GENOMIA
genetic laboratory

Result certificate #102106

Detection of expanded 12-mer repeat in NHLRC1 gene causing Lafora epilepsy in Beagles and Miniature Wirehaired Dachshunds

Sample
Sample: 17-31317
Name: Fanta's brand rule number one
Breed: Beagle
Microchip: 643 095 300 010 030
Reg. number: lof 6 bea 94123/14208
Date of birth: 15.12.2013
Sex: male
Date received: 19.01.2018
Sample type: buccal swab

Customer
Coralie Finck
7 Rue des Sources
68210 Balschwiller
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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 and Miniature Wirehaired Dachshunds was tested. 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: SOPSOP187-Lafora, ASA-PCR of DNA modified template

Report date: 31.01.2018
Responsible person: Mgr. Martina Šafrová, Laboratory Manager

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