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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