Livestock genetics -

examples of current projects

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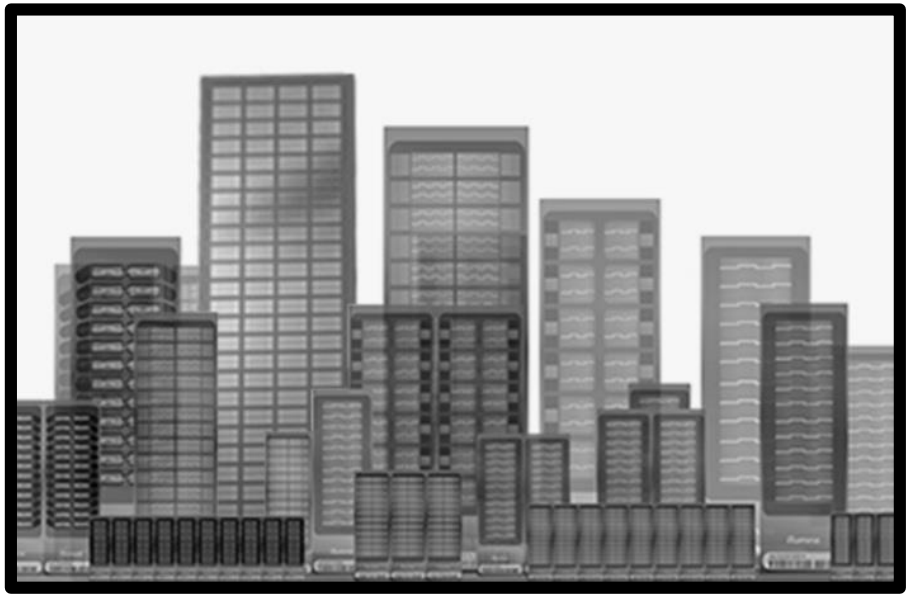
Swiss Animal Breeding Technology Platform, Zurich, 5th June 2019

DNA focused research

(rare) genetic diseases – heritable (morphological/color) traits



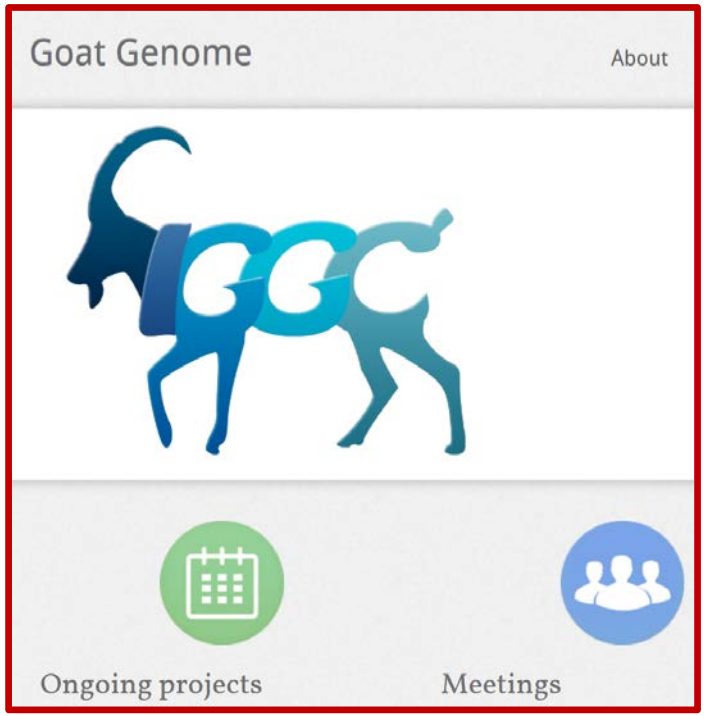
SNP data

WGS data

2019: WGS of hundreds

goat – cattle – sheep

2017: ARS1



2018: ARS-UCD1.2

2019: Oar_rambouillet_v1.0

SNF project 2017 - 2021

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

A comprehensive genetic screen for recessive mutations impairing fertility and rearing success of Swiss cattle

- massive SNP genotyping data of 4 local cattle populations




- collaboration with Qualitas AG (Franz Seefried)

→ Identification of genetic variants causing embryonic lethality and congenital disorders

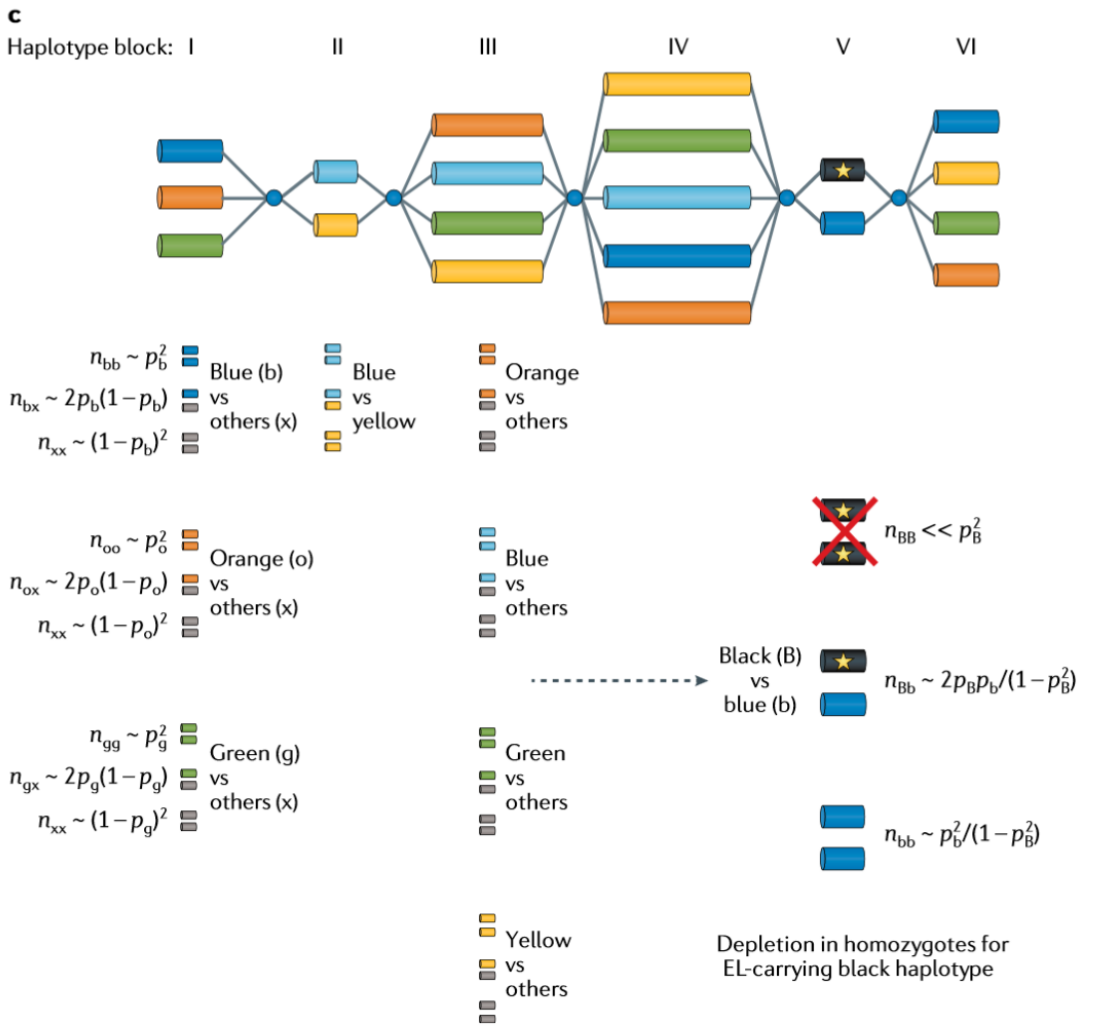
nature REVIEWS GENETICS

Review Article | Published: 04 December 2018

Harnessing genomic information for livestock improvement

Michel Georges ✉, Carole Charlier & Ben Hayes

Nature Reviews Genetics **20**, 135–156 (2019) | Download Citation ⬇

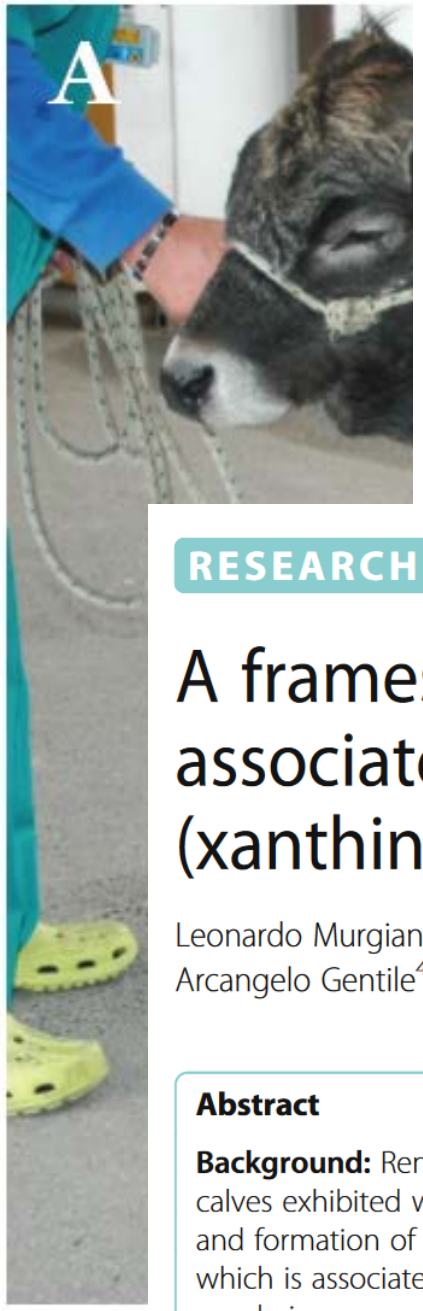


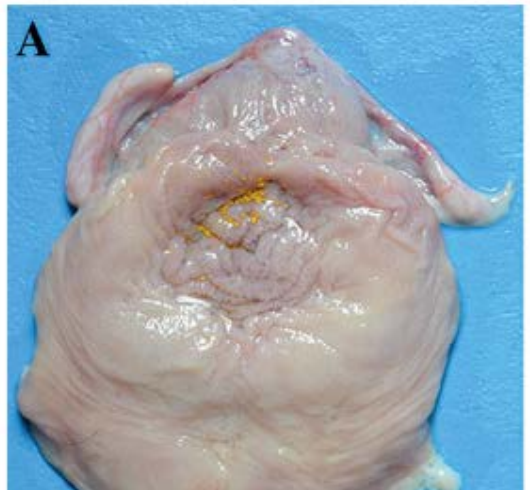
Recessive mutations compromises fertility

Brachyspina in Holstein: rarely the homozygous mutant animals survive to term because this genotype is a relatively common cause of fertility failure

Charlier et al. PLoS One. 2012; 7: e43085

Recessive mutations compromises rearing success





RESEARCH ARTICLE Open Access

CrossMark

A frameshift mutation in MOCOS is associated with familial renal syndrome (xanthinuria) in Tyrolean Grey cattle

Leonardo Murgiano[1], Vidhya Jagannathan[1], Christian Piffer[2], Inmaculada Diez-Prieto[3], Marilena Bolcato[4], Arcangelo Gentile[4] and Cord Drögemüller[1*]

Abstract

Background: Renal syndromes are occasionally reported in domestic animals. Two identical twin Tyrolean Grey calves exhibited weight loss, skeletal abnormalities and delayed development associated with kidney abnormalities and formation of uroliths. These signs resembled inherited renal tubular dysplasia found in Japanese Black cattle which is associated with mutations in the *claudin 16* gene. Despite demonstrating striking phenotypic similarities, no obvious presence of pathogenic variants of this candidate gene were found. Therefore further analysis was

rarely present in Original Braunvieh

Cholesterol deficiency (CD) in Holstein also occur in *APOB* heterozygotes

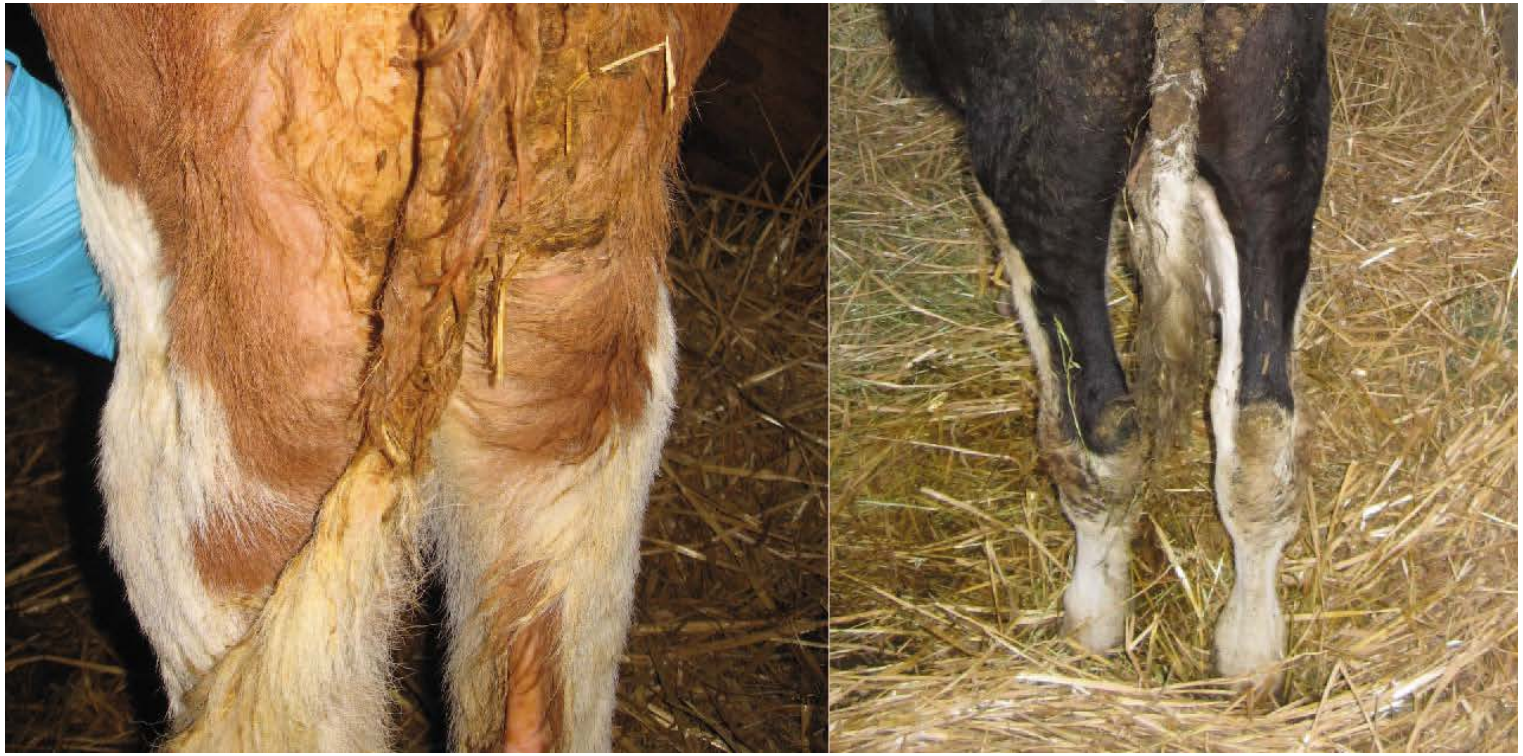
SHORT COMMUNICATION

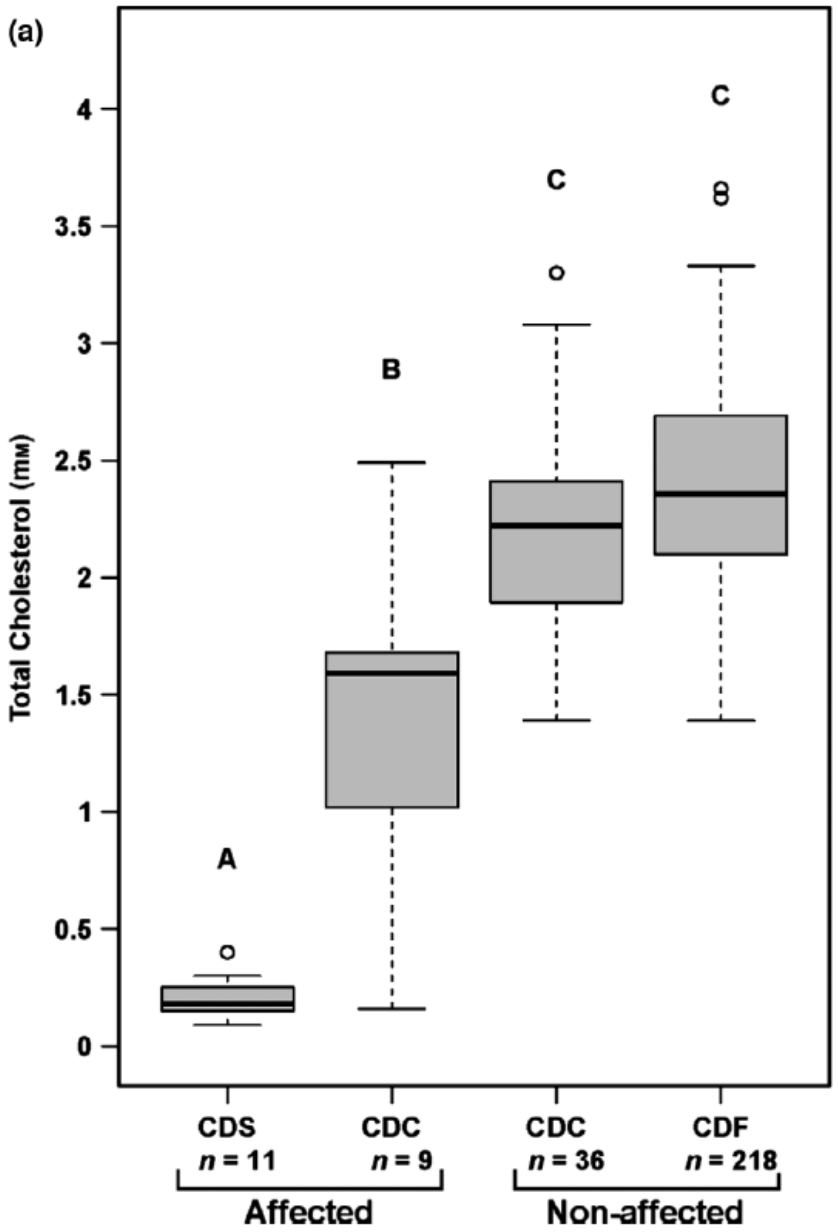
doi: 10.1111/age.12801

APOB-associated cholesterol deficiency in Holstein cattle is not a simple recessive disease

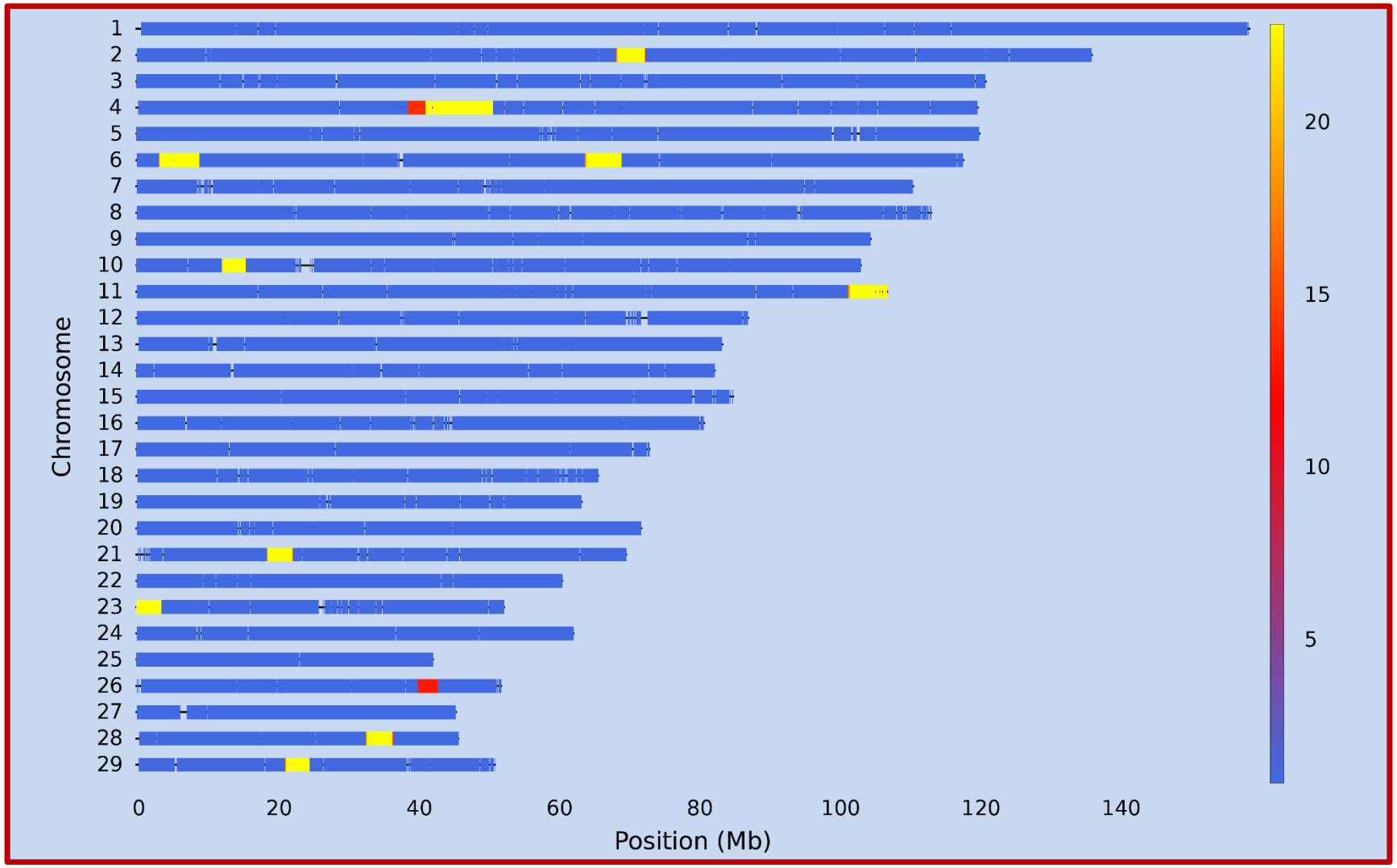
Irene Monika Häfliger*, Sonja Hofstetter*, Thomas Mock†, Manuela Hanna Stettler†, Mireille Meylan†, Kemal Mehinagic‡, Nadine Stokar-Regenscheit‡ and Cord Drögemüller*

*Institute of Genetics, Vetsuisse Faculty, University of Bern, Bern, 3001, Switzerland. †Clinic for Ruminants, Vetsuisse Faculty, University of Bern, Bern 3001, Switzerland. ‡Institute of Animal Pathology, Vetsuisse Faculty, University of Bern, Bern, 3001, Switzerland.





Screening for missing homozygosity in Original Braunvieh

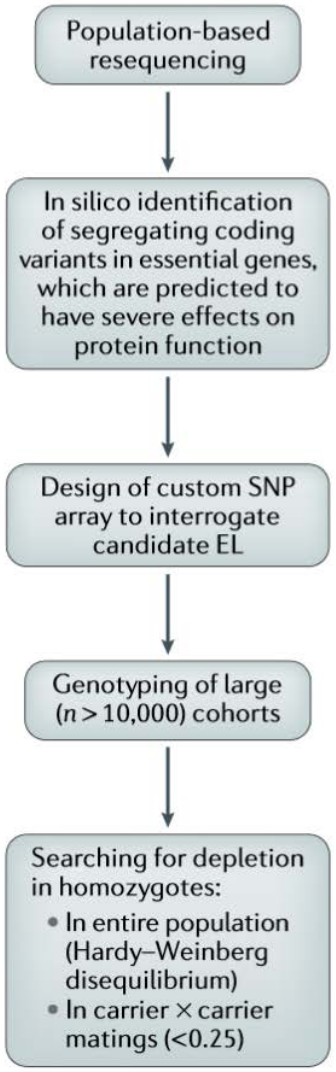


4’892 animals

density: 135K SNPs

window size: 150 SNPs

Workflow focus on SNVs and short indels



focussing on high and moderate impact variants in

- candidate genes for **embryonic lethality**
- genes with high **pLI scores** (pLI ≥ 0.9) are extremely loss-of-function (LoF) intolerant
- developmental disorder genes included in the **DDG2P panel**

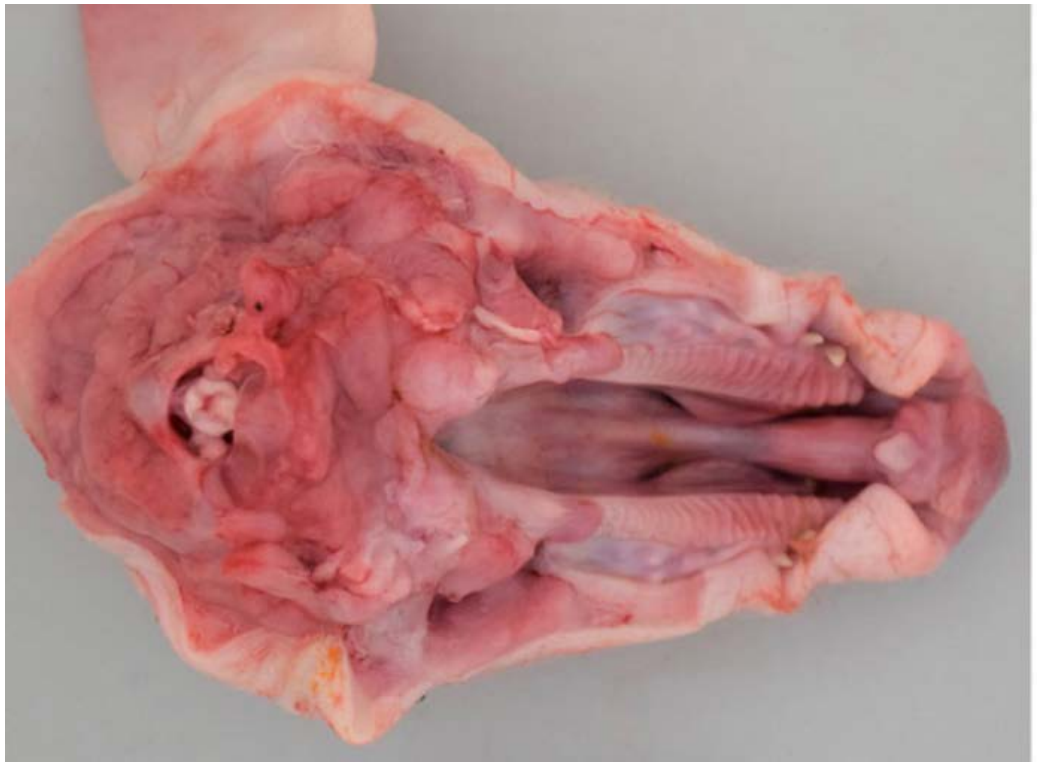
Grahofer *et al. BMC Genomics* (2019) 20:349
https://doi.org/10.1186/s12864-019-5711-4

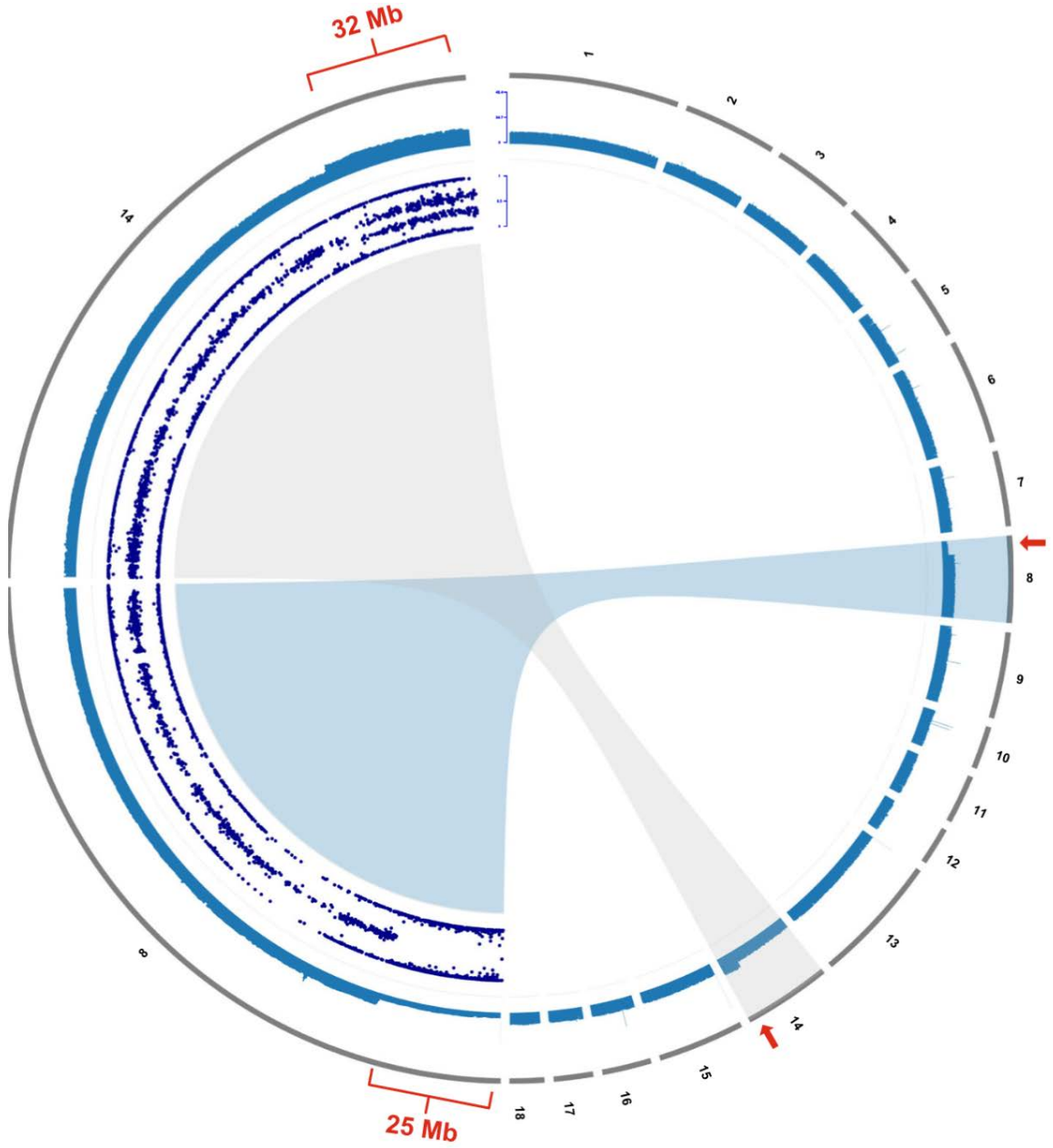
BMC Genomics

RESEARCH ARTICLE Open Access

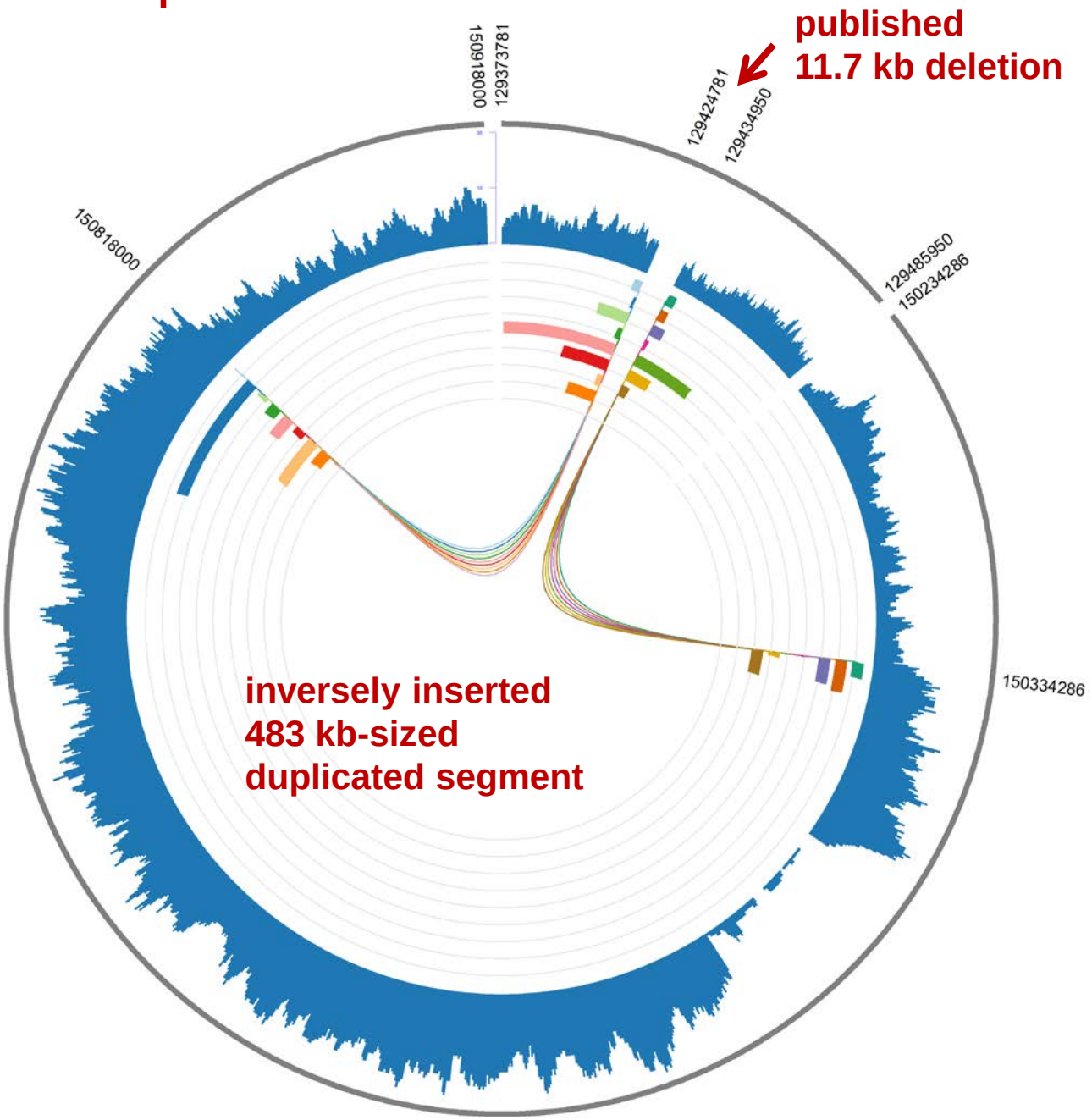
Chromosomal imbalance in pigs showing a syndromic form of cleft palate

Alexander Grahofer[1], Anna Letko[2], Irene Monika Häfliger[2], Vidhya Jagannathan[2], Alain Ducos[3], Olivia Richard[4], Vanessa Peter[5], Heiko Nathues[1] and Cord Drögemüller[2*]





The caprine polled variant: more complex than known before



Take home message

SNV / short indel = only ~half of the variation

CNV / SV = still difficult (and expensive) to detect