GENOMIA
genetic laboratory

Result certificate #010874:

Detection of c.73C>T mutation in VMD2 gene causing CMR1 disease in dogs by PCR-RFLP

Sample
Sample: 11-13712
Name: Beau Brit Od bijelih andjela
Breed: White Swiss Shepherd Dog
Reg. number: HR10016
Microchip: ---
Date of birth: 08.08.2008.
Sex: male
Date received: 21.06.2011
Sample type: buccal swab

Customer
Gordana Horvat
Bukovačka cesta 293
10000 Zagreb
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Result: Mutation was not detected (N/N)

Explanation

Mutation c.73C>T in VMD2 gene was tested. This mutation causes CMR1 disease (Canine Multifocal Retinopathy type 1) in Great Pyrenees, English Mastiffs, Bullmastiffs and related breeds. The mutation forms a premature stopkodon (R25X) in canine VMD2 gene; the gene is responsible for right forming of pigment epithelium in retina. Clinically, rose-grey colored lesions are remarkable in retina. CMR disease usually arises before 4th month of age in an affected puppy. Total blindness usually comes in higher age.

Mutation c.73C>T in VMD2 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 (healthy non-carriers), 25 % P/P (affected), and 50 % N/P (healthy carriers).

Method: SOP40

Report date: 25.06.2011
Responsible person: Mgr. Markéta Dajbychová, Deputy Laboratory Manager

Dajbychová

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