Papillon Club of America
Health & Genetics

PRACTICAL GENETIC COUNSELING ON PURE-BRED POPUØATION

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Genetic defects are controlled by single, or a handful of genes, compared with the estimated 40,000 to 100,000 genes in the dog genome. Prudent breeding practices dictate that you do not throw the puppy out with the bath water in genetic disease control. With the development of gene probe identification of defective genes and tests for carriers of defective genes, practical genetic counseling can be provided to dog breeders.

A recommendation to eliminate all carriers of a defective gene from a gene pool may result in a significant loss of genetic diversity. Additional, previously unknown defective genes could be concentrated through genetic bottlenecking. A recommendation to breed heterozygous carriers to homozygous normal dogs prevents affected dogs,and keeps the gene pool diverse. However, it does not provide selective pressure to decrease the frequency of the defective gene.

The goal of genetic counseling is to control the spread of defective genes effectively, while preserving the health and genetic diversity of the purebred dog population. There are different breeding program recommendations based on several factors, including;

1) Populous breeds versus rare breeds: Genetic selection in a populous breed does not tend to restrict genetic diversity. Recommendations should be geared so that a defective gene does not become widespread in the breeding population. In rare breeds, genetic selection should not be so severe to further restrict genetic diversity in a small gene pool.

2) Widely dispersed versus recently mutated defective genes: Strict control should be instituted against recent mutations, to not allow them to become widespread. Selection against a widely dispersed gene depends on its frequency.

3) High frequency versus low frequency defective genes: High frequency genes require a long term control program that will diminish the frequency, without altering the dynamics of the gene pool. Tests for carriers would be helpful, so that genetically normal dogs will not be selected against. Selection against a low frequency genefocuses on strict control when observed.

4) Single gene versus polygenic disorders: Selection against polygenically controlled disorders must focus on the affected or normal status of the full-sibs (littermates) of the breeding dogs and their parents. Knowledge of breadth of pedigree gives selection information on the possible genetic variation in the individual breeding animals.

5) Disorders with tests for carriers, versus no test for carriers: With a direct gene test, breeders only have to know the results of the dogs they plan on breeding. With phenotypic tests or no tests for carriers, the knowledge of the carrier or affected status of related dogs is important.

Basic protocols for genetic counseling and breeding management of genetic disorders can be based on the known mode of inheritance, and the availability of genetic tests:

Recessive disorders with a test for carriers: Testable disorders allow breeders to use all breeding stock, and should result in no loss of breeding lines or genetic diversity. Quality individuals who test as carriers should be bred to normal testing individuals, preventing additional affected individuals. The offspring should be tested, and the carrier parent should be replaced in the breeding program with a quality, normal offspring. Additional carrier offspring should not be placed in breeding homes; as the goal is to reduce the frequency of the defective gene in the population. As each breeder tests and replaces carriers with normal testing individuals, the problem for the breed as a whole diminishes.

Recessive disorders without a test for carriers: The problem with these disorders is the propagation and dissemination of unapparent carriers in the gene pool. Relative risk pedigree analysis can provide objective risk assessment for prospective breeding animals and planned matings. This requires knowledge of the carrier or affected status of close relatives in the pedigree; which is best accomplished through a breed club supported open health registry. By determining the average carrier risk in the population, breeders can be counseled to attempt matings with risk factors lower than the breed average. They should lower the carrier risk of their breeding stock with each generation, by replacing higher risk individuals with a quality, lower risk offspring. Breeding an individual once and replacing it with an offspring allows breeders to improve their chance of moving away from a defective gene. The number of offspring placed in breeding homes should be limited, as the goal is to lose the defective gene, and not increase the chance of propagating it. A negative aspect of pedigree analysis is that it selects against families, regardless of an individual's normal or carrier status. On the other hand, it allows for the objective risk assessment and continuation of lines that might otherwise be abandoned due to high carrier-risk.

Autosomal dominant disorders :
Managing dominant disorders is usually straightforward, as all individuals carrying the defective gene are affected. Selecting a normal sibling, or parent for future breeding maintains the breeding line. If the disorder shows incomplete penetrance and there is not a genetic test, relative risk analysis and breadth of pedigree analysis (below) can identify individuals with high carrier risk.

Polygenic disorders, or those without a known mode of inheritance :
These disorders require knowledge of the affected or normal status of full-sibs to prospective breeding animals. Individuals whose siblings are normal, and whose parents' siblings are normal have the greatest chance of carrying a low genetic load for the condition. This breadth of pedigree analysis is more important than normalcy in the depth of pedigree (parents and grandparents only.) Affected individuals can be replaced with a normal sib or parent, and bred to a low-liability mate. Breeders can replace the higher risk parent with a quality, lower risk offspring, and repeat the process.

It is distressing to breeders when a genetic disorder is confirmed. Positive and practical genetic counseling recommendations can be offered to maintain breed lines and genetic diversity, and improve the overall health of breeds.

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