Genomia
genetická laboratoř

L 1549

Result report #002444

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

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Details of animal
Sample: 09-34811
Animal: Alfirin Xamba's Yoshi and Us
Breed: Samojed
Reg. number: 177
Microchip: 203098100145901
Year of birth: 09.05.2003
Sex: male
Date received: 15.12.2009
Sample type: buccal swab

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

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, unaccredited method
Sensitivity (probability of correct identification of the defective form of the gene in heterozygous or mutated homozygous) is higher than 99%. Specificity (probability of correct identification of the normal form of the gene in a normal homozygous or heterozygous) is higher than 99%.

Report date: 21.12.2009
Responsible person: Mgr. Martina Šafrová, Laboratory Manager

