GENOMIA
genetic laboratory

Result certificate #172085

Detection of c.2685delA and c.2687_2688insTAGCTA mutations in CNGB1 gene causing Pap-PRA 1 in Papillons and Phalens

Customer: Romana Špottová, Zahradní 124, 25163 Strančice, Czech Republic
Sample:
Sample: 21-06322
Date received: 11.03.2021
Sample type: buccal swab

Information provided by the customer
Name: Iris Mon Amour Emia Alva
Breed: Papillon
Microchip: 203 098 100 430 007
Reg. number: CMKU/ PAP/4413/19/20
Date of birth: 10.1.2019
Date of sampling: 10.03.2021
The identity of the animal has been checked by MVDr. Hana Prausová, KVL 5291

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.2685delA and c.2687_2688insTAGCTA mutations in CNGB1 gene causing Pap-PRA 1 in Papillons and Phalens was tested. Progressive retinal atrophy (PRA) is characterized by a very rapid loss of function of rods followed by a loss of cone function. The primary clinical sign is often vision impairment in a dim light and progressive loss of vision up to complete blindness.

Mutations that cause Pap-PRA1 in Papillon and Phalen are 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: SOP171-papPRA1, fragment analysis

Date of issue: 17.03.2021
Date of testing: 11.03.2021 - 17.03.2021
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