

Detection of c.del2986\_2991 mutation  
in ADAMTS17 gene causing POAG-PLL  
in Shar Pei

**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**

Presence or absence of ENSCAFT00000017051.3: c.del2986\_2991 mutation in ADAMTS17 gene causing POAG-PLL in Shar Pei was tested. The primary open angle glaucoma (POAG) and the primary lens luxation (PLL) are two diseases that are both caused by the same mutation and thus are indistinguishable from the genetics point of view.

Mutation c.del2986\_2991 that causes POAG-PLL 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, 25 % P/P and 50 % N/P.

Method: SOPAgriseq\_canine, ngs

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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