

# VETERINARY TECHNICAL DATASHEET

Congenital Myasthenic Syndrome, (CMS); mutation originally found in Jack Russell Terrier



Mutation Found In :Russell Terrier

## Disorder Type

- Neuromuscular

## Disease Severity

- Mild/moderate

## Background

Several mutations have been found to cause similar clinical signs in various breeds. A mutated CHRNE gene has been identified as causing CMS in the Jack Russell Terrier. The mutation results in a substantial reduction of a muscle related protein called the acetylcholine receptor protein causing affected dogs to show muscle weakness.

## 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

- CHRNE

## 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, Lennon VA, James F, Thoreson JB, Tsai KL, Starr-Moss AN, Humphries HD, Guo LT, Palmer AC, Clark LA, Shelton GD. A CHRNE frameshift mutation causes congenital myasthenic syndrome in young Jack Russell Terriers. Neuromuscul Disord. 2015 Dec;25(12):921-7.