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Result certificate #109738

Detection of c.460_463delAAGA mutation in the exon 5 of RPE65 gene causing CSNB disease in Briard breed

Sample
Sample: 18-11340
Name: Finally the Best Velmond
Breed: Briard (Berger de Brie)
Microchip: 967 000 009 853 165
Reg. number: CMKU/BRI/6662/16
Date of birth: 27.11.2016
Sex: male
Date received: 09.05.2018
Sample type: blood
The identity of the animal has been checked by MVDr. Zbyněk Kratochvíl

Customer
Táňa Holešová
U studánky 449/5
17000 Praha 7
Czech Republic

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.460_463delAAGA mutation in the exon 5 of RPE65 (retina pigment epitelium-specific protein, 65 kDa) causing CSNB (Congenital Stationary Night Blindness) in Briard breed was tested. CSNB disease manifests as slow retina degeneration starting in the age of about six months. During the animal´s life CSNB disease can develop to total blindness.

Mutation that causes CSNB in Briards 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-CSNB, fragment analysis
Sensitivity (probability of correct identification of the defective form of the gene in heterozygous or mutated homozygous) is higher than 99%. Specificity (probability of correct identification of the normal form of the gene in a normal homozygous or heterozygous) is higher than 99%.

Report date: 16.05.2018
Responsible person: Ing. Irena Rusková, Analyst

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