

Result report certificate Detection of mutation in dog PRCD gene

Customer

Gerda van Empel
Dollemanstraat 2
7223 KG Baak
Netherlands

Sample

Sample: 34222
Name: Benclouch Breakdance
Breed: English Cocker Spaniel
Microchip: 981000006339460
Reg. number: 2972017
Date of birth: 10-01-2014
Sex: male
Date received: 30.12.2014
Sample type: buccal swab

Result: N/N

Result codes:

N/N clear (normal homozygote)
N/P carrier (heterozygote)
P/P affected (mutated homozygote)

Explanation

Presence or absence of mutation 1298G>A in PRCD gene in CFA9 (canine chromosome 9) has been examined. This mutation induces PRA-prcd (Progressive Retinal Atrophy form Progressive Rod Cone Degeneration). Disease causes degeneration of retinal cells. Firstly, rods are affected and night blindness develops in the animal. Later, cones degenerate. That results in complete blindness of the animal. The age of onset of disease varies, but, generally, it cannot be recognized before the adulthood of the animal.

Mutation that causes Prcd-PRA 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 (healthy non-carriers), 25 % P/P (affected), and 50 % N/P (healthy carriers).

The PRA-prcd mutation was found in following dog breeds: Am. Eskimo Dog, Austr. Cattle Dogs, Austr. Shepherd (normal, mini), Austr. Stumpy Tail Cattle Dog, Retriever (Chesapeake Bay, Golden, Labrador, Nova Scotia Duck Tolling), Chinese Crested Dog, Cockapoos, Cocker Spaniel (Am., Engl.), Basenji, Poodles (Dwarf, Miniature, Toy), Entlebucher Mountain Dog, Lapphund (Swedish, Finnish), Goldendoodle, Karelian Bear Dog, Kuvasz, Magyar Vizsla, Labradoodle, Lapponian Herder, Norwegian Elkhound, Papillon, Water Dog (Portuguese, Spanish), Terrier (Silky, Yorkshire). With lower probability, other breeds can also suffer from PRA-prcd.

Report date: 02.01.2015

Responsible person: Mgr. Martina Šafrová, Analyst

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Result certificate #054077:

Detection of c.115A>T mutation in exon 3 of COL4A4 gene causing Familial Nephropathy in English Cocker Spaniels by DNA sequencing

Sample

Sample: 14-32118
Name: Bencleuch Breakdance
Breed: English Cocker Spaniel
Microchip: 981000006339460
Reg. number: 2972017
Date of birth: 10-01-2014
Sex: male
Date received: 30.12.2014
Sample type: buccal swab

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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.115A>T mutation in exon 3 of the COL4A4 gene causing familial nephropathy (FN) in English Cocker Spaniels was tested. FN disorder is a fatal renal disease. Kidney failure arises between 6 months and 2 years of age of the dog. The first observed symptoms include vomiting, loss of appetite, excessive water consumption, and weight gain or loss.

Mutation that causes FN is inherited autosomally recessively which means that the disease develops only in those dogs who inherit mutated allele from both parents; 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: SOP19, accredited method

Report date: 02.01.2015

Responsible person: Mgr. Barbora Bláhová, Analyst



Genomia is accredited according to ISO/IEC 17025:2005 under #1549.

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HOUDEN VAN HONDEN



NEDERLANDS JEUGDKAMPIOEN

NAAM HOND

BENCLEUCH BREAKDANCE

STAMBOOMNUMMER

NHSB 2972017

RAS

ENGELSE COCKER SPANIEL

GESLACHT

REU

EVENEMENT

TENTOONSTELLING TE KATWIJK OP 21 DECEMBER 2014

EIGENAAR

G.H. VAN EMPEL



Raad van Beheer, Amsterdam

22 APRIL 2015

Rony Doedijns, Directeur