

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

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

**Sample:**

Sample: 24-22121

Date received: 29.08.2024

Sample type: blood

Information provided by the customer

**Name:** Meg

**Breed:** Border Collie

Tattoo number: N/A

Microchip: 380 260 102 381 321

Reg. number: MKC BOR/06/2024

Date of birth: 21/11/2022

Sex: female

**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: 10.09.2024

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Approved by: Mgr. Martina Šafrová, Laboratory Manager



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