

Result certificate #012345

Detection of c.5855_5862del SPTBN2 in the SPTBN2 gene causing NCCD in Beagles

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.5855_5862del mutation in the SPTBN2 gene causing NCCD (Neonatal cerebellar cortical degeneration) in Beagles was tested. Cerebellar abiotrophy (NCCD) is a neurodegenerative disease characterised with a number of clinical signs causing the dysfunction of the cerebellum, such as dissymmetrical ataxia, wide-based stance, loss of balance and body tremor. In Beagles, the neurological signs are first noticed at around three weeks of age.

Mutation that causes NCCD in Beagles is inherited as an autosomal recessive trait. That means the disease affects dogs with P/P genotype only. The dogs with N/P genotype are considered carriers of the disease (heterozygotes), they are healthy but they can transmit the mutation on their offspring. In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N, 50 % N/P and 25 % P/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



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

