

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

Shirshova			Test number: 02062023-Shi3
<b>Cattery</b> Love Diamante	<b>DOB</b> 20.01.2023	<b>Pedigree</b>	Report date: 06.06.2023
<b>Breed</b> Maine Coon	<b>Cat</b> Felicia, ns 11	<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-Inpep** – Cat is heterozygous for the mutant alleles (one copy of the *LIX1-LNPEP* locus carry the deletion).

**lix1-Inpep/lix1-Inpep** – 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.

Chief of molecular  
diagnostics laboratory  
Bagmanova S. Elena





## Maine coon HCM (hypertrophic cardiomyopathy) test

Shirshova			Test number: 02062023-Shi3
<b>Cattery</b> Love Diamante	<b>DOB</b> 20.01.2023	<b>Pedigree</b>	Report date: 06.06.2023
<b>Breed</b> Maine Coon	<b>Cat</b> Felicia, ns 11	<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).

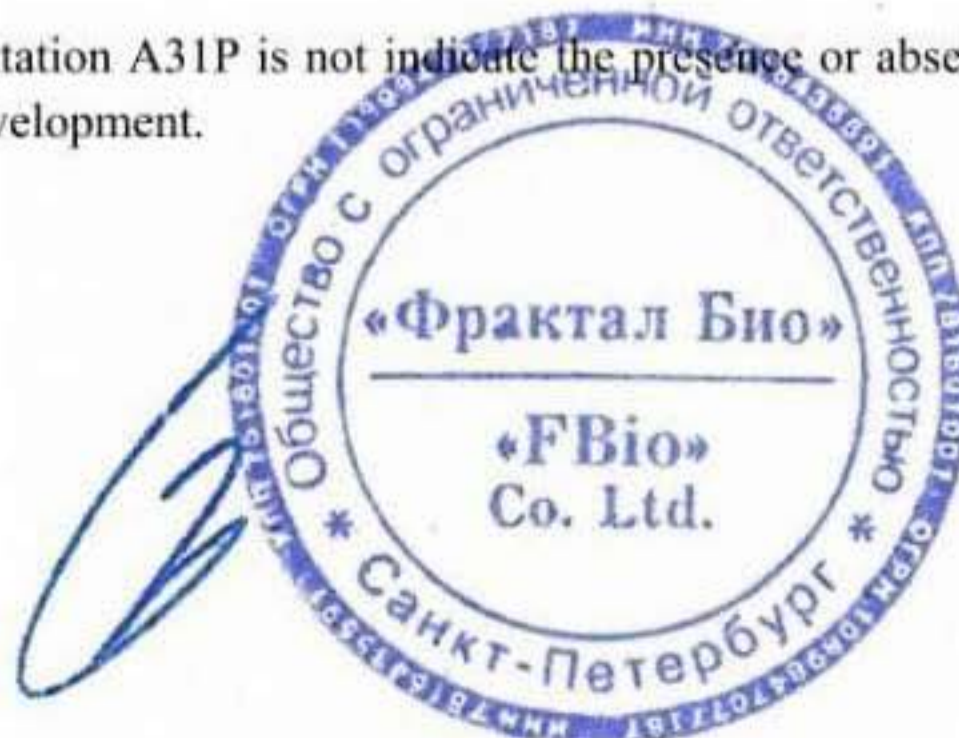
**N/HCMmc** – Cat is heterozygous for the mutant allele (one copy of the *MYBPC3* gene carry the A31P mutation).

**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 Meurs *et 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.

Chief of molecular  
diagnostics laboratory  
Bagmanova S. Elena





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

Natalya Shirshova			Test number: 02062023-Shi3
<b>Cattery</b> Love Diamante	<b>DOB</b> 20.01.2023	<b>Pedigree</b>	Report date: 09.06.2023
<b>Breed</b> Maine Coon	<b>Cat</b> Felicia, ns 11	<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 *PKDI* gene).

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

**pkd1/pkd1** – Cat is homozygous for the 10063C>A mutation (two copies of the *PKDI* 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 *PKDI* gene containing the 10063C>A mutation causes ADPKD.

Chief of molecular  
diagnostics laboratory  
Bagmanova S. Elena

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Исследовательский  
Центр



«FBio» Co. Ltd.  
190020, Russia,  
Saint Petersburg, Bumazhnaya str., 17  
phone: +7 (812)495-96-95  
e-mail: fbiogenetika@mail.ru  
http://vetlaba.ru

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

Shirshova			Test number: 02062023-Shi3
<b>Cattery</b> Love Diamante	<b>DOB</b> 20.01.2023	<b>Pedigree</b>	Report date: 06.06.2023
<b>Breed</b> Maine Coon	<b>Cat</b> Felicia, ns 11	<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.

Chief of molecular  
diagnostics laboratory  
Bagmanova S. Elena

