

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

Sample

Sample: 18-07008
Name: Zorigi Believe in love Beauty Brada
Breed: Beagle
Microchip: 756 098 100 755 417
Reg. number: 743839
Date of birth: 30.05.2016
Sex: female
Date received: 15.03.2018
Sample type: blood
The identity of the animal has been checked by Dr.
med. vet. Fabienne Künzli

Customer

Andrea Wanner
Stegbachstrasse 8
4653 Obergösgen
Switzerland

Result: N₃/N₃

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



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