

**Detection of c.5G>A mutation in TUBB1
gene causing macrothrombocytopenia in
Cairn and Norfolk Terriers****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)**Legend:** N/N = wild-type genotype. N/P = carrier of the mutation. P/P = mutated genotype (individual will be most probably affected with the disease). (N = negative, P = positive)**Explanation**

Presence or absence of c.5G>A mutation in TUBB1 gene causing macrothrombocytopenia (MTC) in Cairn and Norfolk Terriers was tested. Macrothrombocytopenia is a disease of impaired blood clotting, manifested by severe bleeding conditions.

Mutation that causes macrothrombocytopenia is inherited autosomally recessively which means that the disease develops only in those dogs who inherit mutated allele from both parents; disease affects dogs with P/P genotype only. The dogs with N/P genotype are considered carriers of the disease (heterozygotes). In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N, 25 % P/P and 50 % N/P.

Method: SOPAgriseq_canine, ngs**Date of issue:** 06.02.2021**Date of testing:** 01.02.2021 - 06.02.2021**Approved by:** Mgr. Martina Šafrová, Laboratory Manager

Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999

Report verification code is: 12AB-CD34-GENO-MIA0-EFGH. You can verify report online at www.genomia.cz

Without a written consent by the lab, the report must not be reproduced unless as a whole.

The result refers only to the sample as received. Genomia is not responsible for the accuracy of the information provided by the customer.