

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

**Sample**

Sample: 18-22302  
Name: Vermont z Atarova dvora  
Breed: Jack Russell Terrier  
Microchip: 941 000 021 206 144  
Reg. number: ČLP/JRT/4296  
Date of birth: 20.5.2017  
Sex: male  
Date received: 16.08.2018  
Sample type: buccal swab  
The identity of the animal has been checked by  
MVDr. Veverková Katetřina

**Customer**

Ladislav Frnčo  
Bílovice 99  
79841 Kostelec na Hané  
Czech Republic

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: SOP182-PLL, HRMA, accredited method

Report date: 21.08.2018

Responsible person: Mgr. Markéta Dajbychová, Deputy Laboratory Manager

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