

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.

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: SOP188-MPS-canine, MPS, accredited method

Date of issue: 06.02.2021

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Approved by: Mgr. Martina Šafrová, Laboratory Manager



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