# VETERINARY TECHNICAL DATASHEET

Juvenile Myoclonic Epilepsy; mutation originally found in Rhodesian Ridgeback

Mutation Found In :Rhodesian Ridgeback

## Disorder Type

- Nervous system

## Disease Severity

- Moderate/severe

## Background

Juvenile Myoclonic Epilepsy is a specific form of epilepsy that has been described in Rhodesian Ridgebacks. As the name states, clinical signs emerge at a young age. First signs of the disease are muscle jerks/twitches which can progress to generalized seizures. Epileptic activity is triggered usually by resting or by visual stimulus (light).

## Key Signs

- Myoclonic jerks during rest or after visual stimulus

## Clinical Description

Clinical signs of the disease typically emerge around 6 months of age but the age of onset can vary from as young as 6 weeks to 1.5 years of age. Clinical signs begin with myoclonic jerks/twitches or the trunk, forelimbs, and nodding of the head. The appearance can resemble severe startling or even an electric shock. Twitches of varying intensity are most commonly seen in relaxed dogs that are beginning to sleep. The frequency of these twitches can be very high, even 150 twitches per day. The ability to sleep and rest is thus impaired. Affected dogs can also be sensitive to light with visual stimulus triggering epileptic activity. Approximately 38% of affected dogs progress to having generalized tonic-clonic seizures. Biological factors, such as vaccination or heat might affect the frequency and/or initiation of clinical signs.

## Mode of Inheritance

- autosomal recessive

## Gene Name

- DIRAS1

## Next Steps

Antiepileptic drugs are used to reduce epileptic activity. Levitiracetam and potassium bromide seem to achieve a good response. Affected dogs should be kept in a safe environment and in a way that they do not harm themselves during epileptic activity. Seizure control can sometimes be impossible to achieve and euthanasia may be elected on welfare grounds.

## References

Wielaendera F, Sarviaho R, Jamese F, Hytönen M, Cortez M, Klugerh G, Koskinen L, Arumilli M, Kornbergj M, Bathen-Noethenk A, Tipoldl A, Rentmeisterm K, Bhattin S, Hülsmeyera V, Boettchero I, Tästenseno C, Flegelo T, Dietschip E, Leebp T, Matiasekq K, Fischera A, and Lohi H. Generalized myoclonic epilepsy with photosensitivity in juvenile dogs caused by a defective DIRAS family GTPase 1. doi: 10.1073/pnas.1614478114