

Result certificate #289107

Detection of mutation c.228_231del in ABCB1 gene causing drug sensitivity in dogs

Customer: Michal Glaser, Doubek 109, 251 01 Doubek, Czech Republic

Sample:

Sample: 23-15207 Date received: 05.06.2023 Sample type: buccal swab

Information provided by the customer

Name: Emilka z Modrého království

Breed: Collie Smooth

Microchip: 900 085 001 644 611 Reg. number: CMKU/CK/2155/23

Date of birth: 19.4.2023

Sex: female

Date of sampling: 01.06.2023

The identity of the animal has been checked by MVDr. Pavel

Mádr, CSc. KVL4735

Result: Mutation was not detected (N/N)

Explanation

It has been studied the presence and absence of mutation c.228_231del in ABCB1 gene leading to defect 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: SOPAgriseq_canine, ngs, accredited method

Date of issue: 26.02.2025

Date of testing: 12.02.2025 - 26.02.2025

Approved by: Mgr. Martina Šafrová, Laboratory Manager

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