GENOMIA
genetic laboratory

Result certificate #188628

Detection of g.28697542-28705340del7799 mutation in NHEJ1 gene causing CEA in several dog breeds

Customer: Lucie Glaserová, Doubek 109, 25101 Doubek, Czech Republic
Sample:
Sample: 21-27144
Date received: 06.10.2021
Sample type: blood

Information provided by the customer
Name: Bezinka z Modrého království
Breed: Collie Smooth
Microchip: 900 085 001 081 321
Reg. number: CMKU/CK/1647/20
Date of birth: 2.3.2020
Sex: female
Date of sampling: 04.10.2021
The identity of the animal has been checked by MVDr. Michaela Oravská, KVL 5097

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 g.28697542-28705340del7799 mutation in NHEJ1 gene causing Collie eye anomaly (CEA) was tested. CEA is known to affect Australian Shepherd, Border Collie, Boykin Spaniel, Lancashire heeler, Longhaired whippet, Nova Scotia Duck Tolling retriever, Rough and Smooth Collie, Shetland Sheepdog and Silken windhound.

Mutation that causes CEA 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).

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

Method: SOP176-CEA, ASA-PCR

Date of issue: 15.10.2021
Date of testing: 06.10.2021 - 15.10.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document GENOMIA genetic laboratory

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