GENOMIA
genetic laboratory

Result certificate #012345

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

Sample
Sample: 08-12345
Name: Lassie DEMO
Breed: ---
Tattoo number: 1392013
Microchip: 123 456 789 012 345
Reg. number: REGQ12345
Date of birth: 31.12.1909
Sex: female
Date received: 25.11.2008
Sample type: blood
The identity of the animal has been checked.

Customer
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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: SOP175-MDR1, real-time PCR-ASA

Report date: 25.11.2008
Responsible person: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document
GENOMIA genetic laboratory

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