

Detection of c.497G>A mutation in GUSB gene causing mucopolysaccharidosis VII in German Shepherds

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.497G>A mutation in GUSB gene causing mucopolysaccharidosis VII in German Shepherds was tested. Mucopolysaccharidosis VII is a lysosomal storage disease in which mucopolysaccharides accumulate in lysosomes. Clinical signs include facial dysmorphism (noticeable shortening and widening of the muzzle and low-set ears as early as 4 weeks of age in puppies), diffuse corneal opacity, presence of glycosaminoglycans in the urine, bone lesions, lack of body growth relative to the head, later joint problems, and even inability to stand. Various cardiac abnormalities may also occur.

Mutation that causes mucopolysaccharidosis VII is inherited autosomally recessively which means that the disease develops only in those dogs who inherit mutated allele from both parents; 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

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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Report verification code is: 12AB-CD34-GENO-MIA0-EFGH. You can verify report online at www.genomia.cz

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