

Result certificate #027192:

Sample

Sample: 12-32326
Name: Beau Brit Od bijelih andjela
Breed: White Swiss Shepherd Dog
Reg. number: HR 10016
Microchip: 191100000180034
Date of birth: 08.08.2008.
Sex: male
Date received: 07.12.2012
Sample type: buccal swab

Detection of c.1640T>C mutation in RYR1 gene causing malignant hyperthermia in different dog breeds by DNA sequencing

Customer

Gordana Horvat
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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. 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 life without diagnosing the disposition for MH. In case of affected animal the risk of transfer to the offsprings is 50%.

Method: SOP107

Report date: 11.12.2012

Responsible person: Mgr. Markéta Dajbychová, Deputy Laboratory Manager



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