GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.4176dup mutation
in ABCA4 gene causing Stargardt disease
in Labrador Retrievers

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)

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 c.4176dup mutation in ABCA4 gene causing Stargardt disease (STGD) in Labrador Retrievers was tested. Stargardt disease is an inherited degenerative disease of the retina causing visual impairment and even total blindness. It is characterized by deposition of lipofuscin and vitamin A metabolites in the retinal pigment epithelium and weakening of the blood vessels that nourish the retina. This leads to progressive degeneration of rods and cones. The symptoms are particularly evident in older dogs, which have dilated pupils and abnormal reflex responses to glare. Daytime vision and dim light vision are significantly impaired.

Mutation that causes STGD is inherited autosomally recessively which 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: SOPAgriseq_canine, ngs

Date of issue: 06.02.2021
Date of testing: 01.02.2021 - 06.02.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document
GENOMIA genetic laboratory

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