

Detection of the c.452A>C mutation in the JPH2 gene associated with PRA in Shih Tzu Dogs

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)

Explanation

The presence or absence of the c.452A>C mutation in the JPH2 gene, which is likely associated with Progressive Retinal Atrophy (PRA) in Shih Tzu dogs, was examined. Studies in other breeds have not been conducted. Progressive Retinal Atrophy comprises a large group of inherited disorders that represent a major cause of blindness in various dog breeds. Clinical signs typically begin with night blindness, followed by progressive loss of daytime vision and, eventually, blindness under all lighting conditions.

This mutation is inherited in an autosomal recessive manner. This means that only dogs with the P/P genotype, having inherited the mutated allele from both parents, are at risk of developing PRA. Dogs with the N/P genotype are clinically healthy carriers; however, they can pass the mutation on to their offspring. When two heterozygous carriers (N/P) are bred, theoretically 25% of the offspring will be clear (N/N), 50% will be carriers (N/P), and 25% (P/P) will inherit the mutated allele from both parents and therefore be at risk of PRA.

Method: SOP188-MPS-canine, MPS

Date of issue: 06.02.2021

Date of testing: 01.02.2021 - 06.02.2021

Approved by: Mgr. Martina Šafrová, Laboratory Manager



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