

Result certificate #012345

Detection of c.1688_1706dup mutation GLB1 gene causing GM1 in Alaskan Husky

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

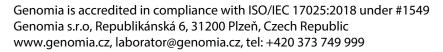
Presence or absence of c.1688_1706dup mutation GLB1 gene causing gangliosidosis type 1 (GM1) in Alaskan Husky was tested. The disease is caused by GM1 ganglioside accumulation in various tissues due. The disease is characterized by progressive neuromuscular dysfunction and growth impairment from an early age. Clinical signs begin to appear around 6 weeks of age and include proportional dwarfism, impaired coordination of movements, head tremors, strabismus, and nystagmus.

Mutation that causes GM1 in Alaskan Husky 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: SOPAgriseq_canine, ngs, accredited method

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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