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REFERENCE NO.: 2024 - 063432/01
OWNER:
STÉPHANIE LECLERC
IMPASSE LES MALPIERRES 3
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FRANCE

NAME/LABEL:
USHUAIA DES POILS ET CARESSES
SPECIES: DOG
BREED: AUSTRALIAN SHEPHERD
SEX: FEMALE
MICROCHIP NO.: 250269610547789
TATOO NO.: NOT PROVIDED
PEDIGREE NO.: COF1 B.AUS 173162

GENETIC REPORT

SAMPLE: BLOOD

SAMPLE TAKEN BY: PIERRICK DE ROOVER (NO. 32750), DVM

REQUESTED TEST: HEREDITARY CATARACT (HSF4) - AUSTRALIAN SHEPHERD TYPE

RESULT: CLEAR (WT/WT)

COMMENT :

The test examines presence or absence of HSF4 gene mutation (g.85286582delC) described as the cause of primary hereditary cataract (HC) in Australian Shepherd. The disease is characterized by opacity of the crystalline lens that leads to blindness. Tested HSF4 gene defect is inherited as an autosomal dominant trait with incomplete penetrance.

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 a mutation, high probability of clinical manifestation
- Affected (mut/mut) - both alleles carry mutations, disease is clinically manifested

Hereditary cataract in Australian Shepherds has autosomal dominant mode of inheritance with incomplete penetrance. That means it is not developed in every heterozygous animal carrying deleterious mutation. Other genetic or environmental factors cannot be excluded in development of the disease. According to the scientific literature, the probability of developing the disease is 17 times higher in heterozygous animal comparing to clear animal. Carriers pass the mutation to their siblings therefore mating of two carrier animals should be avoided as 25% of puppies will be affected. The test cannot exclude other genetic defects, which may be involved in development of hereditary cataract in Australian Shepherds.

AUTHORIZED SIGNATURE:

MARIBOR, 22.05.2024