GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.-6582_*516del mutation
in SELENOP gene causing CACA
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 c.-6582_*516del mutation in SELENOP gene causing central nervous system atrophy with cerebellar ataxia (CACA) in Belgian Shepherds was tested. Clinical signs, such as uncoordinated movements, intention tremors, general elevated muscle tone and reduced swallowing reflex can be observed in puppies at 2 weeks of age. Gradual progression leads to such a degree of severity of clinical signs that the animals must be euthanized. The SELENOP gene encodes selenoprotein P, which is involved in the deposition and transport of selenium to the brain and other organs. Incorporation of selenium into selenoprotein P prevents the toxic effects of free selenium. A deletion in the SELENOP gene results in the complete absence of encoded selenoprotein P and causes a defect in selenium transport to the CNS. Blood selenium concentration is reduced by 70% in affected homozygotes and 30% in heterozygotes compared to healthy dogs.

Mutation that causes CACA 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

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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