

Result certificate #012345

Detection of c.1660_1661insG mutation in ABCB4 gene causing GBM in dogs

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.1660_1661insG mutation in ABCB4 gene causing Gallbladder Mucoceles (GBM) in American and English Cocker Spaniels, Cairn Terriers, Shetland Sheepdogs and Pomeranians was tested. GBM is characterized by an excessive production of bile with a thick gelatinous consistency. The accumulation of this pathologically altered bile in the gallbladder results in its rupture and spillage of bile into the abdominal cavity. The resulting inflammation of the abdominal cavity is very painful for the patient and can be fatal. Common non-specific clinical signs include vomiting, lethargy, abdominal pain, anorexia, jaundice, rapid breathing, thirst and frequent urination, fever, diarrhoea, and bloating.

The inheritance of the mutation causing GBM is still unknown. The mutation was thought to be dominant with incomplete penetrance, nowadays a multifactorial mode of inheritance.

Method: SOPAgriseq_canine, ngs

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Date of testing: 01.02.2021 - 06.02.2021

Approved by: Mgr. Martina Šafrová, Laboratory Manager



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