

# VETERINARY TECHNICAL DATASHEET

Skeletal Dysplasia 2, (SD2)



Mutation Found In :Labrador Retriever

## Disorder Type

- Skeletal

## Disease Severity

- Mild

## Background

Skeletal dysplasia (SD2) is an abnormality of skeletal development that causes mild disproportionate dwarfism or short-leggedness in Labrador Retrievers. The mutation has so far been described in working lines of this breed. The mode of inheritance is autosomal recessive.

## Key Signs

- Mild disproportionate dwarfism

## Clinical Description

Affected dogs have shorter limbs, but otherwise normal build. Forelegs are usually slightly more affected than hind legs. Shoulder height is lower (<50 cm) compared to the international breed standard (54-57 cm), which can be observed after the dog's growth period is finished. In contrast to other skeletal dysplasias, no auditory problems, deafness, or secondary joint problems are associated with SD2. Because the mutation is superimposed on the normal variation seen in the breed, it can be difficult to identify the trait in some individuals.

## Mode of Inheritance

- autosomal recessive

## Gene Name

- COL11A2

## Next Steps

Though an affected dog may have shortened limbs, the condition is not expected to negatively impact their quality of life.

## References

Frischknecht M, Niehof-Oellers H, Jagannathan V, Owczarek-Lipska M, Drögemüller C, Dietschi E, Dolf G, Tellhelm B, Lang J, Tiira K, Lohi H, Leeb T. A COL11A2 mutation in Labrador retrievers with mild disproportionate dwarfism. PLoS One. 2013;8(3):e60149.