

# VETERINARY TECHNICAL DATASHEET

Myotonia Congenita; mutation originally found in Australian Cattle Dog



Mutation Found In :Australian Cattle Dog, Australian Stumpy Tail Cattle Dog

## Disorder Type

- Muscle

## Disease Severity

- Moderate

## Background

Myotonia congenita is a congenital muscular disorder caused by a defect in ion channels of the skeletal muscles which leads to delayed relaxation of skeletal muscles following contractions. The condition causes affected dogs to have hyperexcitable muscles that contract easily. This particular genetic variant has been found in the Australian Cattle Dog.

## Key Signs

- Delayed muscle reaction
- Stiff movements
- Hypertrophic skeletal muscles
- Bunny-hop-like movement

## Clinical Description

The clinical signs can be seen in puppies only a few weeks old. An affected dog suffers from muscle hypertrophy and has stiff movements. It can have difficulties rising after rest and in rapid changes of posture. The disorder is characterized by a bunny-hopping gait. An affected dog may also suffer from superior prognathism (protrusion of one or both jaws), ptyalism (excessive salivation), dental abnormalities, and increased respiratory sounds during exercise. The tongue of affected dogs is enlarged and stiffens when touched.

## Mode of Inheritance

- autosomal recessive

## Gene Name

- CLCN1

## Next Steps

Affected dogs may need assistance after rest or during muscle cramping to allow them to get to their feet and move normally. Some assistance may also be needed for everyday tasks such as stair climbing.

## References

Rhodes TH, Vite CH, Giger U, Patterson DF, Fahlke C, George AL. A missense mutation in canine CIC-1 causes recessive myotonia congenita in the dog. FEBS Lett 456:54-58, 1999.