Detection of mutation c.379_392del14 FUCA1 gene causing fucosidosis in English Springer Spaniel breed

Result certificate #012345

Sample
Sample: 08-12345
Name: Lassie DEMO
Breed: ---
Tattoo number: 1392013
Microchip: 123 456 789 012 345
Reg. number: REGQ12345
Date of birth: 31.12.1909
Sex: female
Date received: 25.11.2008
Sample type: blood
The identity of the animal has been checked.

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Result: Mutation was not detected (N/N)

Legend: N/N = wild-type genotype. N/P = carrier of the mutation. P/P = mutated genotype (individual will be most probably affected with the disease). (N = negative, P = positive)

Explanation
Presence or absence of mutation c.379_392del14 FUCA1 gene causing fucosidosis in English Springer Spaniel breed was tested. This mutation causes a frame shift and formation of stop codon during alpha-L-fucosidase synthesis. Fucosidosis in ESS manifests mainly as neurological disorder. Affected individuals suffer from motoric and mental symptoms (blindness, deafness, motoric and behavioural disorders, vomiting, deglutition problems). First symptoms can be observed between 18 months and four years of age of the dog.

Mutation c.379_392del14 FUCA1 gene is inherited as an autosomal recessive trait. That means the disease affects dogs with P/P genotype only. The dogs with N/P genotype are considered carriers of the disease (heterozygotes). In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N, 25 % P/P and 50 % N/P.

Method: SOP171-FUCO, fragment analysis
Report date: 25.11.2008
Responsible person: Mgr. Martina Šafrová, Laboratory Manager

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