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## DNA ANALYSIS PROTOCOL FOR DETECTION OF HEREDITARY DISEASES Protocol No. D2103010145

## Sandrine BERCIER <br> 821 route des bruyeres <br> 38440 Villeneuve de marc, FR <br> Identity of the animal has not been verified.

Sample type: buccal brush
Date of birth:
Sex: -

Date of sampling: 01.03.2021
Date of receipt: $\quad 09.03 .2021$
Date of analysis: $\quad 22.03 .2021$

| Breed/Name | Tattoo or RFID id <br> Pedigree number | Laboratory code | Type of analysis | Result |
| :---: | :---: | :---: | :---: | :---: |
| Australian Shepherd / RUBIS <br> HABANA DE COLLINES DU <br> LAC DE PALADRU | 250268712844454 |  |  | N/N |
|  |  | $210309 /$ X0537 | HSF4/HC | Non-affected |

The results of analysis are stored in a database under the lab code 210309/X0537.

## Hints:

HSF4 (HC) - Hereditary cataract - deletion/insertion 1 bp in exon 9 of HSF4-gene (Heat shock factor protein 4).
HSF4/HC N/N - NON-AFFECTED (NORMAL), Both genes, inherited from both male and female are unaffected.
HSF4/HC N/A - CARRIER, confirmed heterozygous N/A genotype. Mutation can be transmitted to offspring.
HSF4/HC A/A - AFFECTED by the disease.
The HC disorder in Australian Shepherds has an autosomal dominant mode of inheritance, however with incomplete penetrance, the disease may not develop in every carrier of this deletion. The probability that the binocular HC develops in individuals with one copy of deletion (carriers) is approximately 17 times higher than in dogs clear of the deletion mutation (Mellersh et al. 2009).
Notice: This protocol applies exclusively to the sample and the data that were supplied by the submitter. DNA analysis concerns only the abovementioned disease. No information regarding the customer as well as the purpose and results of the analysis will be provided tothint parties.

In Bratislava 22.03.2021

