GENOMIA
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Result certificate #012345

Detection of c.986T>C mutation in KCNJ10 gene causing SDCA1 disease in Belgian shepherds

Sample
Sample: 08-12345
Name: Lassie DEMO
Breed: ---
Tattoo number: 1392013
Microchip: 123 456 789 012 345
Reg. number: REGQ12345
Date of birth: 31.12.1909
Sex: female
Date received: 25.11.2008
Sample type: blood
The identity of the animal has been checked.

Customer
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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.

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: SOP172-SDCA1, direct DNA sequencing

Report date: 25.11.2008
Responsible person: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document
GENOMIA genetic laboratory

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