

## Primary Ciliary Dyskinesia (PCD)

**Affected breeds:** Old English Sheepdog

Primary Ciliary Dyskinesia (PCD) is an inherited condition characterised by recurrent infections of the respiratory tract and reduced fertility in males. It is caused by reduced motility of the cilia on the surface of the respiratory tract and the flagella that propel sperm cells.

PCD 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 PCD ("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 PCD in an affected dog

### **This test will be reported as:**

**CLEAR** : no evidence of the PCD mutation

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

**AFFECTED** : carries two copies of the defect, causing PCD

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

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

**CARRIER X CLEAR** = 50% CARRIER, 50% CLEAR

**CLEAR X CLEAR** = 100% CLEAR

### **References**

Merveille AC, Davis EE, Becker-Heck A, et al. CCDC39 is required for assembly of inner dynein arms and the dynein regulatory complex and for normal ciliary motility in humans and dogs. Nat Genet. 2011;43(1):72–78.