GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.179G>A mutation GLB1 gene causing gangliosidosis 1 in Portuguese Water Dogs

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.179G>A mutation GLB1 gene causing gangliosidosis 1 (GM1) in Portuguese Water Dogs was tested. The first clinical signs become apparent from 5-6 months of age. The affected dogs suffer from asymmetric growth and progressive neurological degradation caused by incorrect function of cerebellum. Subsequently, progressive loss of coordination, muscle tremors and muscle ataxia occur. The eyes of the dog can be also affected, beginning with squinting and ending by complete loss of vision. The progressive worsening of the health condition results in premature death of the dog or euthanasia.

Mutation that causes GM1 in Portuguese Water Dogs 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

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Report verification code is: 12AB-CD34-GENO-MIA0-EFGH. You can verify report online at www.genomia.cz

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