

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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