

DNA testing for SD2 Skeletal Dysplasia Type 2 in the Labrador (A problem with appearance not a disease)

The DNA tests relevant to our Labrador breed seem to grow in number every few months and one of the interesting things we will also need to keep in mind, particularly as researchers investigate even more subtle but inherited traits, is the effect of what is termed epigenetics (more later). One of the most recent tests, developed by Hannes Lohi's research group of canine genetics at the University of Helsinki and the Folkhalsan Research Centre who identified the causative mutation (gene) and have developed a DNA test offered by Genescope (Norway) and also offered by Laboklin and Animal DNA diagnostics in the UK, is for Skeletal dysplasia type 2 (SD2) or inherited dwarfism in Labradors. This is an abnormality of skeletal development that causes short-leggedness in Labrador Retrievers. The phenotype (appearance) is inherited in an autosomal recessive manner. The mutation has (so far in these countries) only been observed in Labrador Retrievers of working lines, as opposed to in show lines but the incidence of the problem within the UK or USA show Labrador lines is not known. I suspect that we have all at one time or another seen such Labradors who look like a normal dog but on Basset legs. It is becoming clear that some of our top Field Trial Champions in the UK were most probably carriers of SD2.

The University of Bern web site states; "Skeletal dysplasia 2 (SD2) is a mild form of skeletal dysplasia that was recently discovered to affect Labrador Retrievers. The phenotype (appearance) manifests as short legs in combination with normal body length and width. Affected dogs typically have forelegs slightly more affected than hind legs, and a low shoulder height (<50 cm) compared to the international breed standard (54-57 cm). "

Skeletal Dysplasia Type 2 is different to several other skeletal dysplasias, (particularly the severe disease of Retinal Dysplasia Ocular Skeletal Disease which has only so far been identified in the Labrador in the USA where a test is available through Optigen http://www.optigen.com/opt9_rdosd.html.) In SD2 no auditory problems/deafness, eye problems or secondary joint problems have been observed in dogs affected with SD2. Therefore, as the dogs have no health problems we are really looking at a test that purely governs appearance and is not a disease in itself.

The genetic test results for SD2 are reported as follows: Normal: The dog carries two normal copies of the gene and has no or reduced likelihood of developing SD2 during its life. Carrier: The dog has a normal appearance, but carries one copy of the mutation and one copy of the normal gene in its genome. A carrier transfers the disease mutation to approximately 50% of its offspring. Affected: The dog carries two copies of the mutation and is at high risk of developing SD2. An affected animal will pass on the mutation to all of its offspring. All the offspring of an affected dog and a normal dog will be carriers. All offspring of two affected dogs will also be affected."

A paper on SD2 has been published by Frischknecht M., et al.
<http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3603880/>

What is particularly interesting about this condition and shows that we need to start thinking a little differently, about the DNA testing we are undertaking, is explained as a result of my communication with Prof. Dr Tosso Leeb from the Institute of Genetics at the University He writes "According to our data, the "skeletal dysplasia 2 (SD2)"

is inherited as a strictly recessive trait. Thus, we did not see any difference in the shoulder height of heterozygous carriers vs homozygous clear dogs.

We see a very pronounced variability of the phenotype (appearance) of affected dogs. The SD2-affected dog on my webpage (http://www.genetics.unibe.ch/content/rubrik/labrador_dwarfism/index_eng.html) is a very severe case. In most SD2-affected dogs the proportions will be closer to normal than in the dog shown on this picture. One has to keep in mind that size and body proportions are controlled by multiple genes and that you have some variability in the normal Labrador population regarding these traits. Two copies of the SD2 allele “will make the legs shorter” but this happens on a given genetic background. On a dog that otherwise might have had relatively long legs, the result will be a dog with almost normal proportions.

To be more precise, according to our data dogs, which are homozygous (affected) for the SD2 allele, have ~6 cm less shoulder height on average compared to other dogs.”

What do I do if I have a dog or bitch DNA tested as a carrier or affected with SDS?.

If the other factors of your Labrador are all to your liking and other health tests such as Hip Dysplasia and Elbow Scoring Dysplasia results good. Then use a mate who is SD2 DNA tested clear of the condition. Test the puppies in the litter at 4 weeks and ideally keep a clear or carrier puppy to breed on to another SD2 clear Labrador. Bear in mind that it has become clear that some of our very top Field Trial Champion Labradors who were used extensively were most probably carriers of SD2 therefore if you try to eliminate the gene from the Labrador gene pool too quickly by not breeding from your dog or bitch that the gene pool will shrink and other potentially much more serious genetic problems are likely to emerge because inbreeding rates will increase. Also, bear in mind that over the years the simple process of mating a Labrador with shorter legs to a Labrador who is much taller than average will have helped to override the affect of the SD2 gene in many cases and then as today there are a number of Labradors who are affected with SD2 but you may never know from their appearance because they are only a little shorter than some other Labradors. This fact should help you prioritise your undertaking of DNA testing and management within your kennel.

This effect is explained if you look at the study of epigenetics and how scientists are finding that there are other factors such as: environment, other genes on the DNA and indeed the chemical tags linked to genes that switch them on an off that can all influence the final physical result in any given animal or dog. Concerned breeders will also have noticed this effect with other conditions that we are currently DNA testing for. For example, there are many cases emerging of dogs that are affected (have two copies of the mutated gene) for Exercise Induced Collapse (EIC) but have never (and sometimes in spite of regularly picking up on big shoots) shown any symptoms of collapse. This is probably related to other environmental factors such as fitness, nutrition and as yet undiscovered molecular and chemical influences on the gene via its epigenome. If you want to learn more about genetics and the epigenome there is a very useful website that starts from the very basics and is very visual and pictorial at <http://learn.genetics.utah.edu>.

Reporting of SD2. Since the Autumn of 2014 the UK Kennel Club have agreed to a voluntary recording of SD2 which will appear on their mate select site and on documentation. Owners who have had their Labradors DNA tested for the condition can send copies of their certificates to the health department or to Gary Johnson Gary.Johnson@thekennelclub.org.uk. Laboratories will not automatically send on certificates. This should help breeders who are trying to locate clear dogs but is less likely to dissuade breeders from carrying out the test. This is in line with some other conditions, that we DNA test for, particularly where laboratories are located overseas and do not have automatic reporting procedures in place with the Kennel Club.
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