Inqaba Biotechnical Industries (Pty) Ltd
Animal Genetics Division
Co. Reg. No: 2001/011245/07
VAT No: 4150197251

Hypertrophic Cardiomyopathy (HCM)

Client Name:	CrazyCoon Maine Coons		ZO2024/7058/20240308/#66927
Client Address:			
Phone:			
Email:			
Profile:		Species:	Felis catus / Feline / Cat
Name:	CrazyCoon Yennefer		
Breed:	Maine Coon		
Test:	[MYBPC3 (Maine)] Hypertrophic Cardiomyopathy (HCM)		
Results:	c.91G>C	GG	CLEAR

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

[MYBPC3 (Maine)] Hypertrophic Cardiomyopathy (HCM)

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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Spinal Muscular Atrophy (SMA)

Client Name:	CrazyCoon Maine Coons		ZO2024/7058/20240308/#66928
Client Address:			
Phone:			
Email:			
Profile:	FCA2024/66459	Species:	Felis catus / Feline / Cat
Name:	CrazyCoon Yennefer		
Breed:	Maine Coon		
Test:	[LIX1] Spinal Muscular Atrophy (SMA)		
Results:	DEL 140-kb	WT/WT	CLEAR

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

[LIX1] Spinal Muscular Atrophy (SMA)

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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Erythrocyte Pyruvate Kinase Deficiency (PKLR/PKDef)

Client Name:	CrazyCoon Maine Coons		ZO2024/7058/20240308/#66926
Client Address:			
Phone:			
Email:			
Profile:		Species:	Felis catus / Feline / Cat
Name:	CrazyCoon Yennefer		
Breed:	Maine Coon		
Test:	[Fel-PKLR] Erythrocyte Pyruvate Kinase Deficiency (PKLR/PKDef)		
Results:	c.693+304G>A	GG	CLEAR

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

[Fel-PKLR] Erythrocyte Pyruvate Kinase Deficiency (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). THese mutations lead to instability and loss of red blood cells. PK-Def exhibits an inconsistent onset and severity of symptoms in felids, and clinical signs may include severe lethargy, weakness, weight loss, jaundice, and abdominal enlargement.

The genetic test detects a single nucleotide mutation in intron 5 of the PKLR gene (c.693+304G>A). This mutation introduces a 13bp deletion in liver and blood mRNA at the 3' end of exon 5, which ultimately results in an enzyme truncated by nearly 57%.

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

References: Grahn, R.A., Grahn, J.C., Penedo, M.C.T., Helps, C.R., & Lyons, L.A. (2012). Erythrocyte pyruvate kinase deficiency mutation identified in multiple breeds of domestic cats. BMC Veterinary Research, 8, 207. doi: 10.1186/1746-6148-8-207