

# VETERINARY TECHNICAL DATASHEET

Centronuclear Myopathy, (CNM); mutation originally found in Great Dane



Mutation Found In :Great Dane

## Disorder Type

- Muscle

## Disease Severity

- Severe

## Background

With CNM, the severity of the clinical signs and age of onset varies, but common manifestations are muscle weakness and skeletal muscle wasting reducing the dog's exercise capabilities and causing changes in posture.

## Key Signs

- Muscle weakness
- Exercise intolerance
- Ventroflexion
- Skeletal muscle atrophy

## Clinical Description

In Great Danes, typical clinical signs of CNM are general atrophy of the muscles, exercise intolerance, and exercise-related tremors of the muscles. Typically clinical signs appear at 6 to 8 months of age and progress with age. Most affected dogs will need to be euthanized before the age of 18 months due to severe weakness of the muscles.

## Mode of Inheritance

- autosomal recessive

## Gene Name

- BIN1

## Next Steps

Treatment is supportive care. Humane euthanasia for affected dogs is often elected.

## References

Böhm J, Vasli N, Maurer M, Cowling BS, Shelton GD, Kress W, Toussaint A, Prokic I, Schara U, Anderson TJ, Weis J, Tired L, Laporte J. Altered splicing of the BIN1 muscle-specific exon in humans and dogs with highly progressive centronuclear myopathy. *PLoS Genet* 9(6): e1003430, 2013.

Gentilini F, Zambon E, Gandini G, Rosati M, Spadari A, Romagnoli N, Turba ME, Gernone F. Frequency of the allelic variant of the PTPLA gene responsible for centronuclear myopathy in Labrador Retriever dogs as assessed in Italy. *J Vet Diagn Invest* 23:124-6, 2011.

Pelé M, Tired L, Kessler JL, Blot S, Panthier JJ. SINE exonic insertion in the PTPLA gene leads to multiple splicing defects and segregates with the autosomal recessive centronuclear myopathy in dogs. *Hum Mol Genet* n14:1417-27, 2005.

