

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

Customer: Vendula Linková, Valník 373, 783 44 Náměšť na Hané, Czech Republic

Sample:

Sample: 20-17343

Date received: 27.07.2020

Sample type: blood

Information provided by the customer

Name: Adebayor Of Black Seals

Breed: Staffordshire Bull Terrier

Microchip: 941 000 022 996 438

Reg. number: CMKU/SBT/12948/19

Date of birth: 12.5.2019

Sex: male

Date of sampling: 24.07.2020

The identity of the animal has been checked by MVDr. Tomáš

Weidenhöfer, KVL 3804

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: 04.08.2020

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Approved by: Mgr. Markéta Dajbychová, Deputy Laboratory Manager

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