

**Result certificate #069351:**

**Sample**

Sample: 15-40339  
Name: Andorra La Vella Rascal Bull  
Breed: Staffordshire Bull Terrier  
Microchip: 900 182 000 234 795  
Reg. number: SPKP 1045  
Date of birth: 20.10.2012  
Sex: female  
Date received: 14.09.2015  
Sample type: buccal swab  
Sample certified by Vet/Tech or witness.

**Detection of mutation c.[1297T>C;1299C>T] in exon 10 of L2HGDH gene causing L2HGA in Staffordshire bull terriers by allelic discrimination**

**Customer**

Ján Timanik  
Obrancov mieru 19  
064 01 Stará Ľubovňa  
Slovak Republic

Sampling: Ing. Irena Rusková, 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**

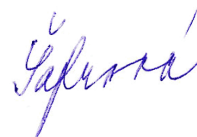
Presence or absence of mutation c.[1297T>C;1299C>T] in exon 10 of L2HGDH gene causing L-2-hydroxyglutaric aciduria in Staffordshire bull terriers were tested. Double nucleotide substitution leads to double amino acid replacement in L-2-hydroxyglutaric dehydrogenase enzyme: leucine to proline in position 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: SOP167

Report date: 22.09.2015

Responsible person: Mgr. Martina Šafrová, Laboratory Manager



Genomia s.r.o, Janáčkova 51, 32300 Plzeň, Czech Republic  
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999