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LABORATORIUM VOOR KLINISCHE DIAGNOSTIEK N.V.

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Report No.:	**2404-N-03578**
Date of arrival:	02.04.2024
Date of report:	17.04.2024
Testing started:	02.04.2024
Testing completed:	17.04.2024
Status of the report:	Final report

Species:	cat
Breed:	Britse Korthaar
Gender:	female
Name:	Tiny Night Damage
Stud book No.:	NCT A 2022-8528
Chip No.:	528210006824117
Date of birth / Age:	07.05.22
Type of sample:	EDTA
Date sample was taken:	29.03.2024
Owner / Animal-ID:	Winckel van de, Wendy
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.

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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

Unremarkable results	Genotype	Gene	Variant
Autoimmune lymphoproliferative Syndrome (ALPS) - PCR	N/N	FASLG	DUPLI
Cystinuria - PCR	N/N	SLC7A9	T>A
FXII deficiency (1321delC)- PCR	N/N	F12	INDEL
FXII deficiency (1631G>C)- PCR	N/N	F12	G>C
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
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
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 (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	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
Polycystic kidney disease (PKD) - PCR	N/N	PKD1	C>A
Primary congenital glaucoma - PCR	N/N	LTBP2	DUPLI
Progressive Retinal Atrophy (PRA-b) - PCR	N/N	KIF3B	C>T
Progressive Retinal Atrophy (pd-PRA) - PCR	N/N	AIPL1	C>T

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BREED NON-SPECIFIC VARIANTS

Unremarkable results	Genotype	Gene	Variant
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	TaM/Tab	TaM > Tab
Coat colour variant Tabby (Mackerel, Blotched) - PCR - S59X	TaM/TaM	TaM > Tab
Coat colour variant Ticked - PCR - TiA (Cys63Tyr)	N/TiA	TiA = TiCK > N
Coat colour variant Ticked - PCR - TiCK (Ala18Val)	N/N	TiA = TiCK > N
Coat colour variant Colourpoint - PCR	C/cs	C dominant over cb, cb dominant over cs
Coat colour variant "Snow" (Bengal) - PCR	C/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	N/N	

M. Bolumburu, DVM MSc

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