

# VETERINARY TECHNICAL DATASHEET

Congenital Myasthenic Syndrome, (CMS); mutation originally found in Labrador Retriever



Mutation Found In :Labrador Retriever

## Disorder Type

- Neuromuscular

## Disease Severity

- Mild/moderate

## Background

Several mutations have been found to cause similar clinical signs in various breeds. This particular mutation was identified in the Labrador Retriever and is associated with a more severe form of CMS and signs are present at a very young age. Muscle weakness results from a defect in the building of an enzyme necessary for the muscles to work properly called acetylcholinesterase.

## Key Signs

- Temporary collapse
- Paralysis induced by exercise

## Clinical Description

Initial signs of congenital myasthenic syndrome can be observed in 12 to 16 weeks old puppies. Affected dogs suffer from exercise intolerance and collapse after 5 to 30 minutes of exercise. Before collapsing, affected dogs will start to take shorter and shorter strides and eventually fall down. Affected dogs are able to recover from the transient paralysis after resting for a few minutes, but the signs reappear if the dog continues to run.

## Mode of Inheritance

- autosomal recessive

## Gene Name

- COLQ

## Next Steps

The clinical signs can be avoided by limiting exercise to a very short period of time and low intensity. If the dog collapses, allow it to rest and recover and then stop further exercise to prevent further difficulties or collapse.

## References

Rinz CJ, Levine J, Minor KM, Humphries HD, Lara R, Starr-Moss AN, Guo LT, Williams DC, Shelton GD, Clark LA. A COLQ missense mutation in Labrador Retrievers having congenital myasthenic syndrome. PLoS One. 2014 Aug 28;9(8):e106425.