

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

**Customer:** Jan Novák, Dlouhá 1, 30000 Plzeň, Czech Republic

**Sample:**

Sample: 21-12345

Date received: 01.02.2021

Sample type: blood

Information provided by the customer

**Name:** Lassie DEMO

**Breed:** Plemeno

Tattoo number: 1392013

Microchip: 123 456 789 012 345

Reg. number: REGQ12345

Date of birth: 1.1.2020

Sex: female

Date of sampling: 01.02.2021

The identity of the animal has been checked.

**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: 06.02.2021

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Approved by: Mgr. Martina Šafrová, Laboratory Manager



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