

## Hereditary Cataract (HSF4)

### Affected breeds:

Boston Terrier, French Bulldog, Staffordshire Bull Terrier

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 type of cataract identified by this test is an Early Onset Hereditary Cataract (EHC), sometimes also referred to as Juvenile Hereditary cataract (JHC). The cataracts develop during the first year of life, symmetrically in both eyes and progressively lead to blindness in the young dog.



Hereditary Cataract (HSF4) 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 suffer from Early-Onset Hereditary Cataract (HSF4) ("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 Hereditary Cataract (HSF4)

### This test will be reported as:

**CLEAR** : no evidence of the Hereditary Cataract (HSF4) mutation

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

**AFFECTED** : carries two copies of the defect, and will develop Hereditary Cataract (HSF4)

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

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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.