

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

**Sample**

Sample: 17-08734  
Name: Jump'N Smile Fifty Shades of Red  
Breed: American Cocker Spaniel  
Microchip: 040 098 100 454 403  
Reg. number: ÖHZB ACS 252  
Date of birth: 25.04.2016  
Sex: female  
Date received: 04.04.2017  
Sample type: buccal swab  
The identity of the animal has been checked by  
Mag.med.vet. Sabine Freysinger

**Customer**

Mag. Damir Dodan & Mag. Martina Dodan  
Beethovengasse 4  
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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 Cocker 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 hemolytic crisis.

Mutation that causes PFK in English Springer Spaniels and American Cocker 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

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Responsible person: Mgr. Martina Šafrová, Laboratory Manager



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