GENOMIA
genetic laboratory

Result certificate #012345

Detection of mutation c.1028_1032delGAGAA in RPGR gene causing XL-PRA in Siberian Husky and Samoyed

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: XmXm

Explanation

Presence or absence of mutation c.1028_1032delGAGAA in exon ORF15 of RPGR gene (retinitis pigmentosa GTP´s regulator) was tested. This mutation causes X-linked progressive retinal atrophy diseases in Siberian Husky and Samoyed breeds. The first symptoms appear by clinical examination in 6 months. Later, rods light receptors begin to appear irregularly damaged. Cones damage arises in final stage of XL-PRA disease. In age of 4 years, affected dogs are usually completely blind.

Females have XX chromosomes so they can have following XL-PRA genotypes:
XnXn – females with two normal X chromosomes = normal phenotype, a healthy female
XnXm – females with one normal X (Xn) and one mutant X (Xm) = a female carrier. Clinical disability of female carriers is individual, depending on the X chromosome inactivation.
XmXm – females with two mutated X chromosomes = an affected female

Males have XY chromosomes so they can have following XL-PRA genotypes:
XnY – normal phenotype, a healthy male
XmY – an affected male; he inherited mutated X chromosome from his mother

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

verified and certified document GENOMIA genetic laboratory

Genomia is accredited in compliance with ISO/IEC 17025:2018 under #1549
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