

Result certificate #012345

Diagnostic test for Copper Toxicity in Bedlington Terriers

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 39,7 kb deletion in exon 2 of canine COMMD1 gene was tested. This deletion causes copper toxicosis (CT) in Bedlington Terriers. Copper toxicosis is a metabolism disease – biliary copper excretion is markedly reduced. As a result copper accumulates in lysosomes of hepatocytes, eventually leading to liver cirrhosis, chronic hepatitis and premature death. CT disease manifestation starts between 2 and 6 years of age. First symptoms are problems with food intake, sickness and weight loss.

Mutation that causes CT disease is 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: SOP176-CT, ASA-PCR

Sensitivity (probability of correct identification of the defective form of the gene in heterozygous or mutated homozygous) is higher than 99%. Specificity (probability of correct identification of the normal form of the gene in a normal homozygous or heterozygous) is higher than 99%.

Date of issue: 06.02.2021 Date of testing: 01.02.2021 - 06.02.2021 Approved by: Mgr. Martina Šafrová, Laboratory Manager



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