

**Result certificate #050776:**

**Sample**

Sample: 14-23808  
Name: Darleyfalls All Zet To Do It  
Breed: Border Collie  
Microchip: 98100004416756  
Reg. number: DK00418/2014  
Date of birth: 20.01.2014  
Sex: female  
Date received: 03.09.2014  
Sample type: buccal swab

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

**Customer**

Jane Elene Christensen  
Ibsensvej 59  
2630 Taastrup  
Denmark

**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: SOP107

Report date: 11.09.2014

Responsible person: Mgr. Martina Šafrová, Laboratory Manager



Genomia s.r.o, Janáčkova 51, 32300 Plzeň, Czech Republic  
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999