GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.286C>T mutation in CCDC39 gene causing primary ciliary dyskinesia in Old English Sheepdog breed

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
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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.286C>T mutation in exon 3 of CCDC39 gene causing primary ciliary dyskinesia (PCD) in Old English Sheepdog breed was tested. PCD is an inherited disorder characterized by insufficient function of cilia of epithelial mucous membrane. Typical symptoms of PCD are recurrent infection of the upper and lower respiratory tract and reduced fertility in males.

Mutation that causes PCD in Old English Sheepdog breed 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: SOP182-PCD, HRMA

Report date: 25.11.2008
Responsible person: Mgr. Martina Šafrová, Laboratory Manager

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