

Retinopathy

Affected breeds: Swedish Vallhund

This is one of a range of progressive inherited degenerative retinal diseases found in dogs. This type of condition always affects both eyes and almost always leads to blindness. The age of on-set of this particular condition, which is so far found exclusively in Swedish Vallhunds, is variable.



Retinopathy 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 retinopathy ("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 retinopathy in an affected dog

This test will be reported as:

CLEAR : no evidence of the retinopathy mutation

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

AFFECTED : carries two copies of the defect, causing retinopathy

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

AFFECTED X AFFECTED	= 100% AFFECTED
AFFECTED X CARRIER	= 50% AFFECTED, 50% CARRIER
CARRIER X CARRIER	= 25% AFFECTED, 50% CARRIER, 25% CLEAR
AFFECTED X CLEAR	= 100% CARRIER
CARRIER X CLEAR	= 50% CARRIER, 50% CLEAR
CLEAR X CLEAR	= 100% CLEAR

References

Everson R, Pettitt L, Forman OP, et al. An intronic LINE-1 insertion in MERTK is strongly associated with retinopathy in Swedish Vallhund dogs. PLoS One. 2017;12(8):e0183021. Published 2017 Aug 16. doi:10.1371/journal.pone.0183021