GENOMIA
genetic laboratory

# Result certificate #012345

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

**Sample**
Sample: 08-12345
Name: Lassie DEMO
Breed: ---
Tattoo number: 1392013
Microchip: 123 456 789 012 345
Reg. number: REGQ12345
Date of birth: 31.12.1909
Sex: female
Date received: 25.11.2008
Sample type: blood
The identity of the animal has been checked.

**Customer**
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## 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: SOP173-PFK, PCR-RFLP

Report date: 25.11.2008
Responsible person: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document
GENOMIA genetic laboratory

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