

# Designer Genes

## Genetic Management or Misery?

*By: Catherine McMillan*

In the early 1970s, Miniature Schnauzer breeders embarked on a program unprecedented and unduplicated in any popular breed: to eliminate the genetic defect that caused juvenile cataracts. Research had established that juvenile cataracts (CJC) were transmitted as autosomal recessive with complete penetrance and were present at birth. Early diagnosis permitted the use of test-breeding, sanctioned by the national breed clubs, in which certified affected dogs were paired with mates whose status was unknown. A litter of normal eyed puppies was known to generate a mathematical probability that the tested dog was clear (the more normals, the better his or her odds), while the diagnosis of a single affected puppy proved the dog a carrier.

There is no argument that the program met its goals. A breed with an estimated 40% carrier rate emerged from two decades of test breeding with show lines cleared of the defect. It was a spectacularly successful example of how a breeding community can come together to eradicate a defect... and cause devastating damage to the gene pool.

### **Enter Stage Left**

It has been written that, as a result of the process to eliminate CJC, over 200 American Champions were retired from breeding. Important kennels quietly closed up shop, taking distinct family branches with them, and bitches were sent exclusively to test-bred stud dogs. It was a lonely time for an untested male.

Around the same time as CJC was defeated, PRA made its entrance. In a few short years, several leading sires were revealed to be carriers and retired. There was no test-breeding program for this late onset defect, so it became a lonely time for the stud dog or bitch with a carrier ancestor. The gene pool contracted again.

Had this been the end of the troubles there may have been time to pause and reflect on what was happening in the big picture, but this was not to be. A novel defect appeared on the scene – a muscular disorder called myotonia congenita. This problem found a solution in short order as a DNA test was developed, allowing breeders to identify carriers with a simple blood test. Those were retired, too.

My choice of the word “retired” has, of course, been deliberately inappropriate here. In the world of dogs, “retired” is usually a euphemism for “sterilized”. As a device for preventing genetic defects, it must rate as one of the most destructive practices ever employed.

In a sensible dog world, quality carriers of genetic disease might be pulled from widespread use, but they’d come out of “retirement” for special occasions (i.e., for research breedings and/or the general advancement and preservation of rarer family lines). However, the dog fancy – and, by extension, breed clubs – have never been famous for our ability to apply knowledge sensibly. There is a common caution against throwing the baby out with the bathwater. In purebred dogs, there is a tendency to gather up the siblings, cousins and parents and throw them into the dust as well. We “improve” our breeds by killing them off one family branch at a time.

When I first began breeding nearly 30 years ago, I accepted the conventional wisdom that largely prevails to this day—that genetic defects are the exception, that carriers should be removed from the gene pool and that health is more important than beauty.

But, as John Maynard Keynes said: “When somebody persuades me that I am wrong, I change my mind. What do you do?”

### **Managing the Unmanageable**

A few years ago, some bright bulb at the Canadian Kennel Club launched a grand scheme to create a Code of Ethics. One of the rules proposed for this set of stone tablets was “Thou shalt not breed a carrier”. I recall writing to one of the Board members at the time to congratulate the CKC for devising an edict that would result in the immediate eradication of a number of breeds. For there are breeds today in which every single member is not merely a carrier, all or nearly all are affected with a genetic defect. The peculiar nature of Dalmatian urine chemistry is the most famous example.

Even in breeds with more moderate disease rates, the policy would have eventually resulted in genetic collapse and extinction. That’s because every normal living being is thought to

carry in the range of 5 disease mutations within their DNA. In breeds with few founders and extreme bottleneck events that average may be much higher. As molecular genetics digs into the DNA of our four footed friends, it is revealing gene frequencies that are nothing short of staggering in some breeds. In English Springer Spaniels, for example, a mutation that elevates the risk of PRA has been identified and a DNA test developed at the University of Missouri-Columbia. Of the dogs tested, only 20% have been found to be clear of the gene while over 40% tested as affected. Dobermans have similar carrier rates for the bleeding disorder, vWD.

### Time for a Change?

The purpose of this article is not to cover the ground of nuts and bolts genetics. There's simply not enough space and I don't have the right letters after my name. There are many good texts available that cover the science, as well as a number of authoritative Internet sources. It is recommended that you seek the most recent material you can find as many of the popular canine genetics books of the past are now obsolete.

What I hope to provoke is an examination of some of our traditionally held beliefs. "Thou shalt not breed a carrier" served us well enough when diagnostics were primitive, most carriers escaped detection, and conditions now known to be inherited were dismissed as environmental or simple bad luck. This is no longer the case.

Unfortunately, a little knowledge can be dangerous. The discovery of extreme carrier rates in a breed has the potential to overwhelm breeders who have always held that their primary goal was to produce healthy dogs. It's depressing to think of how many aspiring breeders accepted as an article of faith that quality foundation stock, good intentions and careful testing would result in good health... only to fail. They'd start over, fail again, become discouraged and move out of the sport. Now we know why.

The bottom line is that much of what we thought was wrong. Now, for the sake of our breeds, we need to change our minds. It is no longer a question of "eliminating" gene defects from a breed. We can only ask which ones, how quickly and should we even try? For this reason, it is imperative that breed clubs take the lead and reform outdated notions about "ethical" breeding practices and the advisability of "retiring" animals before they can leave positive contributions to the gene pool.

### Diversity is Key

One of the most important factors in maintaining a healthy breed population is preserving genetic diversity. Genetic diversity is important for survival and adaptability within species, but dog breeds are not species. They are purpose-bred populations that have undergone selection for specific traits or behaviours. It is not enough to simply survive; they have a job to do. Nonetheless, within closed gene pools, genetic diversity is central to infectious disease resistance and the availability of normal alleles when mutations arise.

There is little disagreement on that point, but there can be great disagreement on the best means to achieve it. One camp believes in outcrossing, de-emphasis of "show ring" traits and performance standards, and even selected infusions of other breeds. Another camp holds that a healthy diversity of successful breeders who work to preserve and develop distinct family lines is the best way to preserve genetic choice. I happen to belong to the latter.

Before one begins, however, one must first define "successful". Or rather, one must understand how success is defined in any breed. It is not a matter of interpretation; it is a matter of record.

A few years after I began showing and breeding Miniature Schnauzers, I realized that no historical archives existed for champion producers in Canada, in the way they have always been catalogued in the US. So, I began gathering the data from old CKC stud books and issues of Dogs In Canada, starting with the first recorded champion in 1933.

Somewhere in the middle of the project, I had an epiphany. Everything that I had been told to believe was wrong: Health is not more important than beauty.

### **Beauty is more important than health.**

Next Issue: It isn't important that we all do the right thing, it is only important that we don't all do the wrong thing. Forcing everyone to do the same thing risks forcing everyone to do the wrong thing.

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