GENOMIA
genetic laboratory

Result certificate #012345

Detection of g.30852988_30902901del mutation in CLN8 gene causing NCL8 in Alpine Dachsbrackes

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 g.30852988_30902901del mutation in CLN8 gene causing NCL8 (Neuronal ceroid lipofuscinosis of type 8) in Alpine Dachsbrackes was tested. The symptoms of this disease seem to be very variable. The NCL usually includes neurological symptoms, such as disorientation, anxiety and aggressiveness, seizures and food intake difficulty. Sudden loss of vision is a common sign. The degree of neurodegeneration increases with the age and psychical abnormalities and spasms develop in each dog. Changes in gait – stumbling gait and limb stiffness can be observed as well.

Mutation that causes NCL8 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 (healthy non-carriers), 25 % P/P (affected), and 50 % N/P (healthy carriers).

Method: SOP176-NCL8, ASA-PCR

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

Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999