GENOMIA
genetic laboratory

# Result certificate #012345

Detection of c.745G>A mutation in TUBB1 gene causing macrothrombocytopenia in several dog breeds

**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.745G>A mutation in TUBB1 gene causing macrothrombocytopenia (MTC) in Cavalier King Charles Spaniels, King Charles Spaniels, Chihuahuas, Russell Terriers, Labrador Retrievers, Maltese Dogs, Poodles and Shih Tzu 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: SOP188-MPS-canine, MPS, accredited method

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

verified and certified document GENOMIA genetic laboratory

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

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