

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

Customer: Ms Jenny Joffer, Jobsbovaegen 21, S-777 33 Smedjebacken, Sweden

Sample:

Sample: 22-08755

Date received: 11.04.2022

Sample type: buccal swab

Information provided by the customer

Name: Joffers Arna Albicilla

Breed: Chinese Crested Dog

Microchip: 941 000 026 015 485

Reg. number: SE17348/2021

Date of birth: 2021 02 09

Sex: female

Date of sampling: 04.04.2022

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

Date of issue: 13.04.2022

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Approved by: Mgr. Martina Šafrová, Laboratory Manager



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