GENOMIA
genetic laboratory

Result certificate #251098

Detection of expanded 12-mer repeat in NHLRC1 gene causing Lafora epilepsy in several dog breeds

Customer: Iain Cruickshank, Mosside Croft, Culsalmond, AB526UE Insch, United Kingdom
Sample:
Sample: 23-36305
Date received: 01.02.2024
Sample type: buccal swab

Information provided by the customer
Name: Burnvale Dazzling Diamond At Culsalmond
Breed: Basset Hound
Microchip: 992 000 000 372 411
Reg. number: AY11464902
Date of birth: 22 August 2021
Sex: female
Date of sampling: 20.01.2024

Result: N_3/N_3

Result codes:

- N_2/N_2, N_3/N_3, N_2/N_3 = negative genotype, dog carrying two or three 12-mers.
- N_2/P, N_3/P = carrier of Lafora epilepsy.
- P/P = dog affected by Lafora epilepsy.

Explanation

Presence or absence of expanded 12-mer repeat in NHLRC1 gene causing Lafora epilepsy in Beagles, Miniature Wirehaired Dachshunds, Basset Hound and Chihuahua was tested. The occurrence of this mutation in other breeds cannot be excluded.

Generally, the clinical signs appear at 5-6 years of age or later. Epileptic seizures include mainly sudden involuntary muscle jerking. Over time seizures are accompanied by other neurological symptoms such as ataxia, twinkling, blindness or dementia. This form of epilepsy is incurable and fatal.

Expanded 12-mer repeat in NHLRC1 gene causing Lafora epilepsy is inherited as an autosomal recessive trait. That means the disease affects dogs with P/P genotype only. Carriers are healthy without symptoms of epilepsy. In offspring of two carriers following genotype distribution can be expected: 25 % N/N, 25 % P/P and 50 % N/P.

Method: SOP187-Lafora, ASA-PCR of DNA modified template, accredited method

Date of issue: 09.02.2024
Date of testing: 01.02.2024 - 09.02.2024
Approved by: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document GENOMIA genetic laboratory

Genomia is accredited in compliance with ISO/IEC 17025:2018 under #1549
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