

Result certificate #012345

Detection of c.784G>A mutation in CNGB3 gene causing Achromatopsia 3 in German Shorthaired Pointers

Customer: Jan Novák, Dlouhá 1, 30000 Plzeň, Czech Republic Sample: Sample: 21-12345 Date received: 01.02.2021

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.784G>A mutation in CNGB3 gene causing Achromatopsia 3 in German Shorthaired Pointers was tested. In achromatopsia, the cones of the retina, which are responsible for colour vision, degenerate.

Mutation that causes Achromatopsia 3 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, 25 % P/P and 50 % N/P.

Method: SOPAgriseq_canine, ngs

Date of issue: 06.02.2021 Date of testing: 01.02.2021 - 06.02.2021 Approved by: Mgr. Martina Šafrová, Laboratory Manager



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