

## Maine coon HCM (hypertrophic cardiomyopathy) test

Vizi Barbara			Test number: 11092019-Viz2
<b>Cattery</b> HU*Diamond Star	<b>DOB</b> 14.04.2018	<b>Electronic chip</b>	Report date: 13.09.2019
<b>Breed</b> Maine Coon	<b>Cat</b> Sina's Little Giants Crispy, black silver classic tabby highwhite	<b>Sex</b> ♂	

**Result**

**N/N**

### Result Codes:

**N/N** – Cat is homozygous for a normal type (i.e. carry two normal copies of the *MYBPC3* gene).

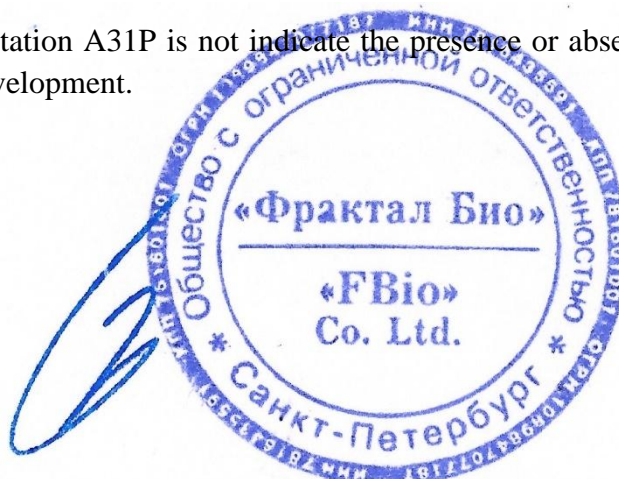
**N/HCMmc** – Cat is heterozygous for the mutant allele (one copy of the *MYBPC3* gene carry the A31P mutation).

**HCMmc/HCMmc** – Cat is homozygous for the A31P mutation (two copies of the *MYBPC3* gene carry the A31P mutation).

This test only detects the A31P mutation associated with HCM in Maine Coon cats and outcrosses as described by Meurset *al.*, 2005.

The presence or absence of the mutation A31P is not indicate the presence or absence of the disease, but only describes the risk of its development.

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**Feline ADPKD test**  
**(autosomal dominant polycystic kidney disease)**

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**Result**

**N/N**

**Result Codes:**

**N/N** – Cat is homozygous for a normal type (i.e. carry two normal copies of the *PKD1* gene).

**N/pkd1** – Cat is heterozygous for the mutant allele (one copy of the *PKD1* gene carry the 10063C>A mutation).

**pkd1/pkd1** – Cat is homozygous for the 10063C>A mutation (two copies of the *PKD1* gene carry the 10063C>A mutation). Homozygote form could be lethal in utero or at a very early age.

This test detects the 10063C>A mutation associated with autosomal dominant polycystic kidney disease in cats as described by Lyons *et al.*, 2004.

The presence of the only one copy of the *PKD1* gene containing the 10063C>A mutation causes ADPKD.

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## Feline PK Deficiency test (Pyruvate Kinase Deficiency in Felines)

Vizi Barbara			Test number: 11092019-Viz2
<b>Cattery</b> HU*Diamond Star	<b>DOB</b> 14.04.2018	<b>Electronic chip</b>	Report date: 13.09.2019
<b>Breed</b> Maine Coon	<b>Cat</b> Sina's Little Giants Crispy, black silver classic tabby highwhite	<b>Sex</b> ♂	

**Result**

**N/N**

### Result Codes:

**N/N** – Cat is homozygous for a normal type (i.e. carry two normal copies of the *PKLR* gene).

**N/k** – Cat is heterozygous for the mutant allele (one copy of the *PKLR* gene carry the mutation).

**k/k** – Cat is homozygous for the mutant allele (two copies of the *PKLR* gene carry the mutation).

This test detects the *c.693+304G>A* mutation associated with pyruvate kinase deficiency in Felines as described by Grahn *et al.*, 2012.

The PKdef disease affects cats with two mutant copies of the *PKLR* gene only. Cats with only one mutant copy of the *PKLR* gene are clinically without any symptoms but are the carriers.

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## Feline SMA test (autosomal recessive spinal muscular atrophy)

Vizi Barbara			Test number: 11092019-Viz2
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<b>Breed</b> Maine Coon	<b>Cat</b> Sina's Little Giants Crispy, black silver classic tabby highwhite	<b>Sex</b> ♂	

**Result**

**N/N**

### Result Codes:

**N/N** – Cat is homozygous for a normal type (i.e. carry two normal copies of the *LIX1-LNPEP* locus).

**N/lix1-lnpep** – Cat is heterozygous for the mutant alleles (one copy of the *LIX1-LNPEP* locus carry the deletion).

**lix1-lnpep/lix1-lnpep** – Cat is homozygous for the mutant alleles (two copies of the *LIX1-LNPEP* locus carry the deletion).

This test detects the 140-kb deletion in the *LIX1-LNPEP* locus associated with autosomal recessive spinal muscular atrophy in domestic cats as described by Fyfe *et al.*, 2006.

The SMA disease affects cats with two mutant copies of the *LIX1-LNPEP* locus only. Cats with only one mutant copy of the *LIX1-LNPEP* locus are clinically without any symptoms.

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