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Result certificate #012345

Detection of c.3149_3150insC mutation in the C17H2orf71 gene causing RCD4 in several dog breeds

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.

Customer
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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 c.3149_3150insC mutation in the C17H2orf71 gene causing rode-cone dysplasia (RCD4) was tested. The mutation is related with RCD4 in breeds: Australian Cattle Dog, English Setter, Gordon Setter, Irish Red & White Setter, Irish Setter, Polish Lowland Sheepdog, Small Munsterlander, Standard Poodle and Tibetan Terrier. RCD4 is a form of progressive retinal atrophy (PRA) characterized by the degeneration of the photoreceptors in the retina. It results in vision loss and eventually complete blindness. The average age of RCD4 diagnosis is 10 years.

Mutation that causes RCD4 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), they are healthy but they can transmit the mutation on their offspring. In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N, 50 % N/P and 25 % P/P.

In individuals with N/N and N/P result might develop any different form of PRA, due to mutations that were not detected by this test.

Method: SOP171-RCD4, fragment analysis

Report date: 25.11.2008
Responsible person: Mgr. Barbora Bláhová, Analyst

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