

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

**Sample**

Sample: 18-00234  
Name: Ululallaluna Lamu'  
Breed: Shetland Sheepdog  
Microchip: 380 260 100 501 822  
Reg. number: DK14377/2017  
Date of birth: 14.10.2016  
Sex: female  
Date received: 19.01.2018  
Sample type: buccal swab

**Customer**

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Result: Mutation was not detected (N/N)

**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 hypersensitivity. 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

Report date: 29.01.2018

Responsible person: Mgr. Markéta Dajbychová, Deputy Laboratory Manager

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

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