GENOMIA
genetic laboratory

Result certificate #012345

Detection of gene variants associated with predisposition to Dermatomyositis in Collies

Customer: Jan Novák, Dlouhá 1, 30000 Plzeň, Czech Republic
Sample:
Sample: 21-12345
Date received: 01.02.2021
Sample type: blood

Information provided by the customer
Name: Lassie DEMO
Breed: Plemeno
Tattoo number: 1392013
Microchip: 123 456 789 012 345
Reg. number: REGQ12345
Date of birth: 1.1.2020
Sex: female
Date of sampling: 01.02.2021
The identity of the animal has been checked.

Result: PREVIEW RESULT LINE

Explanation

Presence or absence of variants of genes PAN2 (g.627760G>A = locus A), MAP3K7CL (c.383_392ACTCCACAAA>GACT = locus B) a DRB1 (DLA-DRB1 002:01 = locus C) associated with predisposition to Dermatomyositis in short-haired and long-haired collies, bearded collies, border collies, and shelties were tested. Dermatomyositis is a multifactorial autoimmune disease. The manifestation of DMS can be caused by both genetic and external factors. In affected individuals, the disease causes inflammatory vasculopathy of the skin and muscles. The immune system attacks the endothelium of small blood vessels, where inflammation subsequently breaks out and a wide range of processes leading to ischemia occur. This results in characteristic atrophy and necrosis of the epidermis and hair follicles and degeneration of muscle fibres.

The combination of these three loci resulted in haplotypes, whose variable combinations classify the risk of developing DMS. Based on the resulting combination of alleles, haplotypes are classified into three groups: haplotypes with a high risk of DMS manifestation, haplotypes with a medium risk of DMS manifestation, and haplotypes with a low risk of DMS manifestation. Each locus can carry either a dominant risk allele (A, B, C) or a recessive non-risk allele (a, b, c).

- Haplotypes with a high risk of disease manifestation (90 – 100 %): AABbCC, AaBBCC, AABBCC, AABBCc
- Haplotypes with a middle risk of DMS manifestation (33 – 50 %): AAbbCC, AAbbCc, aaBBCC, AaBBCc, AABbCc
- Haplotypes with a low risk of DMS manifestation (0 – 5 %): aabbCC, aabbCc, AabbCC, AabbCc, aaBbCC, aaBbCc, AaBbCC, AaBbCc, aaBBCc

The allelic combinations aabbcc, aaBBcc, Aabbcc, AaBbcc, AaBBcc, AAbbcc, AABbcc, and AABBcc cannot currently be assigned to any of the three haplotype groups, because there are no conclusive findings regarding their significance yet.

Method: SOP188-MPS-canine, MPS

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Date of issue: 06.02.2021
Date of testing: 01.02.2021 - 06.02.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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