GENOMIA
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Result certificate #254278

Detection of c.391del mutation HEXB gene causing GM2 in Toy Poodles

Customer: Lenka Orzelová, Bohumínská 437/42, 710 00 Slezská Ostrava, Czech Republic
Sample:
Sample: 24-40118
Date received: 30.01.2024
Sample type: buccal swab

Information provided by the customer
Name: Carramia Royal Glow
Breed: Poodle Standard
Microchip: 941000027192388
Reg. number: CMKU/P/23254/22
Date of birth: 17.7.2022
Sex: female
Date of sampling: 27.01.2024
The identity of the animal has been checked by Ing. Nikola Eretová, Genomia s.r.o.

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.391del mutation HEXB gene causing gangliosidosis type 2 (GM2, Sandhoff-like disease) in Toy Poodles was tested. The disease is caused by GM2 ganglioside accumulation in various tissues due to a deficiency of the enzyme hexosaminidase, which is responsible for breaking down the GM2 ganglioside into its components. The disease is characterized by progressive neuromuscular dysfunction and growth failure at an early age.

Mutation that causes GM2 in Toy Poodles 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: 09.02.2024
Date of testing: 30.01.2024 - 09.02.2024
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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