EVG molekularna diagnostika d.o.o.
Kardeljeva cesta 98 · 2000 Maribor · Slovenia
+ 386 40 566 273 · info@eurovetgene.com
www.eurovetgene.com

REFERENCE NO.: 2026 - 084845/01
OWNER:
SANNA KOPONEN
JUOKSIJANTIE 4
FI-21110 NAANTALI
FINLAND

NAME/LABEL:
TRUCKER'S OMG
SPECIES: DOG
BREED: CIRNECO DELL'ETNA
SEX: FEMALE
MICROCHIP NO.: 978101085474371
TATTOO NO.: NOT PROVIDED
PEDIGREE NO.: FI39364/26

GENETIC REPORT

SAMPLE: BUCCAL SWAB

SAMPLE TAKEN BY: SANNA KOPONEN

REQUESTED TEST: CIRNECO OCULO-NEUROLOGICAL SYNDROME (CONS)

RESULT: CARRIER (MUT/WT)

COMMENT :

The test examines the presence of AMPD2 gene mutation (c.2131del) described as the cause of Cirneco oculo-neurological syndrome (CONS) in Cirneco dell'Etna. The disease is characterized by degeneration of retinal photoreceptors causing a loss of sight, along with paroxysmal neurological episodes consisting of head tremors and involuntary movements of one of the front limbs. AMPD2 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, 07.10.2026

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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.