GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.4151G>A mutation in ATP7B gene causing Wilson disease in dogs

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

Presence or absence of c.4151G>A mutation in ATP7B gene causing Wilson disease in dogs, in Lbarador retrievers and very likely also in some other breeds. The disease is manifested by toxic accumulation of copper in the liver and subsequent hepatitis.

In Labrador Retrievers, there are copper toxicosis modifiers. These variants of the RETN and ATP7A genes do not cause any disease by themselves and have no clinical significance in healthy individuals. In dogs with a homozygous mutation in the ATP7B gene (result ATPB/ATPB), the modifiers reduce the process of copper accumulation and thus lower the risk of liver cell damage. The genotypes N/RETN, RETN/RETN, ATP7A/Y in males, and ATP7A/ATP7A in females have a mitigating effect.

Dogs with a heterozygous mutation in the ATP7B gene (result N/ATPB) are carriers of Wilson's disease and do not show clinical symptoms. Dogs with the result N/N do not carry Wilson's disease.

The ATP7A mutation is inherited as X-linked recessive, the ATP7B mutation is inherited as autosomal recessive, and the RETN mutation is inherited as autosomal dominant. The ATP7A, ATP7B, and RETN genes are each located on a different chromosome, and therefore are inherited completely independently of one another.

Method: SOP188-MPS-canine, MPS

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