GENOMIA
genetic laboratory

Result certificate #173500

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

Customer: Ivana Kubíčková, Želivského 806, 28002 Kolín, Czech Republic
Sample:
Sample: 19-42252
Date received: 16.09.2019
Sample type: buccal swab

Information provided by the customer
Name: Armstrong Siddeley Never Back Down
Breed: Staffordshire Bull Terrier
Microchip: 203098100427064
Reg. number: CMKU/SBT/11374/18
Date of birth: 22.03.2018
Sex: male
Date of sampling: 15.09.2019
The identity of the animal has been checked by Bc. Eva Šandarová, 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 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: 01.04.2021
Date of testing: 24.03.2021 - 01.04.2021
Approved by: Mgr. Lucie Magoči, Analyst

Genomia is accredited in compliance with ISO/IEC 17025:2018 under #1549
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