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Result certificate #176103

Detection of c.1473+1G>A mutation in ADAMTS17 gene causing PLL disease in several dog breeds

Customer: Jenny Joffer, Jobsbovaegen 21, 77733 Smedjebacken, Sweden
Sample:
Sample: 21-11574
Date received: 12.05.2021
Sample type: buccal swab

Information provided by the customer
Name: Jammings' Hola
Breed: Chinese Crested Dog
Microchip: 985 141 001 123 303
Reg. number: SE19640/2019
Date of birth: 2018 November 30
Sex: female
Date of sampling: 05.05.2021

Result: Mutation was not detected (N/N)

Explanation

Presence or absence of c.1473+1G>A mutation in ADAMTS17 gene causing PLL disease (primary lens luxation) in different dog breeds was tested. PLL is an eye defect connected with dislocation of the lens caused by defective fixation of the lens zonules or suspensory fibres. Disease occurs dogs of different age, even results in blinding.

Dog without risk of development of PLL due to mutation c.1473+1G>A has genetic test result N/N (negative in both alleles). Dog in risk of development of PLL due to mutation c.1473+1G>A has genetic test result P/P (positive mutation finding in both alleles). The general inheritance of PLL is considered autosomal recessive and therefore the disease does not develop in the most carriers of c.1473+1G>A mutation (test result N/P) during their lives. Nevertheless, for carriers of c.1473+1G>A mutation the risk of development of PPL is higher. The risk depends on the specific breed or genetic background of the individual dogs. The genetic examination performed neither excludes the possibility of other PLL-form nor reveals the development of the disease in carriers of the mutation.

Method: SOP175-PLL, real-time PCR-ASA, accredited method

Date of issue: 14.05.2021
Date of testing: 12.05.2021 - 14.05.2021
Approved by: Ing. Irena Rusková, Analyst

verified and certified document
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