GENOMIA
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Result certificate #012345

Detection of c.996G&gtA mutation in AGXT gene causing PH I in Coton de Tulear

Sample
Sample: 08-12345
Name: Lassie DEMO
Breed: ---
Tattoo number: 1392013
Microchip: 123 456 789 012 345
Reg. number: REGQ12345
Date of birth: 31.12.1909
Sex: female
Date received: 25.11.2008
Sample type: blood
The identity of the animal has been checked.

Customer
Jan Novák
Dlouhá 1
30000 Plzeň
Czech Republic

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.996G>A mutation in AGXT gene causing PH I (Primary hyperoxaluria type I) in Coton de Tulear breed was tested. The PH disease is caused by insufficient function of a liver-specific enzyme alanine -glyoxalate aminotransferase (AGT, AGXT). PH is characterized by the accumulation of oxalate and subsequent precipitation of calcium oxalate crystals, primarily in the kidneys, leading to progressive kidney failure. If the storage capacity of the kidneys is exhausted, the crystals are accumulated in other tissues, for example in bones, joints, cartilages, retina and muscles.

Mutation that causes PH I in Coton de Tulear 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: SOP173-PHI, PCR-RFLP

Report date: 25.11.2008
Responsible person: 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