

**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)**

**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 c.829T>C mutation in CLN6 gene causing neuronal ceroid lipofuscinosis type 6 (NCL6) in Australian Shepherds was tested. The NCL disorder is characterized by excessive accumulation of waste lipopigment compounds primarily in the cells of the nervous system. The nerve cells in the cortex and the cerebellum and the retinae cells are affected and destroyed due to the high content of lipofuscin and its increasing pressure. The signs may include loss of vision, behavioural changes, worsening of motor and cognitive abilities, seizures which may look like epileptic seizures. The affected dogs start showing signs of NCL6 around a year and a half of age. It ends by premature death usually within one year after the first signs appear.

Mutation that causes NCL6 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), they are healthy but they can transmit the mutation on their offspring. Dogs with N/N genotype are without risk of NCL6. In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N (healthy non-carriers), 50 % N/P (healthy carriers) and 25 % P/P (affected).

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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