Pyruvate Dehydrogenase Phosphatase 1 (PDP1) Deficiency

In brief

Pyruvate dehydrogenase phosphatase 1 (PDP1) deficiency is an inherited metabolic disorder encountered in the Clumber Spaniel and the Sussex Spaniel. This disorder is due to a deficit of the PDP1 enzyme which is normally involved in the activation of the pyruvate dehydrogenase complex in cellular metabolism. PDP1 deficiency is characterised by exercise intolerance and lactic acidosis, but affected dogs can ease symptoms by resting. The disorder is inherited as an autosomal recessive trait.

Clinical overview

PDP1 deficiency affects the energy metabolism of the muscles. Physical exercise causes lactic acid build up leading to low blood pH and lactic acidosis. Affected dogs suffer from exercise intolerance and they may collapse during physical activity. Affected dogs can ease the symptoms of PDP1 deficiency by resting. The exercise intolerance is usually detected in the first year of a dog’s life.

References

Online database
Online Mendelian Inheritance in Animals, OMIA (http://omia.angis.org.au/), Faculty of Veterinary Science, University of Sydney.

Scientific articles

Disease severity
Mild

Clinical signs
- Exercise intolerance
- Lactic acidosis

Mode of Inheritance
Autosomal Recessive

Results of the genetic test are reported as follows:
- Clear
- Carrier
- At risk

Mutation(s) found in:
Clumber Spaniel
Sussex Spaniel