

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 photosensitively 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

Date of issue: 06.02.2021

Date of testing: 01.02.2021 - 06.02.2021

Approved by: Mgr. Martina Šafrová, Laboratory Manager



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