call structural variants? (Laura Dekker)

Guoli Li

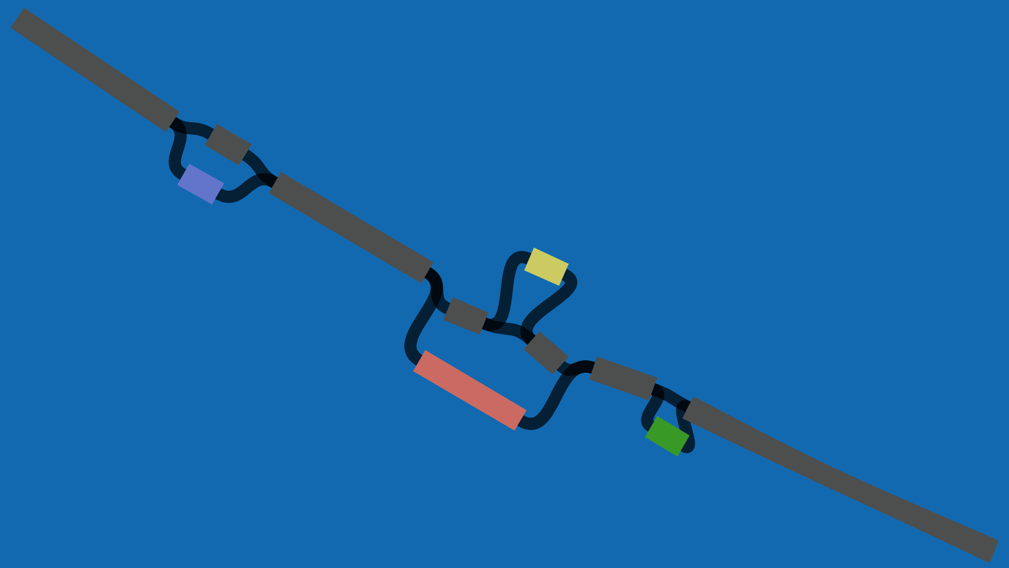
Laura Dekker

Dr. Alexander Leonard

Dr. Tobias van Elst

Sotiria Milia

Structural variant analysis.



How large is the contribution of structural variants to complex trait variability?

Dr. Xena Mapel

- Our mapping cohort consists of 120 Braunvieh bulls
  - 13-fold HiFi coverage
  - 12-fold Illumina coverage
  - 200 million RNA reads from testis tissue

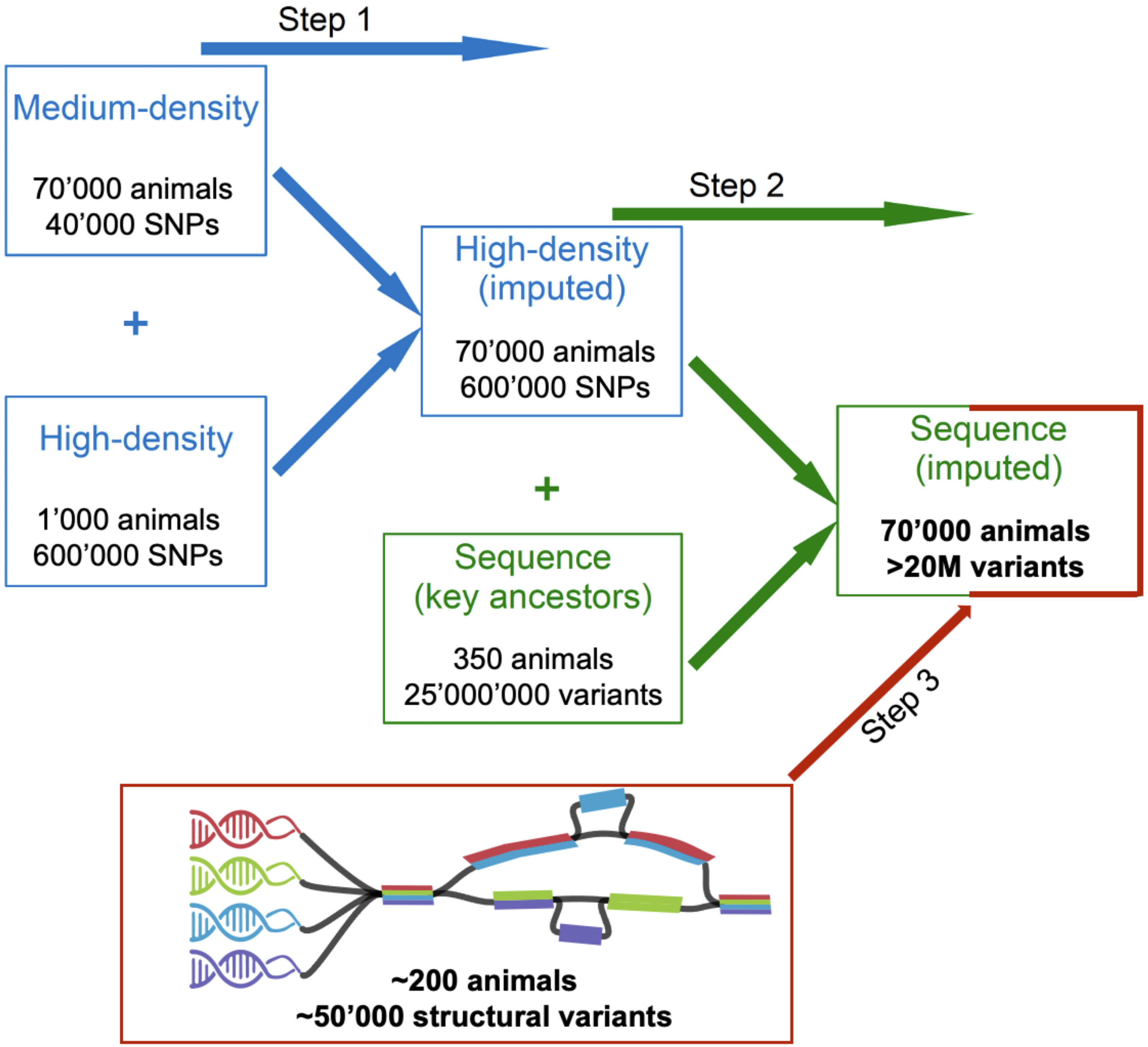
«*We identified 27.3 k molecular QTL (molQTL) including 316 for which SVs were the most significant variant. This corresponds to a 2.1- and 5.6-fold enrichment of SVs among expression and splicing QTL, respectively. When considering SVs in perfect LD with the lead small variant, the enrichment increased to 6.1- and 12-fold for expression and splicing QTL in testis, respectively.*» **Mapel et al., under review**

How large is the contribution of structural variants to complex trait variability? *(Natasha Watson, Naveen Kadri, Alexander Leonard)*

Natasha Watson

Dr. Naveen Kumar Kadri

Dr. Alexander Leonard



Step 1

Medium-density

70’000 animals
40’000 SNPs

+

High-density

1’000 animals
600’000 SNPs

High-density (imputed)

70’000 animals
600’000 SNPs

Step 2

+

Sequence (key ancestors)

350 animals
25’000’000 variants

Sequence (imputed)

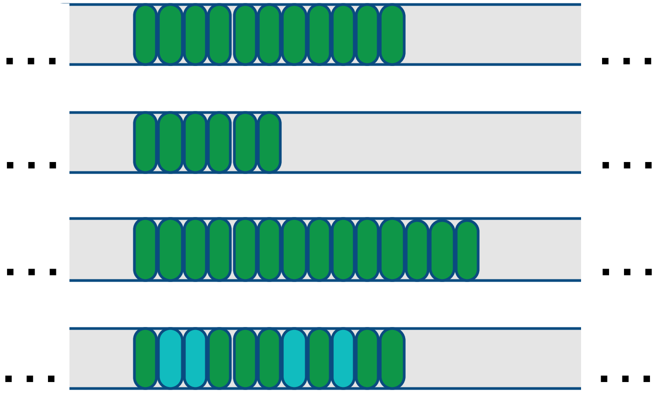
70’000 animals
>20M variants

Step 3

~200 animals
~50’000 structural variants

How prevalent are VNTR in the bovine genome, and how much do they contribute to trait variability? *(Victor Mason)*

Dr. Victor Mason



haplotype assembly #1 … …

haplotype assembly #2 … …

haplotype assembly #3 … …

haplotype assembly #4 … …

Variable **N**umber **T**andem **R**epeats

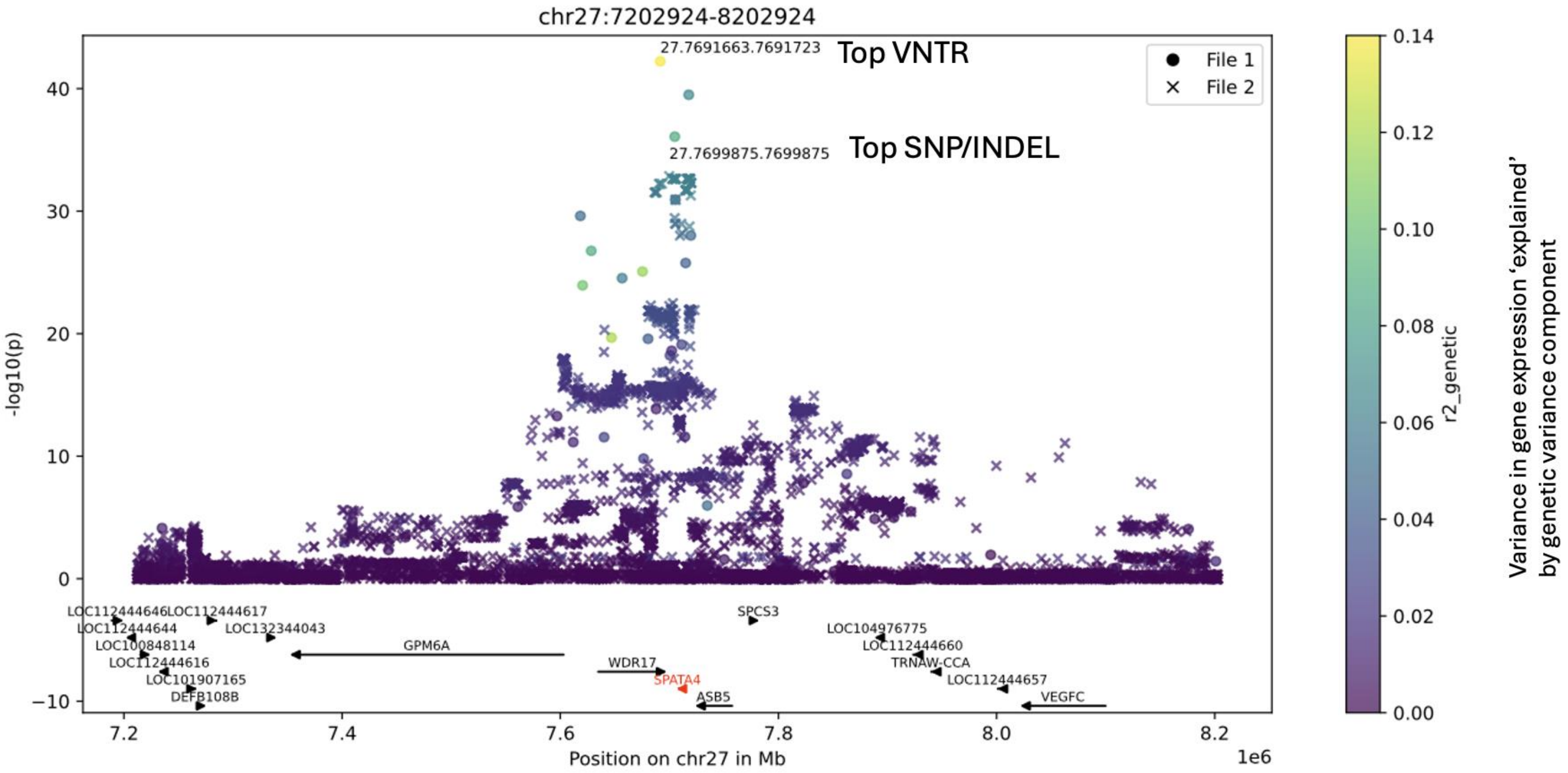
	Repeat counts
This is a **repetitive** motif.	1
This is a **repetitive rapetitove** motif.	2
This is a **repetitive repetitive repetitive** motif.	3
This is a **repatotive rapetitove repetitive** motif.	3
This is a **repatotive repetitive** motif.	2
This is a **repetitive rapetitove repetitive rapetitove** motif.	4

How prevalent are VNTR in the bovine genome, and how much do they contribute to trait variability? *(Victor Mason)*

Dr. Victor Mason

Gene: SPATA4
VNTR: 27.7691663.7691723

O's = VNTR associated with highlighted gene SPATA4
X's = biallelic SNP or INDEL associated with highlighted gene SPATA4



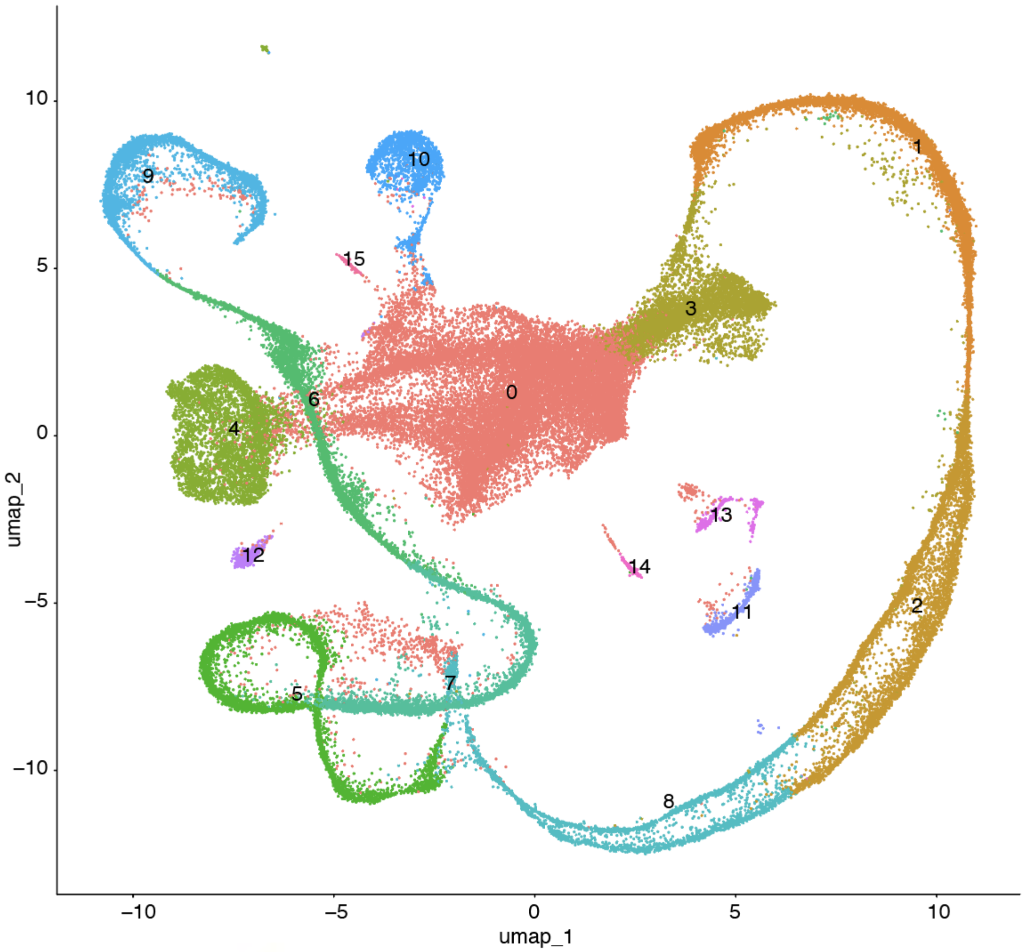
Top VNTR

Top SNP/INDEL

Variance in gene expression 'explained' by genetic variance component

Some further plans and projects

Single-nuclei RNA sequencing

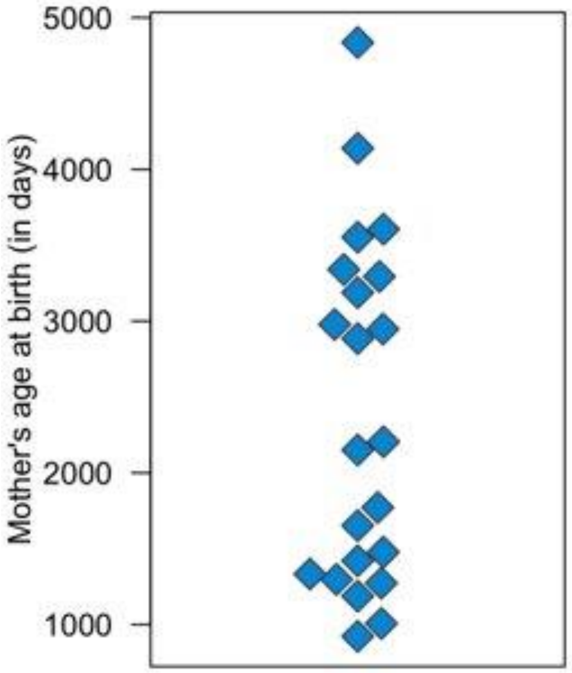


Dr. Xena Mapel

Dr. John O' Grady

SNSF-Project OPISCOW

Origin, **P**revalence, and **I**mpact of complex **S**tructural variation in a large mammalian genome discovered through genome assembly at the population scale



HiFi-sequencing of 50 trios to detect and characterise *de novo* mutations

from January 1st
Damianos Alexandridis
Diego Rosete

Contributors & funding

BRAUNVIEH

Qualitas.

AgroVet Strickhof

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ETH zürich

University of Zurich UZH

Functional Genomics Center Zurich

FNSNF
FONDS NATIONAL SUISSE
SCHWEIZERISCHER NATIONALFONDS
FONDO NAZIONALE SVIZZERO
SWISS NATIONAL SCIENCE FOUNDATION

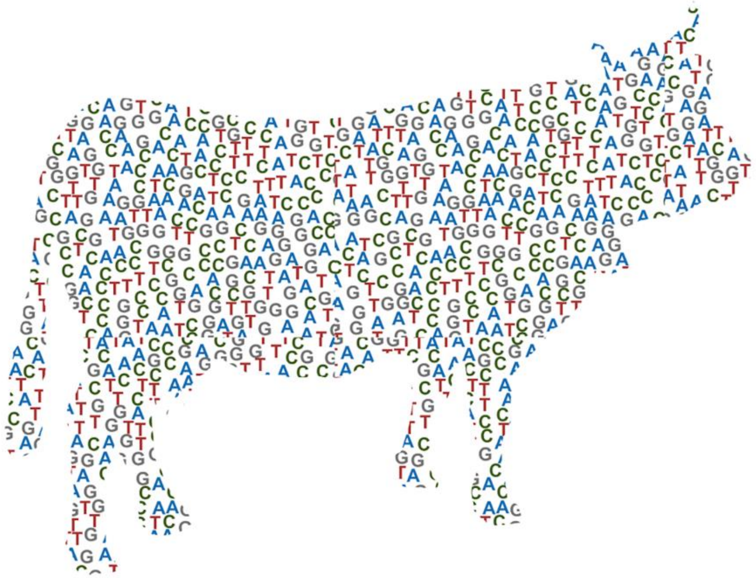
Schweizerische Eidgenossenschaft
Confédération suisse
Confederazione Svizzera
Confederaziun svizra

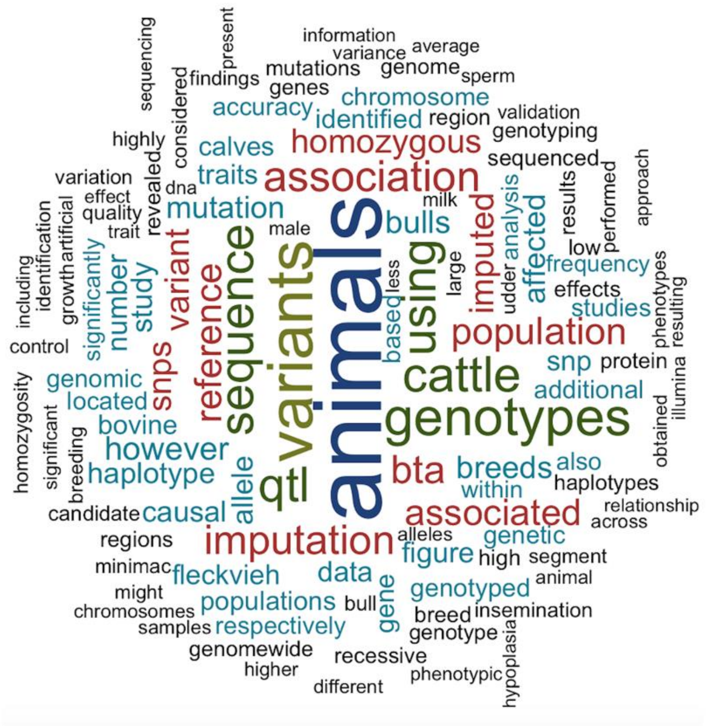
Federal Office for Agriculture FOAG

swissgenetics

PacBio

Thank you for listening!





ETH Zürich
Hubert Pausch
Animal Genomics
Universitätstrasse 2 | LFW B 58.2
8092 Zürich

hubert.pausch@usys.ethz.ch
www.ag.ethz.ch