Golden Retriever Club of America
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From the GRCA Research Facilitator:
Managing Genetic Disease in Golden Retrievers
Managing Genetic Disease Through Research Support
Although we are all deeply saddened that a “new” and devastating disease (neuronal ceroid lipofuscinosis, NCL) is impacting our breed, the ray of light is that at least we have a DNA test that enables us to avoid producing affected puppies. Almost no affected Goldens should be produced going forward, and for that we extend our sincere gratitude to all of the breeders, owners, and researchers who contributed to test development.
It’s a natural extension of that goodwill to want to continue to support researchers and research efforts into other genetic diseases and other aspects of NCL, such as developing therapies to possibly help affected dogs. But as those opportunities arise, it’s wise to proceed carefully and make sure we fully understand and agree with proposed research methods and goals, before providing support (either financial or via samples).
Learning about NCL has also sparked discussions regarding donating frozen semen from carrier or affected dogs to research programs, and it may not be clear to all as to how such semen is generally used. Owners considering donating semen should be aware that the primary use for frozen semen in genetic research is to preserve the ability to create groups of affected dogs for use in investigating disease processes and developing therapies. Commonly, these dogs need to live in research colonies in order to control all aspects of their environment, and it should be noted that the Golden Retriever Foundation does not fund this kind of research.
To make sure participants understand, agree with, and can trust the research that breeders, owners, and donors are asked to support, there are procedures in place – primarily through the AKC Canine Health Foundation (CHF) – that provide GRCA and the GR Foundation with scientific peer-review (expert review) and oversight. Several levels of review and oversight ensure scientific validity and also that dogs are protected by the CHF Humane Use of Animals Policy. Appropriate consent forms that describe the study are provided to owners when samples are submitted, and summaries of the research are openly available.

Of course, CHF and the GR Foundation fund only a portion of research that is vital and very worthy of support, but owners are welcome to contact the Research Facilitator if questions arise regarding sample donations. As always, blood, tissue, or tumor samples provided during an emergency or upon euthanasia are valuable to aid research and are encouraged whenever possible.

Managing Genetic Disease by Protecting the Gene Pool

Once a DNA test for a disease is available, breeders can turn their attention more toward avoiding the disease rather than developing therapies for affected dogs, as these will be minimized. There are excellent websites that describe the mechanics of managing recessive genes (as are our current DNA tests) in individual breeding pairs to avoid producing affected puppies, so that is not the focus here. This is a discussion of managing disease by managing the gene pool.

[It is suggested that Readers briefly pause here and review Figure 1 in the accompanying excerpt from my article “Preserving Genetic Diversity in Golden Retrievers,” originally published in the Nov/Dec 2014 GRNews, before proceeding. The text provides more in-depth background on the concept that as gene pools shrink, previously rare diseases become more common.]

The ever-increasing number of DNA tests adds urgency to the question of how we balance the use of DNA tests to protect the health of individual dogs, while also protecting the health of the gene pool for the benefit of future generations. This is difficult, but hopefully achievable, and often requires putting immediate self-interests aside in order to very carefully keep carriers in the gene pool for more than just a generation or two. A wise guiding principle toward using DNA tests to reduce disease genes is “slowly and safely.”

It starts with recognizing and advocating that carrier dogs very often have value as breeding dogs, even when they carry serious diseases. And the smaller the gene pool, the greater their value. Our breed cannot withstand repeatedly wiping out disease genes in a short few generations, because all the generations that follow will suffer from “new” diseases driven to the surface by continually reducing genetic diversity. Being a responsible breeder is not just about rapidly purging individual lines of all testable recessives, because that almost guarantees that the next genetic disease is right around the corner – and sometimes it’s worse than the initial targeted disease.

Responsible breeders include worthy carrier bitches as brood bitches, and responsible breeders with clear bitches include worthy carrier stud dogs

among their selections. Yes, it means numerous generations of DNA testing puppies, with all the expense and uncertainty that comes with it. Yes, it requires hypervigilance in placing carrier puppies, being completely certain that the disease gene does not “get loose” in any possible breeding population (including doodles, etc). And it also requires keeping carrier puppies as potential breeding dogs in trusted hands, and competition buyers (who may also wish to breed) should be willing to choose a carrier puppy as their pick.

Further, bitch owners also serve the breed when they select stud dogs having “faults” that may reduce competition wins, as long as the fault does not reduce quality of life. Automatically eliminating dogs with missing teeth, or slower agility runs, or less enthusiastic water entry, or many other imperfections, feeds into the popular sire syndrome in the relentless search for “perfection.” Popular sires – more than any other single factor – reduce gene pools and increase risk for dispersal of unknown recessive diseases. Unknown, that is, until a few generations later when those genes pair up and make themselves known, and the gene pool noose tightens again.

For many, it’s a badge of pride to test one’s dog for every conceivable clearance that remotely applies to our breed, often including conditions that are lower on a quality-of-life priority scale. Although this is understandable from the viewpoint of what is best for an individual, it can actually be damaging to the breed. As this all-encompassing testing becomes an expectation and a demand, it exerts enormous pressure on stud dog gene pools – far more than on bitches. Owners of carrier status bitches are typically in control of their breeding options in a way that stud dog owners are not, because owners of carrier status stud dogs are dependent on the choices of bitch owners. This expectation and demand for exhaustive testing further contributes to creating popular sires from among dogs that test “perfect.”

However, stud owners (and bitch owners, for that matter) can help combat this harmful mindset by deciding not to DNA test their dogs for less serious diseases. At this time, ichthyosis (ICH) is an example of an often-unexpressed disease in which DNA testing primarily identifies some dogs as more “perfect” than others. This ultimately does more harm than good because it’s emotionally difficult for breeders to knowingly select “imperfect” breeding dogs; and frankly, it would be better in most cases if there were no ICH test results. Damage to the gene pool can happen in so few generations that breeders who are very familiar with certain pedigrees

can trace the rise of specific serious genetic disease closely tied to avoidance of ICH carriers.

We do not know the reason that many or most US lines do not express clinical ichthyosis even when tested as affected, but among dogs with US lineage, the DNA test is often not a reliable predictor of clinically significant symptoms. In the absence of known close relatives with clinical signs of this generally mild disease, there are good reasons for owners to decline to test their dogs. In this instance, if breeders were to test only when there are known clinically affected close relatives, it would still serve to reduce the incidence of ICH symptomatic dogs, while taking some pressure off of the gene pool. Learning in advance how to manage low impact diseases can serve as a blueprint when DNA tests for other low impact diseases become available in the future. Learning in hindsight has left some wishing that the DNA test for ichthyosis had never been developed.

Making the decision to consider the clinical implications to offspring and the impact on the breed's genetic diversity – rather than strictly the results of a DNA test – may expose responsible breeders to criticism. And it is true that not testing for a less clinically significant disease such as ICH, will sometimes produce a symptomatic puppy. (Remember, we're only considering mild conditions in the category of not routinely DNA testing. Serious diseases would always be tested and carriers bred only with clears.) If a symptomatic puppy is produced, that individual breeder would need to begin DNA testing at least one parent for ICH, but the breed would still benefit from the overall inclusion of more dogs in the gene pool if this test were less widely done. Again, it requires putting the best interests of the breed ahead of the best interests of the individual breeder. It's tough to do, but this too can be a badge of pride.

All of us can also play a role by not criticizing (social media!) those who breed dogs with missing DNA tests (ICH) or carrier status DNA tests (more serious disease such as NCL), and by supporting each other in making these difficult but forward-thinking decisions. Hopefully we can begin to take on the challenge of being responsible not just to our own puppies, but also to the long-term future of the breed, and with vision that sees beyond current generations and current DNA tests. Because if anything is certain about genetic diseases and DNA testing, it is that we have only seen the tip of the iceberg.

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