

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

Customer: Jan Novák, Dlouhá 1, 30000 Plzeň, Czech Republic

Sample:

Sample: 08-12345
Date received: 25.11.2008
Sample type: blood

Information provided by the customer

Name: Lassie DEMO

Breed: ---

Tattoo number: 1392013
Microchip: 123 456 789 012 345
Reg. number: REGQ12345
Date of birth: 31.12.1909
Sex: female
Date of sampling: 25.11.2008
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 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: 30.11.2008

Date of testing: 25.11.2008 - 30.11.2008

Approved by: Mgr. Martina Šafrová, Laboratory Manager



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