

Retinal Dysplasia in the Labrador Retriever

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Of the major eye problems in the Labrador Retriever, retinal dysplasia probably evokes the most questions concerning potential visual problems and breeding decisions. With the other major inherited ocular diseases - progressive retinal atrophy and cataract - the decisions are much more clear and the outcomes much more predictable. Not necessarily so with retinal dysplasia, in which the diagnosis may be less certain, the visual outcome much more variable, and the variations of the disease much more numerous.

So, what is retinal dysplasia? The retina is the light sensitive portion of the eye (like the film in a camera), and dysplasia is an abnormality of development. Sounds pretty simple, and in the extreme case, it is. If the retina fails to develop properly in its entirety the dog is blind at birth, will never see, is easily diagnosed, and decisions can be taken rationally and early. There is one type of retinal dysplasia in Labrador Retrievers which is exactly like this. It was first described in England and Sweden in the early seventies. The retinal dysplasia was complete, the retinas detached from their normal position, and the affected pups blind at birth. The inheritance pattern was quickly determined to be a simple recessive trait without any complicating factors. Test breeding failed to reveal any significant abnormalities other than those of the eye, and all decisions were essentially "no brainers." Both sire and dam were carriers of the trait and should not be used for breeding, the affected pups should be put down, the unaffected pups were probably carriers (two chances out of three) and shouldn't be used as reproductive stock. The disease was much like that seen and described well in other unrelated breeds, the Yorkshire terrier, the Sealyham terrier and the Bedlington terrier. That's the end of the easy stuff!

The problem area occurs with a second type of retinal dysplasia, first described by Drs. Nelson and MacMillan in California field trial Labradors. The group of retrievers they examined had about 20% of the dogs affected. That's much more prevalent than is usual now. This type of retinal dysplasia is completely different genetically than the one described in Europe, and goes under the general name "Retinal and Vitreal Dysplasia with Skeletal Abnormalities (Dwarfism with Retinal Dysplasia)." In this type of retinal dysplasia in Labradors there are three different eye types (phenotypes), and two different skeletal types that can accompany the eye types. The investigators found puppies with normal eyes, puppies with localized retinal dysplasia (retinal folds) and puppies with total retinal detachment (total retinal dysplasia). The two skeletal phenotypes were (simply) affected and normal. Most of the skeletally affected Labrador Retrievers have complete retinal dysplasia, retinal detachment, and an abnormal vitreous body (the gel-like portion of the eye). A few also have a peculiar pattern of pigment in the cornea and varying degrees of cataract (opacity) of the lens. Some dogs with skeletal abnormalities have multiple small retinal folds, without having a complete blinding retinal detachment. Some believe that the occurrence of multiple retinal folds is a lesser manifestation of the more severe retinal disorder.

Visual difficulties in severely affected pups become visible at eight weeks or before, and both visual deficiencies and skeletal abnormalities become more noticeable as the pups grow. The skeletal abnormalities include retarded growth of the front legs and bowing out at the elbows with deformation of the carpus. The hind limbs are straighter than normal, which causes difficulty in rising from the sitting position. There is also retarded growth in the tibia, elbow hypoplasia or dysplasia, hip dysplasia, and delayed development of growth plates of the bones. These severely affected dogs tend to have larger than normal eyes.

Researchers did breeding experiments, outcrossing severely affected (both skeletally and ocularly) Labrador Retrievers to normal beagles, as well as crossing Labrador Retrievers having various degrees of the syndrome. Crosses of dogs with severe ocular abnormalities and abnormal skeletons to normal dogs produced dogs with mild ocular abnormalities and no skeletal deformities. Crosses of dogs with clinically normal skeletons and mild ocular abnormalities to similarly affected dogs produced five types of dog: those without either skeletal or ocular abnormalities, those with normal skeletons and mild ocular abnormalities, those with abnormal skeletons and mild ocular abnormalities, those with abnormal skeletons and severe ocular abnormalities, and those with abnormal skeletons and no ocular abnormalities. Similar results happened by crossing dogs with skeletal abnormalities and severe ocular abnormalities with dogs with no skeletal abnormalities and mild ocular abnormalities. Crossing dogs with both skeletal abnormalities and severe ocular abnormalities to similarly affected dogs resulted in dogs with skeletal abnormalities and either severe or mild ocular abnormalities. The breeding results from crossing either purebred Labrador Retrievers or by outcrossing to beagles were similar.

The breeding experiments indicate that the tendency to develop ocular and skeletal defects are inherited together. The data indicate that skeletal abnormalities segregate as a recessive effect of the gene and that the eye portion of the disease acts like an incomplete dominant trait. The presumption therefore is that any Labrador with any type of retinal dysplasia (severe or mild, detached or with folds) must be a carrier for skeletal defects. The other presumption is that at least one of two parents of a Lab with severe or mild retinal dysplasia is a carrier for skeletal defects. So much for the easy part!

Because of the possibility that dogs with multiple skeletal as well as severe ocular abnormalities may be produced by breeding two mildly affected dogs, the ACVO (American College of Veterinary Ophthalmologists) Genetics Committee advises breeders not to breed affected stock, and CERF (Canine Eye Research Foundation) certificates are not issued for affected animals.

Ophthalmoscopically, retinal folds vary somewhat. They can be dots, lines, or branches of various shapes. Dogs with more severe folds ("geographic") almost invariably also have a severe disturbance of the vitreous humor. When Drs. Nelson and MacMillan reexamined some two year old dogs that had been minimally affected with retinal folds as pups, they found that in some dogs the retinal folds seen previously had disappeared (this happens in collies and cocker spaniels as well), and that a dog previously diagnosed as having retinal folds now appeared normal. This experience has been shared by others and makes control of the disease more complex. So it is necessary to have young Labrador pups checked (ophthalmoscopically) in order to detect the presence of minor dysplasia before it disappears. The best time is eight to ten weeks. Apparently normal older dogs may in fact be affected animals; in some only the presence of an abnormal vitreous humor may reveal the condition. In some Labrador Retrievers which have a peculiar coloration to the back of the eye, it may not be possible to detect minimally affected dogs at any age.

So far as vision is concerned, no one disputes that Labradors affected with complete retinal detachment (complete retinal dysplasia) are blind. But what of those dogs affected with retinal folds? The ACVO Genetics Committee advises that dogs with extensive retinal folds ("geographic") "are associated with vision impairment." My experience, and that of Drs. Nelson and MacMillan, suggests that the practical effect of the presence of retinal folds ("geographic" or simple) varies and is not predictable. Some field trial dogs are apparently not able to see beyond 30 to 40 yards and do not mark well, while others behave normally. One dog, with an estimated 50 abnormal folds in each eye, was stated by Nelson and MacMillan to have perfect vision in field trial work (as well as elsewhere). Interestingly, one researcher found that some Labrador retrievers with the disease also had "axial myopia" - they were near-sighted - this may explain the presence of visual difficulty, rather than the mere presence of folds.

Dogs with retinal folds and vitreous abnormalities are likely to make acceptable pets, but they do have a risk of developing retinal detachment, which may be treatable in the early stages. Therefore ophthalmologic examinations should be performed repeatedly in affected pet animals for at least three years.

To top the whole story off, I believe that there exists in Labradors yet another type of retinal dysplasia, associated neither with blindness nor skeletal disease, resembling the retinal folds commonly seen in cocker spaniels. This is based on the results of a breeding of two dogs with retinal folds. I examined sire, dam (each several times) and all of the pups. There were no skeletally abnormal pups, at least to my view and that of the breeder, and all of the pups had normal eyes. Such a litter would be unlikely to occur if the folds were the result of the presence of the "Retinal and Vitreal Dysplasia with Skeletal Abnormalities (Dwarfism with Retinal Dysplasia)" genes in both parents.

To summarize my recommendations:

- examine pups all pups early
- check breeding stock prior to breeding
- don't make irrevocable decisions if you have an affected pup - at least get another opinion

Editor's note:

Although most of the research regarding retinal dysplasia has been associated with field trial pedigrees, please keep in mind that forms of retinal dysplasia and retinal folds have been observed in Labradors with conformation oriented pedigrees as well.

Suggested Reading

If you would like to do additional reading on this subject the following list may be useful:

- American College of Veterinary Ophthalmologists, *Ocular Disorders Proven or Suspected To Be Hereditary In Dogs*, 1992
- Animal Health Trust, *Hereditary Eye Abnormalities In The Dog*, second edition, 1985
- Barnett, KC, et al, *Hereditary retinal dysplasia in the Labrador Retriever in England and Sweden*, Journal of Small Animal Practice 10:755, 1970
- Carrig, CB, et al, *Retinal dysplasia associated with skeletal abnormalities in Labrador Retrievers*, Journal of the American Veterinary Medical Association, 170:49, 1974
- Carrig, CB, et al, *Inheritance of associated ocular and skeletal dysplasia in Labrador Retrievers*, Journal of the American Veterinary Medical Association, 193:1269, 1988
- Kock, E, *Retinal Dysplasia*, thesis, Stockholm, 1974
- Nelson, D, MacMillan, A, *Multifocal retinal dysplasia in the field trial Labrador Retriever*, Journal of the American Animal Hospital Association 19:388, 1983

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