

## Result certificate #035289:

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

Sample

Sample: 13-16127

Name: BLACK PEARL NAKED OL-A-MI MORAVIA

Breed: American Hairless Terrier Microchip: 972000010017311 Date of birth: 12.5.2010

Sex: female

Date received: 20.06.2013 Sample type: blood

Sample certified by Vet/Tech or witness.

Customer

Lucie Přikrylová Stařechovice 69

79841 Kostelec na Hané

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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.

The general inheritance of PLL is considered autosomal recessive and therefore the disease does not develop in the most carriers of ADAMTS17-mutation 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: SOP106, accredited method

Report date: 24.06.2013

Responsible person: Mgr. Martina Šafrová, Laboratory Manager

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