



DNA ANALYSIS PROTOCOL FOR DETECTION OF HEREDITARY DISEASES

Submitter of analysis: Sabina ACHTIG
Grossreichenbach 17
3931 Schweigggers, AT

Date of sampling: 07.04.2015
Date of samples receipt: 15.04.2015
Date of analysis: 27.04.2015

Breed/name	Tattoo or RFID id/ Certificate of origin	Laboratory code	Type of analysis	Result
Australian Shepherd / Diamond Aire's Make My Day	9810 2000 9810 962	150415/L1140	HSF4/HC	N/N Non-affected

The results of analysis are stored in a database under the lab code 150415/L1140.

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

In Bratislava 27.04.2015
Ing. Marcela Bielíková, PhD.



DNA ANALYSIS PROTOCOL FOR DETECTION OF HEREDITARY DISEASES

Submitter of analysis: Sabina ACHTIG
Grossreichenbach 17
3931 Schweigggers, AT

Date of sampling: 07.04.2015
Date of samples receipt: 15.04.2015
Date of analysis: 16.04.2015

Breed/name	Tattoo or RFID id/ Certificate of origin	Laboratory code	Type of analysis	Result
Australian Shepherd / Diamond Aire's Make My Day	9810 2000 9810 962	150415/L1140	MDR1	N/N (+/+) Non-affected

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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.

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In Bratislava 16.04.2015
Ing. Marcela Bielíková, PhD.



DNA ANALYSIS PROTOCOL FOR DETECTION OF HEREDITARY DISEASES

Submitter of analysis: Sabina ACHTIG
Grossreichenbach 17
3931 Schweiggers, AT

Date of sampling: 07.04.2015
Date of samples receipt: 15.04.2015
Date of analysis: 21.04.2015

Breed/name	Tattoo or RFID id/ Certificate of origin	Laboratory code	Type of analysis	Result
Australian Shepherd / Diamond Aire's Make My Day	9810 2000 9810 962	150415/L1140	PRA-prcd	N/N non-affected

The results of analysis are stored in a database under the lab code 150415/L1140.

Hints:

PRA-prcd - Progressive retinal atrophy (autosomal recessive)

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

PRA-prcd 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 21.04.2015
Ing. Marcela Bielíková, PhD.



PROTOCOL OF DNA ANALYSIS FOR DETECTION OF INHERITED DISEASES

Submitter of analysis: Sabina ACHTIG
Grossreichenbach 17
3931 Schweiggers, AT

Date of sampling: 07.04.2015
Date of samples receipt: 15.04.2015
Date of analysis: 27.04.2015

Breed/name	Tattoo or RFID id/ Certificate of origin	Laboratory code	Type of analysis	Result
Australian Shepherd / Diamond Aire's Make My Day	9810 2000 9810 962	150415/L1140	HUU	N/N Non-affected

The results of analysis are stored in a database under the lab code 150415/L1140.

Hints:

HUU - Hyperuricosuria

Hyperuricosuria is an autosomal recessive inherited disease, characterised by excessive excretion of uric acid into the urine, leading to formation of uric acid stones. Hyperuricosuria in dogs is caused by single nucleotide exchange of c.G563T (p.C188F) in SLC2A9 gene (gene for urate transport) (Bannasch et al. 2008)

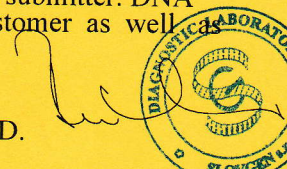
HUU N/N – homozygous individual (non-affected) are genetically clear.

HUU N/A – heterozygous (carrier) are clinically without any symptoms. They are genetically considered carriers of the disease, disease is transmitted to offspring.

HUU A/A – homozygous affected individual (affected)

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In Bratislava 27.04.2015
Ing. Marcela Bielíková, PhD.





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3931 Schweiggers, AT

Date of sampling: 07.04.2015
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Date of analysis: 27.04.2015

Breed/name	Tattoo or RFID id/ Certificate of origin	Laboratory code	Type of analysis	Result
Australian Shepherd / Diamond Aire's Make My Day	9810 2000 9810 962	150415/L1140	MH	N/N Non-affected

The results of analysis are stored in a database under the lab code 150415/L1140.

Hints:

MH- Malignant hyperthermia – autosomal dominant disorder. n dogs, the C >T-substitution was found as the causative mutation leading to substitution of the valine to alanine amino acids at position 547 (p.Val547Ala). Mutation is not bound to specific breeds (Brunson et al. 2004.)

MH 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.

MH N/A – heterozygote – affected. Only one copy of the mutated gene is sufficient for development of clinical signs. In case of affected animal the risk of transfer to the offsprings is 50%.

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In Bratislava 27.04.2015

Ing. Marcela Bielíková, PhD.



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Date of sampling: 07.04.2015
Date of samples receipt: 15.04.2015
Date of analysis: 17.04.2015

Breed/name	Tattoo or RFID id/ Certificate of origin	Laboratory code	Type of analysis	Result
Australian Shepherd / Diamond Aire's Make My Day	9810 2000 9810 962	150415/L1140	CMR1	N/N Non-affected

The results of analysis are stored in a database under the lab code 150415/L1140.

Hints:

CMR1 - Canine Multifocal Retinopathy – is an autosomal recessive eye disorder. This disease is caused by C73T mutation in exon 2 of VMD2 gene. The mutation causes forming of premature stop codon in position 25 (R25X).

CMR1 N/N – homozygous individual non-affected are genetically clear.

CMR1 N/A – heterozygous carrier are clinically without any symptoms. They are genetically considered carriers of the disease, disease is transmitted to offspring..

CMR1 A/A – homozygous affected individual

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In Bratislava 17.04.2015

Ing. Marcela Bielíková, PhD.





Test Report

**Sabina Achtig
Großreichenbach 17
Schweiggen, AT-3931
Austria**

**Optigen Accession #: 15-3461
LBKN#: 1504A15334
Report issued for: Diamond Aire's
Make My Day**

OptiGen Test Certificate

Optigen Accession #: 15-3461

**Test Completed: 04/29/2015
Report Issued: 04/29/2015**

Test Performed: **cd(m) test**

**Result: Normal
Sample Type: DNA - Blood**

Registered Name: **Diamond Aire's Make My Day**

Reg#: OHZB ASH 2355

Breed: **Australian Shepherd**

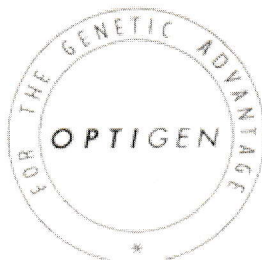
ID#: 981020009810962

Sex: **Male**

Date of Birth: **00/00/**

Owner(s):

Sabina Achtig



Susan Teasdale
OptiGen Authorized Signature

www.optigen.com

Genotype Test Results: Your dog is Normal for the Cone Degeneration (CD) mutation that is known to occur in Alaskan Malamutes, Australian Shepherds and other breeds.

Risk for developing this type of CD: Will never develop this type of CD.

Significance for breeding: Can be bred to any mate and will produce no pups affected by this type of CD.

This interpretation is based on the test result of the DNA test for the specific mutation identified as causing CD (Cone Degeneration) in Alaskan Malamutes, Australian Shepherd and other breeds as of the date on this report.

For further information, please consult the OptiGen website at www.optigen.com.

International DNA Based Genetic Database: To register this result with OFA, make a copy, sign below, mail WITH FEE, to OFA, 2300 E. Nifong Blvd, Columbia, MO 65201-3856 or FAX to 573-875-5073. www.offa.org

I hereby certify that the sample submitted was of the animal described on this application. I authorize the OFA to release all information on the test results thus placing the results in the public domain and I hereby release OFA from any and all liability associated with the release of test information.

Signature of owner or authorized representative: _____

Achtig, Sabina

Zertifikat

über den Gentest auf Neuronale Ceroid Lipofuscinose (NCL)

LABOKLIN-Befund-Nr.: 1504A15334

Hund: Australian Shepherd, männlich,
"Diamond Aire's Make My Day!"

Zuchtbuch-Nummer: ÖHZB ASH 2355

Chip-Nummer: 981020009810962

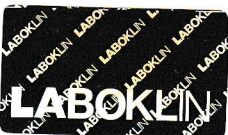
Täto-Nummer: ---

Ergebnis NCL: Genotyp N/N (frei)

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Bad Kissingen, 08-05-2015

Nur gültig mit Originalsiegel



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Schadensersatzverpflichtungen sind, soweit gesetzlich zulässig, auf den Rechnungswert der durchgeführten Untersuchung/en beschränkt.

Achtig, Sabina

Zertifikat

über den Gentest Degenerative Myelopathie (DM - Exon 2)

LABOKLIN-Befund-Nr.: 1504A15334

Hund: Australian Shepherd, männlich,
"Diamond Aire's Make My Day!"

Zuchtbuch-Nummer: ÖHZB ASH 2355

Chip-Nummer: 981020009810962

Täto-Nummer: ---

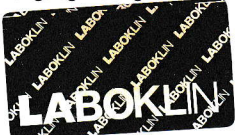
Ergebnis DM - Exon 2: Genotyp N/N (frei)

LABOKLIN

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Bad Kissingen, 08-05-2015

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