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genetic laboratory

Result certificate #106022

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

Sample
Sample: 18-03916
Name: Marylin Auricstaff Fransimo Bohemia
Breed: Staffordshire Bull Terrier
Microchip: 941 000 017 000 929
Date of birth: 18/02/2015
Sex: female
Date received: 19.02.2018
Sample type: buccal swab

Customer
Larisa Anatolyeva
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15521 Praha
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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: 27.02.2018
Responsible person: Mgr. Martina Šafrová, Laboratory Manager

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