GENOMIA
genetic laboratory

# Result certificate #012345

Detection of c.636_637insC mutation in CHRNE gene causing CMS in Russell Terriers

**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.

SAMPLE

## 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 c.636_637insC mutation in CHRNE gene causing Myasthenic syndrome (CMS) in Russell Terriers was tested. Congenital myasthenic syndrome is an inherited disease that affects neuromuscular transmission. It is manifested by fatigable weakness of the skeletal muscles after exercise. The first symptoms can be already observed in puppies at the time of weaning, but the disease progresses rapidly, and most dogs die within 1 year of age.

Mutation that causes CMS 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

verified and certified document
GENOMIA genetic laboratory

Genomia is accredited in compliance with ISO/IEC 17025:2018 under #1549
Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999

ilac-MRA L 1549

Report verification code is: 12AB-CD34-GENO-MIA0-EFGH. You can verify report online at www.genomia.cz
Without a written consent by the lab, the report must not be reproduced unless as a whole.
The result refers only to the tested sample, as received. Genomia is not responsible for the accuracy of the information provided by the customer.