GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.1054G>A in YARS2 gene causing CJM in Belgian Shepherds

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 mutation c.1054G>A in YARS2 gene causing Cardiomyopathy with juvenile mortality (CJM) in Belgian Shepherds was tested. Cardiomyopathy with juvenile mortality is an inherited disease characterized by early death of puppies at birth or at a maximum age of six to eight weeks. The puppies initially develop normally, but then they show unspecific clinical signs like vomiting, uncoordinated movements (trembling, stumbling) and respiratory problems (dyspnea). The puppies die several days after onset of the first clinical signs, usually due to heart failure.

Mutation that causes CJM in Belgian Shepherds is inherited autosomally recessively which means that the disease develops only in those dogs who inherit mutated allele from both parents; 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
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