

Pituitary Dwarfism

Affected breeds: German Shepherd, White Swiss Shepherd Dog

Pups with Pituitary Dwarfism are growth retarded due to a defect in the development of the pituitary gland. In addition to their small stature they have many hidden problems including kidney, liver, cardiovascular and neurological defects. They also develop alopecia (baldness) and are prone to skin infection, giving them a scruffy appearance. Their life expectancy is poor, many only living to 4-5 years. Given their many health problems, affected pups should not be viewed as desirable, and their breeding should be avoided.



Pituitary Dwarfism 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 Pituitary Dwarfism ("AFFECTED").

By identifying carrier dogs (estimated to form about 10% of the population), breeders can use this test to prevent breeding affected puppies.

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 Pituitary Dwarfism in an affected pup

This test will be reported as:

CLEAR : no evidence of the Pituitary Dwarfism 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 Pituitary Dwarfism

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

Test details:

Gene:LHX3 Chromosome:9 Variant detail: p.(N182dup)

References

Voorbij AM, van Steenbeek FG, Vos-Loohuis M, Martens EE, Hanson-Nilsson JM, van Oost BA, Kooistra HS, Leegwater PA. A contracted DNA repeat in LHX3 intron 5 is associated with aberrant splicing and pituitary dwarfism in German shepherd dogs. PLoS One. 2011;6(11):e27940.

Turnaround:

Tests marked * are performed by a partner lab in the USA due to patents in force in the UK; the turnaround for these tests is approximately 3 weeks.

Coat colour panels marked ** are performed by VGL in the USA; the turnaround for these tests is approximately 5 weeks.