

Detection of g.85286582_85286583insC mutation in HSF4 gene causing hereditary cataract in several dog breeds

Customer: Ivana Kubíčková, Želivského 806, 28002 Kolín, Czech Republic

Sample:

Sample: 19-42252

Date received: 16.09.2019

Sample type: buccal swab

Information provided by the customer

Name: Armstrong Siddeley Never Back Down

Breed: Staffordshire Bull Terrier

Microchip: 203098100427064

Reg. number: CMKU/SBT/11374/18

Date of birth: 22.03.2018

Sex: male

Date of sampling: 15.09.2019

The identity of the animal has been checked by Bc. Eva Šandarová, Genomia s.r.o.

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 mutation g.85286582_85286583insC in HSF4 gene causing hereditary cataract (HC) in Staffordshire bull terriers, French bulldogs and early onset HC in Boston Terriers was tested.

Mutation that causes HC in mentioned breeds is inherited in autosomal recessive trait. It means that the disease develops only in those dogs who inherit mutated allele from both parents; 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, 25 % P/P and 50 % N/P.

Method: SOP171-HC, fragment analysis, accredited method

Date of issue: 26.03.2021

Date of testing: 24.03.2021 - 26.03.2021

Approved by: Mgr. Lucie Magoči, Analyst



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