

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

Sample

Sample: 18-06033
Name: Primrose Everdeen
Breed: Beagle
Tattoo number: N/A
Microchip: 380 260 042 669 553
Reg. number: LO16196072
Date of birth: 29.09.2016
Sex: female
Date received: 14.03.2018
Sample type: buccal swab

Customer

Francesco Mennone

Result: N₃/P

Result codes:

- N₂/N₂, N₃/N₃, N₂/N₃ = negative genotype, dog carrying two or three 12-mers.
- N₂/P, N₃/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: SOP187-Lafora, ASA-PCR of DNA modified template

Report date: 20.03.2018

Responsible person: Mgr. Martina Šafrová, Laboratory Manager



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