

**REFERENCE NO.:** 2024 - 067664/01

**OWNER:**

TETIANA TRUFIN  
VIA CESARE BATTISTI 1  
IT-27040 VERRUA PO  
ITALY

**NAME/LABEL:**

ROMA

**SPECIES:** DOG

**BREED:** STAFFORDSHIRE BULL TERRIER

**SEX:** FEMALE

**MICROCHIP NO.:** 380260171529348

**TATOO NO.:** -

**PEDIGREE NO.:** LO22158855

## GENETIC REPORT

**SAMPLE:** ISOLATED DNA

**SAMPLE TAKEN BY:** CHIARA REZZANI, DVM -, -, -, ITALY

**REQUESTED TEST:** L-2-HYDROXYGLUTARIC ACIDURIA (L-2-HGA)

**RESULT:** CLEAR (WT/WT)

**COMMENT :**

The test examines presence or absence of L2HGDH gene mutation (c.1297T>C, c.1299C>T) described as the cause of L-2-hydroxyglutaric aciduria (L-2-HGA) in Staffordshire Bull Terrier. The disease is characterized by accumulation of L-2-hydroxyglutaric acid in urine, plasma and cerebrospinal fluid. It is manifested with a variety of clinical neurological deficits, including psychomotor retardation, seizures and ataxia. Tested L2HGDH gene mutation is inherited as an autosomal recessive trait.

Regarding to the presence of tested mutation animals are classified in three groups:

- Clear (wt/wt) - mutation is not present, normal genotype
- Carrier (mut/wt) - one of two alleles carries tested mutation, disease is not clinically manifested
- Affected (mut/mut) - both alleles carry tested mutation, disease is clinically manifested

For each group different breeding strategies should be followed. Breeding of affected and carrier animals should be avoided. If particularly valuable animal is classified as affected, it should be bred only with clear animal. In such case, all first generation siblings will be carriers. If a carrier is bred with clear animal, 50% of siblings are expected to be clear. In case two carriers are bred, 25% of siblings are expected to be clear and 50% are expected to be carriers. However, 25% of siblings are expected to be affected, therefore such breeding practice is discouraged.

**AUTHORIZED SIGNATURE:**

MARIBOR, 22.11.2024



*Results are valid for laboratory analysed samples only. Accuracy of the data about animal identity is the sole responsibility of the customer/owner. Laboratory is not responsible for false results which arise due to inaccurate animal identity data, false sample labels etc. To the extent the law allows, the maximal compensation for potential false result is limited to the invoiced amount. With the test it is not possible to rule out the presence of other genetic changes which might affect the development of the disease. Testing is performed according to the latest scientific knowledge.*