

Result certificate #130112

Detection of mutation c.295_298delAGAT in ABCB1 gene causing drug sensitivity in dogs

Sample

Sample: 19-13527

Name: Adina Dvůr Bažantnice

Breed: Collie Smooth

Microchip: 972 273 000 004 700 Reg. number: CMKU/CK/1079/16/18

Date of birth: 24.04.2016

Sex: female

Date received: 16.05.2019 Sample type: blood

The identity of the animal has been checked by

MVDr. Michaela Oravská

Customer

Ing. Lucie Glaserová Doubek 109 25101 Doubek Czech Republic

Result: Mutation was not detected (N/N)

Explanation

It has been studied the presence and absence of mutation c.295_298delAGAT in ABCB1 gene leading to shifting of the reading frame and creation of premature stop codon during the synthesis of P-glycoprotein. P-glycoprotein is a membrane drug transporter and a very important component of the blood brain barrier that prevents entry of many potentially toxic compounds into the central nervous system. The dysfunction of P-glycoprotein in dogs can result in potentially fatal neurotoxic reaction, especially following the administration of ivermectin, acepromazin, butorphanol, doramectin, doxorubicin, loperamid, milbemycin, moxidectin, selamectin, vinblastin and vincristin.

The sensitivity to drugs develops in dogs with mutation in both copies of MDR1 gene (P/P). Some dogs that are heterozygotes (N/P) have shown adverse reaction after administration of some drugs. The specific cause of this variation is not known so far – other gene mutations, general health conditions and dosage.

It is not possible to exclude existence of other mutations of ABCB1 gene in various breeds (in Bordier collies, another two mutations have been found). Compound heterozygotes that carry two distinct mutations of ABCB1 gene may occur, where each mutation was inherited from one of the parents. The compound heterozygotes also have defective P-glycoprotein function.

The defect occurs in Collies, Longhaired Whippets, Australian Shepherds, Miniature Australian Shepherds, McNab Shepherd dogs, Silken windhounds, English sheepdogs, Shelties, German shepherd dogs, Bobtails, Border Collies and herding breed cross.

Method: SOP171-MDR1, fragment analysis, accredited method

Report date: 20.05.2019

Responsible person: Mgr. Martina Šafrová, Laboratory Manager

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

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