

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

Customer: Jan Novák, Dlouhá 1, 30000 Plzeň, Czech Republic

Sample:

Sample: 21-12345

Date received: 01.02.2021

Sample type: blood

Information provided by the customer

Name: Lassie DEMO

Breed: Plemeno

Tattoo number: 1392013

Microchip: 123 456 789 012 345

Reg. number: REGQ12345

Date of birth: 1.1.2020

Sex: female

Date of sampling: 01.02.2021

The identity of the animal has been checked.

Result: Mutation was not detected (N/N)

Explanation

Presence or absence of mutation g.85286582delC in HSF4 gene causing hereditary cataract (HC) in Australian Shepherds was tested. Presence of deletion is connected with development of binocular cataract in different age of the dog. Generally, the mutation is inherited in autosomal dominant trait with incomplete penetration. It means that carriers do not need to be affected with HC; there is also possibility involving other genetic or environmental factors.

Individuals with one deleted allele (result N/P, negative/positive) have approximately 17-time higher risk of binocular cataract than the individuals without any deleted allele (result N/N). Heterozygous individuals (N/P) transfer the mutation to their offspring.

This test does not exclude existence of any other unknown mutation of HSF4 gene nor different gene responsible for hereditary cataract.

Method: SOP171-HC, fragment analysis, accredited method

Date of issue: 06.02.2021

Date of testing: 01.02.2021 - 06.02.2021

Approved by: Mgr. Martina Šafrová, Laboratory Manager



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

Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic

www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999



Report verification code is: 12AB-CD34-GENO-MIA0-EFGH. You can verify report online at www.genomia.cz

Without a written consent by the lab, the report must not be reproduced unless as a whole.

The result refers only to the sample as received. Genomia is not responsible for the accuracy of the information provided by the customer.