GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.2623G>A mutation in MTBP gene associated with Periodic Fever Syndrome 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 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: SOP188-MPS-canine, MPS, accredited method

Date of issue: 06.02.2021
Date of testing: 01.02.2021 - 06.02.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document GENOMIA genetic laboratory

Genomia is accredited in compliance with ISO/IEC 17025:2018 under #1549
Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999

ilac-MRA L 1549