

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

**Customer:** Renée Kryšpín Hájková, Prostřední Staré Buky 94, 54101 Staré Buky, Czech Republic

**Sample:**

Sample: 23-13708

Date received: 18.05.2023

Sample type: blood

Information provided by the customer

**Name:** El Mariachi Bagheera Bulls

**Breed:** Staffordshire Bull Terrier

Microchip: 203 098 100 528 713

Reg. number: CMKU/SBT/17404/22

Date of birth: 18.04.2022

Sex: male

Date of sampling: 17.05.2023

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

Sova, KVL 7127

**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: 22.05.2023

Date of testing: 18.05.2023 - 22.05.2023

Approved by: Ing. Nikola Eretová, Analyst

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Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic  
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999



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