GENOMIA
genetic laboratory

Result certificate #044281:

Detection of g.85286582insC and g.85286582delC mutations in HSF4 gene causing hereditary cataract in several dog breeds by fragment analysis

Sample
Sample: 14-06610
Name: Mazie
Breed: Miniature Australian Shepherd
Date of birth: 1/15/2014
Sex: female
Date received: 18.03.2014
Sample type: buccal swab

Customer
Erin Litton
1194 Old grubby Road
24592 South Boston
United States

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: SOP25, accredited method

Report date: 25.03.2014
Responsible person: Mgr. Martina Šafrová, Laboratory Manager

Genomia is accredited according to ISO/IEC 17025:2005 under #1549.

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