



Result

Detection of g.3( mutation in CL

**Sample**

Sample: 18-33626  
Name: Lasi Majerka  
Breed: Alpine Dachsbracke  
Tattoo number: 4839  
Reg. number: ABJ/4839  
Date of birth: 20.04.2013  
Sex: female  
Date received: 03.12.2018  
Sample type: buccal swab  
The identity of the animal has been checked by M. Vodnárková, ČKCHABJ

**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 with the disease). (N = negative, P = positive)

**Explanation**

Presence or absence of g.30852988\_30902901del mutation in CLN8 gene causing lipofuscinosis of type 8) in Alpine Dachsbrackes was tested. The symptoms of the disease are variable. The NCL usually includes neurological symptoms, such as disorientation, aggressiveness, seizures and food intake difficulty. Sudden loss of vision is a common symptom. As neurodegeneration increases with the age and psychical abnormalities and spasms. Changes in gait – stumbling gait and limb stiffness can be observed as well.

Mutation that causes NCL8 is inherited as an autosomal recessive trait. That means that the disease occurs only with P/P genotype. The dogs with N/P genotype are considered carriers of the mutation. In offspring of two heterozygous animals following genotype distribution carrier (healthy non-carriers), 25 % P/P (affected), and 50 % N/P (healthy carriers).

Method: SOP176-NCL8, ASA-PCR

Report date: 07.12.2018

Responsible person: Mgr. Martina Šafrová, Laboratory Manager