

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

**Sample**

Sample: 19-13527  
Name: Adina Dvůr Bažantnice  
Breed: Collie Smooth  
Microchip: 972 273 000 004 700  
Reg. number: CMKU/CK/1079/16/18  
Date of birth: 24.04.2016  
Sex: female  
Date received: 16.05.2019  
Sample type: blood  
The identity of the animal has been checked by  
MVDr. Michaela Oravská

**Customer**

Ing. Lucie Glaserová  
Doubek 109  
25101 Doubek  
Czech Republic

**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

Report date: 22.05.2019

Responsible person: Mgr. Martina Šafrová, Laboratory Manager



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