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Result certificate #117206

Detection of (c.799C>T, c.848T>C, c. 994G>A) mutations in PK-LR gene causing PK deficiency in Labrador Retrievers, Beagles and Pugs

Sample
Sample: 18-08120
Name: Hasco vom Ammeler Forst
Breed: Beagle
Microchip: 756 097 200 034 766
Reg. number: 675986
Date of birth: 10.09.2008
Sex: male
Date received: 28.03.2018
Sample type: blood
The identity of the animal has been checked by Dr. med. vet. Fabienne Künzli

Customer
Andrea Wanner
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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 (c.799C>T, c.848T>C, c. 994G>A) mutations in PK-LR gene causing deficiency of pyruvate kinase (PK deficiency) in Labrador Retrievers, Beagles and Pugs was tested. The deficiency of this enzyme causes insufficient production of ATP, which results in erythrocyte lysis or their premature destruction in the spleen. The increased destruction of erythrocytes expresses itself clinically as anaemia.

Mutations that cause PK deficiency in Labrador Retriever, Beagle and Pug breeds are 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: SOP172-PKdef, direct DNA sequencing

Report date: 10.10.2018
Responsible person: Ing. Irena Rusková, Analyst

verified and certified document
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