Juvenile Myoclonic Epilepsy (JME)

Affected breeds: Rhodesian Ridgeback

JME is a unique form of early onset epilepsy in the Rhodesian Ridgeback (first signs appear at 6-18months of age), which is characterised initially by frequent jerks and twitches, and progresses into generalised tonic-clonic seizures after several months.

The mutation is recessive, which means that dogs which carry the mutation ("CARRIERS") are normal but will pass the mutation on to an average of 50% of their offspring. Puppies which inherit two copies of the mutation will develop Juvenile Myoclonic Epilepsy

This test is particularly useful for breeders:

- To identify carriers among their breeding stock so that they can avoid CARRIER X CARRIER mating combinations which would risk AFFECTED puppies.
- To conclusively confirm Juvenile Myoclonic Epilepsy

This test will be reported as:

CLEAR : no evidence of the Juvenile Myoclonic Epilepsy mutation
CARRIER : carries one copy of the defect, which will be passed to 50% of offspring
AFFECTED : carries two copies of the defect, and will develop Juvenile Myoclonic Epilepsy

The genetic status of dogs can be used to predict breeding outcomes when different combinations are mated:

CLEAR X CLEAR = 100% CLEAR
CARRIER X CLEAR = 50% CARRIER, 50% CLEAR
CARRIER X CARRIER = 25% AFFECTED, 50% CARRIER, 25% CLEAR

References

Wielaender F, Sarviaho R, James F, Hytönen MK, Cortez MA, Kluger G, Koskinen LL, Arumilli M, Kornberg M, Bathen-Noethen A, Tipold A, Rentmeister K, Bhatti SF, Hülsmeyer V, Boettcher IC, Tästensen C, Flegel T, Dietschi E, Leeb T, Matiasek K, Fischer A, Lohi H. Generalized myoclonic epilepsy with photosensitivity in juvenile dogs caused by a defective DIRAS family GTPase 1. Proc Natl Acad Sci U S A. 2017 Mar 7;114(10):2669-2674. doi: 10.1073/pnas.1614478114. Epub 2017 Feb 21. PMID: 28223533; PMCID: PMC5347561.