

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

**Customer:** Jan Novák, Dlouhá 1, 30000 Plzeň, Czech Republic

**Sample:**

Sample: 21-12345

Date received: 01.02.2021

Sample type: blood

Information provided by the customer

**Name:** Lassie DEMO

**Breed:** Plemeno

Tattoo number: 1392013

Microchip: 123 456 789 012 345

Reg. number: REGQ12345

Date of birth: 1.1.2020

Sex: female

Date of sampling: 01.02.2021

The identity of the animal has been checked.

**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: 06.02.2021

Date of testing: 01.02.2021 - 06.02.2021

Approved by: Mgr. Martina Šafrová, Laboratory Manager



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Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic

www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999



Report verification code is: 12AB-CD34-GENO-MIA0-EFGH. You can verify report online at [www.genomia.cz](http://www.genomia.cz)

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