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Result certificate #183325

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

Customer: Jana Kočová, Pod Chvojinkou 230, 26703 Nový Jáchymov, Czech Republic
Sample:
Sample: 21-20040
Date received: 29.07.2021
Sample type: buccal swab

Information provided by the customer
Name: Jeremy Heritage
Breed: Briard (Berger de Brie)
Microchip: 945 000 006 141 097
Reg. number: CMKU/BRI/7088/19
Date of birth: 05.06.2019
Sex: male
Date of sampling: 28.07.2021
The identity of the animal has been checked by MVDr. Lucie Musilová, KVL 4695

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%.

Date of issue: 10.08.2021
Date of testing: 29.07.2021 - 10.08.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999