

REFERENCE NO.: 2018 - 20302

OWNER:

JENNIFER ZIESERL
ÜBERSBACH 199
AT-8280 ÜBERSBACH
AUSTRIA

NAME/LABEL:

CLAN ABBY DANGER ZONE
SPECIES: DOG
BREED: BORDER COLLIE
SEX: MALE
MICROCHIP NO.: 953010001322295
TATOO NO.: NOT PROVIDED
PEDIGREE NO.: ANKC 3100347128

GENETIC REPORT

SAMPLE: BUCCAL SWAB

SAMPLE TAKEN BY: OWNER

REQUESTED TEST: IMERSLUND-GRASBECK SYNDROME (IGS)

RESULT: CLEAR

COMMENT :

The test examines presence or absence of CUBN gene mutation (c.8392delC) described as the cause of Imerslund-Gräsbeck syndrome (IGS) in Border Collie. The disease is characterized by cobalamin malabsorption that leads to vitamin B12 deficiency and consequently causes dyshematopoiesis, lethargy, failure to thrive, and life-threatening metabolic disruption in juvenile period of life. CUBN gene defect is inherited as an autosomal recessive trait.

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

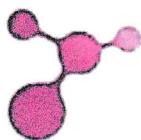
- Clear (wt/wt) - mutation is not present, normal genotype
- Carrier (mut/wt) - one of two alleles carries tested mutation, disease is not clinically manifested
- Affected (mut/mut) - both alleles carry tested mutation, disease is clinically manifested

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, 17.07.2018

Molekularna diagnostika



EVG

Molecular diagnostics

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TATOO NO.: NOT PROVIDED

PEDIGREE NO.: ANKC 3100347128

GENETIC REPORT

SAMPLE: BUCCAL SWAB

SAMPLE TAKEN BY: OWNER

REQUESTED TEST: TRAPPED NEUTROPHIL SYNDROME (TNS)

RESULT: CARRIER

COMMENT :

The test examines presence or absence of VPS13B gene mutation (g.4411950_4411953delGTTT) described as the cause of inherited neutropenia called trapped neutrophil syndrome (TNS) in Border Collie. Consequence of tested mutation is severe neutropenia, which leads to severe life-threatening infections. Trapped neutrophil syndrome 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.

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EVG molekularna diagnostika

EVG d.o.o., Taborska ulica 8, SI-2000 Maribor

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GENETIC REPORT

SAMPLE: BUCCAL SWAB

SAMPLE TAKEN BY: OWNER

REQUESTED TEST: MULTI DRUG RESISTANCE (IVERMECTIN SENSITIVITY, MDR1)

RESULT: CLEAR

COMMENT :

The test examines presence or absence of MDR1/ABCB1 gene mutation (c.295_298del) described as the cause of multi drug resistance (MDR) in several dog breeds. The condition is characterized by increased susceptibility to neurotoxic side effects of several drugs including Ivermectin. MDR1 gene defect 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.

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GENETIC REPORT

SAMPLE: BUCCAL SWAB

SAMPLE TAKEN BY: OWNER

REQUESTED TEST: NEURONAL CEROID LIPOFUSCINOSIS (NCL5)

RESULT: CLEAR

COMMENT :

The test examines presence or absence of CLN5 gene mutation (c.619C>T) described as the cause of neuronal ceroid lipofuscinosis (NCL5) in Border Collie. The disease is characterized by neurodegeneration, which causes psychological abnormalities and ataxia. Neuronal ceroid lipofuscinosis 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.

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GENETIC REPORT

SAMPLE: BUCCAL SWAB

SAMPLE TAKEN BY: OWNER

REQUESTED TEST: COLLIE EYE ANOMALY (CEA)

RESULT: CLEAR

COMMENT :

The test examines presence or absence of NHEJ1 gene mutation (c.588+462_588+8260del7799bp) described as the cause for collie eye anomaly (CEA) in several dog breeds. The disease is characterized by different level of impairment of retina and choroid sclera that occurs during development of the eye. Collie eye anomaly 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.

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GENETIC REPORT

SAMPLE: BUCCAL SWAB

SAMPLE TAKEN BY: OWNER

REQUESTED TEST: MALIGNANT HYPERTHERMIA (MH)

RESULT: CLEAR

COMMENT :

The test examines presence or absence of RYR1 gene mutation (c.1640T>C) described as the cause of malignant hyperthermia (MH) in many dog breeds. The disease is a pharmacogenetic disorder of skeletal muscle elicited by exposure to volatile anaesthetics and depolarizing muscle relaxants. Malignant hyperthermia is inherited as an autosomal dominant trait.

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

- Clear (wt/wt) - mutation is not present, normal genotype
- Single affected (mut/wt) - one of two alleles carries a mutation, disease is clinically manifested
- Double affected (mut/mut) - both alleles carry mutations, disease is clinically manifested

Because of autosomal dominant mode of inheritance the disease is clinically manifested in all animals that carry a mutation (one or both affected alleles). When double positive animal is bred with clear animal all siblings will be single affected with clinical manifestation of the disease. When single positive and clear animals are bred 50% of siblings will be clear and 50% will be single affected. With the aim of disease eradication and prevention of possible animal suffering it is advised to avoid breeding of double affected and single affected animals.

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GENETIC REPORT

SAMPLE: BUCCAL SWAB

SAMPLE TAKEN BY: OWNER

REQUESTED TEST: GONIODYSGENESIS AND GLAUCOMA (GG)

RESULT: CLEAR

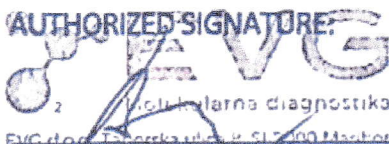
COMMENT :

The test examines presence or absence of OLFML3 gene mutation (c.590G>A) described as the cause of goniodysgenesis and glaucoma (GG) in Border Collie. The disease is characterized by developmental abnormality of eye ligaments that interfere with the flow of the eye fluid and lead to an increased eye pressure and consequently blindness. OLFML3 gene defect 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:

EVG d.o.o. Molekularna diagnostika
Taborska ulica 8, SI-2000 Maribor

MARIBOR, 12.07.2018

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