

## **Health and welfare information about your cat from Vetlexicon Felis.**



### **Pyruvate kinase deficiency**

Pyruvate kinase deficiency is an inherited disease that was first documented in Abyssinian, Somali and some domestic short-hair cats in the early 1990s. The genetic mutation that results in this disorder has been identified in various breeds of cat, including the Bengal, Egyptian Mau, Savannah, Norwegian Forest Cat, Singapura, Maine Coon, La Perm, and Siberian.

#### **What is Pyruvate kinase deficiency?**

Pyruvate kinase (PK) is an enzyme found in red blood cells. If this enzyme is lacking, the lifespan of the red blood cells is reduced and this results in anaemia (a reduction in the number of red blood cells in the circulation).

#### **What are the signs of PK deficiency?**

The main sign of PK deficiency is anaemia. This is usually mild and develops over a long period of time so that your cat may not show any typical signs. The anaemia may also come and go which may cause further problems with diagnosis.

The clinical signs of anaemia are quite variable. Some animals will appear tired and weak, others will lose their appetite, and lose weight. Some cats may have swollen bellies due to an enlargement of the spleen. Most affected cats cope relatively well, but occasionally a life-threatening anaemia may develop causing lethargy, increased breathing effort or even seizures.

The age of onset of signs is quite variable and may depend on environmental factors. The youngest age at which an affected cat has been diagnosed with anaemia is 6 months of age. The oldest recorded age of diagnosis is 12 years old and in that case the disease was only found when apparently healthy cats were being screened for PK deficiency.

### **What tests will my vet do?**

The diagnostic test for PK deficiency itself is a DNA test which can be performed in a blood sample, or on a sample of cells collected on a cheek swab.

If anaemia is detected then your vet will also want to do some other tests to rule out more common causes. A careful search for acquired diseases, such as infections (*Mycoplasma haemofelis*, FeLV, FIV, and FIP infections), toxins (drugs, onions), and immune mediated haemolytic anaemia may be carried out. Some cats may appear to respond to treatment for *M. haemofelis* and immune disease, however, affected cats have very low PK activity in red blood cells.

### **What causes PK Deficiency?**

PK deficiency is a genetic abnormality that has been recognised in Abyssinians, Somalis, Bengal, Egyptian Mau, La Perm, Maine Coon, Norwegian Forest Cat, Savannah, Siberian, Singapura, and domestic shorthair cats. It is caused by a defective gene and affected cats have 2 copies of this gene. Carrier cats have only one copy of the defective gene and do not show signs of disease. The disease occurs when two carrier cats mate with each other and

produce a kitten with two defective genes (on average one-quarter of the offspring of two carriers will be affected). Affected cats can live to old age so if they are not tested for the disease at an early age they may have produced very many litters of kittens before they are identified as carriers of the disease.

### **Can PK deficiency be treated?**

As it is caused by a genetic mutation, there is no treatment or cure for the underlying condition. However, the episodes of anaemia may be managed.

### **How can PK deficiency be prevented?**

PK deficiency can only be prevented if it is known which cats are affected and which are carriers so that breeding from these cats can be avoided. Cats can be tested for PK deficiency. A blood sample or cheek swab can be submitted for DNA testing.

It is recommended that at risk breeds of cat be tested for the disease before they are used for breeding. It is especially important to test:

- Anaemic Abyssinian and Somali cats.
- Relatives of cats affected by PK deficiency.
- Relatives of cats that are unaffected carriers of the PK deficiency gene.
- Any Abyssinian or Somali cat that is to be bred from unless the parents of this cat have been found to be clear of the PK deficiency gene.

The Governing Council of the Cat Fancy (GCCF), one of the pedigree cat registration bodies in the UK, has now introduced a requirement that all Somali cats registered with the GCCF must be tested for PK deficiency.

For further information, visit the following websites:

<https://icatcare.org/advice/pyruvate-kinase-pk-deficiency/>

<http://www.vgl.ucdavis.edu/services/pkdeficiency.php>  
<http://www.vet.upenn.edu/penngen>  
<https://www.langfordvets.co.uk/diagnostic-laboratories/>