

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

**Sample**

Sample: 18-06032  
Name: Lucky Girl  
Breed: Beagle  
Microchip: 380 260 042 623 758  
Reg. number: LO1659228  
Date of birth: 04.02.2016  
Sex: female  
Date received: 14.03.2018  
Sample type: buccal swab

**Customer**

Francesco Mennone

**Result: N<sub>3</sub>/N<sub>3</sub>**

**Result codes:**

- N<sub>2</sub>/N<sub>2</sub>, N<sub>3</sub>/N<sub>3</sub>, N<sub>2</sub>/N<sub>3</sub> = negative genotype, dog carrying two or three 12-mers.
- N<sub>2</sub>/P, N<sub>3</sub>/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