

Detection of c.118G>A mutation
in SOD1 gene causing
degenerative myelopathy in dogs

Customer: Dr. Josette Debrincat, 20, Animal Doctors, Guze Bajada, HMR 2131 Hamrun, Malta

Sample:

Sample: 24-26623

Date received: 01.10.2024

Sample type: blood

Information provided by the customer

Name: Scott Dell'Antica Contrada

Breed: Border Collie

Microchip: 380 206 044 889 554

Reg. number: MKC BOR/14/2024

Date of birth: 18/12/2022

Sex: male

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.118G>A mutation in SOD1 gene causing degenerative myelopathy in many canine breeds was tested. This mutation is sometimes referred to as SOD1A. Affected dogs have progressive loss of movement and gradual worsening of the condition up to complete paralysis. The age of disease onset and symptoms severity vary among the breeds.

Mutation SOD1A 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 (healthy non-carriers), 25 % P/P (affected), and 50 % N/P (healthy carriers).

The test does not exclude existence of another, nowadays unknown, mutation which can cause DM. In Bernese Mountain Dogs, there has been identified also SOD1B mutation responsible for DM - this test does not refer about SOD1B.

Analysis was performed by the partner laboratory. Genomia guarantees the quality of its partner's services.

Method: SOPAgriseq_canine, ngs

Date of issue: 13.10.2024

Date of testing: 01.10.2024 - 13.10.2024

Approved by: Mgr. Martina Šafrová, Laboratory Manager



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