

Pyruvate Kinase (PK) Deficiency

Description:

Pyruvate kinase (PK) is an enzyme critical to the anaerobic glycolytic pathway of energy production in the erythrocyte. If erythrocytes are deficient in PK they are unable to sustain normal cell metabolism and hence are destroyed prematurely. This deficiency manifests as an hemolytic anemia of variable severity with a strong regenerative response. In dogs, the anemia is always severe (PCV 10-20%) whereas in cats the anemia shows a regenerative response. Also associated with the disease in dogs but not cats is a progressive myelofibrosis and osteosclerosis of unknown etiology and this feature, along with liver failure, is the major cause of death in affected dogs.

The life expectancy of affected dogs is shortened and most die before 4 years of age.

PK deficiency has been recognized in both dogs and cats. The dog breeds involved are the Basenji, Beagle, Dachshund, Eskimo, West Highland White Terriers and the Beagle. In cats, PK deficiency has been described in Abyssinian and Somali cats, as well as DSH cats. The feline disease differs from the canine disease in that affected cats can have a normal life span, only intermittently have anemia, and do not seem to develop either osteosclerosis or liver failure. In all breeds the disease is inherited as an autosomal recessive condition. Heterozygotes (carriers) do not have any clinical signs of disease and lead normal lives. They are able to propagate the mutant allele throughout the population however and it is therefore important that carrier animals be detected prior to breeding.

PK deficiency can be detected, using molecular genetic testing techniques, in the Basenji, Beagle, Dachshund, Eskimo, West Highland White and Cairn Terriers and the Beagle. These tests identify both affected and carrier animals.

Clinical Signs:

The clinical signs of disease reflect the anemic status of the animal and include exercise intolerance, weakness, heart murmur and splenomegaly. The anemia is macrocytic, hypochromic and highly regenerative in dogs. Radiographs reveal generalized abnormalities in bone density including intramedullary mineralisation of the long bones suggestive of progressive osteosclerosis in dogs.

Affected Breeds:

Basenji, Beagle, Eskimo, Chihuahua, Dachshund, West Highland White & Cairn Terrier
Abyssinians, Somalis, DSH

Required Samples:

EDTA Blood, 1-2 mL or 2 Buccal Swabs

Cost:

Please see PennGen Price List for current DNA Test pricing

*To submit a sample for PK testing please use our **DNA Submission Form***