GENOMIA
genetic laboratory

**Result certificate #012345**

Detection of c.130_131ins227 mutation in ATP1B2 gene causing SDCA2 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.130_131ins227 mutation in ATP1B2 gene causing Spongy cerebellar degeneration with cerebellar ataxia subtype 2 (SDCA2) 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. SDCA2 is relatively variable as to disease onset, severity and histopathological lesions. The cerebellar dysfunction occurs at the age of 4 to 6 weeks. The main symptoms are ataxic gait, balance loss and insufficient movement coordination. All affected puppies show wide-based ataxic gait, which is more obvious in the hind limbs. In this way the puppies try to keep stability and improve the movement coordination. The prognosis is poor and usually ends by euthanizing the dog.

Mutation that causes SDCA2 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: SOP176-SDCA2, ASA-PCR

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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