

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

Sample

Sample: 18-41637
Name: Angry Axel Harpagon
Breed: Staffordshire Bull Terrier
Microchip: 900164001446100
Reg. number: CMKU/SBT/10393/-17/16
Date of birth: 22.12.2016
Sex: male
Date received: 17.09.2018
Sample type: buccal swab
The identity of the animal has been checked by Mgr. Martina Šafrová, Genomia s.r.o.

Customer

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Ordered: 17.09.2019

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

Report date: 19.09.2019

Responsible person: Mgr. Martina Šafrová, Laboratory Manager

Genomia is accredited according to ISO/IEC 17025:2005 under #1549.

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