

Result certificate #012345

Detection of c.743delC mutation in RAB3GAP1 gene causing JLPP in Rottweilers and Black Russian Terriers

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.743delC mutation in RAB3GAP1 gene causing Juvenile laryngeal paralysis and polyneuropathy (JLPP) in Rottweilers and Black Russian Terriers was tested. The affected dogs by JLPP have breathing problems leading to laryngeal paralysis. The other symptoms may be ataxia, distal weakness and reduced spinal reflexes. In some affected dogs, the eyes were abrnomally small and had cataracts or modified bark as a result of atrophy of the laryngeal muscles. The first symptoms appear around 3 months of age. The affected dogs must be in most cases euthanized by 6 month of age.

Mutation that causes JLPP 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), they are healthy but they can transmit the mutation on their offspring. Dogs with N/N genotype are without risk of JLPP. 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: 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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