GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.473A>C mutation in GALC gene causing GLD in in West Highland White Terriers and Cairn 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.473A>C mutation in GALC gene causing GLD (Globoid cell leukodystrophy) in West Highland White Terriers and Cairn Terriers was tested. GLD is a severe lysosomal inherited storage disease caused by insufficient function of galactocerebrosidase (GALC). GLD occurs in puppies as early as 1 to 3 months after birth. The first symptoms include tremor of limbs, subsequent muscle atrophy and neurological degeneration of brain white matter and spinal cord. Dogs can survive 8 to 9 months when the signs become unbearable and the dogs are euthanized.

Mutation that causes GLD 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). Dogs with N/N genotype are without risk of GLD. In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N (healthy non-carriers), 25 % P/P (affected), and 50 % N/P (healthy carriers).

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

Genomia is accredited in compliance with ISO/IEC 17025:2018 under #1549
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