

Result certificate #012345

Detection of c.128+1_128+4delGTAA mutation in SERAC1 gene causing CMSD in Chinese Crested dogs

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.128+1_128+4delGTAA mutation in the SERAC1 gene causing CMSD (Multiple System degeneration in dogs) in Chinese Crested dogs was tested. CSMD is a neurodegenerative disorder. It is characterized by degeneration of neurons in one of the midbrain region and loss of cerebellar cells. The affected dogs suffer primarily from movement disorders due to motor system degeneration. Symptoms are for example unnatural tremor of the head, late reaction, convulsiveness and progressive loss of movement coordination.

Mutation that causes CMSD in Chinese Crested dogs 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. In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N, 50 % N/P and 25 % P/P.

Method: SOPAgriseq_canine, ngs, accredited method

Date of issue: 06.02.20<mark>21</mark> Date of testing: 01.0<mark>2.2</mark>021 - 06.02.2021 Approved by: Mgr. Martina Šafrová, Laboratory Manager



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