# GENETICS OF BREEDING BENGALS

Tests for genetic and infectious diseases of cats.

Our Cattery tests its producers on:

1. Genetic Diseases
2. Heart Disease

## 1. GENETIC DISEASES

## PK DEF - PYRUVATE KINASE DEFICIENCY

Pyruvate Kinase Deficiency (PK Deficiency) is an inherited hemolytic anemia caused by insufficient activity of this regulatory enzyme which results in instability and loss of red blood cells. The anemia is intermittent, the age of onset is variable and clinical signs are also variable. Symptoms of this anemia can include: severe lethargy, weakness, weight loss, jaundice, and abdominal enlargement. This condition is inherited as an autosomal recessive.

INTERPRETATION OF RESULTS

N/N - Normal no copies of PK deficiency, cat is normal

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

N/K - Carrier 1 copy of PK deficiency, cat is normal but is a carrier.

(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.)

K/K - Affected 2 copies of PK deficiency, cat is or will be affected. Severity of symptoms cannot be predicted.

(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).

## PRA-B -BENGAL PROGRESSIVE RETINAL ATROPHY

Bengal PRA-b causes loss of photoreceptors in the eye and ultimately results in blindness. Clinical signs typically become evident between 8 and 20 weeks of age and the disease progresses so that by around one year of age complete retinal degeneration is apparent in most cats1. At this age the affected cats also show behavioural signs of blindness. Affected cats can have more difficulty at night, their pupils are usually more dilated and they show marked tapetal hyperreflectivity1. As with blind cats in general, PRA-b affected Bengals can negotiate their home environment relatively easily and are mobile and active.

The mutation is inherited as an autosomal recessive trait, meaning that cats with only one copy of the mutant gene (heterozygous or carrier) have normal vision, but they can pass the mutation to their offspring. Cats with two copies of the mutant gene (Affected) will develop PRA.

INTERPRETATION OF RESULTS

N/N - Normal No copies of the PRA-b mutations

(Normal result on the PRA-b genetic assay means that the cat does not have the PRA-b genetic mutation. It is possible that some cats may go on to develop retinal atrophy due to other, as yet unidentified, genetic mutations)

N/PRA - Carrier - 1 Copy of the PRA-b mutations; vision will be normal (Carrier result on the PRA-b genetic assay means that the cat carries one copy of the PRA-b genetic mutation. This cat is a carrier of PRA-b and will not develop retinal atrophy due to the PRA-b mutation, but can pass the mutation to its offspring. It is possible that some cats may go on to develop retinal atrophy due to other, as yet unidentified, genetic mutations)

PRA/PRA - Affected – 2 copies of the PRA-b mutations; cat will develop clinical signs of Bengal PRA

(An Affected result on the PRA-b genetic assay means that the cat has two copies of the PRA-b genetic mutation and will be affected by Bengal PRA)

Each certificate we issue will specify whether the cat is Normal, Carrier or Affected for the Bengal PRA-b mutation.

The gene test will help breeders decide whether or not to use cats for breeding. Generally Affected cats should not be used for breeding because they are certain to pass on the genetic mutation. There is a 25% probability of two Carrier cats producing Affected kittens. Breeding Carrier and Normal cats will produce around 50% Normal and 50% Carrier kittens.

## PKD - POLYCYSTIC KIDNEY DISEASE

Polycystic Kidney Disease - PKD. This is a progressive genetic disease.

PKD is characterized by the formation of multiple cysts in both kidneys. The progressive nature of the disease is manifested in the fact that cysts, hardly noticeable at birth, increase with age, filling the entire kidney.

The disease manifests itself at the age from 3 to 10 years old, average at 7 years old. Until this age, the disease cannot show itself at all and cannot be detected during ordinary examination. At kittens and young cats, cysts are quite small and can vary in size from 1 mm to 1 cm or more. With age, their size and quantity increases up to the complete replacement of the normal structure of the kidney with various calibrated cavities.

The reduction of kidneys function is expressed by symptoms of chronic renal failure (CRF). The intoxication of the organism is developed - poisoning with the products of metabolism, especially with urea. Clinically, CRF is characterized by loss of appetite, lethargy and depressed state of the cat, progressive weight loss, oliguria (decrease of daily urine amount). Vomiting is often observed. These symptoms can be manifested with different frequency and intensify as the disease develops.

One of the main issues associated with PKD is the impossibility to predict which cat with positive PKD will live full life, and which one will die at an early age. So far, scientific researches are not able to say why the disease progresses rapidly at some cats, and slowly at others.

Unfortunately, at present there is no specific treatment of this genetically determined disease. The disease will progressively develop in any case and will lead to the death of the animal from CRF.

The effective method of diagnosis is ultrasound scan (ultrasound study). Diagnosis of PKD is real at the earliest stages of the disease, already at the eight-week-old kitten.

At the age of 8-10 months, renal cysts are well seen. The most reliable and highly effective (98% of cases of correct detection of PKD) is diagnosis at the age of 10 months. Thus, ultrasound scan of the kidneys should be included in the compulsory examination before breeding cats.

Ideally, only PKD-negative male and female cats are allowed for breeding.

## 2. HEART DISEASE HCM - HYPERTROPHIC CARDIOMYOPATHY

What is Feline Hypertrophic Cardiomyopathy (HCM)?

Hypertrophic Cardiomyopathy, HCM is a heart disease characterizing by thickening (hypertrophy) of papillary muscles and the walls of the predominantly left ventricle. The consequence of walls hypertrophy is the decrease of the diastolic volume of the ventricle that leads to the violation of the cardiac contractility. HCM is one of the most common heart diseases that causes sudden death among young cats, and in most cases has hereditary nature.

The cat suffering from HCM can have an active way of life up to irreversible changes in the heart muscle. Therefore, HCM is associated with a high risk of sudden death and diseases such as heart failure and arterial thromboembolism. The main signs of HCM are difficult breathing, fainting, apathy and lethargy of the animal, heart palpitations, at late stages of disease - accumulation of fluid in the thoracic and abdominal cavities, pulmonary edema. Also it may happen paralysis of the hind limbs due to clotting of arteries with a thrombus. Hypertrophy of the heart walls can be caused by the presence of another disease - arterial hypertension (high blood pressure). In this case, there is a non-hereditary secondary HCM.

With the help of echocardiography method (ECG) the hypertrophy of the ventricular wall can be diagnosed from the first year of life.

HCM treatment of cats is medicamentous and directly depends on the timeliness of the diagnosis. Mostly, ACE blockers and selective beta 1- adrenoblockers are used. In case of development of thromboembolism additionally antiplatelet agents are included, at the late stages of the disease a diuretic is prescribed for the removal of edemas.