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Result certificate #012345

Detection of c.1169_1170dup mutation in PNPLA8 gene causing Progressive Degenerative Myeloencephalopathy in the Australian 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 c.1169_1170dup mutation in PNPLA8 gene causing Progressive Degenerative Myeloencephalopathy in the Australian Shepherds was tested. Progressive degenerative myeloencephalopathy is a neurological disease that affects the spinal cord and brain. The first signs of the disease appear in dogs between 4 and 19 months of age and include hypermetria (overreach of limbs while walking), a wobbly or stiff gait and difficulty walking up stairs and getting up. Ataxia worsens over time and the disease leads to an inability to walk and deficits in proprioception.

Mutation that causes Progressive Degenerative Myeloencephalopathy 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
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