

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 detected in heterozygous status (N/P)

Explanation

Presence or absence of c.110A>C mutation in PLP1 gene causing Shaking puppy syndrom (SPS, Tremor) in Springer Spaniels was tested. The disease characterized by uncontrollable shaking of the puppies' entire body shortly after birth and often leads to premature death.

The mutation causing SPS is located on the X chromosome. Males have one X and one Y chromosome and therefore can only be either healthy (X_n/Y) or affected (X_m/Y). Females have two X chromosomes and can therefore be healthy (X_n/X_n), carriers (X_n/X_m), or affected (X_m/X_m). Carrier females do not show clinical signs but are capable of passing the mutated allele to their offspring.

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



Genomia is accredited in compliance with ISO/IEC 17025:2018 under #1549

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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