GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.986T>C mutation in KCNJ10 gene causing SDCA1 disease in Belgian and Dutch 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.986T>C mutation in KCNJ10 gene causing Spongy cerebellar degeneration with cerebellar ataxia subtype 1 (SDCA1) in Belgian shepherds was tested. The occurrence of this mutation in breeds that were cross-bred with Belgian shepherds in the past, for example in some lineages of Dutch shepherds, cannot be excluded.

KCNJ10 gene encodes potassium channels (K+ channels) that are present in central nervous system, eyes, internal ear and kidneys. Function of K+ channel in cerebellar cortex altered due to this mutation results in extracellular accumulation of potassium, reduction of membrane potential and subsequent occurrence of neurological attacks.The first signs appear before the age of two months.

Mutation that causes SDCA1 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 (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

verified and certified document
GENOMIA genetic laboratory

Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999