

**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, accredited method

Date of issue: 06.02.2021

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Approved by: Mgr. Martina Šafrová, Laboratory Manager



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