

Detection of mutations associated with an increasing risk of Dilated Cardiomyopathy (DCM) in Dobermans

**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: PREVIEW RESULT LINE

**Explanation**

The presence or absence of mutations in the PDK4 gene (DCM1), TTN gene (DCM2) and mutations on chromosome 5 (DCM3 and DCM4) were examined. The discovery of other variants is expected. DCM1 and DCM2 variants were found in the American Doberman population, DCM3 and DCM4 variants are typical for the European Doberman population. Dilated cardiomyopathy is accompanied by symptoms such as cardiac arrhythmias, myocardial dysfunction or subsequent congestive heart failure. In Dobermans, the disease manifests itself in adulthood.

The inheritance of dilated cardiomyopathy DCM1, DCM2, DCM3 and DCM4 in Dobermans is characterized by incomplete penetrance. The resulting genotypes may be as follows:

- N/N; N/DCM4: the individual carries none of the previously known genetic variants or carries one copy of the DCM4 genetic variant → is healthy
- N/DCM1; N/DCM2; N/DCM3: the individual carries one copy of the risk genetic variant → risk of developing dilated cardiomyopathy
- DCM1/DCM1; DCM2/DCM2; DCM3/DCM3; DCM4/DCM4: individual carries two copies of the risk genetic variant → high risk of developing dilated cardiomyopathy

If an individual carries a combination of multiple risk genotypes, the likelihood of developing DCM increases.

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



Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic  
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999

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