GENOMIA
genetic laboratory

Result certificate #012345

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: 08-12345
Date received: 25.11.2008
Sample type: blood

Information provided by the customer
Name: Lassie DEMO
Breed: ---
Tattoo number: 1392013
Microchip: 123 456 789 012 345
Reg. number: REGQ12345
Date of birth: 31.12.1909
Sex: female
Date of sampling: 25.11.2008
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: 30.11.2008
Date of testing: 25.11.2008 - 30.11.2008
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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