GENOMIA
genetic laboratory

Result certificate #067599:

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

Sample
Sample: 15-20216
Name: Aklaro Heart Breaker for Orleansnow
Breed: Samoyed
Microchip: 688 010 000 052 034
Reg. number: CMKU/S/2530/-14/14
Date of birth: 29.04.2014
Sex: male
Date received: 22.07.2015
Sample type: buccal swab
Sample certified by Vet/Tech or witness.

Customer
Marcela Luxová
Sportovní 545
56169 Králíky
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Result: Based on mutation examination genotype was determined Xn/Y

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: SOP24, accredited method

Report date: 30.07.2015
Responsible person: Mgr. Martina Šafrová, Laboratory Manager

Genomia is accredited according to ISO/IEC 17025:2005 under #1549.

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