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## DNA ANALYSIS PROTOCOL FOR DETECTION OF HEREDITARY DISEASES

Submitter of analysis: Sandrine BERCIER Date of sampling: 07.11.2017  
821 route des bruyeres Date of samples receipt: 14.11.2017  
38440 Villeneuve de marc, FR Date of analysis: 15.11.2017

Breed/name	Tattoo or RFID id/ Certificate of origin	Laboratory code	Type of analysis	Result
Australian Shepherd / LENACK		171114/P1912	MDR1	N/N (+/+) Non-affected

The results of analysis are stored in a database under the lab code 171114/P1912.

### Hints:

MDR1 -Multidrug resistance gene - nt230(del4), (autosomal recessive)

MDR1 +/+ or N/N (non-affected): Both genes, inherited from both mother and father are undamaged (healthy).

MDR1 +/- or N/P (carrier): Subjects with confirmed heterozygous genotype are carriers. Defective gene can be transmitted to offspring.

Unwanted side effects are unlikely to occur but cannot be excluded.

MDR1 -/- or P/P (affected): Particular caution is necessary in case an individual is diagnosed MDR1 -/- genotype. Treatment with certain drugs in this case can cause significant problems in some cases lethal neurotoxic reaction.

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 purpose and results of the analysis will be provided to third parties.

In Bratislava 15.11.2017

Ing. Marcela Bielíková, PhD.



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## DNA ANALYSIS PROTOCOL FOR DETECTION OF HEREDITARY DISEASES

Submitter of analysis: Sandrine BERCIER  
821 route des bruyeres  
38440 Villeneuve de marc, FR

Date of sampling: 07.11.2017  
Date of samples receipt: 14.11.2017  
Date of analysis: 16.11.2017

Breed/name	Tattoo or RFID id/ Certificate of origin	Laboratory code	Type of analysis	Result
Australian Shepherd / LENACK		171114/P1912	PRA-pred	N/N non-affected

The results of analysis are stored in a database under the lab code 171114/P1912.

Hints:

PRA-pred - Progressive retinal atrophy (autosomal recessive)

PRA-pred N/N – healthy subject – non-affected. Both genes, inherited from both male and female are unaffected. That means that the subject has both alleles healthy.

PRA-pred N/A – carrier. Subjects with confirmed heterozygous N/A genotype are carriers. Gene mutation can be transmitted to offspring.

PRA-pred A/A – affected subject. The subject is a homozygote with A/A genotype, which inherited the affected allele from both parents and thus is affected by the disease.

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In Bratislava 16.11.2017

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## DNA ANALYSIS PROTOCOL FOR DETECTION OF HEREDITARY DISEASES

Submitter of analysis: Sandrine BERCIER  
821 route des bruyeres  
38440 Villeneuve de marc, FR

Date of sampling: 07.11.2017  
Date of samples receipt: 14.11.2017  
Date of analysis: 22.11.2017

Breed/name	Tattoo or RFID id/ Certificate of origin	Laboratory code	Type of analysis	Result
Australian Shepherd / LENACK		171114/P1912	CEA-EVG	MUT/WT CARRIER

The results of analysis are stored in a database under the lab code 171114/P1912.

### Hints:

CEA - collie eye anomalie (autosomal recessive) - performed by laboratory EVG (MARIBOR)

WT/WT - healthy subject - non-affected. Both genes, inherited from both male and female are unaffected. That means that the subject has both alleles healthy.

MUT/WT - carrier. Subjects with confirmed heterozygous CEA R/r genotype are carriers. Gene mutation can be transmitted to offspring.

MUT/MUT - affected subject. The subject is a homozygote with r/r genotype, which inherited the affected allele from both parents and thus is affected by the disease.

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In Bratislava 22.11.2017

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## DNA ANALYSIS PROTOCOL FOR DETECTION OF HEREDITARY DISEASES

Submitter of analysis: Sandrine BERCIER

821 route des bruyeres

38440 Villeneuve de marc, FR

Date of sampling: 07.11.2017

Date of samples receipt: 14.11.2017

Date of analysis: 05.12.2017

Breed/name	Tattoo or RFID id/ Certificate of origin	Laboratory code	Type of analysis	Result
Australian Shepherd / LENACK		171114/P1912	HSF4/HC	N/A carrier

The results of analysis are stored in a database under the lab code 171114/P1912.

### 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).

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In Bratislava 05.12.2017

Ing. Marcela Bielíková, PhD.