GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.754C>T mutation in PDP1 gene causing [pyruvatedehydrogenase] phosphatase deficiency in Clumber and Sussex Spaniels

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.754C>T in PDP1 gene was tested. The mutation forms a premature stopkodon that leads to [pyruvatedehydrogenase] phosphatase 1 deficiency disease in Clumber and Sussex Spaniels.

Clinically, PDP1 deficiency is manifested as exercise intolerance that can cause total collapse of the affected dog. The disease is often accompanied by cardiac, pulmonary and neurological problems. So the affected dogs often die prematurely. Disease symptoms can be reduced by the timely initiation of ketogenic diet.

Mutation that causes PDP1 deficiency 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: SOPAgriseq_canine, ngs
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

verified and certified document
GENOMIA genetic laboratory

Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999