English version

Dear Ladies and Gentlemen

We are pleased to inform you about research progress regarding cerebellar dysfunction in Belgian Shepherd dogs. After intensive research in an international consortium we were able to identify a genetic defect in Belgian Shepherd dogs, especially in Malinois and Tervueren dogs, which causes a severe neurological disease with monogenic autosomal recessive inheritance. The disease was termed "Spongy Degeneration with Cerebellar Ataxia Subtype 1" (**SDCA1**).
Thanks to the results of our study, genetic testing for **SDCA1** is now possible at specialized laboratories, which helps to avoid the non-intentional breeding of affected puppies. The genetic test is currently offered by LABOKLIN (https://shop.labogen.com/en) und OPTIGEN (http://www.optigen.com/opt9_sdca1_test.html). The Institute of Genetics of the University of Bern does not provide a genetic test for SDCA1.

It is important to note that the identified genetic defect mutation cannot explain all forms of cerebellar dysfunction in Belgian Shepherd dogs. We are aware of clinically very similar, additional forms of cerebellar dysfunction in Belgian Shepherd dogs, which are caused by other currently unidentified genetic defects. We would like to identify these additional genetic defects in the future.

Explanation of the genetic test result:

There are two copies of each gene in the genome of a dog. One copy is inherited from the father and one from the mother. If a trait is inherited in an autosomal recessive manner, it means that an animal will only get the disease if it receives defective gene copies from both the father and the mother. Thus to produce an affected puppy, both parents (father and mother) must carry the defective gene. However, the carriers with only one copy of the defect will not be affected themselves.

Spongy Degeneration with Cerebellar Ataxia Subtype 1 (SDCA1) - Autosomal recessive inheritance		
Genotype: **TT** (clear)	Genotype: **TC** (carrier)	Genotype: **CC** (affected)
This animal does not carry the genetic defect and has no risk of developing SDCA1. The dog cannot pass the genetic defect to its offspring.	This animal carries one copy of the defective gene. The dog has no risk of developing SDCA1. However, this defect will be passed to its offspring with a probability of 50%. Such an animal should only be mated to a clear animal.	This animal carries two copies of the defective gene and is affected by SDCA1. Most SDCA1 affected dogs are euthanized by the 17th week of life because of severe neurological symptoms and poor quality of life.

Carriers have a 50% probability of passing the defective gene copy to their offspring. If two carriers are mated, there is a risk that 25% of the offspring will be affected by SDCA1. Therefore, the mating of two carriers should be strictly avoided (also legally forbidden in many countries).
Carriers do not have to be categorically excluded from breeding. However, carriers should only be mated to clear dogs so that no homozygous affected puppies will be produced.

Detailed information about our study can be found here:

- Link to the scientific publication: http://www.g3journal.org/content/early/2016/12/19/g3.116.038455.full.pdf+html
- Link to our website for general information and for sending blood samples for future research: http://www.genetics.unibe.ch/research/documents_dogs/cerebellar_ataxia_in_the_malinois/index_eng.html

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