GENOMIA
genetic laboratory

Result certificate #026316:

Detection of c.227_230delATAG mutation in the MDR1 gene causing drug sensitivity in dogs by fragmentation analysis

Sample
Sample: 12-29737
Name: KissMe Its All About Andy
Breed: Miniature American Shepherd
Reg. number: DN32398201
Date of birth: Aug 8, 2011
Sex: male
Date received: 07.02.2013
Sample type: buccal swab

Customer
Judy Challinger
PO Box 140
96092 Vina
United States

Result: Mutation was not detected (N/N)

Explanation

Presence or absence of AF045016.1: c.227_230delATAG mutation in MDR1 gene was tested. This mutation causes a frame shift and formation of a stop codon during P-glycoprotein synthesis. P-glykoprotein, an ATP-dependent transporter of various substrates, is contained in cells lining the blood vessels in the brain. In P-glycoprotein defective animals, administering of ivermectin or similar drug can lead to elevated levels of drug in the CNS, resulting in potentially lethal neurotoxic reaction. These drugs include, but are not limited to: Acepromazine, Butorphanol, Doramectin, Doxorubicin, Ivermectin, Loperamide, Milbemycin, Moxidectin, Selamectin, Vinblastine, Vincristine.

Mutation that causes MDR1 related drug hypersensitivity is inherited as an autosomal recessive trait. That means the defect affects dogs with P/P (positive / positive) genotype only. The dogs with N/P (negative / positive) genotype are considered carriers of the deletion (heterozygotes). The dogs with N/N genotype are not endangered with MDR1 related drug hypersenzitivity. Multiple drug hypersensitivity based on MDR1 gene mutation was proved in following breeds: Rough Collie, Smooth Collie, Shetland Sheepdog, Australian Sheepdog, White Swiss Shepherd Dog, Wäller, Bobtail, Border Collie and others.

Method: SOP04, accredited 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: 13.02.2013
Responsible person: Mgr. Martina Šafrová, Laboratory Manager

Genomia is accredited according to ISO 17025 under #1549.
Genomia s.r.o, Janáčkova 51, 32300 Plzeň, Czech Republic, VAT#: CZ25212991
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999

ilac-MRA L 1549

