

REFERENCE NO.: 2022 - 046653/01

OWNER:

ALEXANDRE KOHLER
IMPASSE DE LA RIETTA 10
CH-1673 RUE
SWITZERLAND

NAME/LABEL:

MIKUNI AKIKO NO KIBOO GO IHOKU HAKURENSOU
SPECIES: DOG
BREED: SHIKOKU
SEX: FEMALE
MICROCHIP NO.: 380260101534747
TATOO NO.: NOT PROVIDED
PEDIGREE NO.: SHSB N° 779058 / NIPPO N° R2-511

GENETIC REPORT

SAMPLE: BUCCAL SWAB

SAMPLE TAKEN BY: OWNER

REQUESTED TEST: CYSTINURIA TYPE III

RESULT: CLEAR (c.574 A/A, c.2092 A/A, c.649 G/G)

COMMENT :

The test examines presence or absence of two SLC3A1 gene mutations (c.574A>G, c.2092A>G) and one mutation in SLC7A9 gene (c.649G>A) described as the cause for cystinuria in English and French bulldog. The disease is characterized by formation of cystine stones in kidneys or bladder. Mutations in SLC3A1 gene are inherited in autosomal recessive manner. The mutation in SLC7A9 gene is inherited in incompletely recessive manner, which means that some heterozygous animals can develop the disease.

Regarding to the presence of tested mutations animals are classified in four groups:

- Clear - mutations are not present (574A/A, 2092A/A, 649G/G), normal genotype
- Carrier at risk - always heterozygous for allele c.649G>A (649G/A); alleles c.574A>G and c.2092A>G can be homozygous without the mutation (574A/A, 2092A/A) or heterozygous (574A/G, 2092A/G);
- Carrier - always homozygous without the mutation for allele c.649G>A (649G/G); one or both of the alleles c.574A>G and c.2092A>G can be heterozygous (574A/G, 2092A/G)
- Affected - one, two or three tested alleles are homozygous for the mutation (574G/G, 2092G/G, 649A/A)

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. Animals classified as carriers at risk might also develop the disease therefore we recommend breeding only with clear animals.

AUTHORIZED SIGNATURE:

MARIBOR, 10.03.2022



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.