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Result certificate #230266

Detection of mutation c.[1297T>C;1299C>T] of L2HGDH gene causing L2HGA in Staffordshire bull terriers

Customer: Renée Kryšpín Hájková, Prostřední Staré Buky 94, 54101 Staré Buky, Czech Republic
Sample:
Sample: 23-13708
Date received: 18.05.2023
Sample type: blood

Information provided by the customer
Name: El Mariachi Bagheera Bulls
Breed: Staffordshire Bull Terrier
Microchip: 203 098 100 528 713
Reg. number: CMKU/SBT/17404/22
Date of birth: 18.04.2022
Sex: male
Date of sampling: 17.05.2023
The identity of the animal has been checked by MVDr. Jakub Sova, KVL 7127

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 mutation c.[1297T>C;1299C>T] in exon 10 of L2HGDH gene causing L-2-hydroxyglutaric aciduria in Staffordshire bull terriers were tested. Double nucleotide substitution leads to double amino acid replacement in L2-hydroglutaric dehydrogenase enzyme: leucine to proline in position 433 and histidine to tyrosine in position 434. These mutations cause L-2-hydroxyglutaric aciduria disease (L-2-HGA) in Staffordshire bull terriers. L-2-HGA is a neurological disorder manifested by psychomotor retardation, seizures and ataxia. Accumulation of L-2-hydroxyglutaric acid in cerebrospinal fluid, plasma and urine is very typical.

Mutation that causes L-2-HGA 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: 30.05.2023
Date of testing: 18.05.2023 - 30.05.2023
Approved by: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document
GENOMIA genetic laboratory

Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999