GENOMIA
genetic laboratory

Result report #010580:

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

Sample
Sample: 11-12221
Name: AVRIL ALBA ORLEANSNOW
Breed: Samoyed
Reg. number: 1828
Microchip: -
Date of birth: 26.10.2006
Sex: female
Date received: 03.05.2011
Sample type: buccal swab
Sampling confirmed by veterinarian.

Customer
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Result: Based on mutation examination genotype was determined Xn/Xn

Explanation

Mutation 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: 12.05.2011
Responsible person: Mgr. Markéta Dajbychová, Veterinary Laboratory Manager

Dajbychová

