

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

**Customer:** Renée Kryšpín Hájková, Prostřední Staré Buky 94, 54101 Staré Buky, Czech Republic

**Sample:**

Sample: 23-13708

Date received: 18.05.2023

Sample type: blood

Information provided by the customer

**Name:** El Mariachi Bagheera Bulls

**Breed:** Staffordshire Bull Terrier

Microchip: 203 098 100 528 713

Reg. number: CMKU/SBT/17404/22

Date of birth: 18.04.2022

Sex: male

Date of sampling: 17.05.2023

The identity of the animal has been checked by MVDr. Jakub

Sova, KVL 7127

**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

Date of issue: 12.04.2024

Date of testing: 05.04.2024 - 12.04.2024

Approved by: Ing. Irena Rusková, Analyst



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