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DNA ANALYSIS PROTOCOL FOR DETECTION OF HEREDITARY DISEASES

Protocol No. D2103010145

Sandrine BERCIER
821 route des bruyeres
38440 Villenauve de mare

38440 Villeneuve de marc, FR

e de <mark>marc, FR</mark>

Sample type: buccal brush Date of birth: -

Sex: -

Date of sampling: 01.03.2021

Date of receipt: 09.03.2021
Date of analysis: 22.03.2021

Identity of the animal has not been verified.

Breed/Name	Tattoo or RFID id Pedigree number	Laboratory code	Type of analysis	Result
Australian Shepherd / RUB <mark>IS</mark>	250268712844454	87	100	N/N
H <mark>A</mark> BANA DE COLLINES <mark>DU</mark> LAC DE PALADRU		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 above-mentioned disease. No information regarding the customer as well as the purpose and results of the analysis will be provided to third parties.



In Bratislava 22.03.2021

Ing. Marcela Bieliková, PhD.