Pyruvate Kinase Deficiency

About the disease

Pyruvate kinase deficiency is an inherited disease of Abyssinian and Somali cats. Pyruvate kinase is a red blood cell enzyme important in red blood cell energy metabolism. Therefore, if this enzyme is lacking, a haemolytic anaemia can result.

However, the anaemia may only be mild and intermittently detectable, or may not become evident until the cat is older. A rapid severe life-threatening anaemia can also develop. The disease is inherited as an autosomal recessive trait, so only cats with two copies of the defective gene are affected.

Carrier cats are clinically healthy but can pass the defective gene to their offspring.

About the test

The Molecular Diagnostic Unit offers a genetic test to diagnose autosomal-recessive pyruvate kinase deficiency (PKDef) in cats. This genetic test is a PCR-based pyrosequencing assay that can reliably distinguish between Affected, Carrier and Normal cats.

It is highly recommended that all Abyssinian and Somali cats used for breeding are tested for the defective gene, as well as cats of these breeds showing signs of haemolytic anaemia. For Somali cats, testing for PKDef is now compulsory if cats are to be registered on the GCCF (Governing Council of the Cat Fancy) active register and used for breeding.

Please note: The test detects the Normal and Mutant PKDef genes found in domestic cats. The test also works in Bengal and Savannah cats, and detects the Asian Leopard Cat or Serval gene if present (i.e. in F1/F2 cats). There is no point in testing Asian Leopard Cats and Servals for PKDef since the mutant gene came from the domestic cat population.

Reception Hours
Mon-Fri 9am - 5pm

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Interpretation of results

A **Normal** autosomal recessive PKDef genetic test result means that the cat does not have the genetic mutation causing pyruvate kinase deficiency.

A **Carrier** autosomal recessive PKDef genetic test result means that the cat has one copy of the mutation. The cat will not have pyruvate kinase deficiency but may pass the mutation to their offspring.

An **Affected** autosomal recessive PKDef genetic test result means that the cat has two copies of the mutation. The cat will have pyruvate kinase deficiency.

Each certificate we issue will specify whether the cat is Normal, Carrier or Affected for the autosomal recessive pyruvate kinase mutation.

FAQs

Can my cat be included on the GCCF active register?

Yes it can! PK Def is now compulsory in some breeds (Asian, Bengal, Somali and Tonkinese) to be registered on the GCCF (Governing Council of the Cat Fancy) active register and used for breeding.

For Inclusion on the register, the sample submitted for PKDef testing MUST be taken by a veterinary surgeon and labelled with the cat's microchip number. You and your vet must complete a submission form, which must be included when your vet sends us the swabs.

The GCCF also require PKDef for cats being 'imported' (i.e. imported to the GCCF from a different registry body) for Australian Mist, Aztec/Ocicat, Bengal, Russian, Somali and Tonkinese.

Which breeds are affected?

Abyssinian	Bengal	Egyptian Mau	La Perm
Maine Coon	Norwegian Forest	Savannah	Siberian
Singapura	Somali	Toyger	Tonkinese

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Australian Mist	Asian	Aztec/Ocicat	Russian

My cat has been diagnosed with PKDef, should I worry?

The resulting anaemia is often mild and intermittently detectable or may not become evident until the cat is older, so there is not usually cause for immediate concern as the cat can adapt to the anaemia and not show any obvious symptoms. Indeed some cats never show clinical signs associated with PKDef. Signs to look out for are lethargy and inappetence.

A rapid severe life-threatening anaemia can also develop but this appears to be relatively rare from the cases we are aware of; in these cases it is possible that stressful situations may predispose to the development of clinical signs.

Although PKDef is congenital, since the anaemia is usually mild and clinical signs may not be obvious, the anaemia may not be noticed until the cat is quite old.

If life threatening anaemia is rare, why should we worry?

Pyruvate kinase deficiency is inherited and although predominantly a problem in the US it is being reported increasingly in cats throughout Australia, New Zealand and Europe, including the UK.

As described above, the clinical signs that develop can be serious and life threatening.

Because initial clinical signs can be mild or go unnoticed, Affected cats may not be identified until after they have had large numbers of offspring.

Carrier cats do not show any clinical signs at all. The disease occurs when two Carrier cats are mated with each other. This is important because a large number of Carrier cats can arise in a population before PKDef is even noticed.

With any genetic disease, by the time the disease becomes an obvious problem within a population it is much more difficult to control and involves a lot more expense and heartbreak.

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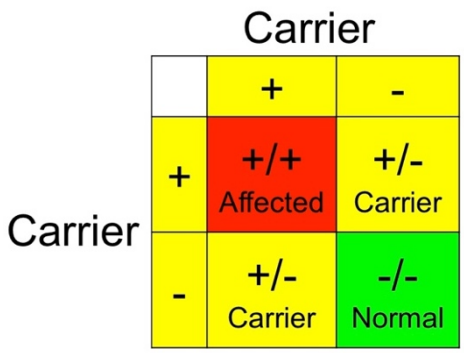
How is PKDef inherited?



Autosomal recessive

The disease is inherited as an autosomal recessive trait, so only cats with two copies of the defective gene are Affected. Clinical signs due to PKDef will only occur in Affected cats, but, as described above, they are variable and some cats will not show signs of PKDef at all. Carrier cats (with only one copy of the defective gene) are clinically healthy and are not affected by the disease, but they can pass the defective gene to their offspring.

My cat is Affected, should I neuter it?

No! It is important to remember that genetic diseases are more likely to arise when the gene pool within a breed is small. This is typically the case in breeds with low numbers of cats that are related and often bred together. If a significant proportion of cats carry the defective gene for PKDef, neutering would only reduce the number available for breeding and, therefore, reduce the gene pool even further. This increases the chances of other genetic diseases arising.

Should we breed from Affected cats?

Whilst it is preferable not to breed from Affected cats in the medium to longer term, in order to preserve the 'good genes' from Affected cats it is permissible to breed Affected cats with Normal cats (no copies of the defective gene). This does not risk producing any Affected kittens, but will produce Carrier cats. These Carrier cats can then be bred to other Normal cats, thereby producing more cats that are Normal and do not carry the defective gene. If this method is used to preserve breeding lines, it is preferable to use Affected male cats where possible as Affected females could become ill during pregnancy with risk of anaemia developing.

Should we stop breeding?

There is no need to stop breeding, but it is important to ensure that you test for PKDef before breeding, and careful breeding programmes will be needed in order to gradually eliminate the disorder from the breed.

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How do we breed safely then?

Strategic and controlled breeding of Affected and Carrier cats to Normal cats is important for preserving the gene pool and retaining important lines.

It is preferable if the Affected cat is a male since a female could become ill during pregnancy.

Cats have two copies of the PK gene, so can be Normal (-/-), Carrier (+/-) or Affected (+/+) depending on how many copies of the defective PK gene (+) they have. Below are shown various matings between Normal, Carrier and Affected cats.

Matings to avoid:

Affected	Affected	+	+
	+	+/+ Affected	+/+ Affected
	+	+/+ Affected	+/+ Affected

Carrier	Affected	+	+
	+	+/+ Affected	+/+ Affected
	-	+/- Carrier	+/- Carrier

Carrier	Carrier	+	-
	+	+/+ Affected	+/- Carrier
	-	+/- Carrier	-/- Normal

Matings that can be safely carried out:

Normal	Normal	-	-
	-	-/- Normal	-/- Normal
	-	-/- Normal	-/- Normal

Normal	Carrier	+	-
	-	+/- Carrier	-/- Normal
	-	+/- Carrier	-/- Normal

Normal	Affected	+	+
	-	+/- Carrier	+/- Carrier
	-	+/- Carrier	+/- Carrier

Information Correct May 2021

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