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Inqaba Biotechnical Industries (Pty) Ltd
Animal Genetics Division

Co. Reg. No: 2001/011245/07
VAT No: 4150197251

Hypertrophic Cardiomyopathy

Client Name:	CrazyCoon Maine Coons		ZO2022/3541/20220125/#19510
Client Address:			
Phone:			
Email:			
Profile:		Species:	Felis catus / Feline / Cat
Name:	Y* Xavier Gracia Felina	Microchip #:	900215001295119
Breed:	Maine Coon	Registration #:	KZERK-2124-87273/21
Test:	[MYBPC3 (Maine)] Hypertrophic Cardiomyopathy		
Results:	c.91G>C	GG	CLEAR

Sample Type: Buccal Swab (Cats Only) Extraction Method: DNA Extraction: D4069 Test Type: Genetic Health

[MYBPC3 (Maine)] Hypertrophic Cardiomyopathy

Hypertrophic cardiomyopathy (HCM) is the most common cause of heart failure in felines. HCM has been reported in felines of all ages. This disease is most common to Maine Coons and Ragdolls.

A SNP mutation at c.91G>C in the MYBPC3 gene, has been shown to be associated with HCM in Maine Coons.

HCM is an autosomal dominant disorder that requires one copy of the mutant allele to cause the disease.

References: Meurs, et al., 2005. A cardiac myosin binding protein C mutation in the Maine coon cat with familial hypertrophic cardiomyopathy, Hum. Mol. Genet. 14, 3587-3593.

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Erythrocyte Pyruvate Kinase Deficiency (Fel. PKLR/PKDef)

Client Name:	CrazvCoon Maine Coons		ZO2022/3541/20220124/#19509
Client Address:			
Phone:			
Email:			
Profile:		Species:	Felis catus / Feline / Cat
Name:	Y* Xavier Gracia Felina	Microchip #:	900215001295119
Breed:	Maine Coon	Registration #:	KZERK-2124-87273/21

Test:	[PKLR] Erythrocyte Pyruvate Kinase Deficiency (Fel. PKLR/PKDef)		
Results:	c.693+304G>A	GG	CLEAR

Sample Type: Buccal Swab (Cats Only) Extraction Method: DNA Extraction: D4069 Test Type: Genetic Health

[PKLR] Erythrocyte Pyruvate Kinase Deficiency (Fel. PKLR/PKDef)

Erythrocyte pyruvate kinase deficiency (PK-Def) is a form of hemolytic anaemia caused by mutations in PKLR, the gene encoding the regulatory glycolytic enzyme pyruvate kinase (PK). PK-Def exhibits an inconsistency in onset and severity of symptoms in felids.

The genetic test detects a 13 bp deletion at c.693+304G>A in the PKLR gene. The deletion results in a truncated enzyme.

PK-Def is an autosomal recessive disease which requires two copies of the mutant allele to cause PK-Def.

References: Grahn et al 2012. Erythrocyte Pyruvate Kinase Deficiency mutation identified in multiple breeds of domestic cats. BMC Veterinary Research 8, 207.

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Spinal Muscular Atrophy

Client Name:	CrazyCoon Maine Coons		ZO2022/3541/20220124/#19512
Client Address:			
Phone:			
Email:			
Profile:		Species:	Felis catus / Feline / Cat
Name:	Y* Xavier Gracia Felina	Microchip #:	900215001295119
Breed:	Maine Coon	Registration #:	KZERK-2124-87273/21

Test:	[LIX1] Spinal Muscular Atrophy		
Results:	DEL 140-kb	WT/WT	CLEAR

Sample Type: Buccal Swab (Cats Only) Extraction Method: DNA Extraction: D4069 Test Type: Genetic Health

[LIX1] Spinal Muscular Atrophy

Spinal muscular atrophy is a genetic heterogenous group of disorders defined by the degeneration of motor neurons of the spinal cord and vary by severity and symptoms.

Juvenile onset SMA in Maine Coon cats is caused by a large deletion of approximately 140kb of the LIX1 and LNPEP genes. LIX1 gene function is restricted to the central nervous system; the large deletion disrupts the function of LIX1.

Spinal muscular atrophy exhibits an autosomal recessive pattern of inheritance. The individual requires deletions on both chromosomal regions to present with SMA.

References: Fyfe et al 2006. An ~140-kb deletion associated with feline spinal muscular atrophy implies an essential LIX1 function for motor neuron survival. Genome Research 16, p1084-1090.

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Polycystic Kidney Disease (PKD)

Client Name:	CrazyCoon Maine Coons		ZO2022/3541/20220124/#19511
Client Address:			
Phone:			
Email:			
Profile:		Species:	Felis catus / Feline / Cat
Name:	Y* Xavier Gracia Felina	Microchip #:	900215001295119
Breed:	Maine Coon	Registration #:	KZERK-2124-87273/21

Test:	[PKD] Polycystic Kidney Disease (PKD)		
Results:	c.10063C>A	CC	CLEAR

Sample Type: Buccal Swab (Cats Only) Extraction Method: DNA Extraction: D4069 Test Type: Genetic Health

[PKD] Polycystic Kidney Disease (PKD)

PKD is a kidney disorder commonly diagnosed in humans and Persian cats. PKD is characterised by the development of fluid-filled cysts on the kidneys, pancreas and liver. These cysts can ultimately lead to renal failure.

PKD is caused by the a SNP mutation c.10063C>A in the PKD1 gene.

PKD is an autosomal dominant disease that affects males and females equally. An autosomal dominant disease implies that only one copy of the disease mutation needs to be present in order for the animal to exhibit symptoms.

References: Lyons et al 2004. Feline Polycystic Kidney Disease mutation identified in PKD1. J. Am. Soc. Nephrol. 15, pp 2548 â€“ 2555.

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A Locus (ASIP)

Client Name:	CrazyCoon Maine Coons der Merwe		ZO2022/3541/20220124/#19513
Client Address:			
Phone:			
Email:			
Profile:		Species:	Felis catus / Feline / Cat
Name:	Y* Xavier Gracia Felina	Microchip #:	900215001295119
Breed:	Maine Coon	Registration #:	KZERK-2124-87273/21

Test:	[A Locus (Fel)] A Locus (ASIP)		
Results:	c.123_124delCA	CA/-	Aa
	c.142T>C	TT	AA
	Final Conclusion	Agouti Banded	

Sample Type: Buccal Swab (Cats Only) Extraction Method: DNA Extraction: D4069 Test Type: Genetic Colour

[A Locus (Fel)] A Locus (ASIP)

The agouti signaling peptide (ASIP) is responsible for the production of phaeomelanin, the red pigment and is associated with melanism (black/brown markings) in the feline coat. A homozygous mutation in the ASIP gene causes solid coat colour (non-agouti coat) in felines.

This test detects the absence or presence of the non-agouti (a) deletion (c.123_124delCA) and Asian Leopard Ca (APbe) SNP mutation c.142T>C in exon 2 of the ASIP gene.

a is autosomal recessive and require two copies of the mutation allele(s) to confer solid colour. The charcoal presentation in Bengals is caused by a compound heterozygote (APbe/a) of the leopard cat agouti allele (APbe) and the domestic cat non-agouti melanism allele (a). The relationship between APbe, A and a is not fully understood and further studies are needed to determine the mode of inheritance.

References: Gershony et al 2014. The Asian leopard cats Agouti (ASIP) allele likely affects coat colour phenotype in the Bengal cat breed. Animal Genetics 45, p893-897. Schneider et al 2012. How the leopard hides its spots: ASIP mutations and melanism in wild cats. PLoS ONE 7, e50386.

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Dilution, MLPH (D Locus)

Client Name:	CrazyCoon Maine Coons		ZO2022/3541/20220125/#19508
Client Address:			
Phone:			
Email:			
Profile:		Species:	Felis catus / Feline / Cat
Name:	Y* Xavier Gracia Felina	Microchip #:	900215001295119
Breed:	Maine Coon	Registration #:	KZERK-2124-87273/21
Test:	[D Locus (Fel)] Dilution, MLPH (D Locus)		
Results:	c.83delT T/-		Dd (Carrier)

Sample Type: Buccal Swab (Cats Only) Extraction Method: DNA Extraction: D4069 Test Type: Genetic Colour

[D Locus (Fel)] Dilution, MLPH (D Locus)

Melanophillin (MLPH) aka D Locus is the protein responsible for the transportation of melanin. Melanophillin is encoded by the MLPH gene. A mutation in the MLPH gene reduces the distribution of eumelanin and phaeomelanin resulting in a dilute phenotype.

The test detects the absence or presence of the 1-base pair deletion in exon 2 of the MLPH gene.

The wildtype allele is non-dilute D and will produce an intense colour. The mutant allele d is recessive, therefore the genotype dd is required to produce dilute colour. Colour dilution will present as follows: dilution of black to blue, chocolate to lilac, cinnamon to fawn or red/orange to cream.

References: Ishida et al 2006. A homozygous single-base deletion in MLPH causes the dilute coat color phenotype in the domestic cat. Genomics 88, p698-705.