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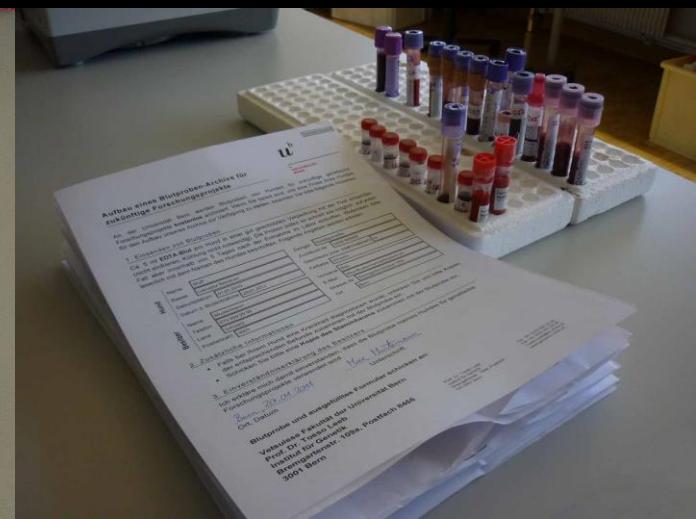
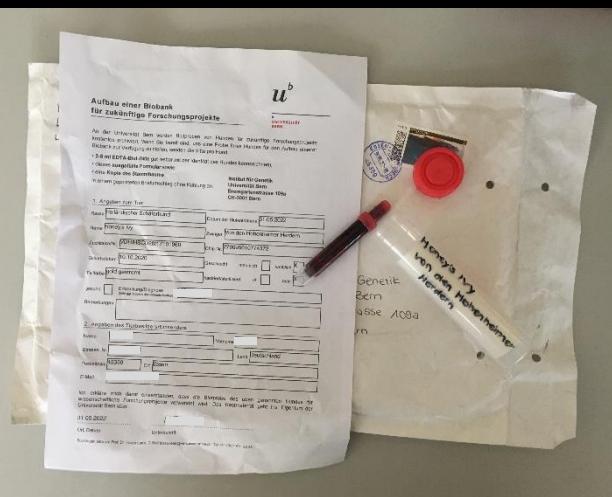
The Vetsuisse Biobank and New Insights on the Genetic Diversity of Bernese Mountain Dogs

Universität Bern | Universität Zürich

vetsuisse-fakultät

Tosso Leeb

Research requires biological samples



The Vetsuisse Biobank



The Vetsuisse Biobank

Bern

VET-GEN-BERN

VET-NEURO-BERN

VET-PATH-BERN

> 1 million samples

Zurich

VET-LAB-ZURICH

VET-PATH-ZURICH

The Vetsuisse Biobank

VET-GEN-BERN

> 100'000 samples



Owner Consent

1. Animal information

Breed	Date of sample taking		
Name	Kennel		
Registry ID	Chip No.		
Date of birth	Sex	male	female
Coat color	Sterilized	yes	no
Healthy	Disease/diagnostics (if applicable please add copies of the diagnostic findings)		
Remarks			

2. Owner information

Name	First name
Street, No.	Country
Postal code	City
E-mail	

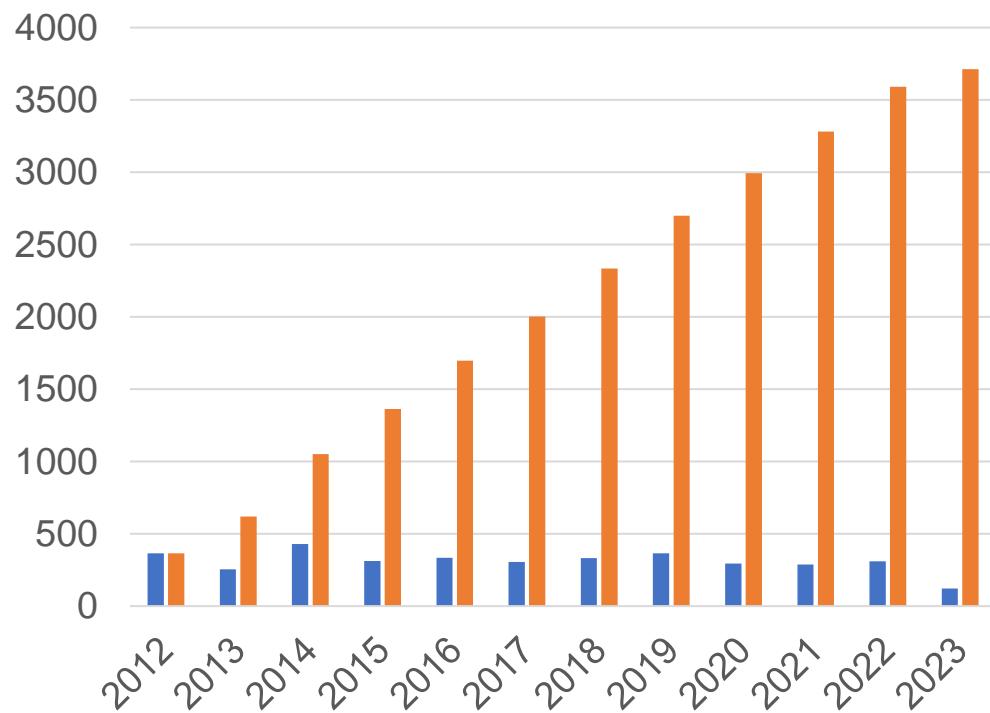
I agree that the sample material of the above-mentioned animal may be used for scientific research projects. The leftover sample material will belong to the University of Bern.

.....
City, date

.....
Signature

Bernese Mountain Dog Samples

Year	New samples	Total samples
≤ 2012	365	365
2013	255	620
2014	430	1050
2015	312	1362
2016	335	1697
2017	306	2003
2018	331	2334
2019	364	2698
2020	294	2992
2021	288	3280
2022	310	3590
2023	122	3712



SOD1 Variants: Allele & Genotype Frequencies

243 Swiss BMDs, birth year 2019

Allele Frequency		Genotype Frequency		Dogs at risk:
N	72.0%	N/N	47.7%	DM1/DM2 6
DM1 (SOD1A)	23.3%	N/DM	48.6%	DM1/DM1 3
DM2 (SOD1B)	4.7%] 28.0%	DM/DM	3.7%	Total 9

Hardy-Weinberg Equilibrium: N/N N/DM DM/DM

expected	126	98	19
observed	116	118	9

P_{HWE} = 0.001



Article

Genomic Diversity and Runs of Homozygosity in Bernese Mountain Dogs



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Genes (Basel) 2023, 14:650

Study Cohort

12 U.S.A.



7 France



7 Sweden



7 Switzerland



33 Total

year of birth: 1991 ... 2019

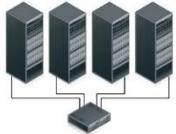


illumina whole genome sequence data
23x average coverage (9.38x ... 50.61x)

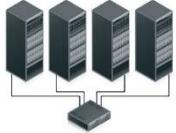
Data Analysis



- raw sequence data

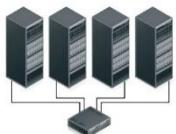


Alignment to
UU_CFam_GSD_1.0 reference genome (2.4 Gb)



Calling of
Sequence variants

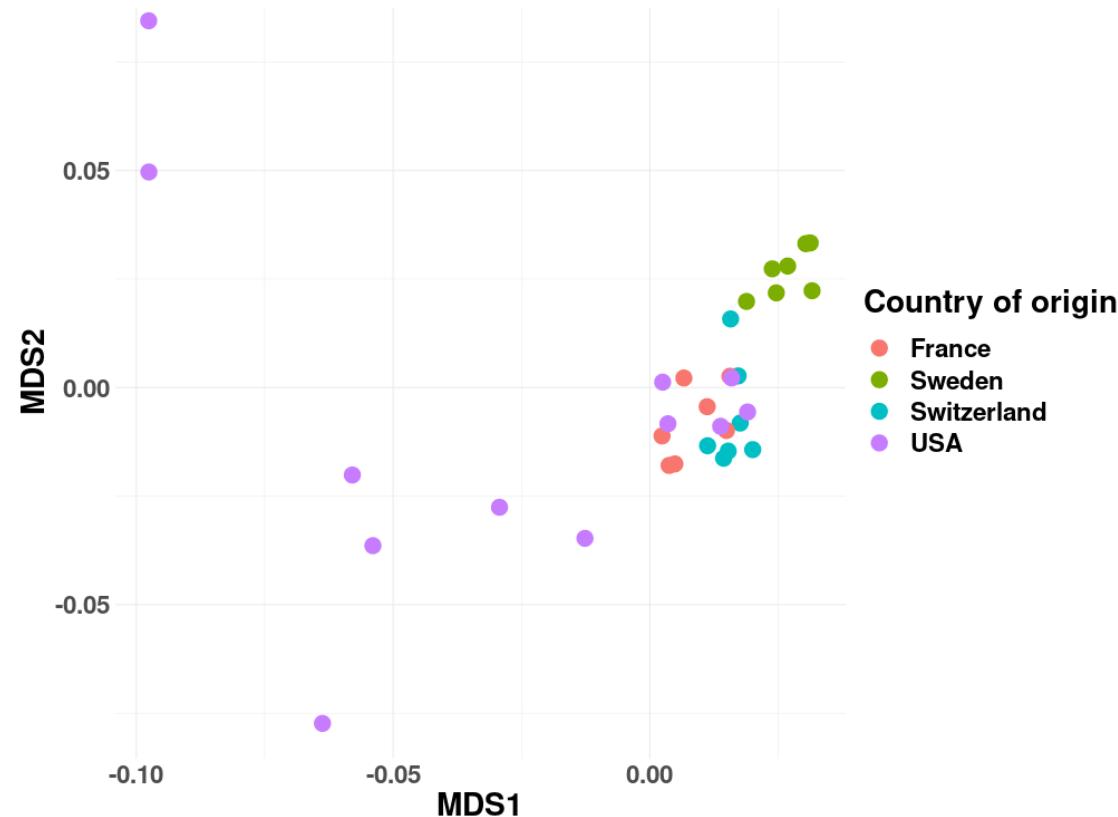
12,814,168 variants



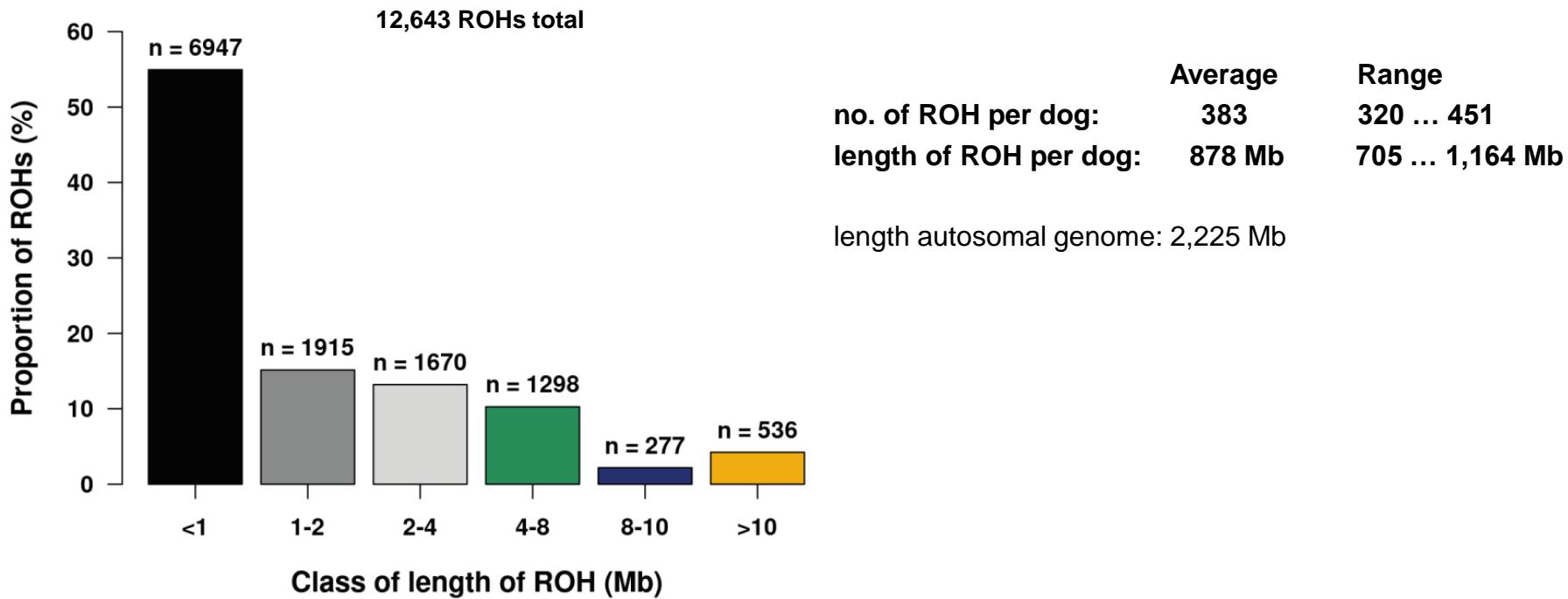
Quality control, pruning of variants

6,344,366 high quality, bi-allelic SNVs on autosomes

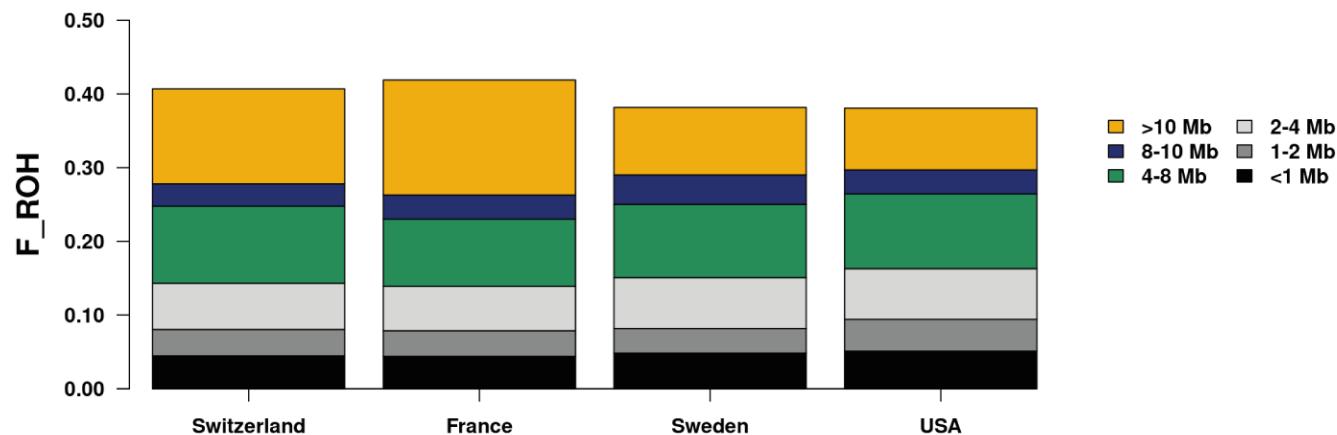
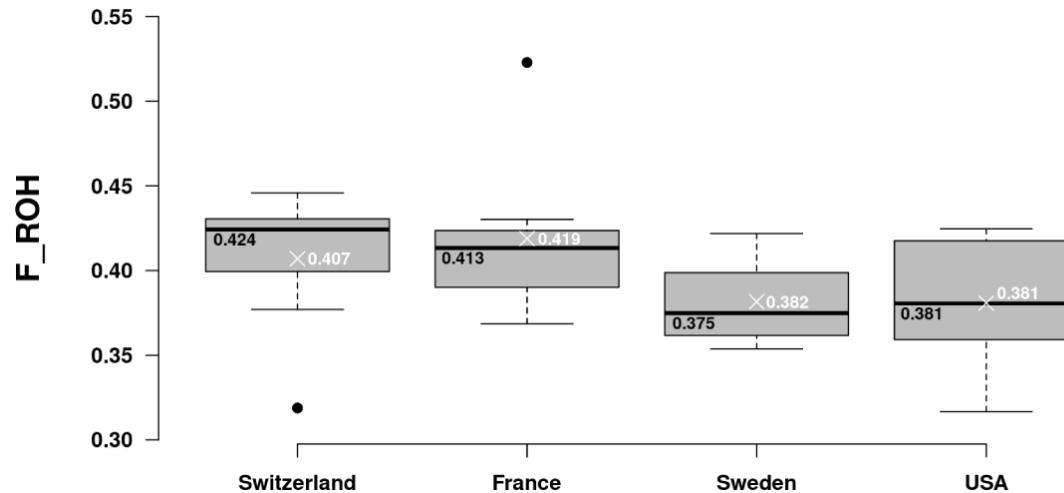
Population structure



Runs of Homozygosity (ROH)



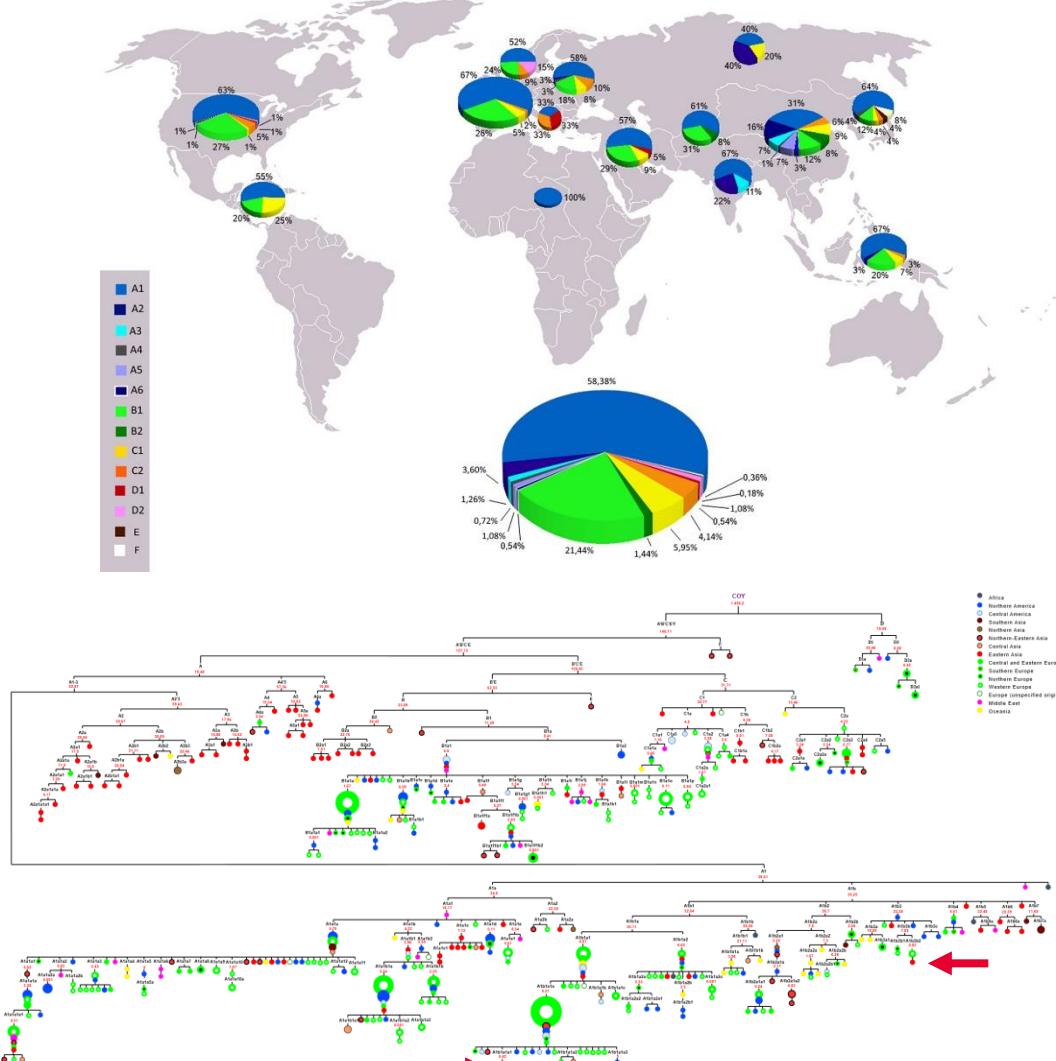
Runs of Homozygosity (ROH)



Mitochondrial Haplotypes

Haplotype	Dogs
A1b2a1a1	7
A1b3b2	26

→ only 2 mitochondrial haplotypes



Functional Variants

Disease/Trait	Gene	Allele Frequency	Genotype Counts		
			Ref/Ref	Ref/A _l t	A _l t/A _l t
Degenerative Myelopathy	<i>SOD1</i>				
DM1 (SOD1A)		37.9%	10	21	2
DM2 (SOD1B)		1.5%] 39.4%	32	1	-
Height	<i>IGF1</i>	3.0%	31	2	-
Coat Color Black Back (VP2-HCP3)	<i>ASIP</i>	100.0%	-	-	33
Hair Length	<i>FGF5</i>	100.0%	-	-	33

Questions ?



Bildquelle: <https://www.bernernennenhund.ch/>