

What is PK Def and why testing is so important?

An article by Danielle, co-owner of Charmedabys Cattery

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Pyruvate Kinase Deficiency is a genetic illness present in some breeds of dogs and cats, especially within the Abyssinian and Somalis. PK is an important factor in the regulation of sugar in the blood stream. A deficiency manifest itself in a lack of energy production by the degradation of sugar in the red blood cells that leads to instability of the red blood cell that results in anaemia. In cats, that anaemia can be intermittent and easily misdiagnosed if testing is not done.

The clinical signs are various and variable. Some animals may develop a severe lethargy, weakness and lack of appetite and thereby lose weight. The mucus membranes will appear pale, they may also experience some abdominal enlargement due to the swelling of the spleen (splenomegaly). PK Def is an autosomal recessive inherited disease where affected animals have 2 mutated genes and carriers that are asymptomatic have one mutant gene and one normal gene.

PK Def (Pyruvate Kinase deficiency) is easily confused with PKD (Polycystic Kidney disease) not known to occur among Abyssinians or Somalis. There are two effective ways to collect DNA for PK Def testing:

Blood sample:

Between 1 and 2 ml of EDTA blood are needed to perform a molecular genetic testing prior to breeding the cat.

Swab sample (2 buccal swabs):

On the website of the California University, there is a little video that shows how you should take the samples. It is easy and less expensive since you can do it yourself at home without the help of the veterinarian. <http://www.vgl.ucdavis.edu/service/cat/index.html>

Which cats should be tested?

Anaemic Abyssinian and Somali cats

Relatives of affected PK def cats

Relatives of carrier cats

Prior to breeding any Abyssinian or Somali cats unless the parents are clear

The possible results are:

normal (clear): doesn't have the mutant gene

carrier: one mutant gene but can't develop the sickness

Affected: two mutants genes, the cat will develop the sickness at one level or an other.

To establish the probability of results during the mating:

Normal (clear) X normal (clear) = normal offsprings (no sick cats)

Normal (clear) X carrier = 50% normal, 50% carriers (no sick cats)

Carrier X carrier = 25% normal, 50% carrier and 25% affected (25% sick cats)

Affected X carrier = 50% carrier and 50% affected (50% sick cats)

Affected X affected = 100% affected (100% sick cats)

Where could we have the tests done?

Pennsylvania University

Dr Urs GIGER/PK

Veterinary Hospital / Room 4006

University of Pennsylvania

3850 Spruce Street

Philadelphia, PA 19104-6010

USA

UC Davis (University of California)

Veterinary Genetics Laboratory

One Shields Avenue

Davis, CA 95616-8744

Phone: (530) 752-2211

Fax : (530) 752-3556

Je donne cependant les références pour les deux labos :

- Laboklin :

Laboklin GmbH & Co KG

z.Hd. Herr Dasch

Prinzregentenstrasse 3

97688 Bad Kissingen

Allemagne

tel +49 971 7202 0

fax +49 971 68 546

email mueller@laboklin.de

<http://www.laboklin.de/>

Please note that this is a partial list of some of the common labs that can perform the DNA testing for PK Deficiency.