GENOMIA
genetic laboratory

Result certificate #012345

Detection of deletion in NPHP4 gene causing CORD2 in dogs

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 180bp deletion in exon / intron 5 of NPHP4 gene (nephronophthisis 4, also known as nephroretinin) was tested. This deletion causes CORD2 disease (cone-rod dystrophy type 2), specific type of progressive retinal degeneration, in standard wire-haired dachshund. Mutated nephroretinin proteine preserves a binding domain interacting with nephrocystin-1 in kidneys, but lacks a domain interacting with RPGRIP1 (retinitis pigmentosa GTPase regulator interacting protein) in retina. Therefore, due to 180 bp deletion in NPHN4 gene, standard wire-haired dachshunds suffer only from retina disorder not from kidney disease. The first clinical symptoms of CORD2-PRA in standard wire-haired dachshunds appear between 10 months and 3 years. Total retinal atrophy occurs around the 6th year of life, when CORD2-PRA is manifested daily blindness.

Deletion in NPHP4 gene 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). In offspring of two heterozygous animals following genotype distribution can be expected: 25% N/N, 50% N/P and 25% P/P.

Method: SOPAgriseq_canine, ngs
Sensitivity (probability of correct identification of the defective form of the gene in heterozygous or mutated homozygous) is higher than 99%. Specificity (probability of correct identification of the normal form of the gene in a normal homozygous or heterozygous) is higher than 99%.

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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