GENOMIA
genetic laboratory

Result certificate #289110

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

Customer: Michal Glaser, Doubek 109, 251 01 Doubek, Czech Republic
Sample:
Sample: 23-15207
Date received: 05.06.2023
Sample type: buccal swab

Information provided by the customer
Name: Emilka z Modrého království
Breed: Collie Smooth
Microchip: 900 085 001 644 611
Reg. number: CMKU/CK/2155/23
Date of birth: 19.4.2023
Sex: female
Date of sampling: 01.06.2023
The identity of the animal has been checked by MVDr. Pavel Mádr, CSc. KVL4735

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: SOPAgriseq_canine, ngs

Date of issue: 27.02.2025
Date of testing: 12.02.2025 - 27.02.2025
Approved by: Mgr. Markéta Dajbychová, Deputy Laboratory Manager

verified and certified document
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