

PK Def Meeting by Langford Veterinary Services (LVS) at the Veterinary School, Langford, University of Bristol, 31st July 2011

It's good to know that breeders can turn to experts for advice and support when their breed has to face a new problem, and I'm sure Bengal and Singapura people attending that Sunday were grateful to those who gave their time to help. Angie Hibbert, Andrea Harvey, Chris Helps, Kathleen Tennant and Séverine Tasker not only shared their knowledge but also provided the opportunity for discussion and one to one questioning, which I'm sure those with particular concerns found invaluable.

I went as a GCCF Genetics Committee representative, because I've always had an interest in inherited disease and genetic testing since the Korats were the first breed to be given the opportunity to eradicate two forms of a disease fatal to kittens. My intention is to pass new information back to GC colleagues and write this report so it can be shared with those who couldn't make it to the meeting, but who would have liked to be there.

Just in case anyone is concerned, it's not the Genetics Committee's role to tell you what to do. Any of us will certainly help if asked, but breeding choices are still down to breeders, and the clubs and BACs have important supportive roles, plus responsibility in the BAC for developing a registration policy which can include a testing scheme to encourage eradication of the defective gene in cats used for breeding.

My points to remember from the day are:

- Don't panic - you can work through this without necessarily having sick cats
- Don't rush to neuter - careful assessment is needed; other genetic problems can arise if the gene pool is made too small (this reduces genetic diversity) by excessive neutering
- Do test - make future breeding decisions with knowledge of your cats' PK def status
- Do co-operate - share results with other breeders and plan future matings together; and clubs should look at a programme of support and encouragement, possibly with fund raising

What is Pyruvate Kinase Deficiency?

Detailed information from the talks is available to download from the Langford site:

http://www.langfordvets.co.uk/lab_pcr_bengal_singapura_pkdef.htm

It's enough for me to say that PK is an enzyme that maintains the health of red blood cells so that a deficiency can cause their premature breakdown (haemolysis), resulting in anaemia

What does that mean for the cat?

We learned that in some breeds of dog PK Def is a very serious and often fatal disease. In cats the course of PK Def disease is a lot more variable. A significant number of affected cats show no outward signs. Indeed, many cats are not anaemic since their bone marrow is able to compensate for any haemolysis by producing new red blood cells at an increased rate. Others do show signs such as lethargy but what cat doesn't have its 'flaked out' moments, so this could pass unnoticed. There could be pallor of the cat's mucous membranes (a sign of anaemia) or jaundice (yellow discolouration to the mucous membranes due to the release of pigment from haemolysed red blood cells if this is severe). Sometimes clinical support is required for PK Def cats, with blood transfusions and other supportive care. Occasionally excessive pigment release from haemolysed red blood cells can lead to the formation of gall stones which can cause blockage in the cat's bile tracts.

Why DNA test?

To date no sick Bengals or Singapuras have come to the attention of the veterinary scientists.

At present no homozygous Bengals have been found via the LVS testing, and two homozygous Singapuras who have been blood tested for anaemia were completely normal. So perhaps some are thinking that testing is just not worth bothering about because this is something that doesn't necessarily make cats ill. However, the potential for serious consequences exist for any homozygous cat or kitten. Stress and other health problem may cause clinical signs of PK Def to become apparent. As responsible breeders we all want the best for every single kitten who leaves us and PK Def is just so easy to prevent.

As Korat breeders in 1997 we had never seen gangliosidosis here. It would have been so easy to consider it a USA or European problem and bury our heads in the sand and ignore the testing offered. It meant having blood samples taken by a vet, and the DNA had to be extracted at Liverpool before going out to America as no blood products were allowed from UK because of mad cow disease (remember that?). The breed club organised testing in batches - all very complicated, but so very worth it as we proved that it was disease we didn't have and now, by screening imports, we could ensure we would never see it.

What persuaded Korat breeders to test? We had recessive colour genes in the breed and we all knew just how quickly they spread, and popped up in unexpected places, even after intensive pedigree study. We imported for fresh bloodlines and found we still had more of the same. We had proved to ourselves that recessives can go on forever, and so they did until the advent of DNA testing. Since then to the best of my knowledge no Korat kitten has died of gangliosidosis anywhere in the cat fancying world.

[You can so easily ensure that no Bengal or Singapura ever becomes sick with PK Def by all working together and testing prior to breeding. The Somali Cat Club is an excellent example of how this can be achieved, just as we heard that Sunday.](#)

How to test

In the early days a blood sample was required. Now it's a simple matter of cheek swabs, which you do with a little stick similar to a cotton wool bud. You don't need even to open the cat's mouth, just slide in along the inside of its cheek.

http://www.langfordvets.co.uk/laboratory_owners.htm

Dr Lyons always gives the reminder that this should not be done immediately after your cat has eaten even if it's at its most co-operative then. No point in testing tuna cells for PK Def or anything else!

Yes, you'll have to pay for the service, but contact your breed club and you should be given a code for a discount. It is a very modest charge for peace of mind for your kittens, and planning your future breeding, and remember it is short term. Korat breeders (GM) and Somali breeders (PK Def) are now screening imports only as they know via their original testing and their GCCF registration policy that their own cats are clear.

Breeding Choices

http://www.langfordvets.co.uk/lab_pcr_bengal_singapura_pkdef.htm

The genetics of PK Def, testing and results -Chris Helps

This is the presentation to download and study. Note that carriers, and even affected homozygous cats may have to be bred from initially to preserve all bloodlines. Affected males, rather than females, are preferably used in such breeding, as the stress of pregnancy and kitting in queens can worsen PK Def signs. The aim is to avoid producing affected cats, even if initially you are producing more carriers. It doesn't matter as carriers cannot get sick and this is a way of preserving genetic diversity and lines despite the initial presence of PK Def.

For the future

<http://www.somalibac.co.uk/policy.html>

Scroll to the testing scheme to see how a GCCF registration policy can be used as a permanent record of results. If some breeders choose not to test (as is their right) their future progeny will be on the Genetic rather than the Active register as the risk from breeding with a cat with unknown PK Def status is as great (if not more so) as breeding from one who is known to be a carrier or affected. It acts as an alert and also provides an incentive to test.

Additions to the Singapura and Bengal registration policies will be a matter for the respective BACs. The conditions and wording do not have to be exactly as the Somali BAC have decided, but the Genetics Committee is keen to see testing schemes in policies for breeds where tests are available and appropriate.

Jen Lacey