GENOMIA
genetic laboratory

Result certificate #037918:

Detection of c.586C>T mutation in exon 2 of SLC3A1 gene causing cystinuria in Newfoundland and Landseer breeds by PCR-RFLP

Sample
Sample: 13-24001
Name: Daffy Alvemia
Breed: Landseer
Reg. number: 477
Microchip: 20309810028133
Date of birth: 21.10.2009
Sex: female
Date received: 30.08.2013
Sample type: blood

Customer
Renata Cepková
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Result: Mutation was not detected (N/N)

Explanation

Presence or absence of c.586C>T mutation in exon 2 of SLC3A1 gene causing cystinuria in Newfoundland and Landseer breeds was tested. Due to high concentration of cystine, hard crystals of cystine can be formed in the urine of affected individuals. The crystals can cause uncomfortable health complications e.g. urinary deposits and/or kidney stones, which can ultimately lead to the obstruction of the urinary tract.

Mutation that causes cystinuria is inherited as an autosomal recessive trait. That means the disease affects dogs with P/P (positive/positive) genotype only. The dogs with N/P (negative / positive) genotype are considered carriers of the disease (heterozygotes). In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N (healthy non-carriers), 25 % P/P (affected), and 50 % N/P (healthy carriers).

Method: SOP03, accredited method
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%.

Report date: 06.09.2013
Responsible person: Mgr. Markéta Dajbychová, Deputy Laboratory Manager

Dajbychová