GENOMIA
genetic laboratory

Result certificate #012345

Detection of deletion in CNGB3 gene causing Cone Degeneration in several dog breeds

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 deletion in CNGB3 gene causing congenital achromatopsia, so-called day blindness, in Alaskan Malamute, Australian Shepherd, German Short-haired Pointing Dog, Miniature Australian Shepherd, Samoyed and Siberian Husky was tested. Achromatopsia is a rare vision disorder leading to retinal cone degeneration, therefore the disease is also called CD. The affected dogs are not able to localize the approaching objects at daylight, if they do not hear or smell them. This disease can already be observed in puppies from 8 to 12 weeks of age.

Mutation that causes CD 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), they are healthy but they can transmit the mutation on their offspring. Dogs with N/N genotype are without risk of CD. In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N (healthy non-carriers), 50 % N/P (healthy carriers) and 25 % P/P (affected).

Method: SOP176-CD, ASA-PCR

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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