

## Result certificate #012345

Detection of c.719G>A mutation in GFAP gene causing Alexander 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)

## **Explanation**

Presence or absence of c.719G>A mutation in GFAP gene causing Alexander disease in Labrador Retrievers was tested. Alexander's disease, or fibrinoid leukodystrophy, is a progressive fatal neurodegenerative disorder in which myelin formation is impaired and protein aggregates accumulate in astrocytes. Clinical signs are usually seen in dogs as young as twelve months of age and initially include impaired coordination, head tilt, nystagmus (oscillatory eye movements) and intolerance to touch.

Mutation that causes Alexander disease is inherited as an autosomal dominant trait. This means that it only takes one copy of the mutated gene inherited from one parent to cause symptoms of the disease. Affected dogs carry genotypes N/P or P/P, healthy dogs carry genotype N/N.

Method: SOPAgriseq\_canine, ngs, accredited method

Date of issue: 06.02.2021

Date of testing: 01.02.2021 - 06.02.2021

Approved by: Mgr. Martina Šafrová, Laboratory Manager



Genomia is accredited in compliance with ISO/IEC 17025:2018 under #1549 Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999

