Livestock genetics at UniBE

News from the Institute of Genetics, Vetsuisse Bern

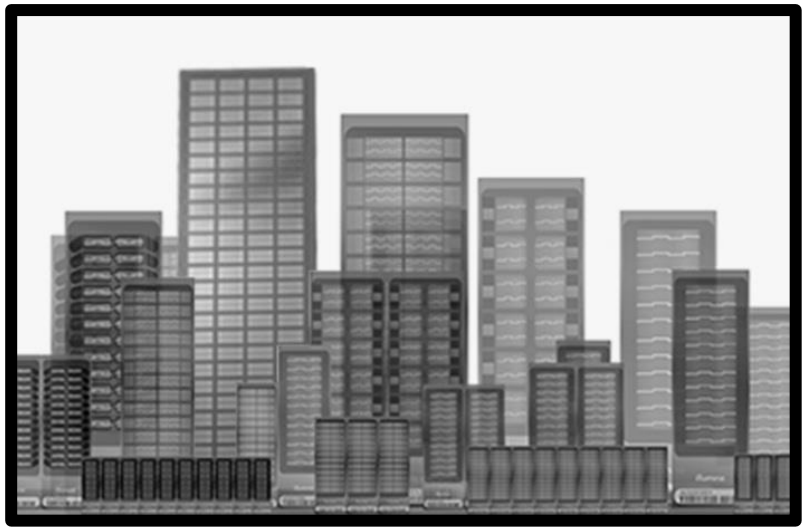
Cord Drögemüller, Institute of Genetics, Vetsuisse faculty, University of Bern

Swiss Animal Breeding Technology Platform, Zurich, 11th January 2024

DNA focused research

rare (visible/hidden) disorders – heritable (morphological/color) traits

phenotypes



SNP data

WGS data

WGS established as affordable approach

nature

Milestones | 10 February 2021

Milestones in Genomic Sequencing

Genomes go platinum

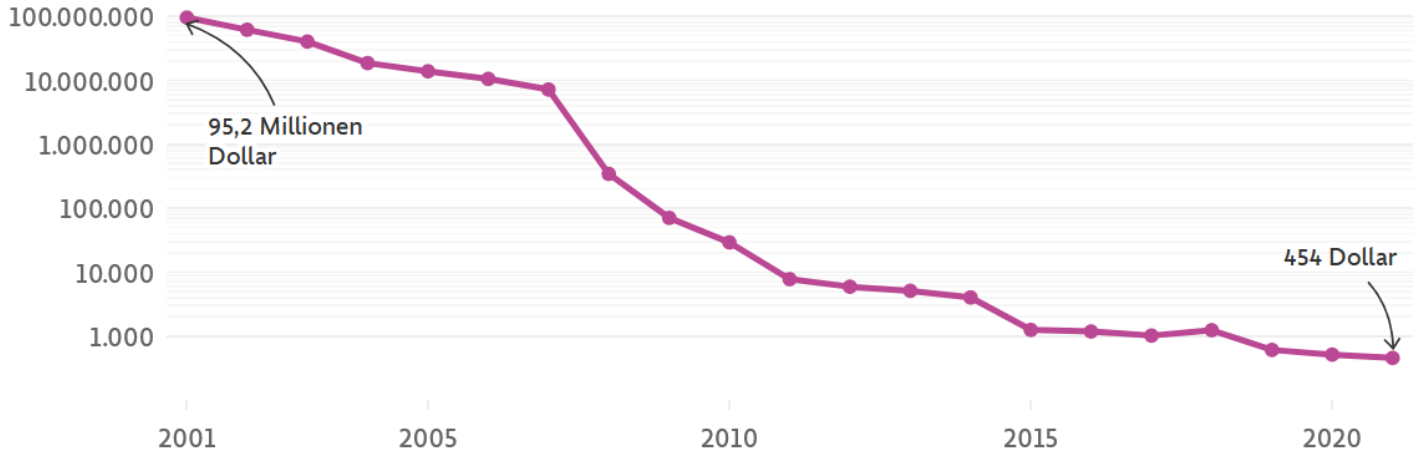
By Brooke LaFlamme

Credit: Ian M Butterfield (Concepts) / Alamy Stock Photo

Sequencing a vertebrate genome had almost become routine by 2017, but, with very few exceptions, assemblies of most diploid genomes remained highly fragmented and incomplete. The domestic goat genome ARS1 created a new standard for de novo assemblies of complex genomes.

3 Milliarden Basenpaare für weniger als 500 Dollar

Kosten für die Sequenzierung eines vollständigen menschlichen Genoms, in Dollar*



*) Daten bis 2021 und nicht inflationsbereinigt

Grafik: omer. / Quelle: Nationales Humangenom-Institut, Our World in Data

https://www.nature.com/immersive/d42859-020-00099-0/index.html

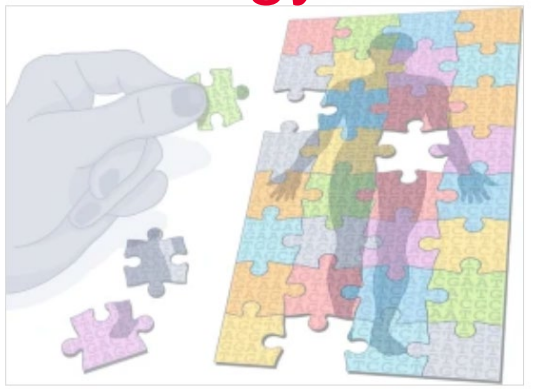
UniBE NGSP provides access to the latest technology

Method of the Year 2022: long-read sequencing

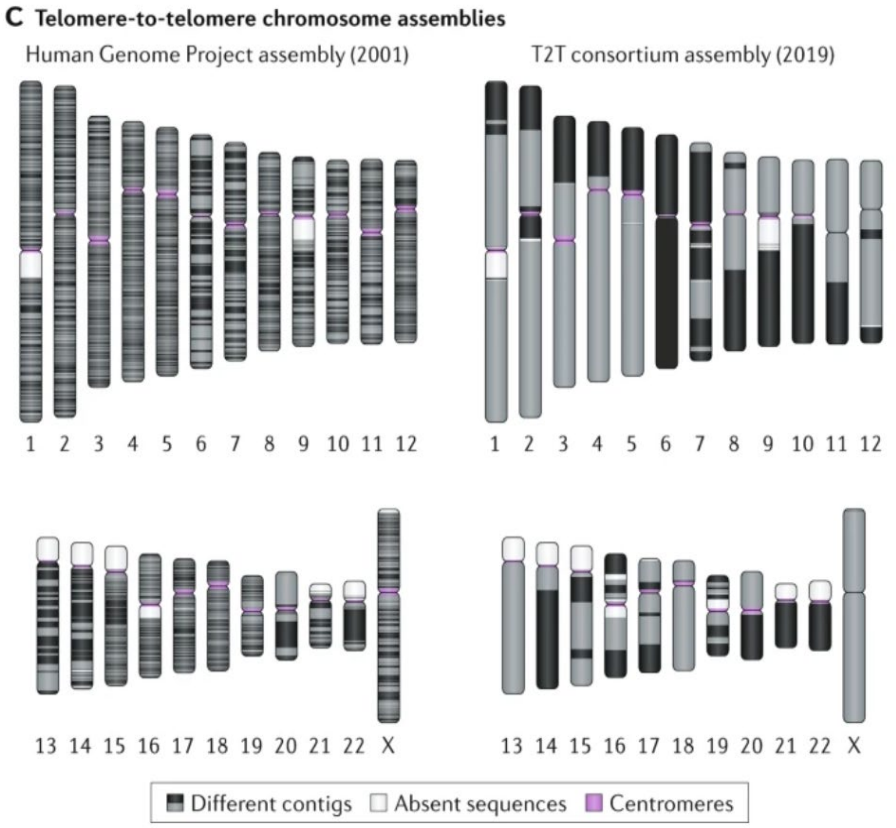
Nature Methods **20**, 1 (2023) | Cite this article

Long-read sequencing powers a more complete reading of genomic information.

Long reads are a way for many labs to tackle gnarly genome sections such as complex genome rearrangements and the many types of repetitive elements. Credit: T. Phillips, Springer Nature



Credit: P. Morgan/Springer Nature Limited



https://www.nature.com/articles/s41576-020-0236-x

UniBE participates in global efforts

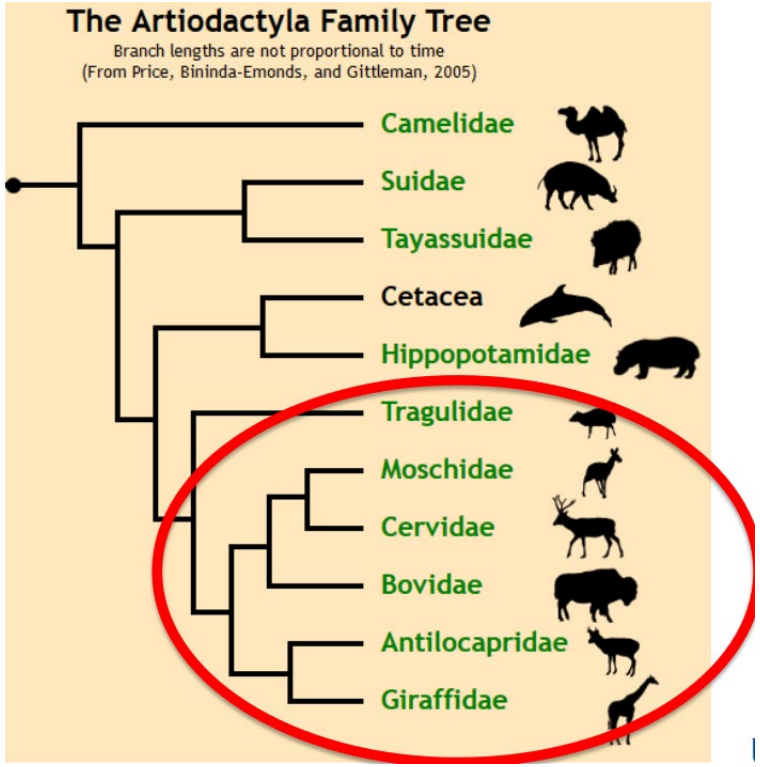
The Bovine Pangenome Consortium

https://bovinepangenome.github.io/

The Ruminant T2T Consortium

Tim Smith, USDA – Brenda Murdoch, U. Idaho – Stephanie McKay, U. Missouri

Ben Rosen (ben.rosen@usda.gov)
Animal Genomics and Improvement Laboratory
USDA-ARS, Beltsville, MD



Increasing knowledge on inherited disorders

if we find a new causal variant, it will appear here

THE UNIVERSITY OF SYDNEY

OMIA - ONLINE MENDELIAN INHERITANCE IN ANIMALS

OMIA | SYDNEY SCHOOL OF VETERINARY SCIENCE | UNIVERSITY HOME | CONTACTS | Enter search terms | SEARCH

Home | Donate | Browse | Search | Landmarks, Reviews, Maps | PMIA | Download | Curate | Contact | Citing OMIA | News | Acknowledgements | Links

You are here: OMIA / Home

WELCOME TO OMIA

Online Mendelian Inheritance in Animals (**OMIA**) is a catalogue/compendium of inherited disorders, other (single-locus) traits, and associated genes and variants in 503 animal species (other than human and mouse and rats and zebrafish and western clawed frog, which have their own resources) co-authored by Professor Frank Nicholas and Associate Professor Imke Tammen of the University of Sydney, Australia, with help from many people over the years. OMIA information is stored in a database that contains textual information and references, as well as links to relevant PubMed and Gene records at the NCBI, to OMIM, Ensembl, Mondo Disease Ontology and the Vertebrate Breed Ontology (VBO).

OMIA is manually curated by a team of specialists. If you see an error or wish to submit an entry, please contact us.

To join the OMIA Support Group, register at OMIA Support Group.

From 1st September 2011, the OMIA ID is binomial. To better conform to global standards, in March 2023 the binomial format was changed slightly by replacing a space with a colon, i.e. OMIA:xxxxxx-yyyy.., where xxxxxx is the 6-digit number for a trait/disorder, and yyyy.. is the NCBI species taxonomy id (usually four digits, but sometimes longer).

OMIA has been a free internet resource for over 25 years, with much of the crucial curation work done on a volunteer basis. Please consider donating to help pay for maintenance, upgrades and curation time.

i We have recently launched the Pioneers of Mendelian Inheritance in Animals project (PMIA), an exploration of the history of research into Mendelian inheritance in animals.

Summary

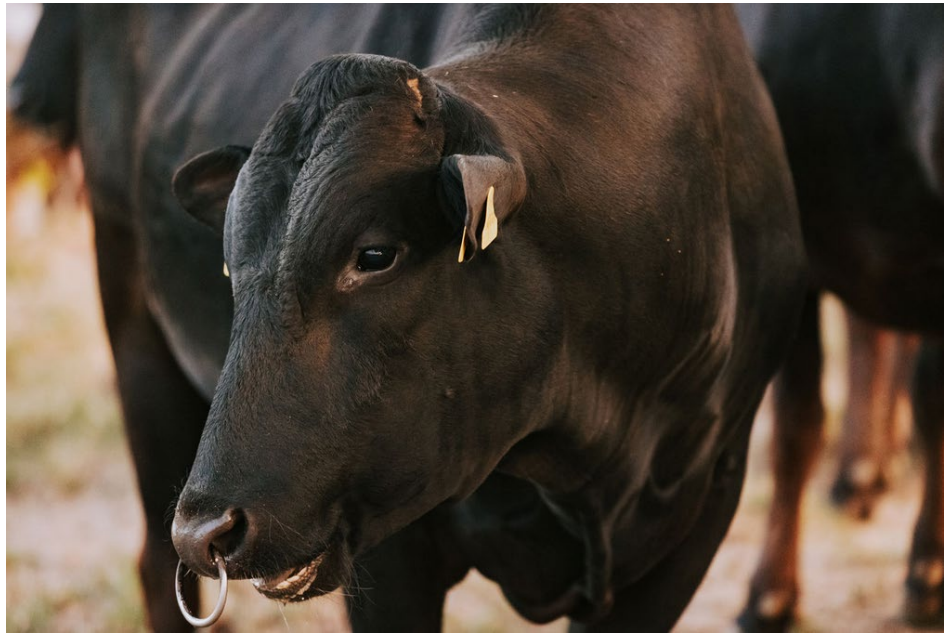
	dog	taurine cattle	cat	pig	sheep	horse	chicken	rabbit	goat	Other	TOTAL
TOTAL TRAITS/DISORDERS	923	670	431	383	317	281	256	131	124	1294	4913
Mendelian trait/disorder	418	304	142	138	125	62	137	78	26	397	1896
Mendelian trait/disorder; likely causal variant(s) known	355	209	110	69	62	49	59	21	18	230	1200
Likely causal variants	528	273	188	72	93	109	72	20	30	197	1601
Potential models for human traits	590	337	273	212	137	153	88	77	56	700	2671

OMIA
Online Mendelian Inheritance in Animals
THE UNIVERSITY OF SYDNEY

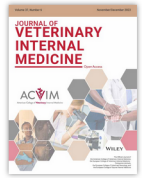
RECENT NEWS

Giraffe with no spots: OMIA entry awaiting results!:
Many OMIA users will have seen that the spotless giraffe born on 31 July in Brights Zoo, Tennessee, was yesterday given a name: Kipekee. Imke has created a new entry in OMIA for this phene in giraffes (https://omia.org/OMIA000200/9894/). We assume that someone somewhere is already investigating whether there is a likely causal variant for this phene. If there is, we look forward to entering it in OMIA. (posted 7 September 2023)

Reporting likely causal genotypes from sequencing data:
Boeykens et al. (BMC Bioinformatics (2023) 24:305) have created an R-package



ACVIM | JOURNAL OF VETERINARY INTERNAL MEDICINE Open Access



Volume 37, Issue 6
November/December 2023
Pages 2631-2637

CASE REPORT | Open Access

A missense variant in *DGKG* as a recessive functional variant for hepatic fibrinogen storage disease in Wagyu cattle

Joana G. P. Jacinto, Peter Wohlsein, Irene M. Häfliger, Michael Karl, Michael Pohlers, Lutz Plobner, Walter Grünberg, Cord Drögemüller

First published: 08 September 2023 | https://doi.org/10.1111/jvim.16865

Figures | References | Related | Information

Recommended

Cattle genetics

A successful change has taken place: SWISScow genotyping since 2020

"Züchterische Förderung und Erhaltung der genetischen Variabilität und der Gesundheit des Rätischen Grauviehs"

BLW funded 2022-2025 including massive SNP genotyping

Partner in a collaboration of

Rätisches Grauvieh Schweiz

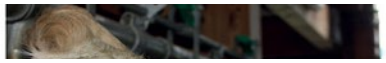
pro specie rara – Schweizerische Stiftung für die kulturhistorische und genetische Vielfalt von Pflanzen und Tieren

Berner Fachhochschule

J. Dairy Sci. 106:8969–8978
https://doi.org/10.3168/jds.2023-23315

WNT10B: A locus increasing risk of brachygnathia inferior in Brown Swiss cattle

Sarah Widmer,[1,2] Franz R. Seefried,[2] Irene M. Häfliger,[1] Heidi Signer-Hasler,[3] Christine Flury,[3] and Cord Drögemüller[1]*
[1]Institute of Genetics, Vetsuisse Faculty, University of Bern, 3012 Bern, Switzerland
[2]Qualitas AG, 6300 Zug, Switzerland
[3]School of Agricultural, Forest and Food Sciences, Bern University of Applied Sciences, 3052 Zollikofen, Switzerland




21.5-fold increased risk of brachygnathia inferior in homozygous carriers of the *WNT10B* variant

Table 1. Overview of Brown Swiss study cohorts and genotyping results for the *WNT10B* variant (c.910dupC) in Swiss dairy cattle

				Genotype[2]		
Group	Subgroup 1	Subgroup 2[1]	Quantity	Ref/ref	Ref/var	Var/var
Controls[3]	SNP data		509			
		With WGS data (sharing ROH)	10 (1)			
Cases[3]	SNP data		145			
		With WGS data (sharing ROH)	12 (11)			
	PCR validation		81	16	13	52
Population	Custom array data[4]		41,335			
		Brown Swiss	16,617	7,063	7,663	1,891
		Original Braunvieh	2,837	2,834	3	
		Holstein	19,045	19.026	19	
		Simmental	2,836	2,835	1	
	Imputed genotypes Brown Swiss[5]		29,498			
		Affected[6]	187	32	37	118
		Unaffected[6]	29,311	16,222	10,933	2,156

SWISScow Chip

ANIMAL GENETICS
Immunogenetics, Molecular Genetics and Functional Genomics

BRIEF REPORT | Open Access

MYH3-associated non-syndromic palatoschisis (cleft palate, CP) in Limousine cattle

frequency of the variant *MYH3* allele in 1167 genotyped LM cattle was 1.5%

Joana G. P. Jacinto, Eliana Schiavon, Irene M. Häfliger, Patrizio Coli, Franz R. Seefried, Cord Drögemüller

First published: 26 March 2023 | https://doi.org/10.

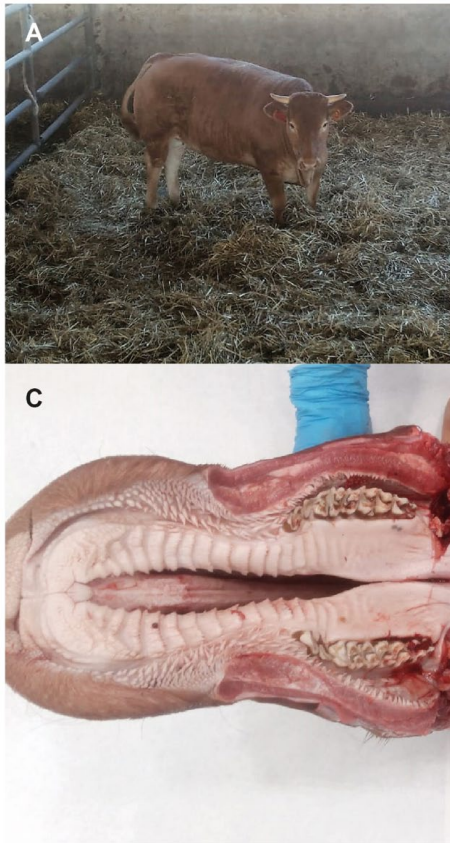


Figure S1. ***MYH3*-associated non-syndron**
Affected heifer; note the reduced body size. (**I**
palate involving both the soft and hard palates

	***MYH3* genotype**		
	Homozygous reference (CPF)	**Heterozygous carrier (CPC)**	**Homozygous variant (affected) (CPS)**
Palatoschisis case from Italy			1
Swiss Limousine population control cohort [a]	1133	34	
Sequenced Limousine cattle [a,b]	115	5	2 [c]
Sequenced control cattle from various breeds [a,b]	5364		
Total (Limousine)	6612 (1248)	39 (39)	3 (3)

JOURNAL OF VETERINARY INTERNAL MEDICINE Open Access

STANDARD ARTICLE | Open Access

MOCOS-associated renal syndrome in a Brown Swiss cattle

Joana G. P. Jacinto, Leonore Bettina Küchler, Laureen M. Peters, Elke Van der Vekens, Corinne Gurtner, Franz R. Seefried, Mireille Meylan, Cord Drögemüller

First published: 07 September 2023 | https://doi.org/10.1111/jvim.16856

no other homozygotes were observed in > 24k BS and >4k OB cattle

frequency of the variant *MOCOS* allele: 0.7 % in BS and 0.2 % in OB

TABLE 1. Occurrence of the deleterious *MOCOS* allele in Swiss dairy cattle.

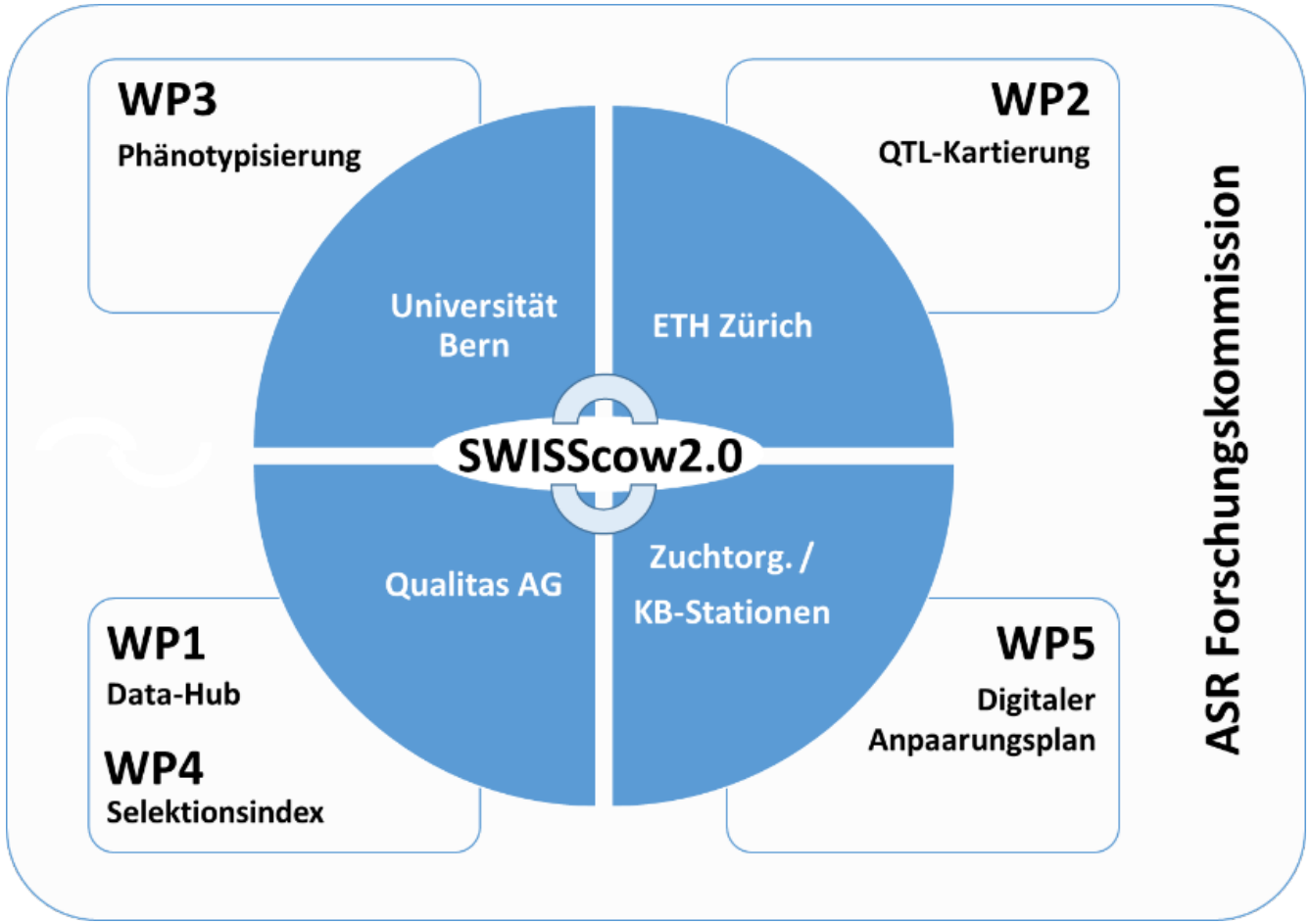
	MOCOS genotype	
	Ref/ref	Ref/var
Brown Swiss	23 995	342 [a]
Original Braunvieh	4085	19
Grauvieh	806	8
Simmental	3926	0
Swiss Fleckvieh	4035	0
Holstein	28 227	0
Total	65 074	369

SWISScow2.0 - an exciting joining of forces

Optimierung von Aufzucht und Reproduktion unter Berücksichtigung genetischer Besonderheiten

2022 - 2026

ASR & BLW funded



SWISScow2.0

WP3: Präzise Phänotypisierung und gezielte Sequenzierung genetischer Besonderheiten

BLIRD (Bovine Lymphocyte Intestinal Retention Defect) in Holsteins

frequency of the variant *ITGB7* allele in >90k genotyped Swiss HO cattle is 3.0%

homozygotes for the variant allele (in most cases) show chronic diarrhoea and significant changes in haematological parameters, resulting in a reduced life expectancy

SWISScow2.0

WP3: Präzise Phänotypisierung und gezielte Sequenzierung genetischer Besonderheiten

04.03.2023

KALB DER WOCHE

«Die Druckerfarbe ist ausgegangen»

«Ja, da staunt man nicht schlecht, wenn plötzlich ein schneeweisses Kalb hervorguckt», sagt Peter Küng über die Geburt seines Simmentaler-Munis Astor. Das Kalb, das am 9. Januar 2023 zur Welt gekommen ist, ist ein waschechter Albino – etwas, das man nicht alle Tage sieht. «Also ich sehe das täglich, schliesslich steht Astor bei mir im Stall», witzelt Züchter Küng. Seine Simmentaler-Mutterkühe sind allesamt hornlos, das hat er durch das Einkreuzen von Deutschem Fleckvieh erreicht. Astors Mutter Edelweiss ist trotz ihres stolzen Alters noch gut «zwäg»; sie ist 18 Jahre alt und hat in dieser Zeit ebenso viele Kälber geboren, zweimal waren es Zwillinge. «Edelweiss hat jedes Jahr gekalbt, ihre Zwischenkalbezeit beträgt 368 Tage. Bei Astor ist es nun vermutlich so, dass ihr einfach die Druckerfarbe ausgegangen ist», meint Peter Küng und lacht herzlich. Astors Geburt habe Edelweiss etwas mitgenommen, meint er dann, «wie es bei so alten Kühen eben ist». Sie erhole sich aber gut und könne im Sommer wohl mit allen anderen Kühen ganz normal z Alp. «Wir alpen am Nüegg hier im Diemtigtal», erzählt Peter Küng. Auch Astor wird dann mit von der Partie sein. Der Muni wurde bereits kastriert; Küng hofft, dass der kleine Ochse künftig ein waschechtes Hofmaskottchen wird. «Wenn er denn die Sonne erträgt, denn er ist wirklich tauhell. Aber wir sind zuversichtlich und freuen uns über unseren Astor», schliesst Peter Küng und schmunzelt noch immer.

lja

Der schneeweisse Albino-Simmentaler Astor ist ein Kalb mit Seltenheitswert.

BAUERNZEITUNG

***A novel tyrosinase(TYR)*-related form of albinism in Original Simmental**

monogenic recessive missense variant
detected by WGS in a single case

population prevalence will be assessed with the next version of the SWISScow LD routine array
(expected for Q2/2024)

New group member (starting Feb 2024): Joana Jacinto, DVM, PhD, Dip. ECBHM

Habilitationsstelle at Nutztierklinik & Institut für Genetik,

funded by "Faculty Clinical Research Platform"

Hoof health improvement through breeding

UNIVERSIDADE LUSÓFONA

ALMA MATER STUDIORUM A.D. 1088

Bovine hoof disease and genetics

Reference

Lameness prevalence: small changes between 1989 and 2020

Mean prevalence: at cow-level 23 %

at herd-level from 0 % to 88 %

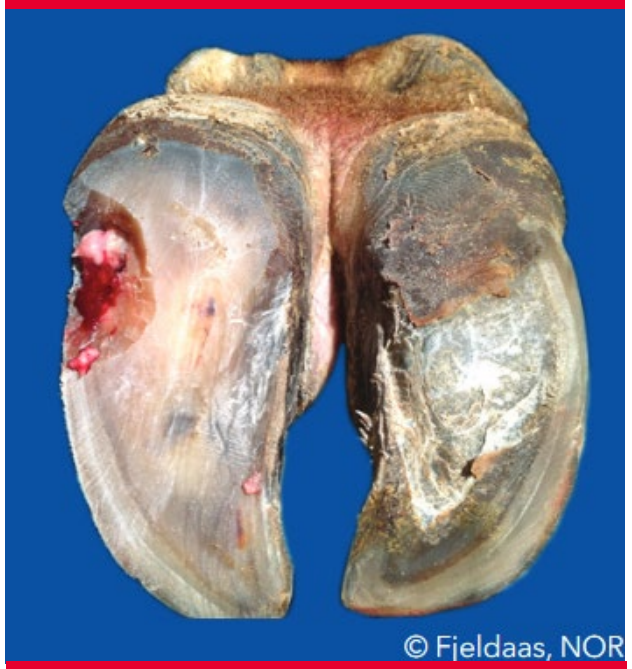
15 %

81 %

Digital dermatitis (DD)

h^2 = 0.1 to 0.4

White line disease (WL)

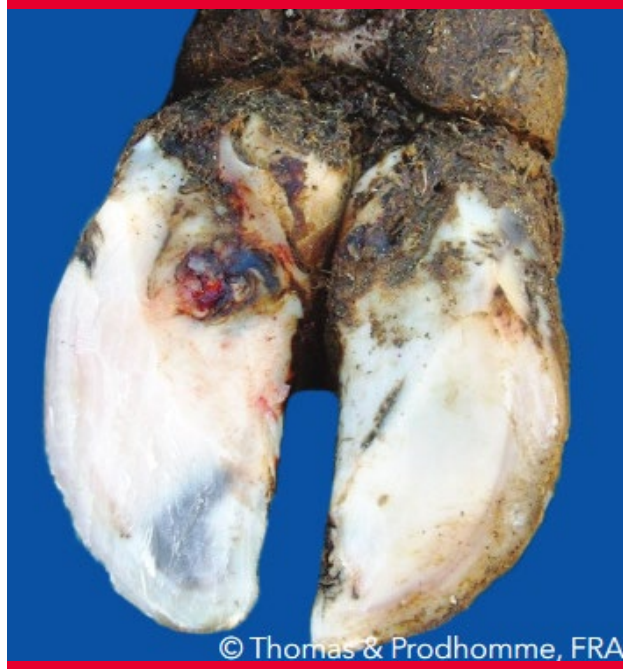


h^2 = 0.24 to 0.30

Sole hemorrhage (SH)

h^2 = 0.12 to 0.27

Sole ulcer (SU)



h^2 = 0.1 to 0.30

Sheep genetics

Breeding for shorter tails: evaluation of a *HOXB13* variant

Does the ancestral allele occur in Swiss sheep populations?

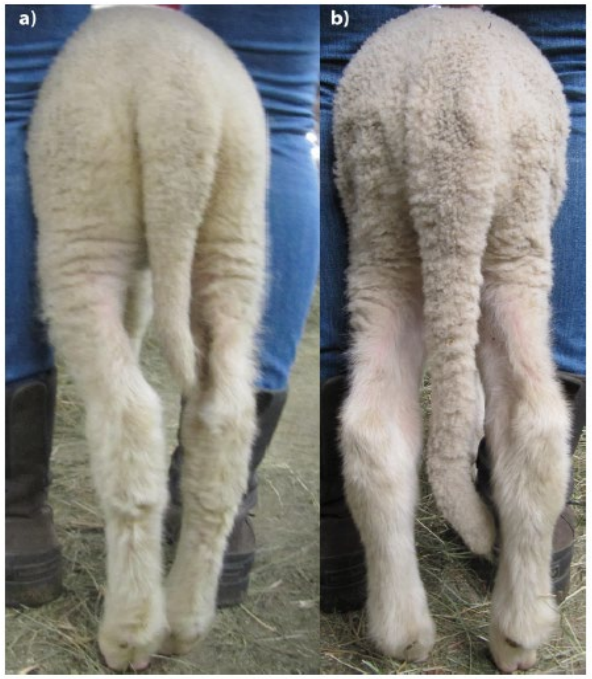
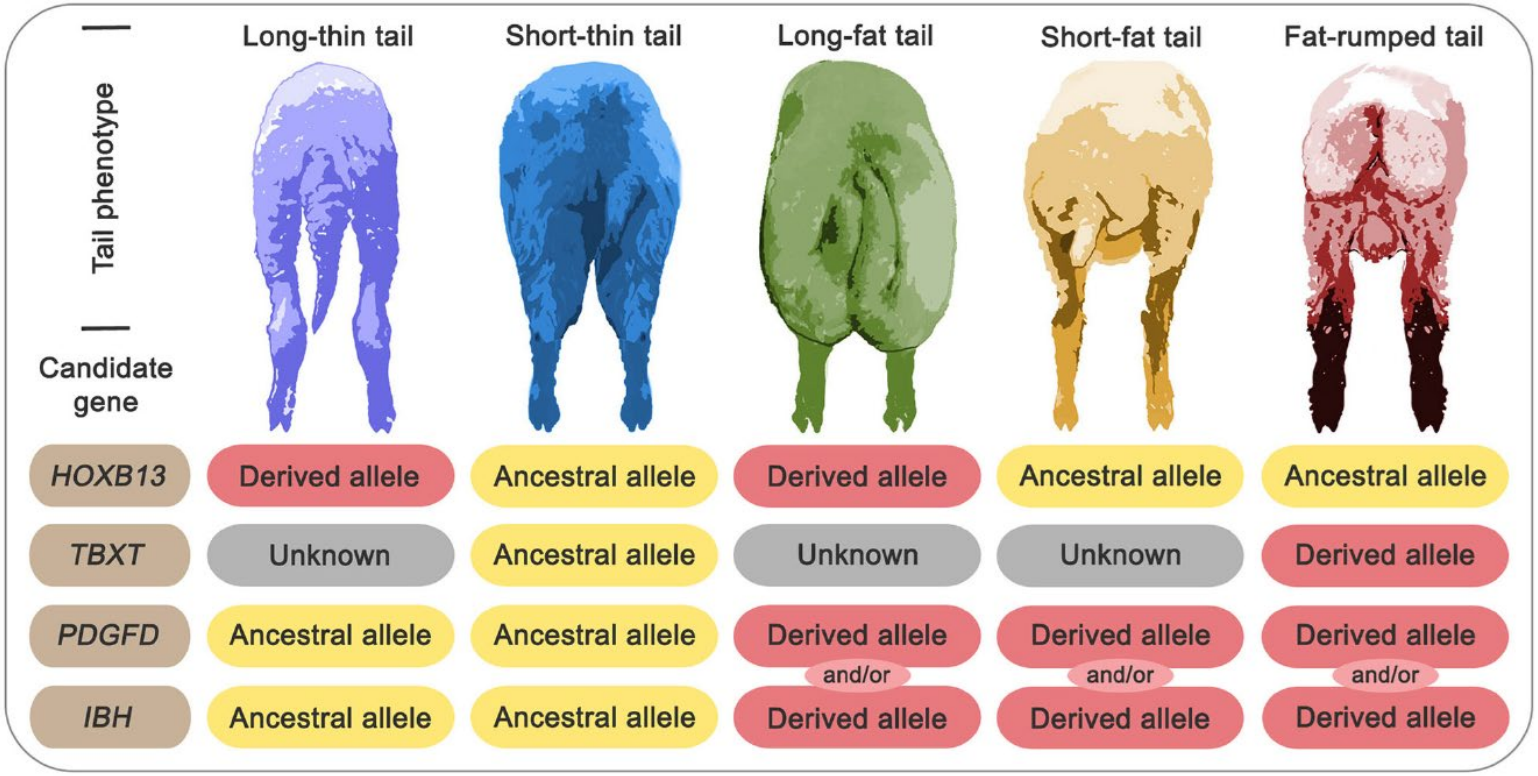


Fig. 4 Natural tail length variation in Merinolandschaf. Photo of a short-tailed (**a**) and a long-tailed (**b**) lamb, both lambs were ~5 weeks old and not docked.

DOI: 10.1038/s42003-022-03854-3



DOI: 10.1038/s42003-022-04199-7

Goat genetics

SNP genotyping of Swiss goats successfully implemented

forum KLEINWIEDERKÄUER PETITS RUMINANTS

Eiweissvarianten in der Ziegenmilch: ein Update

Heidi Signer-Hasler / Stefanie Rohn /
Erika Bangerter / Cord Drögemüller

Alpha-S1-Kasein-Genotypenkategorie Catégorie de génotype de caséine alpha-S1	Mittelwert Eiweissgehalt Teneur moyenne en protéine	Anzahl Tiere Nombre d'animaux	Mittelwert Eiweisszuchtwert Valeur d'élevage moyenne pour la protéine	Anzahl Tiere Nombre d'animaux
++	3.20	15	102	17
+-	3.17	47	101	53
--	2.94	476	92	489

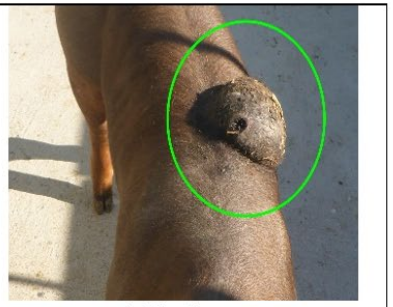
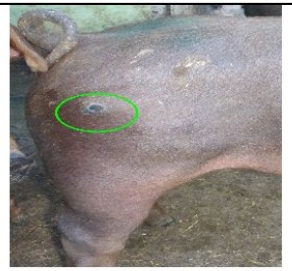
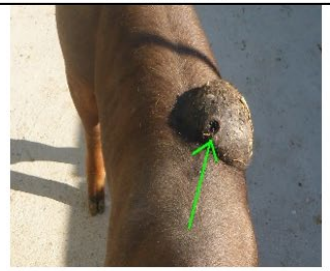
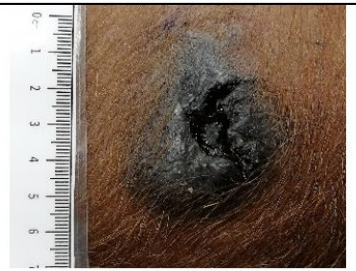
collaboration with Heidi Signer-Hasler and Erika Bangerter

FH Berner Fachhochschule

SZZV FSEC FSAC

Swine genetics

Melanoma in Duroc

Oberfläche (flach/ erhaben)	 Flach	 Erhaben	 Erhaben
Ulzeration/ Blutung (Ja/Nein)	 Nein	 Ja	 Ja

Sample collection ongoing

GWAS planned

Partner in a collaboration of

Universität Bern | Universität Zürich

vetsuisse-fakultät

SUISAG

Swine genetics

Reducing losses of pigs due to Haemorrhagic Intestinal Syndrome (HIS)

Holenweger *et al. Porcine Health Management* (2023) 9:44
https://doi.org/10.1186/s40813-023-00340-y

Porcine Health Management

RESEARCH Open Access

Housing and management factors and breed predisposition for haemorrhagic bowel syndrome in swine

Fabienne Holenweger[1], Gertraud Schüpbach[2], Andreas Hofer[3], Xaver Sidler[4] and Alexander Grahofer[1*]

Partner in a collaboration of

ETH zürich

Universität Bern | Universität Zürich

vetsuisse-fakultät

SUISAG

Food and Agriculture Organization of the United Nations

INTERNATIONAL YEAR OF

CAMELIDS

2024

Camelid genetics

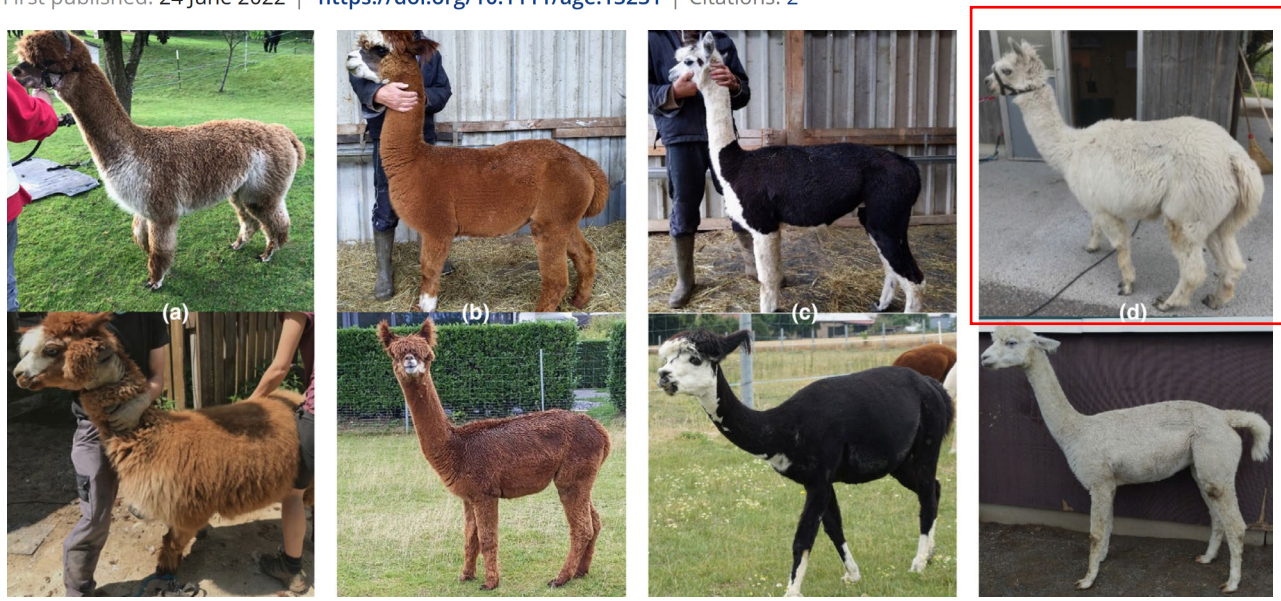
ANIMAL GENETICS
Immunogenetics, Molecular Genetics and Functional Genomics

BRIEF REPORT | Open Access

The *KIT*:c.376G>A variant in German and Swiss alpacas (*Vicugna pacos*) with different coat colors

Kirsty Tan, Mia Roy, Eberhard Manz, Henrik Wagner, Patrik Zanolari, Cord Drögemüller, Gesine Lühken

First published: 24 June 2022 | https://doi.org/10.1111/age.13231 | Citations: 2



absence of the *KIT* c.376 A allele in an alpaca with proven BEW phenotype opens questions

GWAS for blue-eyed white (BEW) planned

Partner in a collaboration of

JUSTUS-LIEBIG-UNIVERSITÄT GIESSEN

NeuweltkamelidenSchweiz NWKS

Thanks for listening

UNIVERSITÄT BERN

Cord Drögemüller, Institute of Genetics, Vetsuisse faculty, University of Bern

Swiss Animal Breeding Technology Platform, Zurich, 11th January 2024