

Customer: Darina Šablíj, Nováková 972, 29301 Mladá Boleslav, Czech Republic**Sample:**

Sample: 22-29646

Date received: 26.10.2022

Sample type: buccal swab

Information provided by the customer

Name: CAPTAIN AMERICA AT Mystic Fire**Breed:** Australian Shepherd

Microchip: 203 098 100 577 554

Reg. number: CMKU/AUO/7703/21

Date of birth: 17.05.2021

Sex: male

Date of sampling: 21.10.2022

The identity of the animal has been checked by MVDr. Petra

Orthová, KVL 4347

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 retinal 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: 20.12.2023

Date of testing: 08.12.2023 - 20.12.2023

Approved by: Mgr. Martina Šafrová, Laboratory Manager

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