

Detection of c.5G>A mutation in PRCD gene causing PRA-prcd in many 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.5G>A mutation in PRCD gene causing PRA-prcd (Progressive Retinal Atrophy) was tested. Disease causes degeneration of retinal cells. Firstly, rods are affected and night blindness develops in the animal. Later, cones degenerate. That results in complete blindness of the animal. The age of onset of disease varies, but, generally, it cannot be recognized before the adulthood of the animal.

Mutation that causes PRA-prcd 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.

It is necessary to be aware that not all retinal diseases belong in progressive retina atrophy group of disorders and not all are a variant of PRA-prcd. In many breeds, the cause of PRA inheritance has not been still clarified. It is also possible that several mutations can be responsible for retinal atrophy in one breed. Therefore, we recommend that an eye examination by a veterinary ophthalmologist is performed every year.

The analysis was carried out by the PrcdTest Laboratory, branch office of Genomia.

Method: SOP122, real-time PCR-ASA

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Responsible person: Mgr. Martina Šafrová, Laboratory Manager



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