

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)**Legend:** N/N = wild-type genotype. N/P = carrier of the mutation. P/P = mutated genotype (individual will be most probably affected with the disease). (N = negative, P = positive)**Explanation**

Presence or absence of mutation c.241C>T in AMHR2 gene causing Persistent Mullerian duct syndrome (PMDS) in Miniature Schnauzers was tested.

Persistent Mullerian duct syndrome (PMDS) is a disorder of sexual development that affects males. Males with this disorder also develop a part of female reproductive organs. The affected males can also have a uterus, oviducts, ectocervix and a part of vagina that extends to the prostate gland. This anomaly results in unilateral or bilateral cryptorchidism (undescended testis into the scrotum) and its consequences such as infertility and an increased risk for testicular cancer. This syndrome is often accompanied with inflammation, e.g., pyometra (infection in the uterus) and associated effects, like polydipsia, polyuria, and loss of appetite.

Mutation that causes PMDS in Miniature Schnauzers is inherited autosomally recessively which means that the disease develops only in those dogs who inherit mutated allele from both parents; disease affects dogs with P/P genotype only. The dogs with N/P genotype are considered carriers of the disease (heterozygotes). In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N, 25 % P/P and 50 % N/P.

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



Genomia is accredited in compliance with ISO/IEC 17025:2018 under #1549

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