GENOMIA
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Result certificate #111181

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

Sample
Sample: 18-40732
Name: Míša Vltavské Údolí
Breed: Alpine Dachsbracke
Tattoo number: 4973
Microchip: 941000019592909
Reg. number: ABJ/4973
Date of birth: 21.05.2014
Sex: female
Date received: 24.05.2018
Sample type: buccal swab
The identity of the animal has been checked by Jolana Michalcová, ČKCHABJ

Customer
Hanzlík Libor Ing.
Hartmanice 76
34201 Sušice
Czech Republic

Result: Mutation was detected in heterozygous status (N/P)

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

Report date: 29.05.2018
Responsible person: Mgr. Martina Šafrová, Laboratory Manager

Genomia s.r.o, Janáčkova 51, 32300 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999