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REFERENCE NO.: 2022 - 052418/01
OWNER:
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PT-8600 013 LAGOS
PORTUGAL

NAME/LABEL:
ALZAWAIA ALMIR FAMOUS NOBLE
SPECIES: DOG
BREED: DACHSHUND SMOOTH MINIATURE
SEX: MALE
MICROCHIP NO.: 620098102104154
TATOO NO.: NOT PROVIDED
PEDIGREE NO.: LOP570452

GENETIC REPORT

SAMPLE: BLOOD

SAMPLE TAKEN BY: OWNER

REQUESTED TEST: NEURONAL CEROID LIPOFUSCINOSIS 2 (NCL 2)

RESULT: CLEAR (WT/WT)

COMMENT :
The test examines presence or absence of TPP1 gene mutation (c.325delC) described as the cause of neuronal ceroid lipofuscinosis 2 (NCL2) in Dachshund. The disease is characterized by progressive neuronal degeneration and variety of neurological clinical signs. TPP1 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, 29.11.2022

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PEDIGREE NO.: LOP570452

GENETIC REPORT

SAMPLE: BLOOD

SAMPLE TAKEN BY: OWNER

REQUESTED TEST: CHONDRODYSPLASIA, CHONDRODYSTROPHY AND DEGENERATION OF INTERVERTEBRAL DISCS (CDPA/CDDY – IVDD)

RESULT:

CDPA:	CDPA/CDPA
CDDY-IVDD:	CDDY/CDDY

COMMENT :

The test examines presence or absence of two insertions of FGF4 gene on chromosome 18 and on chromosome 12 described as the cause for chondrodysplasia (CDPA), chondrodistrophy (CDDY) and intervertebral disc disease (IVDD) in several dog breeds. The condition is characterized by dysplastic, shortened legs and premature degeneration and calcification of intervertebral discs leading to disc herniation. CDPA is associated with FGF4 gene insertion on chromosome 18 and is inherited in autosomal dominant manner. CDDY is associated with FGF4 gene insertion on chromosome 12 and is inherited in semi-dominant manner (dogs with 2 copies of the mutation have shorter legs than dogs with one copy of the mutation). IVDD is associated with the same FGF4 gene insertion on chromosome 12 as CDDY and is inherited in autosomal dominant manner.
Regarding to the presence of tested mutation for CDPA animals are classified in three groups:

- **N/N** - mutation is not present, normal genotype
- **N/CDPA** - one of two alleles carries tested mutation, shorter legs compared to normal genotype
- **CDPA/CDPA** - both alleles carry tested mutation, shorter legs compared to normal genotype

Regarding to the presence of tested mutation for CDDY-IVDD animals are classified in three groups:

- **N/N** - mutation is not present, normal genotype
- **N/CDDY** - one of two alleles carries tested mutation, shorter legs compared to normal genotype, at risk for IVDD
- **CDDY/CDDY** - both alleles carry tested mutation, shorter legs compared to normal genotype, at risk for IVDD

Because of autosomal dominant mode of inheritance, the CDPA, CDDY and IVDD are manifested in all animals that carry a mutation (one or both affected alleles). When a dog with normal CDPA genotype (N/N) is bred to a heterozygous dog (CDPA/N) 50% of puppies will have shortened legs. When a dog with normal CDPA genotype (N/N) is bred to a dog homozygous for the mutation (CDPA/CDPA) all puppies will have shortened legs. When a dog with normal CDDY genotype (N/N) is bred to a heterozygous dog (CDDY/N) 50% of puppies will have shortened legs, all will be at risk for IVDD. When a dog with normal CDDY genotype (N/N) is bred to a dog homozygous for the mutation (CDDY/CDDY) all puppies will have shortened legs and will be at risk for IVDD.

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PEDIGREE NO.: LOP570452

GENETIC REPORT

SAMPLE: BLOOD

SAMPLE TAKEN BY: OWNER

REQUESTED TEST: OSTEOGENESIS IMPERFECTA (OI)

RESULT: CLEAR (WT/WT)

COMMENT :
The test examines presence or absence of SERPINH1 gene mutation (c.977 T>C) described as the cause for osteogenesis imperfecta (OI) in Dachshund. The disease is characterized by reduction of bone and tooth mass formation (osteopenia and dentinopenia) due to a defect in collagen type I. Collagen type I represents 90% of the organic material of bones, tendons and teeth. While it gives structure and elasticity to these organs, the defects in collagen lead to fragility. Osteogenesis imperfecta 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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PEDIGREE NO.: LOP570452

GENETIC REPORT

SAMPLE: BLOOD

SAMPLE TAKEN BY: OWNER

REQUESTED TEST: CONE-ROD DYSTROPHY 2 (CORD2 - PRA)

RESULT: CLEAR (WT/WT)

COMMENT :
The test examines presence or absence of NPHP4 gene mutation (180 bp deletion in exon 5) described as the cause of cone-rod dystrophy 2 (cord2 - PRA) in wire haired Dachshund. The disease is characterized by a predominant loss of cone function, with relative preservation of rod function. Clinical signs include day blindness and appear between 10 months and 3 years of age. NPHP4 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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PEDIGREE NO.: LOP570452

GENETIC REPORT

SAMPLE: BLOOD

SAMPLE TAKEN BY: OWNER

REQUESTED TEST: NEURONAL CEROID LIPOFUSCINOSIS (NCL 1)

RESULT: CLEAR (WT/WT)

COMMENT :
The test examines presence or absence of PPT1 gene mutation (c.736_737insC) described as the cause of neuronal ceroid lipofuscinosis 1 (NCL1) in Dachshund. The disease is characterized by progressive neuronal degeneration and variety of neurological clinical signs. PPT1 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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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.