

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

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

Presence or absence of c.1640T>C mutation in RYR1 gene causing malignant hyperthermia (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: 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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