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Ms.
Hilde Viktoria Hagavei
Hagebyen 46
8050 Tverlandet
Norwegen

Report No.: **2311-W-83266**
Date of arrival: 21.11.2023
Date of report: 06.12.2023
Testing started: 21.11.2023
Testing completed: 06.12.2023
Status of the report: Final report

Species:	Cat
Breed:	Ragdoll
Gender:	Female
Name:	(N) Nordlæningen's Trulte av Eugene
Stud book No.:	(NO) NRR LO 212345
Chip No.:	900217000732751
Date of birth / Age:	21.08.2023
Type of sample:	EDTA-Blood
Date sample was taken:	17.11.2023
Sampler:	Trine Braendmo
Owner / Animal-ID:	Hagavei, Hilde Viktoria
IT No. / Report-ID:	---



In genetic testing, we analyse the genetic variants associated with hereditary diseases or genetic traits. The results of these genetic tests always show both alleles of the animal for the variant that has been tested. The symbol "N" indicates the presence of the wild-type allele, while the variant alleles are designated according to the associated diseases (in the example referred to as 'mut').

Possible results:

- N/N: The genetic variant associated with the disease is absent.
- N/mut: The tested animal carries one copy of the analysed variant.
- mut/mut: The tested animal carries two copies of the analysed variant.

It is important to note that solely relying on this genetic information cannot provide definitive insight into whether, when, or to what extent a disease may manifest. For certain diseases, the severity of the condition is influenced by additional factors, some of which are not detectable through genetic testing. Variable penetrance, which involves varying degrees of severity, also frequently plays a role. In cases of recessive hereditary diseases, the disease usually only manifests when an individual possesses two copies of the investigated variant. In contrast, for dominant hereditary diseases, the presence of a single copy of the variant already influences the likelihood of disease occurrence.

For more comprehensive information regarding specific hereditary diseases, please refer to our website.

GENETIC DETERMINATION OF BLOODGROUP - PCR

Result: Genotype N/N

Interpretation: The examined animal is homozygous for the N allele. It does not carry the causative genetic variant found in correlation with the serologic blood group B and AB (C) so far.

The test detects three genetic variants (268T>A, 179G>T, 1322delT) for the alleles b and one variant for c (364C>T).

Allelic series: N>c>b

BREED SPECIFIC VARIANTS

Noticeable results	Genotype	Trait of inheritance	Gene	Variant
FXII deficiency (1631G>C)- PCR	FXII/FXII	autosomal recessive	F12	G>C
Unremarkable results	Genotype		Gene	Variant
Cystinuria - PCR	N/N		SLC7A9	T>A
FXII deficiency (1321delC)- PCR	N/N		F12	INDEL
Hypertrophic Cardiomyopathy (HCM3) Ragdoll - PCR	N/N		MYBPC3	G>A
MDR1 gene variant (MDR) - PCR	N/N		ABCB1	INDEL
Mucopolysaccharidosis type VII (MPS VII) - PCR	N/N		GUSB	G>A
Myotonia congenita - PCR	N/N		CLCN1	G>T
Polycystic kidney disease (PKD) - PCR	N/N		PKD1	C>A
Progressive Retinal Atrophy (rdAc-PRA) - PCR	N/N		CEP290	A>C

BREED NON-SPECIFIC VARIANTS

Unremarkable results	Genotype	Gene	Variant
Acrodermatitis enteropathica (AE) - PCR	N/N	SLC39A4	C>G
Alpha-Mannosidosis (AMD) - PCR	N/N	MAN2B1	INDEL
Autoimmune lymphoproliferative Syndrome (ALPS) - PCR	N/N	FASLG	DUPLI
Factor XI Deficiency - PCR	N/N	F11	G>A
Gangliosidosis (GM1) - PCR	N/N	GLB1	C>G
GM2-Gangliosidosis - PCR	N/N	HEXB	INDEL
Gangliosidosis (GM2) - PCR	N/N	HEXB	INDEL
Glycogen storage disease (GSDIV) - PCR	N/N	GBE1	INDEL
Head Defect - PCR	N/N	ALX1	INDEL
Hypertrophic cardiomyopathy (HCM1) Maine Coon - PCR	N/N	MYBPC3	C>G
Hypertrophic cardiomyopathy (HCM4) Sphynx - PCR	N/N	ALMS1	G>C
Hypokalemia - PCR	N/N	WNK4	C>T
Hypotrichosis/Short Life Expectancy - PCR	N/N	FOXN1	INDEL
Congenital myasthenic syndrom (CMS) - PCR	N/N	COLQ	C>T
Mucopolysaccharidosis type VI (MPS VI) - PCR	N/N	ARSB	A>G, C>T
Osteochondrodysplasie - PCR	N/N	TRPV4	C>A
Primary congenital glaucoma - PCR	N/N	LTBP2	DUPLI
Progressive Retinal Atrophy (PRA-b) - PCR	N/N	KIF3B	C>T

BREED NON-SPECIFIC VARIANTS

Unremarkable results	Genotype	Gene	Variant
Progressive Retinal Atrophy (pd-PRA) - PCR	N/N	AIPL1	C>T
Pyruvatkinase Deficiency:	N/N	PKLR	G>A
Feline Spinal Muscular Atrophy (SMA) - PCR	N/N	LIX1	INDEL

COAT COLORS & COAT CHARACTERISTICS

Genetic test	Genotype	Allelic series
Coat colour brown - PCR	B/B	B dominant over b, b dominant over bl
Coat colour Variant Dilution - PCR	d/d	D dominant over d
Coat colour Variant Agouti - PCR	a/a	A dominant over a
Coat colour Charcoal - PCR	a/a	A dominant over a
Coat colour variant Tabby (Mackerel, Blotched) - PCR - W841X	Tab/Tab	TaM > Tab
Coat colour variant Tabby (Mackerel, Blotched) - PCR - S59X	TaM/TaM	TaM > Tab
Coat colour variant Ticked - PCR - TiA (Cys63Tyr)	N/N	TiA = TiCK > N
Coat colour variant Ticked - PCR - TiCK (Ala18Val)	N/N	TiA = TiCK > N
Coat colour variant Colourpoint - PCR	cs/cs	C dominant over cb, cb dominant over cs
Coat colour variant "Snow" (Bengal) - PCR	cs/cs	C dominant over cb, cb dominant over cs
Coat colour Amber - PCR	E/E	E dominant over e
Coat colour Copal - PCR	E/E	E dominant over ec
Coat colour Russet - PCR	E/E	E dominant over er
Coat colour Variant Gold (Copper) - PCR	N/N	N dominant over wbBSH
Coat Colour Variant Gold (Sunshine) - PCR	N/N	N dominant over wbSib
Coat colour Variant Gold (extreme sunshine) - PCR	N/N	N > wbeSib > wbSib
Coat variant Curly - PCR	N/N	
Hairless/Curly Coat (SPH/DRX) - PCR	N/N	N dominant over hr, hr dominant over re
Coat Length - PCR	M1/M4	

Breeding club discounts were granted for discountable services!

These results are based on the sample material submitted to our laboratory.

This was suitable if not stated otherwise. The submitter is responsible for the accuracy of the information regarding the sample. This report can only be transmitted in toto and unchanged. Doing otherwise requires written permission from Laboklin GmbH & Co. KG.

LABOKLIN is an officially accredited laboratory according to DIN EN ISO/IEC 17025:2018, DAkkS No. D-PL-13186-01-01 and D-PL-13186-1-02. The accreditation applies to all test procedures listed in the accreditation certificate.



Fr. Nadine Gaenstaller
Abt. Molekularbiologie

***** END of report *****



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