# GENETIC TEST INTERPRETATION FORM

Reference No.: 772/2022



### **Owner Information**

Last Name:	Rassl	First name:	Manuela
City:	Wien	Address:	Sickenberggase $7/10$
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Phone:	+43	E-mail:	${f manuela.rassl@chello.at}$

## **Dog** information

Registered Name:	Legendary Team Firestorm Mexx	Registration No.:	Met.Labr 3266/22
Breed:	Labrador Retriever	Microchip No.:	95300 00105 89266
Gender:	Male	Date of Birth:	22.01.2022
Coat Colour/Length:	$\mathbf{Yellow}/\mathbf{Short}$	Sample No.:	772/2022
Sample Information			
Date of sample collection:	17.03.2022	Date of sample processing	: <b>17.02.2023</b>
0 1 1 1 1	11	0 1 1	

Date of sample collection:	17.03.2022	Date of sample processing:	17.02.2023
Sample label:	blue	Sample received as:	EDTA-Blood
Sample taken by (name of vete	erinarian):	Dr. Kagerer Lisa	

### Results

GENETIC TEST(S)	GENOTYPE
B-locus (Loc-B)	$\mathbf{B}/\mathbf{B}$
D-locus: Coat color dilution (Loc-D)	$\mathbf{D}/\mathbf{D}$
Malignant hyperthermia (MH)	N/N
Oculo-sceletal dysplasia (OSD)	N/N
Canine coat lenght (PL)	N/N
Alexander disease (AxD)	N/N
Laryngeal Paralysis and Polyneuropathy (LPPN)	N/N
Achromatopsia (Day blindness) Labrador retriever type (ACHM-L)	N/N
Degenerative myelopathy exon-2 (DM-exon2)	N/N

# Genotype Interpretation Form for Loc-B

### Genotype B/B

The dog carries both copies of the dominant B allele (this dog will always pass on the B allele to any offspring). The dog is black in locus B and does not carry brown colour.

### Genotype $\mathbf{B}/\mathbf{b^c}$ or $\mathbf{B}/\mathbf{b^s}$ or $\mathbf{B}/\mathbf{b^d}$

The dog has one copy of the dominant B allele and one copy of the mutated gene (gene mutation can be transmitted on to half of their offspring). The dog is black in locus B and carries brown colour.

# Genotype $\mathbf{b^c}/\mathbf{b^c}$ or $\mathbf{b^s}/\mathbf{b^s}$ or $\mathbf{b^d}/\mathbf{b^d}$

The dog has two copies of the mutated gene (gene mutation will always be transmitted to any o spring). The dog is brown in locus B.

### Genotype b° b° / bs b° / bd bd

The dog has all three mutated alleles in homozygous form. (All three mutated alleles will be transmitted to all offsprings) The

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	ISAG ID number of the lab: 118494.		
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dog is brown in B locus.

### Genotype $\mathbf{B}/\mathbf{b^c}\ /\ \mathbf{B}/\mathbf{b^s}/\ \mathbf{B}/\mathbf{b^d}$

The dog has three heterozygous alleles. The dog has one copy of the dominant B allele and three copies of the mutated gene (gene mutation can be transmitted on to half of their offspring).

### Genotype B $\mathbf{b^c}/\mathbf{B} \mathbf{b^s}$ or B $\mathbf{b^c}/\mathbf{B} \mathbf{b^d}$ or B $\mathbf{b^s}/\mathbf{B} \mathbf{b^d}$

The dog has two heterozygous alleles. The dog has one copy of the dominant B allele and two copies of the mutated gene (gene mutation can be transmitted on to half of their offsrping).

### Genotype Interpretation Form for Loc-D

### $\mathbf{Genotype} \ \mathbf{D}/\mathbf{D}$

The dog carries both copies of the dominant  $\mathbf{D}$  allele (this dog will always pass on the  $\mathbf{D}$  allele to any offspring). The dog does not carry dilute.

### $Genotype \ D/d$

The dog has one copy of the dominant  $\mathbf{D}$  allele and one copy of the mutated gene (gene mutation can be transmitted on to half of their offspring). The dog is a dilute carrier.

#### $Genotype \; d/d$

The dog has two copies of the mutated gene (gene mutation will always be transmitted to any offspring). The dog has dilute phenotype.

### Genotype Interpretation Form for MH

#### Genotype N/N

The clear dog does not carry the mutation (a clear dog has two copies of the normal gene and will not pass on the mutation to their offspring).

#### Genotype N/A

Mutation that causes MH is inhered as an autosomal dominant trait. Only one copy of the mutated gene is sufficient for development of clinical signs. The carrier dog has one copy of the normal form of the gene and one copy of the mutated gene (this is also referred to as being heterozygous). Gene mutation can be transmitted on to half of their off spring. Inform your veterinarian and consult with him/her regarding test result.

#### Genotype $\mathbf{A}/\mathbf{A}$

The affected dog has two copies of the gene mutation (this is also referred to as being homozygous sick). Affected dogs will pass one copy of this mutation on to all of their offspring and should not be used for breeding. Inform your veterinarian and consult with him/her regarding this test result.

### Genotype Interpretation Form

### Genotype N/N – CLEAR/NON-AFFECTED

The clear dog does not carry the mutation (a clear dog has two copies of the normal gene and will not pass on the mutation to their offspring).

#### Genotype N/A – CARRIER

The carrier dog has one copy of the normal form of the gene and one copy of the mutated gene (this is also referred to as being heterozygous), but it will not exhibit disease symptoms. Carriers will not have medical problems as a result. Gene mutation can be transmitted on to half of their offspring. Inform your veterinarian and consult with him/her regarding test result.

#### Genotype A/A – AFFECTED

The affected dog has two copies of the gene mutation (this is also referred to as being homozygous sick). Affected dogs will pass one copy of this mutation on to all of their offspring and should not be used for breeding. Inform your veterinarian and consult with him/her regarding this test result.

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### Genotype Interpretation Form for PL

#### Genotype N/N

The dog carries both copies of the normal allele (this dog will always pass on the normal allele to any offspring). The dog has short hair.

#### Genotype N/M1

The carrier dog has one copy of the normal form of the gene and one copy of the mutated gene (this is also referred to as being heterozygous). Gene mutation can be transmitted on to half of their offspring. The dog is short-haired. Inform your veterinarian and consult with him/her regarding test result.

### Genotype M1/M1

The dog has two copies of the mutated gene (gene mutation will always be transmitted to any offspring). The dog is long-haired. Inform your veterinarian and consult with him/her regarding this test result.

### Genotype Interpretation Form for AxD

#### $Genotype \ N/N$

The clear dog does not carry the mutation (a clear dog has two copies of the normal gene and will not pass on the mutation to their offspring).

#### $\mathbf{Genotype} \ \mathbf{N}/\mathbf{A}$

Mutation that causes AxD is inhered as an autosomal dominant trait. Only one copy of the mutated gene is sufficient for development of clinical signs. The carrier dog has one copy of the normal form of the gene and one copy of the mutated gene (this is also referred to as being heterozygous). Gene mutation can be transmitted on to half of their off spring. Inform your veterinarian and consult with him/her regarding test result.

#### Genotype A/A

The affected dog has two copies of the gene mutation (this is also referred to as being homozygous sick). Affected dogs will pass one copy of this mutation on to all of their offspring and should not be used for breeding. Inform your veterinarian and consult with him/her regarding this test result.

### Genotype Interpretation Form

### ${\bf Genotype} \ {\bf N/N-CLEAR/NON-AFFECTED}$

The clear dog does not carry the mutation (a clear dog has two copies of the normal gene and will not pass on the mutation to their offspring).

#### Genotype N/A – CARRIER

The carrier dog has one copy of the normal form of the gene and one copy of the mutated gene (this is also referred to as being heterozygous), but it will not exhibit disease symptoms. Carriers will not have medical problems as a result. Gene mutation can be transmitted on to half of their offspring. Inform your veterinarian and consult with him/her regarding test result.

#### Genotype A/A – AFFECTED

The affected dog has two copies of the gene mutation (this is also referred to as being homozygous sick). Affected dogs will pass one copy of this mutation on to all of their offspring and should not be used for breeding. Inform your veterinarian and consult with him/her regarding this test result.

### Notes

Loc-B: presence or absence of the c.121T>A ( $b^c$ ), c.991C>T ( $b^s$ ) and c.1033-1036 delCCT ( $b^d$ ) mutations in TYRP1 gene were determined by sequencing.

Loc-D: presence or absence of the c.-22G>A mutation in MLPH gene was determined by sequencing.

MH: presence or absence of the c.1640T>C mutation in RYR1 gene was determined by sequencing.

OSD: presence or absence of an insG mutation in COL9A3 gene was determined by sequencing.

PL: presence or absence of the c.284G>T mutation in FGF5 gene was determined by sequencing.

AxD: presence or absence of the c.719 G>A in GFAP gene was determined by sequencing.

LPPN: presence or absence of the c.2810 G>Ain CNTNAP Igene was determined by sequencing.

ACHM-L: presence or absence of the deletion mutation c.1931\_1933delTGG in CNGA3gene was determined by sequencing.

DM-exon2: presence or absence of the c.118G>A mutation in SOD1 gene was determined by SNP Genotyping Assay using qPCR.

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