Spongy Degeneration with Cerebellar Ataxia subtype 1 (SDCA1)

Affected breeds: Belgian Shepherd (Malinois)

Spongy Degeneration with Cerebellar Ataxia subtype 1 (SDCA1) is a severe neurodegenerative disease found in the Malinois variety of the Belgian Shepherd. Affected puppies show a rapidly progressing ataxia (difficulty walking) accompanied by other progressive symptoms associated with neurodegeneration. Euthanasia is required at a young age.
There are other cerebellar ataxia conditions in this breed that are caused by other as yet unidentified mutations.

SDCA1 is caused by a recessive genetic mutation. This means that dogs which carry the mutation ("CARRIERS") are normal but will pass the mutation on to an average of 50% of their offspring. Puppies which inherit two copies of the mutation will develop SDCA1 ("AFFECTED").

This test is particularly useful for breeders:

- To identify carriers among their breeding stock so that they can avoid CARRIER X CARRIER mating combinations which would risk AFFECTED puppies.
- To conclusively confirm SDCA1 in an affected dog

This test will be reported as:

CLEAR : no evidence of the SDCA1 mutation
CARRIER : carries one copy of the defect, which will be passed to 50% of offspring
AFFECTED : carries two copies of the defect, causing SDCA1

The genetic status of dogs can be used to predict breeding outcomes when different combinations are mated:

CLEAR X CLEAR = 100% CLEAR
CARRIER X CLEAR = 50% CARRIER, 50% CLEAR
CARRIER X CARRIER = 25% AFFECTED, 50% CARRIER, 25% CLEAR

References

Mauri, N., Kleiter, M., Leschnik, M., Högler, S., Dietschi, E., Wiedmer, M., … Leeb, T. (2017). A Missense Variant in KCNJ10 in Belgian Shepherd Dogs Affected by Spongy Degeneration with Cerebellar Ataxia (SDCA1). G3: Genes|Genomes|Genetics, 7(2), 663–669. http://doi.org/10.1534/g3.116.038455