GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.2262_c.2263insA mutation in CCDC66 gene causing EO PRA in Portuguese Water 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.

SAMPLE

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.2262_c.2263insA mutation in CCDC66 gene causing early onset variant (EO PRA) in Portuguese Water Dogs was tested. Clinical signs of EO PRA begin to appear as early as between 2 and 3 years of age. The disease manifests itself with progressively worsening vision, both in dim light and in good light conditions. Tracking of moving objects is also difficult. In affected dogs, ophthalmoscopic changes such as tapetal hyperreflectivity, diffuse vascular attenuation, optic disc pallor, and multifocal depigmentation in the non-tapetal fundus are gradually observed.

Mutation that causes EO PRA 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

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