

Primary Lens Luxation

Affected breeds: Australian Cattle Dog, Chinese Crested Dog, Jack Russell Terrier, Lancashire Heeler, Miniature Bull Terrier, Parson Russell Terrier, Patterdale Terrier, Sealyham Terrier, Tibetan Terrier, Welsh Terrier, Wire-haired Fox Terrier, Yorkshire Terrier

Primary lens luxation (PLL) is an inherited eye disease which leads to blindness. In affected dogs, the tiny fibres which hold the lens in place in the eyeball start to break down and the lens can fall out of position and cause glaucoma and blindness. This can be extremely painful. Typically an affected dog would develop signs of PLL at 3 - 8 years of age.

The condition is caused by an inherited mutation in a gene which is required for the healthy construction of the lens fibres. A puppy which inherits the mutation from both of his parents is extremely likely to develop PLL. His parents, on the other hand, who each have only one copy of the mutation, are at a much reduced risk of developing PLL; this is estimated at 2 - 20%.

This test is particularly useful for breeders:

- To identify CARRIER and AFFECTED individuals among their dogs; these individuals should be regularly examined by a veterinary ophthalmologist.
- To identify CARRIER and AFFECTED individuals among their breeding stock so that they can avoid mating combinations which would risk AFFECTED puppies.

This test will be reported as:

CLEAR : free of the PLL mutation, and unlikely to develop primary lens luxation. The possibility remains of developing lens luxation due to a non-inherited cause such as trauma.

CARRIER : has one copy of the mutation and has a low risk of developing PLL during their lifetime; this is estimated at 2 - 20%.

AFFECTED : has two copies of the mutation and is at extremely high risk of PLL.

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
CLEAR X AFFECTED	= 100% CARRIER
CARRIER X AFFECTED	= 50% AFFECTED, 50% CARRIER
AFFECTED X AFFECTED	= 100% AFFECTED

Due to the high levels of carriers in many breeds, carriers should not be excluded from mating - this approach would reduce the genetic diversity within the breed and increase the risk of new inherited diseases.

References:

Farias FHG et al., (2010) An ADAMTS17 splice donor site mutation in dogs with primary lens luxation. *IVOS* 51(9): 4716-4721

Gould D et al., (2011) ADAMTS17 mutation associated with primary lens luxation is widespread among breeds. *Vet Ophthalmol* 14(6): 378-384