

Detection of c.1552A>T mutation in LGI2
gene causing juvenile epilepsy in Lagotto
Romagnolo

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.1552A>T mutation in LGI2 gene causing juvenile epilepsy in Lagotto Romagnolo was tested. In the Lagotto Romagnolo canine breed, the epileptic syndromes occur at 5 to 9 weeks of age. The seizures manifest by tremor of the entire body sometimes connected with short loss of consciousness. The puppies with most severe seizures show also signs of neurological disease including generalized ataxia and hyperthermia. The epileptic seizures resolve spontaneously by 8 to 13 weeks of age.

Mutation that causes juvenile epilepsy in Lagotto Romagnolo dogs is inherited as an autosomal recessive trait. That means the disease affects dogs with P/P (positive/positive) genotype only. The dogs with N/P (negative/positive) genotype are considered carriers of the disease (heterozygotes), they are healthy but they can transmit the mutation on their offspring. In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N (healthy non-carriers), 50 % N/P (healthy carriers) and 25 % P/P (affected).

Method: SOPagrisseq_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



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