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Result certificate #231887

Detection of insertion in exon 4 of RD3 gene causing RCD2 disease in collies

Customer: Michal Glaser, Doubek 109, 25101 Doubek, Czech Republic
Sample:
Sample: 23-15207
Date received: 05.06.2023
Sample type: buccal swab

Information provided by the customer
Name: Emilka z Modrého království
Breed: Collie Smooth
Microchip: 900 085 001 644 611
Reg. number: CMKU/CK/2155/23
Date of birth: 19.4.2023
Sex: female
Date of sampling: 01.06.2023
The identity of the animal has been checked by MVDr. Pavel Mádr, CSc. KVL4735

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 22 bp insertion in exon 4 of RD3 gene causing RCD2 disease (rode-cone dysplasia 2) in rough and smooth collies was tested. RCD2 is manifested by retina degeneration that leads to early onset night blindness in puppies about 6 weeks of age. RCD2 affected dogs are usually totally blind in one year of age.

Insertion in RD3 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-RCD2, fragment analysis

Date of issue: 16.06.2023
Date of testing: 05.06.2023 - 16.06.2023
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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