

Hereditary Cataract (Australian Shepherd type)

Affected breeds:

Australian Shepherd

Cataracts lead to blindness. They are caused when proteins in the lens of the eye change and become opaque, restricting the amount of light entering the eye and reaching the light-sensitive retina. Cataracts can either be inherited or age related. The mutation identified in the Australian Shepherd is associated with a 12 times increased risk of developing cataracts. The age at which these develop is 2–7 years.



Hereditary Cataract (Australian Shepherd type) is caused by a dominant genetic mutation. This means that dogs which have either one or two copies of the mutation are at increased risk of cataracts. It may be that dogs with two copies of the mutation are likely to develop cataracts at an earlier age than those with only one.

This test is particularly useful for breeders:

- To identify dogs which carry none, one or two copies of the mutation so that sensible breeding plans can be made.

This test will be reported as:

CLEAR : no evidence of the HC mutation
1 COPY : carries one copy of the defect – the defect will be passed to 50% of offspring
2 COPIES : carries two copies of the defect – the defect will be passed to all offspring

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

2 COPIES X 2 COPIES	= 100% 2 COPIES
2 COPIES X 1 COPY	= 50% 2 COPIES, 50% 1 COPY
2 COPIES X CLEAR	= 100% 1 COPY
1 COPY X 1 COPY	= 25% 2 COPIES, 50% 1 COPY, 25% CLEAR
1 COPY X CLEAR	= 50% 1 COPY, 50% CLEAR
CLEAR X CLEAR	= 100% CLEAR

References

Identification of mutations in HSF4 in dogs of three different breeds with hereditary cataracts. Mellersh CS, Pettitt L, Forman OP, Vaudin M, Barnett KC. *Vet Ophthalmol.* 2006 Sep-Oct;9(5):369-78.

Mutation in HSF4 associated with early but not late-onset hereditary cataract in the Boston Terrier. Mellersh CS, Graves KT, McLaughlin B, Ennis RB, Pettitt L, Vaudin M, Barnett KC. *J Hered.* 2007;98(5):531-3. Epub 2007 Jul 4.

Mutation in HSF4 is associated with hereditary cataract in the Australian Shepherd. Mellersh CS, McLaughlin B, Ahonen S, Pettitt L, Lohi H, Barnett KC. *Vet Ophthalmol.* 2009 Nov-Dec;12(6):372-8. doi: 10.1111/j.1463-5224.2009.00735.x.