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

Result: \( N_2/N_3 \)

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, twinking, 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: 25.11.2008

Responsible person: Mgr. Martina Šafrová, Laboratory Manager

Genomia s.r.o., Republikánská 6, 31200 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999

Report verification code is: 12AB-CD34-GENO-MIA0-EFGH. You can verify report online at www.genomia.cz
Without a written consent by the lab, the report must not be reproduced unless as a whole. This result is valid only for the submitted sample.