

Result certificate #026318:

Sample

Sample: 12-29737
Name: KissMe Its All About Andy
Breed: Miniature American Shepherd
Reg. number: DN32398201
Date of birth: Aug 8, 2011
Sex: male
Date received: 07.02.2013
Sample type: buccal swab

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

Customer

Judy Challinger
PO Box 140
96092 Vina
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) are in higher risk of HC disease and they 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: 13.02.2013

Responsible person: Mgr. Martina Šafrová, Laboratory Manager

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Genomia s.r.o, Janáčkova 51, 32300 Plzeň, Czech Republic, VAT#: CZ25212991
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999

