

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 c.2623G>A mutation in MTBP gene associated with Periodic Fever Syndrome, also called Shar-Pei autoinflammatory disease (SPAID), was tested. The disease is characterised by fever, arthritis, vesicular hyaluronosis (accumulation of hyaluronic acid in the tissues), otitis (otitis of middle ear) and amyloidosis (deposition of amyloid in the tissues). Recurrent fever syndrome is associated with mutations in the HAS2 and MTBP genes. Mutation in MTBP gene was tested.

The inheritance of this mutation is autosomal incompletely dominant. The disease manifests in affected homozygotes who have inherited the mutant allele from both parents. Heterozygotes will have an increased likelihood of developing the disease compared to healthy unaffected individuals.

Method: SOPAgriseq_canine, ngs, accredited method

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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