GENOMIA
genetic laboratory

**Result certificate #240185**

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

**Customer**: Jaroslav Zavadil, Vlasatice 355, 69130 Vlasatice , Czech Republic
**Sample:**
Sample: 23-42420
Date received: 06.09.2023
Sample type: buccal swab

Information provided by the customer
**Name: Viktory z Hambalek Bušín**
**Breed: Staffordshire Bull Terrier**
Microchip: 203164000137927
Reg. number: CMKU/SBT/16390/21
Date of birth: 27.07.2021
Sex: female
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: 13.09.2023
Date of testing: 06.09.2023 - 13.09.2023
Approved by: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document GENOMIA genetic laboratory

Genomia is accredited in compliance with ISO/IEC 17025:2018 under #1549
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