GENOMIA
genetic laboratory

Result certificate #065850:

Detection of c.2228G>A mutation in PFK gene causing Pyruvate kinase deficiency in several dog breeds by PCR-RFLP

Sample
Sample: 15-15810
Name: Jump'n Smile Didgeridoo
Breed: American Cocker Spaniel
Microchip: 040 098 100 437 393
Reg. number: ÖHZB/ACS 241
Date of birth: 24.2.2015
Sex: female
Date received: 08.06.2015
Sample type: buccal swab
Sample certified by Vet/Tech or witness.

Customer
Mag. Damir Dodan
Beethovengasse 4
3420 Kritzendorf
Austria

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.2228G>A in exon of 21 PFK gene causing Pyruvate kinase deficiency (PFK) in English Springer Spaniels and American Cockers was tested. The deficiency of the muscle phosphofructokinase belongs to the group of glycogenoses (Inherited Glycogen Storage Disease). The main clinical features are especially muscle fatigue, weakness and exercise intolerance. The clinical symptoms may occur in the first months of the life; however, they may be relatively bad recognisable and some cases go unrecognised. The life quality of the affected animal can be improved, if you avoid exercises that stimulate the occurrence of hemalytic crisis.

Mutation that causes PFK in English Springer Spaniels and American Cockers is inherited autosomally recessively which means that the disease develops only in those dogs who inherit mutated allele from both parents; 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: SOP60

Report date: 17.06.2015
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