

Detection of c.563G>T mutation in SLC2A9 gene causing hyperuricosuria in several dog breeds

**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.563G>T mutation in SLC2A9 gene causing hyperuricosuria in several dog breeds was tested. Disease affects for example Dalmatian dog, Bulldog, Black Russian Terrier, American Staffordshire Terrier, Retriever, Parson Russell Terrier, South African Boerboel, Waimaraner, Big Munsterland Pointer, German shepherd; occurrence in other dog breed is not excluded. Disease is characterised by excessive excretion of uric acid into the urine. Affected dogs suffer from formation of uric acid stones.

Mutation that causes hyperuricosuria 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

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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