GENOMIA
genetic laboratory

Result certificate #170952

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

Customer: Pavla Hašková, V Pařezinách 547, 19012 Praha, Czech Republic
Sample:
Sample: 21-04924
Date received: 03.03.2021
Sample type: blood

Information provided by the customer
Name: Charlie Tango Chesway
Breed: Staffordshire Bull Terrier
Microchip: 203 098 100 504 285
Reg. number: CMKU/SBT/13640/19
Date of birth: 2.11.2019
Sex: male
Date of sampling: 26.02.2021
The identity of the animal has been checked by MVDr. Radek Musil, KVL 3848

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: SOP175-L2HGA, real-time PCR-ASA, accredited method

Date of issue: 05.03.2021
Date of testing: 03.03.2021 - 05.03.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

Genomia is accredited in compliance with ISO/IEC 17025:2018 under #1549
Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999

ilac-MRA L 1549