

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

**Customer:** Jan Novák, Dlouhá 1, 30000 Plzeň, Czech Republic

**Sample:**

Sample: 21-12345

Date received: 01.02.2021

Sample type: blood

Information provided by the customer

**Name:** Lassie DEMO

**Breed:** Plemeno

Tattoo number: 1392013

Microchip: 123 456 789 012 345

Reg. number: REGQ12345

Date of birth: 1.1.2020

Sex: female

Date of sampling: 01.02.2021

The identity of the animal has been checked.

**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 Cocker Spaniels was tested. The deficiency of the muscle phosphofruktokinase 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 hemolytic crisis.

Mutation that causes PFK in English Springer Spaniels and American Cocker Spaniels 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: SOPAgriSeq\_canine, ngs, accredited method

Date of issue: 06.02.2021

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Approved by: Mgr. Martina Šafrová, Laboratory Manager



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Report verification code is: 12AB-CD34-GENO-MIA0-EFGH. You can verify report online at [www.genomia.cz](http://www.genomia.cz)

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