GENOMIA
genetic laboratory

# Result certificate #012345

Detection of c.241C>T in AMHR2 gene causing PMDS in Miniature Schnauzers

**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: SOPAgriseq_canine, ngs

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Approved by: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document
GENOMIA genetic laboratory

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