

The Vetsuisse Biobank and new Insights on the Genetic Diversity of Bernese Mountain Dogs
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Biological samples needed for research are stored with the Vetsuisse Biobank at the University of Bern and at the University of Zurich. More than a million samples from many animal species are stored in five different collections. Dr. Leeb was drawing his information from the more than 100,000 blood samples stored in the Vet-Gen-Bern collection that are used for genetic research. As of the time of the presentation, there were 3,712 Berner samples in their repository. These samples are predominantly from Swiss dogs and the Swiss BMD population is nearly completely biobanked since 2012.

Looking at the SOD1 variants for DM, they tested 243 Swiss dogs born in 2019. The results of that analysis were:

Allele Frequency:		Genotype Frequency:	
Normal	72.0%	Clear	47.7%
SOD1-A	23.3%	Carrier	48.6%
SOD1-B	4.7%	At Risk	3.7%

Nine at-risk dogs were found, three had two copies of the SOD1-A mutation, and six were compound heterozygotes having one copy of the SOD1-A mutation, and one copy of the SOD1-B mutation. These results were significantly different from what would be expected from a random distribution using the Hardy-Weinberg Equilibrium calculation. In random breeding, 19 dogs with two SOD1 alleles would have been expected, but only 9 were observed. This indicates that breeders are using the SOD1 genetic tests to avoid matings that are at risk to produce puppies with two mutant SOD1 alleles. However, there is still room for improvement. In every mating, one of the two breeding dogs should be clear for SOD1-A and SOD1-B.

Research using whole genome sequence data from 33 Bernese (seven each from France, Sweden, and Switzerland, and 12 from the USA) were used to assess the genetic diversity in the breed. The year of birth for these dogs ranged from 1991 to 2019. A map of the results of genetic variation found a great deal of similarity among the European dogs, with some of the USA dogs clustered with them, but more of the USA dogs distributed away from the European cluster. See figure 1 in the article link below.

The full canine genome consists of 2.4 billion base pairs, and the first look at the analysis of these 33 dogs found nearly 13 million variants – so the breed is not completely homozygous. But looking at the data in detail and identifying so-called runs of homozygosity, the estimated coefficient of inbreeding averaged as follows (see figure 2 in the article link below):

Sweden –	37.5%
USA –	38.1%
France –	41.3%
Switzerland –	42.4%

This indicates that in each sequenced Berner, about a third of the genome is homozygous. Only 2 mitochondrial haplotypes were found, and both belong to haplogroup A. This is the most common haplogroup in domestic dogs.

See the full article here: <https://www.mdpi.com/2073-4425/14/3/650>