GENOMIA
genetic laboratory

Result certificate #289108

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

Customer: Michal Glaser, Doubek 109, 251 01 Doubek, Czech Republic
Sample:
Sample: 23-15207
Date received: 05.06.2023
Sample type: buccal swab

Information provided by the customer
Name: Emilka z Modrého království
Breed: Collie Smooth
Microchip: 900 085 001 644 611
Reg. number: CMKU/CK/2155/23
Date of birth: 19.4.2023
Sex: female
Date of sampling: 01.06.2023
The identity of the animal has been checked by MVDr. Pavel Mádr, CSc. KVL4735

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, accredited method

Date of issue: 26.02.2025
Date of testing: 12.02.2025 - 26.02.2025
Approved by: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document GENOMIA genetic laboratory