

Pyruvate Dehydrogenase Phosphatase Deficiency (PDP1)

Affected breeds: Clumber Spaniel, Sussex Spaniel

PDP1 results in exercise intolerance and post-exercise collapse resulting from an inherited defect causing elevated lactate and pyruvate concentrations.

PDP1 is caused by a recessive genetic mutation. This means that dogs which carry the mutation ("CARRIERS") are normal but will pass the mutation on to an average of 50% of their offspring. Puppies which inherit two copies of the mutation will develop PDP1 ("AFFECTED").



This test is particularly useful for breeders:

- To identify carriers among their breeding stock so that they can avoid CARRIER X CARRIER mating combinations which would risk AFFECTED puppies.
- To conclusively confirm PDP1 in an affected dog

This test will be reported as:

CLEAR : no evidence of the PDP1 mutation

CARRIER : carries one copy of the defect, which will be passed to 50% of offspring

AFFECTED : carries two copies of the defect, causing PDP1

The genetic status of dogs can be used to predict breeding outcomes when different combinations are mated:

CLEAR X CLEAR = 100% CLEAR

CARRIER X CLEAR = 50% CARRIER, 50% CLEAR

CARRIER X CARRIER = 25% AFFECTED, 50% CARRIER, 25% CLEAR

References

Identification of a canine model of pyruvate dehydrogenase phosphatase 1 deficiency
Jessie M. Cameron, Mary C. Maj, Valeriy Levandovskiy, Neviana MacKay, G. Diane Shelton, Brian H. Robinson. Molecular Genetics and Metabolism Volume 90, Issue 1, January 2007, Pages 15-23