Skeletal Dysplasia 2 (SD2)

Affected breeds: Labrador retriever

Skeletal dysplasia 2 is a mild form of dwarfism in Labrador retrievers in which affected dogs have short legs when compared to their body length and width. Unlike some other forms of dwarfism, there do not seem to be any obvious secondary health problems (such as joint defects or eye defects) associated with the condition.

Specifically this condition appears in working lines of the Labrador retriever.

Affected male dogs typically have shoulder heights of less than 55cm, and females less than 50cm, while the breed standard calls for heights of 56-57cm in males and 54-56cm in females respectively. However, the height of affected dogs is quite variable, and there is an overlap in height between small clear dogs and the taller of the affected dogs.

SD2 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 have this short-legged appearance ("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 Skeletal Dysplasia 2

This test will be reported as:

CLEAR : no evidence of the Skeletal dysplasia 2 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 have Skeletal dysplasia 2

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

References

Frischknecht M, Niehof-Oellers H, Jagannathan V, Owczarek-Lipska M, Drögemüller C, et al. (2013) A *COL11A2* Mutation in Labrador Retrievers with Mild Disproportionate Dwarfism. PLoS ONE 8(3): e60149. doi:10.1371/journal.pone.0060149