Исследовательский Центр



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## Feline PK Deficiency test

## (Pyruvate Kinase Deficiency in Felines)

Vizi Barbara			Test number: 21102019-Viz1
Cattery	DOB	Electronic chip	
HU*Diamond Star	07.09.2019		
Breed	Cat	Sex	Report date:
Maine Coon	HU*Diamond Star Reckless Kelly, black silver mc. torbie with white	Ŷ	23.10.2019

## 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).

 $\mathbf{k}/\mathbf{k}$  – 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

