

Centronuclear Myopathy (CNM)

Affected breeds: Labrador retriever

Centronuclear Myopathy is an inherited condition found in Labradors which results in muscle weakness and exercise intolerance. At birth the puppies appear normal, but from about two weeks of age they begin to lose weight. By 2 – 5 months they develop an awkward movement and become weak due to poor muscle strength. Although the condition is not fatal, it is a serious debilitating disease which requires on-going treatment and should be avoided at all costs.

CNM has also been referred to as Hereditary Myopathy of the Labrador Retriever (HMLR), Autosomal Recessive Myopathy (ARMD), Type II deficiency Myopathy and Labrador Muscular Myopathy.

CNM is caused by a recessive genetic mutation. This means that dogs which carry the mutation ("CARRIERS") are healthy but will pass the mutation on to an average of 50% of their offspring. Puppies which inherit two copies of the mutation will suffer from CNM ("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 diagnosis of Centronuclear Myopathy

This test will be reported as:

CLEAR : no evidence of the Centronuclear Myopathy 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 develop Centronuclear Myopathy

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

Pele M, Turet L, Kessler J-L, Blot S, Panthier J-J (2005) SINE exonic insertion in the *PTPLA* gene leads to multiple splicing defects and segregates with the autosomal recessive centronuclear myopathy in dogs. *Human Molecular Genetics* 14(11): 1417-1427

Maurer M, Mary J, Guillaud L, Fender M, Pelé M, et al. (2012) Centronuclear Myopathy in Labrador Retrievers: A Recent Founder Mutation in the *PTPLA* Gene Has Rapidly Disseminated Worldwide. *PLoS ONE* 7(10): e46408. doi:10.1371/journal.pone.0046408