

Detection of mutations in the SH3TC2,
MTMR2, and MPZ genes causing
hypomyelinating polyneuropathy in Golden
Retrievers

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 the c.1924C>T mutation in the SH3TC2 gene, the c.1479+1G>A mutation in the MTMR2 gene, and the c.434T>C mutation in the MPZ gene associated with hypomyelinating polyneuropathy in Golden Retrievers was examined. Hypomyelinating polyneuropathy is a genetically determined disorder affecting the peripheral nervous system. It involves insufficient production of the myelin sheath, leading to muscle weakness, loss of reflexes, and impaired coordination. The SH3TC2, MTMR2, and MPZ genes are inherited independently of each other.

The inheritance pattern of the c.1924C>T mutation in the SH3TC2 gene and the c.1479+1G>A mutation in the MTMR2 gene is most likely autosomal recessive. This means that the disease develops only in individuals that inherit the mutated gene from both parents (P/P). Carriers (N/P) are clinically healthy but can pass the mutation on to their offspring. In a mating between two heterozygous individuals (N/P), the theoretical distribution of offspring is 25% completely healthy (N/N), 50% carriers (N/P), and 25% affected (P/P).

The inheritance of the c.434T>C mutation in the MPZ gene is most likely autosomal dominant. This means that a single copy of the mutated gene inherited from one parent is sufficient for the disease to manifest. Individuals with genotypes N/P and P/P are affected, while only individuals with genotype N/N are considered healthy. In a mating between two heterozygous individuals (N/P), the theoretical distribution of offspring is 25% completely healthy (N/N), 50% inheriting one copy of the mutated gene, and 25% inheriting two copies of the mutated gene (P/P), all of whom will be affected by the disease.

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

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