GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.2653+1G>A mutation
in SNX14 gene causing NCCD
in Hungarian Vizslas

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.2653+1G>A mutation in SNX14 gene causing Neonatal Cerebellar Cortical Degeneration (NCCD), or cerebellar abiotrophy, in Hungarian Vizslas was tested. NCCD is a neurodegenerative disease. The characteristic feature of the disease is cerebellar ataxia, that begins to develop at around three months of age and the progression is relatively fast. The affected dogs suffer from general lack of coordination, intention tremor and head tremor, insufficient response to threat and symptoms of dysfunction of balance organ incl. nystagmus.

Mutation that causes NCCD in Hungarian Vizslas 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, 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

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