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Agroscope

Projects of Animal GenoPhenomics

Claudia Kasper

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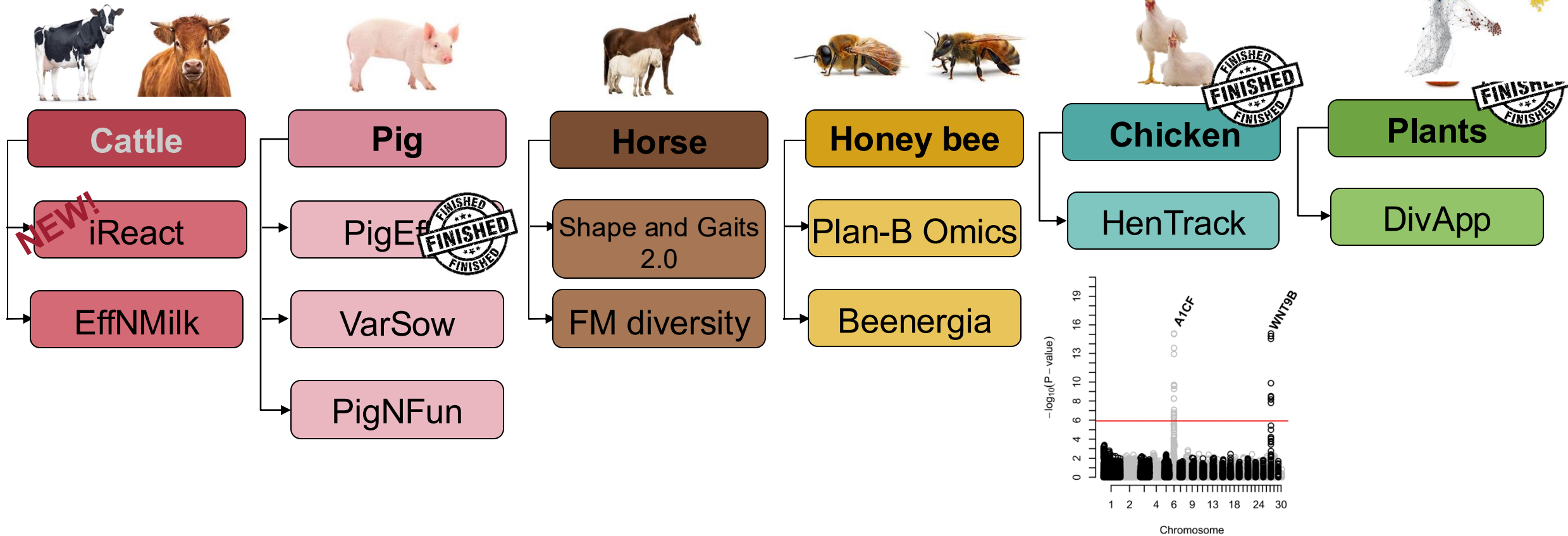
Team

- Markus Neuditschko (head of group)
- Claudia Kasper (deputy)
- Olga Wellnitz (currently researcher pigs)
- Maria Frizzarin (postdoc cattle)
- Victor Rossier (postdoc honey bees)
- Bastien Monney (research assistant pigs)





Animal GenoPhenomics





Cattle: iReact

- **≈ 0.8 Mio CHF**
- International collaboration with INRAE
- Duration: 2026-2030
- **Aim:** Understanding bovine adaptive physiology in a context of low-carbon meat production limiting feed-food competition
 - **Animal GenoPhenomics:** Identification of candidate genes for fat-lean ratio, adaptive traits, and resilience features using low-pass sequencing data





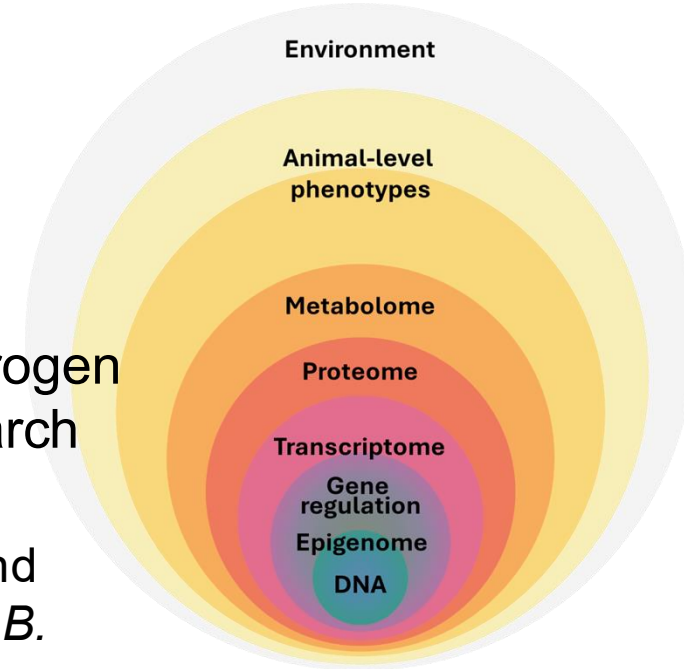
Cattle: EffNMilk

- Data collection Jan 2022 – Feb 2025
- > 2,300 cows sampled at 5 cantonal and >30 private farms
- milk (NIRS and MIRS), faeces (NIRS), blood, feed
- **Aim:** genetic analysis of nitrogen use efficiency and methane traits in Swiss dairy cows
- analysis within new Agroscope work program (Project GENESIS-Cattle)
- follow-up/related projects:
 - **MIRage:** calibration models for MIR spectra to predict nitrogen use efficiency and methane emissions (*M. Frizzarin*)
 - **GREEN-BREED** proposal submitted to Horizon Europe in September with international consortium;
Animal GenoPhenomics: leader of WP2 (Phenotyping)
 - **FutureCow** – Water use efficiency and heat stress resilience; submitted to SNF Ambizione (*M. Frizzarin*)
 - **MilkMetrics** - Using mid-infrared spectroscopy for farm-level environmental monitoring; submitted to Innosuisse Bridge (*M. Frizzarin*)



Pigs: PigNFun

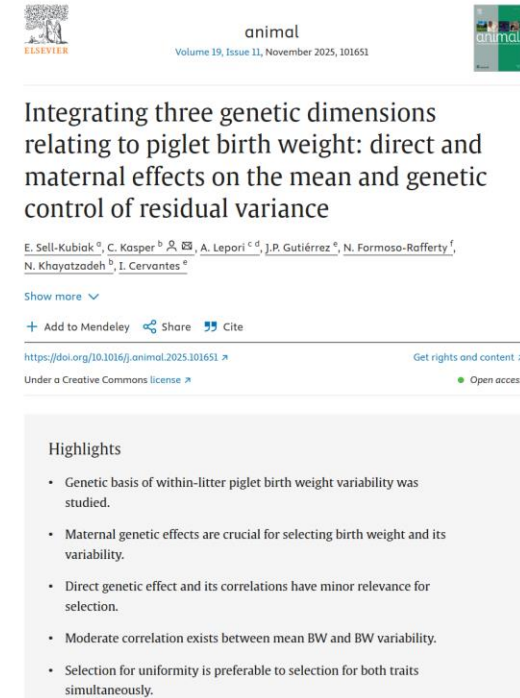
- ongoing
- **Aim:** Explore the biological functions of genes underlying nitrogen use efficiency (NUE) in pigs through multi-omics (basic research and applied interest).
 - **MuLi** (transcriptomics): distinct gene expression profiles in liver and muscle of pigs with low and high NUE (manuscript in preparation; *B. Monney*)
 - **PigCircus** ('mobilomics'): characterize mobile elements on extrachromosomal circular DNA and their link to NUE (collaboration with Crop Genome Dynamics research group – Etienne Bucher)
 - **N-FiXD:** Genomic insights into nitrogen use efficiency in pigs: harnessing gene expression across the lifespan – ongoing Spark





Pigs: VarSow

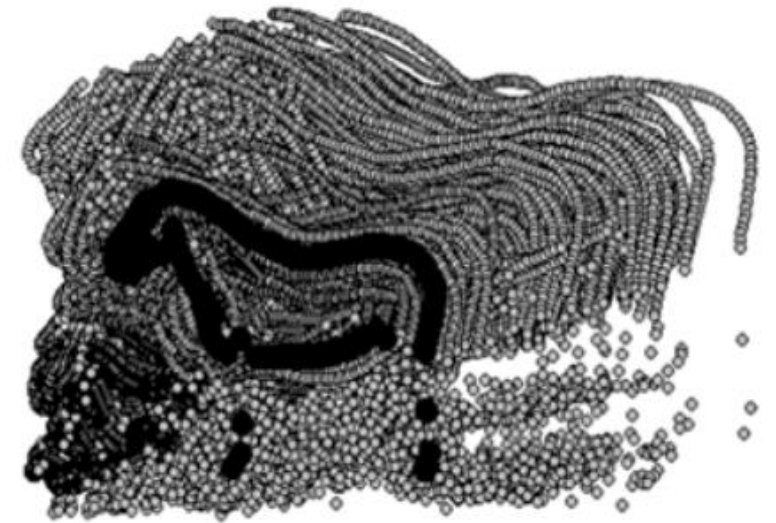
- ongoing long-term project
- **Aim:** genetic analysis of within-litter birth-weight uniformity
- **Results:** genetic component of residual variance at sow level ($h^2 = 0.22 \pm 0.02$), direct genetic effect is negligible – **maternal trait**
- currently analysis of parent-of-origin effects (collaboration with U Guelph & FBN Dummerstorf)
- building large database
 - birth weights of individual piglets (since Dec 2004; > 52,400 records from > 3,800 litters and > 1,100 sows; around 2,600 new piglets each year)
 - sequence data (since 2022; goal ~1,000 sows)





Horses: Shape and Gaits 2.0

- Collaboration with University Zürich
- Duration: 2020-2024
- **Aim:** Objective assessment of gait and conformation traits
- **Results:**
 - Conformation traits derived from **Shape space** data show **higher heritability** compared to subjective evaluations
 - **Horse breeds** (FM, Warmblood, Lusitano) can be differentiated based on their **movement patterns** (EquiMoves) – Data and GWAS paper (in preparation)





Horses: FM diversity

- Collaboration with Vetsuisse Bern (Institute of Genetics and ISME)
- **Aim:** Preservation of the Franche-Montagne horse
- **Results:**
 - Identification of a **causal genetic variant** responsible for hypertriglyceridemia-induced **pancreatitis** (HIP)
 - Identification of a **putative candidate gene** (*HPGD*) for **lameness** through an admixture-based GWAS, which we term **TRACE** (Tracking **R**ecombination and **A**dmixture **C**aptures **E**volution) - Abstract submitted to WCGALP

scientific reports

OPEN *LMF1* frameshift deletion in Franches-Montagnes horses with hypertriglyceridemia-induced pancreatitis

Michaela Drögemüller¹, Nathalie Fouché², Michelle Wyler², Corinne Gurtner³, Seraina L. Meister⁴, Markus Neuditschko⁵, Vidhya Jagannathan⁶, Vincenz Gerber² & Tasso Leeb¹✉

Hypertriglyceridemia (HTG) may be inherited and caused by variants in genes encoding enzymes of lipid metabolism. This study was prompted by the observation of eight Franches-Montagnes (FM) foals showing elevated plasma triglyceride levels and episodes of fatal acute pancreatitis. We termed this phenotype hypertriglyceridemia-induced pancreatitis (HIP). The affected foals were distantly related and inbred to a prominent stallion suggesting autosomal recessive inheritance. Whole genome sequencing of an affected foal identified a homozygous loss of function variant in *LMF1* encoding lipase maturation factor 1. The variant, XM_023616679.1:c.369_373delinsTC, leads to an early frameshift and is predicted to alter or truncate 79% of the *LMF1* coding sequence. We genotyped the variant in a cohort of 2122 FM horses and identified 11 homozygous mutant animals including all eight foals that had initially been identified based on their clinical presentation. The three additional homozygous mutant animals had a comparable phenotype and were inbred to the same stallion. We concluded that all 11 had been affected by the same disease. Thus, we found perfect genotype-phenotype association in the tested cohort. The carrier frequency in the 2111 unaffected FM horses was 15.0%. Our findings enable genetic testing to prevent the unintentional breeding of further HIP-affected foals.

Keywords: Equus Caballus, Metabolism, Lipid, Combined lipase deficiency, Precision medicine, Breeding, Inbreeding, Animal model



Honey bees: Plan-B Omics



- **≈ 1.4 Mio CHF**
- International Collaboration (Uni Bern, Uni Lausanne, Uni Laval – Canada)
- **Aim:** Using metagenomics to improve the selection of *Varroa destructor* resistant honey bees
- Structured in 4 modules – **Animal GenoPhenomics** responsible for **M3-Modulisation**
 - Optimization of current applied animal models in honey bee breeding by including **metagenomic** data, e.g. parasites (Varroa), bacteria, viruses, etc.





Honey bees: Beenergia



- **≈ 4.0 Mio CHF**

- International collaboration (Uni Bern, Uni Neuchâtel, State Museum of Natural History Karlsruhe)
- **Aim:** Assessing the interactions between managed and wild bees
- Structured in 4 modules (M1-Food competition, M2-Shared Pathogen, **Animal GenoPhenomics: M3-Genomics**, M4-Socioeconomics)
 - Compare genomes of museum and contemporary bees to assess conservation relevant parameters (e.g. gene flow, N_e , etc.)





Thank you for your attention

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