GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.1640T>C mutation in RYR1 gene causing malignant hypertermia after exposure to a chemical trigger in different dog breeds

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

Presence or absence of c.1640T>C mutation in RYR1 gene causing malignant hypertermia (MH), which develops after exposure to a chemical trigger, was tested. MH is a severe complication during the general anaesthesia that can be even fatal. MH is a pharmacogenetic disease of skeletal muscles characterized by hypercapnia, tachycardia and hyperthermia, which occur in response to some chemical drugs, in this case anaesthetics. The affected dogs have no clinical symptoms unless they are exposed to these drugs inducing this condition.

Mutation that causes MH is inherited as an autosomal dominant trait. Only one copy of the mutated gene is sufficient for development of clinical signs (result N/P (negative / positive) or P/P (positive / positive)). With regard to the fact that affected animals are without clinical signs, provided they are not exposed to the trigger substance, some animals can live their whole lives without diagnosing the disposition for MH. In case of affected animal the risk of transfer to the offsprings is 50%.

Method: SOP188-MPS-canine, MPS, accredited method

Date of issue: 06.02.2021
Date of testing: 01.02.2021 - 06.02.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document GENOMIA genetic laboratory

SAMPLE

Genomia is accredited in compliance with ISO/IEC 17025:2018 under #1549
Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999

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