

Neuroaxonal Dystrophy (NAD)

Affected breeds: Papillon

NAD is a neurodegenerative disorder which is progressive and fatal. Affected pups first show signs at a few months of age and these include loss of sight, tremours and movement abnormality followed by paralysis. Euthanasia is ultimately required.



The mutation is recessive, which 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 NAD ("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 NAD

This test will be reported as:

CLEAR : no evidence of the NAD mutation

CARRIER : carries one copy of the defect, which will be passed to 50% of offspring

AFFECTED : carries two copies of the defect, and will have NAD

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

Tsuboi M, Watanabe M, Nibe K, et al. Identification of the PLA2G6 c.1579G>A Missense Mutation in Papillon Dog Neuroaxonal Dystrophy Using Whole Exome Sequencing Analysis. PLoS One. 2017;12(1):e0169002. Published 2017 Jan 20. doi:10.1371/journal.pone.0169002