

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

**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: 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: SOPAgriseq\_canine, ngs

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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