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Result certificate #319825

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

Customer: Serhii Korenchenko, Mladé gardy 2781, 8, 434 01 Most, Czech Republic
Sample:
Sample: 25-33529
Date received: 02.01.2026
Sample type: buccal swab

Information provided by the customer
Name: DANALIYA FLY FINKI
Breed: Papillon
Microchip: 900 233 001 513 475
Reg. number: CMKU/PAP/6014/-25/24
Date of birth: 22.05.2024
Sex: female
Date of sampling: 22.12.2025
The identity of the animal has been checked by MVDr. Alice Šimková, KVL 7073

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: SOP188-MPS-canine, MPS, accredited method

Date of issue: 08.01.2026
Date of testing: 02.01.2026 - 08.01.2026
Approved by: Mgr. Martina Šafrová, Laboratory Manager



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