GENOMIA
genetic laboratory

Result certificate #108623

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

Sample
Sample: 17-23340
Name: Blanka One Bull Staff
Breed: Staffordshire Bull Terrier
Microchip: 203 098 100 405 976
Reg. number: CMKU/SBT/10609/17
Date of birth: 10.6.2017
Sex: female
Date received: 22.08.2017
Sample type: buccal swab
The identity of the animal has been checked by Mgr. Lucie Pinďáková, Genomia s.r.o.

Customer
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Ordered on April 3, 2018.

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

Report date: 06.04.2018
Responsible person: Mgr. Markéta Dajbychová, Deputy Laboratory Manager

Genomia is accredited according to ISO/IEC 17025:2005 under #1549.

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