GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.388C>T mutation in EHBP1L1 gene causing DAMS in Labrador Retrievers

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.388C>T mutation in EHBP1L1 gene causing Dyserythropoietic anemia and myopathy syndrome (DAMS) in the Labrador Retrievers was tested. Dyserythropoietic anemia is characterized by a defect in the production of red blood cells (erythrocytes) in the bone marrow. In Labrador retrievers, there is a marked microcytosis (reduced erythrocytes) leading to anaemia, which is manifested by weakness, shortness of breath, fatigue and dizziness. Myopathic syndrome is a clinical condition that arises due to muscle fibre disorder and leads to muscle weakness, fatigue and muscle pain. There is also noticeable muscle atrophy in affected dogs.

Mutation that causes DAMS 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

verified and certified document
GENOMIA genetic laboratory

Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999