GENOMIA
genetic laboratory

Result certificate #032176:

Detection of 487-490del AAGA mutation in the exon 5 of RPE65 gene causing CSNB disease in Briard breed by fragmentation analysis

Sample
Sample: 13-08614
Name: Smooth Breeze Catherine Fabienne
Breed: Berger de Brie
Microchip: 981 098 102 650 201
Date of birth: 12.04.2011
Sex: female
Date received: 28.03.2013
Sample type: blood
Sample certified by Vet/Tech or witness.

Customer
Alena Bendová
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Result: Mutation was not detected (N/N)

Explanation

Four base pair deletion in exon 5 of canine RPE65 gene (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.

CSNB 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 (healthy non-carriers), 25 % P/P (affected), and 50 % N/P (healthy carriers).

Method: SOP05
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: 02.04.2013
Responsible person: Mgr. Martina Šafrová, Laboratory Manager

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