LABOKLIN NV, Industriestraat 29, 6433 JW Hoensbroek

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LABORATORIUM VOOR KLINISCHE DIAGNOSTIEK N.V.

Mevr.
Bettie G. Smaling

Utrecht
Nederland

Report No.:	**2406-N-07487**
Date of arrival:	27.06.2024
Date of report:	08.07.2024
Testing started:	27.06.2024
Testing completed:	08.07.2024
Status of the report:	Final report

Species:	cat
Breed:	Siberische Kat
Gender:	female
Name:	Ziazan La Nouba
Stud book No.:	(FI)SK LO 2031032
Chip No.:	985113003333566
Date of birth / Age:	25.10.20
Type of sample:	eNAT
Date sample was taken:	25.06.2024
Owner / Animal-ID:	Smaling, Bettie
IT No. / Report-ID:	---

LABO XXL Genetics

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.

Sample ID: 2406-N-07487

Genetic determination of bloodgroup - PCR

Result: Genotype N/b

Interpretation: The examined animal is heterozygous for one of the causative genetic variants found in correlation with the serologic blood group B so far.

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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

Unremarkable results	Genotype	Gene	Variant
Congenital hypothyroidism (CH) - PCR	N/N	TPO	C-T
Cystinuria - PCR	N/N	SLC7A9	T-A
FXII deficiency (1321delC)- PCR	N/N	F12	DEL
FXII deficiency (1631G>C)- PCR	N/N	F12	DEL
MDR1 gene variant (MDR) - PCR	N/N	ABCB1	DEL
Mucopolysaccharidosis type VII (MPS VII) - PCR	N/N	GUSB	G-A
Myotonia congenita - PCR	N/N	CLCN1	G-T
Polydactyly - Hw variant - PCR	N/N	LMBR1	T-C
Polydactyly - UK1 variant - PCR	N/N	LMBR1	C-G
Polydactyly - UK2 variant - PCR	N/N	LMBR1	T-A
Pyruvatkinase Deficiency:	N/N	PKLR	G-A

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	DEL
Autoimmune lymphoproliferative Syndrome (ALPS) - PCR	N/N	FASLG	INS
Factor XI Deficiency - PCR	N/N	F11	G-A
Gangliosidosis (GM1) - PCR	N/N	GLB1	C-G
GM2-Gangliosidosis - PCR	N/N	HEXB	DEL
Gangliosidosis (GM2) - PCR	N/N	HEXB	DEL
Glycogen storage disease (GSDIV) - PCR	N/N	GBE1	COMPLEX
Head Defect - PCR	N/N	ALX1	DEL
Hypertrophic cardiomyopathy (HCM1) Maine Coon - PCR	N/N	MYBPC3	C-G
Hypertrophic Cardiomyopathy (HCM3) Ragdoll - PCR	N/N	MYBPC3	G-A
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	DEL
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
Polycystic kidney disease (PKD) - PCR	N/N	PKD1	C-A
Primary congenital glaucoma - PCR	N/N	LTBP2	INS
Progressive Retinal Atrophy (PRA-b) - PCR	N/N	KIF3B	C-T
Progressive Retinal Atrophy (pd-PRA) - PCR	N/N	AIPL1	C-T
Progressive Retinal Atrophy (rdAc-PRA) - PCR	N/N	CEP290	A-C
Feline Spinal Muscular Atrophy (SMA) - PCR	N/N	LIX1	COMPLEX

Sample ID: 2406-N-07487

COAT COLORS & COAT CHARACTERISTICS

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Genetic test

	Genotype	Allelic series
Coat colour brown - PCR	B/B	B>b>bl
Coat colour Variant Dilution - PCR	D/D	D>d
Coat colour Variant Agouti - PCR	A/A	A>a
Coat colour Charcoal - PCR	A/A	A>a
Tabby (W841X)	Tab/Tab	TaM > Tab
Tabby (S59X)	TaM/TaM	TaM > Tab
TiA (Cys63Tyr)	N/N	TiA = TiCK > N
TiCK (Ala18Val)	N/N	TiA = TiCK > N
Coat colour variant Colourpoint - PCR	C/C	C>cb>cs
Coat colour variant "Snow" (Bengal) - PCR	C/C	C>cb>cs
Coat colour Amber - PCR	E/E	E>e
Coat colour Copal - PCR	E/E	E>ec
Coat colour Russet - PCR	E/E	E>er
Coat colour Variant Gold (Copper) - PCR	N/N	N>wbBSH
Coat Colour Variant Gold (Sunshine) - PCR	N/wbSib	N>wbSib
Coat colour Variant Gold (extreme sunshine) - PCR	N/N	N>wbeSIB>wbSib
Coat variant Curly - PCR	N/N	Cu>N
Hairless/Curly Coat (SPH/DRX) - PCR	N/N	N>hr>re
Coat Length - PCR	M2/M4	

The current result is only valid for the sample submitted to our laboratory. The sender is responsible for the correct information regarding the sample material.The laboratory can not be made liable. Furthermore, any obligation for compensation is limited to the value of the tests performed.

There is a possibility that other mutations may have caused the disease/phenotype. The analysis was performed according to the latest knowledge and technology.

The laboratory is accredited for the performed tests according to DIN EN ISO/IEC 17025:2018. (except partner lab tests).

Explanations on coat colour genetics

Help for interpreting the genetic variants can be found here: https://shop.labogen.com/coat_colour_genetics_cat

Breeding club discounts were granted for discountable services!

Robin Maes, DVM MSc

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