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Result certificate #080589

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

Sample
Sample: 16-18528
Name: Foula´s Mystery Of The Dark
Breed: Shetland Sheepdog
Microchip: 208 250 000 080 603
Reg. number: DK11254/2016
Date of birth: 24.06.2016
Sex: female
Date received: 13.07.2016
Sample type: buccal swab

Customer
Pia Skøtt Hansen
Rejnstrupvej 2a
4250 Fuglebjerg
Denmark

Result: Mutation was detected in heterozygous status (N/P)

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: SOP171-MDR1, fragment analysis, 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: 20.07.2016
Responsible person: Mgr. Barbora Bláhová, Analyst

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

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