GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.564_567delAGAC mutation in DIRAS1 gene causing JME in Rhodesian ridgebacks

Customer: Jan Novák, Dlouhá 1, 30000 Plzeň, Czech Republic
Sample:
Sample: 21-12345
Date received: 01.02.2021
Sample type: blood

Information provided by the customer
Name: Lassie DEMO
Breed: Plemeno
Tattoo number: 1392013
Microchip: 123 456 789 012 345
Reg. number: REGQ12345
Date of birth: 1.1.2020
Sex: female
Date of sampling: 01.02.2021
The identity of the animal has been checked.

Result: Mutation was not detected (N/N)

Legend: N/N = wild-type genotype. N/P = carrier of the mutation. P/P = mutated genotype (individual will be most probably affected with the disease). (N = negative, P = positive)

Explanation

Presence or absence of c.564_567delAGAC mutation in DIRAS1 gene causing juvenile myoclonic epilepsy (JME) in Rhodesian ridgebacks was tested. Disease is characterized by sudden short and uncontrolled muscle jerks or twitches. The epileptic seizures affect mainly the muscles of proximal limbs and trunk, cervical muscles, head muscles and facial muscles. The disease occurs in young dogs, usually around 6 months of age. The seizures occur daily or almost daily. The frequency and intensity of the seizures can differ among the individual dogs or among the seizures of an individual dog respectively. The seizures occur most commonly when the animals are relaxed, drowsy or in the first stages of sleep. Some affected dogs show photosensitivly induced seizures.

Mutation c.564_567delAGAC mutation in DIRAS1 gene causing JME in Rhodesian ridgebacks is inherited as an autosomal recessive trait. That means the disease affects dogs with P/P genotype only. The dogs with N/P genotype are considered carriers of the disease (heterozygotes). In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N, 25 % P/P and 50 % N/P.

Method: SOPAgriseq_canine, ngs, accredited method

Date of issue: 06.02.2021
Date of testing: 01.02.2021 - 06.02.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document GENOMIA genetic laboratory

Genomia is accredited in compliance with ISO/IEC 17025:2018 under #1549
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