Basic Genetics

Test breeding does not help for a polygenic trait like HD.

Polygenes: The problem that dog breeders have with using ideas of dominant and recessive genes in breeding dogs is that most traits don't appear in discreet intervals, but instead are continuously distributed over a range of values. For instance, dogs don't come in just short, medium, and tall; they come in all sizes. Even within a breed, height is normally distributed in a bell curve. This is because many important traits are the result of many pairs of genes acting together. In these cases, the extent of a trait is determined by gene dosage, which is the number of particular alleles present in a genotype.

Imagine that height is controlled by incompletely dominant alleles at three different loci, A, B, and C, with A+, B+, and C+ all coding for an additional half inch of height. A dog with the genotype A+A+, B+B+, C+C+ would be three inches taller than one with the genotype A-A-,B-B-,C-C-. In fact, 27 different genotypic combinations are possible in this example, resulting in seven different heights. The more loci involved, the greater the number of possible genotypes and phenotypes, until the phenotypes become so numerous that they appear to be continuously distributed. This blending is further influenced by environmental factors. Hip dyspalsia is thought to be polygenic.

Linkage and Linkage Disequalibrium: In a highly inbred population, genetic defects can become fixed rather rapidly if they happen to be on the same chromosome as a gene that codes for a desirable trait. The closer they are physically on the chromosome the tighter they are linked. These genes and their respective alleles will be inherited together unless they become unlinked in a procedure called crossing over or recombination. This is a process that occurs during the formation of gametes, whereby homologous chromosome pairs exchange segments of their DNA structure. Such closely linked genes are said to be in a state of linkage disequilibrium. When a breeder selects for or against a specific gene trait, he or she is also choosing those traits or not, which are located on the same chromosome. One should remember this when making a breeding decision. Severe selection pressure against an unwanted trait, could result in throwing the baby out with the bathwater and the permanent loss of a necessary or desirable attribute.

Sex Linkage: A special case of linkage exists when genes are located on the sex chromosomes. Unlike the other 38 pairs of chromosomes, the sex chromosomes are not always paired in a homologous fashion. This is simply because sex is determined by whether an individual has two X-chromosomes (XX=female) or an X and a Y chromosome (XY=male). The Y chromosome is a very small chromosome and until recently there were doubts that any significant information was contained on it. The X chromosome is larger and is known to carry on it genes that code for several important traits. Genes on the X chromosome are not

matched by genes on the Y chromosome, negating the possibility of allelic pairs. In the male, whatever alleles are on his single X-chromosome will be expressed (a condition known as hemizygous). In the female, the situation is still a little different from what is seen in the other autosomal (non-sex) chromosomes.

For many years it was assumed that these X-linked alleles acted just the same as autosomal alleles. But they don't. Instead of acting in a standard dominant-recessive way, these alleles act more like co-dominant alleles. In placental mammals, one of the two X-chromosomes is randomly inactivated in each cell of the body. The remnants of these inactivated chromosomes can be seen as dark spots (Barr bodies) in almost every cell of a normal (XX) female, but not in normal (XY) males. Very early in embryonic development, both X-chromosomes are apparently active, but then one of the two is rendered dysfunctional by staying tightly condensed in the heterochromitin state. It is entirely a matter of chance whether it is the paternally or maternally derived X-chromosome, but once inactivated, all subsequent cells derived from that cell will continue to have the same inactivated X-chromosome. In individuals with visible sex-linked traits, the results can be clearly seen as patches of paternally and maternally derived traits.

Thus, all female mammals are mosaics. The best known example is the calico cat, which is almost always female (the few males are abnormal XXY individuals) and which displays a patchwork of black and orange colors, each patch representing a clone of an original cell that randomly inactivated either the X-chromosome with an allele for orange fur or the X-chromosome with the allele for black fur.

In dogs, we have to look a little more carefully for such evidence. Examples include X-linked muscular dystrophy in Golden retrievers and X-linked hereditary nephritis. Because these female carriers are mosaics for the abnormalities seen in these diseases, they may exhibit attenuated signs of the disorder, with the severity depending upon the proportion of the mosaic derived from the X-chromosome that carried the abnormal allele.

Sex-linked traits will be passed from dams to sons via one of her X-chromosome. Because sires can only pass their X-chromosomes to their daughters, in order for the trait to be fully expressed in a female, she must have an affected sire and carrier (or affected) dam. The degree of mosaicism that the dam expresses is random and does not affect the chances of her offspring being affected or the severity of that trait, if affected.

Misunderstandings about sex-linked inheritance have given rise to many breeding myths, the most widespread of which (1) place greater emphasis on the "sire line" (sire to grandsire) in the belief that "what you see is what you get," due to the single X-chromosome, as well as the belief that important breed attributes are carried on the Y-chromosome; or (2) contend that, whether an ancestor is on the dam's versus the sire's side of the pedigree is of prime importance. These theories neglect the fact that the Y-chromosome contains little, if any,

identified genes apart from those involved with male reproduction, and that the sex chromosomes are but one of 39 pairs of chromosomes. These ideas served the 19th century breeder well, but they have no place in the 21st century breeder's arsenal.

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