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Result certificate #012345

Detection of g.32373916_32373923del8 mutation in STRN gene used for indirect diagnostics of ARVC

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 g.32373916_32373923del8 mutation in STRN gene was tested. The mutation is in genetic linkage with the development of right ventricular arrhythmogenic cardiomyopathy (ARVC) and is used for indirect diagnostics of ARVC.

ARVC is a multifactorial degenerative heart disease characterized by atrophy of muscle cells of the heart muscle and deposition of fat in the heart cells. ARVC affects predominantly the right ventricle. It is a life-threatening disease and may lead a sudden death.

The STRN mutation increases the severity of the clinical symptoms. With almost 100% probability, dogs homozygous for STRN mutation will be ARVC-affected and the clinical signs will occur at earlier age than in heterozygous dogs. The mutation has autosomal dominant pattern of inheritance with incomplete penetration. Whereas the STRN-mutation is clearly associated with ARVC in boxers, the studies have shown that also other unknown mutations play their role in this disease.

STRN is an important marker of a not yet identified gene locus located on the same chromosome that is directly responsible for development of ARVC in boxers.

Method: SOP171-ARVC, fragment analysis

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

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