

# VETERINARY TECHNICAL DATASHEET

Pyruvate Dehydrogenase Phosphatase 1 (PDP1) Deficiency



Mutation Found In :Clumber Spaniel, Sussex Spaniel

## Disorder Type

- Metabolic

## Disease Severity

- Mild/moderate

## Background

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. The disorder is inherited as an autosomal recessive trait.

## Key Signs

- Exercise intolerance
- Lactic acidosis

## Clinical Description

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.

## Mode of Inheritance

- autosomal recessive

## Gene Name

- PDP1

## Next Steps

Therapy is limited to symptomatic treatments and general supportive care. There is no cure.

## References

Cameron JM, Maj MC, Levandovskiy V, MacKay N, Shelton GD, Robinson BH. Identification of a canine model of pyruvate dehydrogenase phosphatase 1 deficiency. *Mol Genet Metab* 90:15-23, 2007.