

Detection of g.9459\_9460ins236 mutation  
in PLPLA gene causing CNM in Labrador  
Retrievers

**Sample**

Sample: 17-07916  
Name: Silver Skydiver Advocate  
Breed: Labrador Retriever  
Microchip: 804 098 100 099 956  
Reg. number: ČLP/LR/32361  
Date of birth: 18.10.2016  
Sex: male  
Date received: 28.03.2017  
Sample type: buccal swab

**Customer**

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**Result: Mutation was not detected (N/N)**

**Legend:** N/N = wild-type genotype. N/P = carrier of the mutation. P/P = mutated genotype (individual will be most probably affected with the disease). (N = negative, P = positive)

**Explanation**

Presence or absence of g.9459\_9460ins236 mutation in PLPLA gene causing CNM (Centronuclear Myopathy or also HMLR (Hereditary Myopathy in Labrador Retrievers)) in Labrador Retrievers was tested. CNM is a genetically conditioned defect of muscle fibres development. The disease manifests in two weeks of puppies age, muscle atrophy gradually affects movement and swallowing muscles. Affected individuals die during the first weeks or months of life.

Mutation that causes CNM is inherited as an autosomal recessive trait. That means the disease affects dogs with P/P genotype only. The dogs with N/P genotype are considered carriers of the disease (heterozygotes). In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N, 25 % P/P and 50 % N/P.

Method: SOP176-CNM, ASA-PCR

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Responsible person: Mgr. Markéta Dajbychová, Deputy Laboratory Manager



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