Popular-Sire Syndrome: Keeping watch over health and quality issues in purebreds

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An important issue in dog breeding is the popular-sire syndrome. This occurs when a stud dog is used extensively for breeding, spreading his genes quickly throughout the gene pool. There are two problems caused by the popular-sire syndrome. One is that any detrimental genes which the sire carries will significantly increase in frequency – possibly establishing new breed-related genetic disorders. Second, as there are only a certain number of bitches bred each year, overuse of a popular sire excludes the use of other quality males, thus narrowing the diversity of the gene pool.

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The popular-sire syndrome is not limited to breeds with small populations. Some of the most populous breeds have had problems with this syndrome. Compounding this, there are several instances where a popular sire is replaced with a son, and even later a grandson. This creates a genetic bottleneck in the breeding population, narrowing the variety of genes available.

Every breed has its prominent dogs in the genetic background of the breed. But most of these dogs become influential based on several significant offspring that spread different combinations of the dog's genes over several generations. The desirable and undesirable characteristics of the dog were passed on, expressed, evaluated by breeders, and determined if they were worthy of continuing in future generations.

The Challenges

The problem with the popular-sire syndrome is that the dog's genes are spread widely and quickly - without evaluation of the long-term effects of his genetic contribution. By the time the dog's genetic attributes can be evaluated through offspring and grand-offspring, his genes have already been distributed widely, and his effect on the gene pool may not be easily changed.

In almost all instances, popular sires are show dogs. They obviously have phenotypic qualities that are desirable, and as everyone sees these winning dogs, they are considered desirable mates for breeding. What breeders and especially stud-dog owners must consider is the effect of their mating selection on the gene pool. At what point does the cumulative genetic contribution of a stud dog outweigh its positive attributes? A popular sire may only produce a small proportion of the total number of litters registered. However, if the litters are all out of top-quality, winning bitches, then his influence and the loss of influence of other quality males may have a significant narrowing effect on the gene pool.

In some European countries, dog-breeding legislation is being considered that limits the lifetime number of litters a dog can sire or produce. If, however, certain matings

produce only pet-quality dogs, but no quality breeding prospects, should the dog be restricted from siring a litter from a different line? The popular sire's effect on the gene pool is on the number of offspring that are used for breeding in the next generation, and how extensively they are being used. This cannot be legislated.

At what point does a stud-dog owner determine that their dog has been bred enough? It can be difficult to deny stud service when asked, but the genetic effect of a dog on the whole breed must be considered. If everyone is breeding to a certain stud dog, the intelligent decision may be to wait and see what is produced from these matings. If you still desire what the stud dog produces, it is possible that you can find an offspring who has those positive attributes, and also a genetic contribution from its dam that you may find desirable. If a popular stud dog deserves to make a significant genetic contribution to the breed, doing so through multiple offspring, and therefore getting a mixed compliment of his genes, is better than focusing on a single offspring.

Wait-and-See Approach

All breeding dogs should be health tested for the conditions seen in the breed. If your breed has enrolled in the AKC-Canine Health Foundation/Orthopedic Foundation for Animals CHIC program (www.caninehealthinfo.org), prospective breeding dogs and bitches should complete the recommended breed-specific health testing prior to breeding. These may include hip radiographs, CERF eye examinations, or specific genetic tests.

It is important to monitor the positive and negative characteristics being produced by popular sires. While it is satisfying to own a popular stud dog, a true measure of a breeder's dedication is how negative health information in the offspring is made available. All dogs carry some undesirable traits. Based on the variety of pedigree background of bitches who are usually brought to popular sires, there is a greater chance that some undesirable traits could be expressed in the offspring. It is up to the stud-dog owner to keep in touch with bitch owners, and check on the characteristics that are being produced.

Some breeders will argue that the strength of a breed is in its bitches, but the fact remains that the stud dogs potentially have the greatest cumulative influence on the gene pool. There will always be popular sires, and that is not necessarily bad for a breed. But a dog's influence on a breed should be gradual, and based on proven production and health testing. Maintaining surveillance of health and quality issues in breeding dogs and their offspring, and preserving the genetic diversity of the gene pool, should allow a sound future for purebred dogs.

Maneuvering the Maze of Genetic Tests: Interpretation and Utilization

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(Presented at the 5th Tufts' Canine and Feline Breeding & Genetics Conference)

Genetic tests vary on what they are able to identify, and therefore how they can be used in managing genetic disease. To understand how we can use genetic tests, we have to understand the types of tests that are available, what they can tell us, and their limitations.

Phenotypic tests: Some tests measure the phenotype, or what can be seen in the animal. This may not directly relate to the genotype, or the genes regulating the defect that you are trying to manage. Screening for cataracts, ausculting for heart murmurs, hip and elbow radiographs, thyroid profiles, urinalysis for crystals or metabolites, skin biopsy for sebaceous adenitis, and observations on behavioral traits are all tests of the phenotype. Most tests of the phenotype only identify affected individuals, and not carriers of disease liability genes.

Linked-marker based tests: Some defective genes can be linked to a genetic marker, which could be tested for. Linked-marker based tests do not identify the defective gene, but a marker that lies close on the chromosome. If a crossover occurs between the marker and the defective gene during reproduction, the marker will no longer be linked to the defective gene. False positive and false negative results will occur. Due to this phenomenon, linkage test results must be compared with results from other family members to determine whether they correlate with the known genotype of relatives. Linked marker tests include those for cerebellar ataxia in Italian Spinone and primary hyperparathyroidism in Keeshond.

Direct mutation based tests: Direct gene tests are specific for mutations and are a direct measurement of the genotype. They can identify affected, carrier, and normal individuals. These can be run at any age, regardless of the age of onset of the disorder. Most direct gene tests identify a mutation that is causative for a genetic disorder. These genes are 100% penetrant, and an affected genetic test result is 100% correlated with clinical disease. However, some direct genetic tests identify a mutation that causes an increased susceptibility for genetic disease. These **susceptibility alleles** can be part of polygenic/complexly inherited traits, or the cause of incomplete penetrance of (assumed) simple Mendelian traits.

Degenerative myelopathy is considered a complexly (polygenic) inherited disease. An autosomal recessive susceptibility gene has been identified that is homozygous (two abnormal copies) in all DM affected dogs. However, a large proportion of individuals in these breeds are homozygous for this gene and do not become affected. They are considered "at risk", but not genetically affected. In the Wire Fox Terrier, there is a 91% allele frequency in the breed; however no Wire Fox Terrier has even been diagnosed with degenerative myelopathy. In the Boxer, less than 0.5% of dogs develop the disease. Testing Boxers for the DM susceptibility gene shows 39% testing carrier, and 43% testing homozygous "at risk" for the susceptibility gene. This is an example of a genetic test with **low penetrance**; indicating that the homozygous state is poorly predictive of clinical disease. There are additional (unidentified) genes that must also be present to produce clinical DM. This test is useful in ruling out a diagnosis of DM in homozygous normal and carrier dogs. However, selecting against 82% ("at risk" and carrier dogs) of the Boxer gene pool when making breeding decisions - when the vast majority will not produce the disorder - is detrimental to the genetic diversity of the breed. Similar situations occur in other breeds susceptible to DM. In these breeds, breeding dogs should NOT be selected against or have their mating choices altered due to carrier or homozygous "at risk" status of DM unless there is knowledge of close (first or second degree) relatives diagnosed with clinical degenerative myelopathy.

Some breeders feel that any carrier or "at risk" dogs should only be breed to homozygous normal testing dogs. However, requiring that all mating be performed with dogs from only 18% of the population (following the Boxer example) would tremendously skew the breed's gene pool and restrict genetic diversity. This is unnecessary for an extremely low prevalence disease. With genetic tests for lowly penetrant defective genes, selection should only be considered for dogs with families that contain clinically affected individuals. This recommendation should significantly reduce the frequency of clinical disease, as well as the frequencies of other contributory alleles.

Other examples of low prevalence but high allele frequency diseases are cord1 PRA in English Springer Spaniels (42% "at risk", 38% carrier, ~1-2% disease prevalence) and Miniature Dachshunds, and rcd4 PRA in Gordon Setters and Irish Setters. With rcd4 PRA, the average age of clinical diagnosis of the disease is around 10 years of age. However, this is the average age that clinically affected dogs are recognized; not the average age when all homozygous dogs become affected. The actual average "age of onset" of this late-onset disease may be in the teens; when many dogs will already be deceased. Now that Gordon Setters are being tested worldwide, many dogs who are homozygous for the defective gene with normal vision are being identified. With an approximately 30% carrier rate in the small Gordon Setter gene pool, selection against the gene must necessarily be gradual so as to not restrict the breed's genetic diversity.

Other susceptibility genes are found to occur at a greater frequency in affected animals, but **are not present in all**

affected animals. An example is the susceptibility gene for perianal fistula/anal furunculosis in German Shepherd Dogs. Dogs with the susceptibility haplotype (specific sequence of 3 DLA genes) have a 5.0X odds ratio for the disease versus those without the haplotype. This risk factor occurs whether the susceptibility haplotype is heterozygous or homozygous; though homozygous dogs develop the disease at an earlier age. Another example is the genetic test for Pug Dog Encephalitis, a painful, fatal disease affecting 1-2% of Pugs. Dogs homozygous for a susceptibility haplotype have a 15.6X odds ratio for developing the disease, but dogs heterozygous for the susceptibility haplotype have no greater risk.

Utilizing Genetic Tests

We need to be knowledgeable about what genetic tests are available, and in which individuals they should be run. Dogs from breeds with an incidence of von Willebrand disease should be tested early in life, so that measures can be taken to prevent excessive hemorrhage during surgery or injury. Dogs in breeds at risk of carrying the mdr-1 drug sensitivity mutation should be tested early in life, before drug treatment. In high risk breeds, individual animals should be genetically tested (or verified results documented for parents) before purchase. These include Maine Coon and Ragdoll cats for the autosomal dominant hypertrophic cardiomyopathy gene, Persian and Himalayan cats for autosomal dominant polycystic kidney disease, Boxers for arrythmogenic right ventricular cardiomyopathy, and Doberman Pinschers for dilated cardiomyopathy.

We need to understand the temporal periods when genetic testing will be most accurate, and allow for intervention. Puppy hips should be palpated with a gentle Ortolani procedure at each vaccine visit, and again at spaying or neutering under anesthesia. Juvenile interventional surgery will only benefit those with significant subluxability prior to major growth (for pubic symphysiodesis) or the development of osteoarthritic changes (for triple pelvic osteotomy). Genetic testing for inherited hypothyroidism (autoimmune thyroiditis) is based on the presence of thyroglobulin autoantibodies. A dog with normal TgAA levels on two tests at least two years apart between two and six years of age is phenotypically normal. However, TgAA levels should not be measured within 2-3 months post-vaccination, as a transient iatrogenic rise can occur during this period.

For most genetic diseases, we know how to either prevent their occurrence, or at least lessen the possibility of producing offspring with genetic disease. This can occur through the genotypic testing of the parents (identification of parents carrying liability genes for genetic disease), phenotypic testing of the parents (identification of parents affected with genetic disease), or pedigree analysis (identification of carrier and affected risk based on the knowledge of carrier or affected relatives).

The genetic improvement of cats and dogs will only occur through selective breeding. Inherent in this point is the acknowledgement that breeding without genetic testing is irresponsible, and unethical. **Genetic testing is health quality control.** It is no longer acceptable for a breeder to choose two individuals and breed them together without regard to genetic disease control. The responsibility for genetic improvement lies not just with the breeder; but also with the breed organizations, veterinarians, and the general public. Breeders must perform genetic testing on prospective breeding stock before breeding. Breed organizations must identify breed specific health issues through regular breed health surveys, fund research for breed specific disorders, and recommend genetic testing for breeding animals. Veterinarians must counsel prospective owners and breeders on breed specific health issues. They should provide the necessary genetic testing, or direct owners to specialists (ophthalmologists, cardiologists, etc.) that can perform the testing. The general public must become knowledgeable about what genetic tests are needed on parents of prospective kitten and puppy purchases, and how to verify testing status.

The most important goal of managing genetic disease is to avoid producing affected individuals. The secondary goal is to reduce the frequency of carriers of defective genes in the population. At the same time, recommendations should allow perpetuation of breeding lines, in order to preserve the genetic diversity of the population. With each new generation, breeders ask, “How can I continue my line and improve it?” Aside from selecting for conformation, behavior and general health, breeders must consider how they are going to reduce the incidence of whichever genetic disorders are present in their breed. There are no answers that will fit every situation. There are, however, guidelines to preserve breeding lines and genetic diversity while reducing the risk of producing animals that carry defective genes, or are affected with genetic disorders.

Autosomal Recessive Disorders

In the case of a simple autosomal recessive disorder for which a *direct genetic test for carriers* is available, the recommendation is to test breeding-quality stock, and breed normals to normals, or carriers to normal-testing individuals. This prevents affected offspring from being produced.

Breeders are the custodians of their breed's past and future. "Above all, do no harm" is a primary oath of all medical professionals. Genetic tests are powerful tools, and their use can cause significant positive or negative changes to breed gene pools. Once a genetic test is developed that allows breeders to determine if an animal is a carrier of a defective gene, many owners are likely to simply eliminate carriers from breeding. Although doing so is human nature, this temptation must be overcome. If an owner would breed an individual if it tested normal for a genetic disease, then a carrier result should not change that decision. A direct

genetic test should not alter WHO gets bred, only WHO THEY GET BRED TO. One defective gene that can be identified through a genetic test out of tens of thousands of genes is not a reason to stop breeding. A genetic test that should be used to help maintain breed quality and diversity should not result in limiting it.

We know that most individuals carry some unfavorable recessive genes. The more genetic tests that are developed, the greater chance there is of identifying an undesirable gene in a breeding animal. History has shown that breeders can be successful in reducing breed-wide genetic disease through testing and making informed breeding choices. However, there are also examples of breeds that have actually experienced more problems as a result of unwarranted culling and restriction of their gene pools. These problems include: 1) Reducing the incidence of one disease and increasing the incidence of another by repeated use of males known to be clear of the gene that causes the first condition. 2) Creating bottlenecks and diminishing diversity by eliminating all carriers of a gene from the breeding pool, instead of breeding and replacing them. 3) Concentrating on the presence or absence of a single gene and not the quality of the whole animal.

The aim is to replace the carrier breeding-animal with a normal-testing offspring that equals or exceeds it in quality. Additional carrier testing 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 carrier animals with normal-testing offspring, the problem for the breed as a whole diminishes, while not restricting gene pool diversity.

The problem with a simple autosomal recessive disorder for which *no carrier test exists* is the propagation and dissemination of unapparent carriers in the gene pool. A quality individual that is found to be a carrier of a recessive gene can be retired from breeding and replaced with a quality relative or prior-born offspring. The genes of the retired individual can thus be preserved through the selected relative, but the carrier risk can be cut in half. To further limit the spread of the defective gene, the offspring should be used in only a limited number of carefully planned matings, and then should also be replaced with one or two representative offspring. The rest of the litter should be placed in non-breeding (pet) homes. With this mating scheme, you are maintaining the good genes of the line, reducing the carrier risk with each generation, and replacing, not adding to the overall carrier risk in the breeding population.

Breeders must assess the carrier risk of each individual animal in their breeding program. An open health registry that is supported by the parent club makes it easier for breeders to objectively assess these matters. An example is the genetic disease control program for cerebellar abiotrophy by the Scottish Terrier Club of America (http://www.stca.biz/health-registries/ca-registry). By determining the average carrier-risk for the breeding population, breeders can select matings that have a projected risk that is lower than the breed average. Relative risk assessments only take into account the identified carrier and affected individuals in the pedigree. Therefore, these estimates determine the minimum risk based on the information available. If additional affected relatives to the pedigree are diagnosed, the computed risk will rise. The relative risk pedigree calculator on the Scottish Terrier website can be used by any breed to compute carrier and affected risk for any simple autosomal recessive disorder.

If a quality breeding animal is at high risk of being a carrier, the best advice is to breed to an individual that has a low risk. Using relative-risk assessment as a tool, breeders should replace higher-risk breeding animals with lower-risk offspring that are equal to or better than their parents in quality. 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

Autosomal dominant genetic disorders are usually easy to manage. Each affected animal has at least one affected parent, but it can be expected that half of the offspring of an affected animal will be free of the defective gene. With disorders that cause death or discomfort, the recommendation is to not breed affected animals. To produce the next generation of a line; a normal full sibling to an affected animal, a normal close relative, or the parent that is normal can be used.

If the defective gene is at a high frequency in the gene pool, eliminating all affected breeding animals in one generation may have a significant negative impact on genetic diversity. When a high frequency autosomal dominant disorder is first identified, some quality, affected animals may have to be bred, and replaced with quality, normal testing offspring. This occurred with the identification of the gene for polycystic kidney disease in Persian cats (38% affected), and hypertrophic cardiomyopathy in Maine Coon Cats (33% affected). However, once a few generations have gone by and breeders have had the opportunity to replace affected with normal individuals, the continued breeding of affected animals is not ethical.

A problem with some autosomal dominant disorders is incomplete penetrance; where some animals with the defective gene may not show the disorder. Roughly half their offspring, however, may be affected. If a genetic test is available, this is not a problem. Otherwise, pedigree analysis and relative-risk assessment can identify which animals are at risk of carrying incompletely penetrant dominant genes.

Sex-Linked Disorders

For sex-linked (also known as x-linked) recessive defective genes for which carrier tests exist, breeders should follow the same "breed and replace" recommendations as are outlined above in the discussion of autosomal recessive disorders. If there is no test, the defective gene can be traced through the pedigree. Selecting a normal male for breeding loses the defective gene in one generation, regardless of his relationship to affected and carrier relatives. Carrier, affected, or high risk females should not be used, due to the high risk of producing affected male offspring. If a male is affected, he would have received the defective gene from his carrier mother. All of his daughters will be carriers, but none of his sons. Without a test for carriers, you can use relative-risk assessment to breed him to a female that is at low risk of being a carrier. This minimizes the chance of producing affected offspring, and a quality son can be selected for replacement. Rare sex-linked dominant disorders are managed the same way as autosomal dominant disorders. The difference is that affected males will always produce all affected daughters.

Polygenic disorders/Complex Inheritance

Polygenic disorders are those caused by more than one pair of genes. A number of liability genes must combine to cross a threshold and produce an affected individual. Most polygenic disorders have no tests for carriers, but they do have phenotypic tests that can identify affected individuals. Controlling polygenically inherited disorders involves; 1) identifying traits that more closely represent genes being selected against, 2) the standardization of nuisance factors (such as environment) that can limit your selective pressure against the genes and 3) selecting for breadth of pedigree as well as depth of pedigree.

In polygenic disorders, the phenotype of the individual does not directly represent its genotype. If phenotypically normal parents produce affected offspring, both should be considered to carry a genetic load of liability genes that combined to cause the disorder. Breeders must break down affected phenotypes into traits that more directly represent the genes that control them. For example, in hip dysplasia these can include clinical signs of lameness, shallow hip sockets, subluxation or remodeling on an extended leg view, and radiographic distractibility on a PennHIP view. If a quality individual is to be bred, but has shallow hip sockets, it should be bred to an individual with deep hip sockets. You need to select for enough genes influencing normal development, to get below the threshold where dysplasia develops.

The environment has a role in the expression of polygenic disorders. Plane of nutrition and environmental stress, especially during critical growth periods can alter the expression of some inherited musculoskeletal disorders. You do not want to overly protect or overly stress the development of prospective breeding animals. Breeders should evaluate prospective breeding individuals raised under fairly uniform conditions, which will not mask or alter the expression of genetic disease.

Polygenic disorders require knowledge of the affected or normal status of full-siblings to prospective breeding animals. Individuals whose siblings are normal and whose parents' sibs 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.) This is why it is important to screen both pet and breeding animals from litters for polygenic disorders, and report the results in open health registries, such as the not-for profit Orthopedic Foundation for Animals (www.offa.org). Breadth of pedigree results can be visualized by clicking on vertical pedigrees on the individual dog's OFA page.

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. In addition, the offspring of breeding dogs should be monitored to see which are passing the disorder with higher frequency.

Undetermined Mode of Inheritance

For disorders without a known mode of inheritance or carrier test, breeders should be counseled to use the same control methods as with polygenic disorders. Animals with a low genetic load for the disorder should be selected for breeding, through the results of examinations of first-degree relatives (littermates, parents, and offspring). If there are multiple generations of normalcy in the breadth of the pedigree, then you can have some confidence that there is less risk that liability genes are being carried.

It is distressing when a genetic disorder is confirmed. Positive and practical genetic counseling recommendations can be made to maintain breed lines and genetic diversity, and improve the overall health of breeds. The total elimination of defective genes will probably be impossible for most breeds. The use of these guidelines can assist breeders in making objective breeding decisions for genetic disease management, while continuing their breeding lines. The individual breeder can use genetic tests to; 1) identify carriers, 2) work to breed away from the defective gene(s), and 3) ensure (through testing) that the defective gene(s) is not reintroduced in future matings. Each breeder will have their own rate of progress, depending on the frequency of the defective gene(s) in their own breeding animals, and which desirable individuals carry liability genes.

Improving the Genetic Health of Your Puppies

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Many breeders use the HGH method of genetic disease control. This stands for “Hope for Good Health”. The most important concern of the public on purchasing a puppy is on its health.

RESPONSIBILITY; *noun.* Duty, obligation, burden.

What is the responsibility when someone decides to breed two dogs together to produce a litter? Everyone knows that genetic disease can occur in any dog. A dog with genetic disease suffers morbidity and/or mortality. For most genetic diseases, we know how to either prevent their occurrence, or at least lessen the possibility of producing offspring with genetic disease. This can occur through the genotypic testing of the parents (identification of parents carrying liability genes for genetic disease), phenotypic testing of the parents (identification of parents affected with genetic disease), or pedigree analysis (identification of carrier risk based on the knowledge of carrier or affected relatives).

It is the ethical responsibility and obligation of all breeders to perform the available required pre-breeding genetic health tests on prospective breeding stock. Nowhere has it ever been stated that breeders can; take a pass, ignore, excuse themselves, or exempt their breeding stock from genetic health testing. Genetic disease does not “just happen”. It is predictable, and for the most part preventable. Genetic testing is a requirement, not a choice.

A breeder is anyone that plans a mating between two dogs. These include matings between two members of the same breed, or crosses between two members of different breeds (designer matings). The fallacy that mixed-breed dogs have less genetic disease is disproven every day in veterinary hospitals. The most common genetic diseases, including; canine hip dysplasia, valvular heart disease, patella luxation, and hypothyroidism occur at similar frequencies in mixed-breed versus pure-bred populations. If two dogs are purposely bred, then the breed specific genetic testing for each parent is required.

Many genetic disorders cannot be absolutely prevented. They have complex, multi-gene inheritance, and liability genes have not been identified. However, for most of these inherited disorders, a method of diagnosing affected individuals is documented. It is also documented which breeds have a higher liability for these inherited disorders. Breeding decisions by health conscious breeders based on test results or diagnoses is a powerful tool for improved genetic health.

Most genetic tests only need to be done once in the prospective breeding dog’s lifetime. Others (eye examinations, phenotypic heart examinations, thyroid profile, etc.) should be repeated, depending on the breed specific age of onset of the disorder, and age requirement for diagnosis. If you are not willing or able to have the prescribed pre-breeding genetic tests performed, then you should find a different hobby or profession. Dogs are living beings. It is not ethical to forgo the obligation of genetic testing.

Prospective breeding dogs should have their health testing completed prior to being bred. Breed specific pre-breeding health test requirements are available on the Canine Health

Information Center (CHIC) website: www.caninehealthinfo.org/breeds.html. It is not necessary that your breeding stock pass all of the required health tests. Dogs become CHIC certified by completing the health requirements, regardless of the test results. This shows health consciousness. If your breed club has not yet joined CHIC by identifying the breed specific pre-breeding tests, then your dog should undergo screening for the common musculoskeletal diseases, heart disease, eye disease, thyroid disease, and any specific genetic diseases seen at an increased frequency in the breed. Using the "Statistics and Data" tool on the OFA website (http://www.offa.org/stats.html) for instance, shows that Pugs are the #2 breed with hip dysplasia (62.4% affected) and #15 breed with patella luxation (7.8% affected).

Many health tests can be performed during an examination with your veterinarian, or obtained inexpensively at local health screening clinics. These can be found on the OFA website: http://www.offa.org/clinics.html, or on an excellently maintained Cavalier King Charles Spaniel website: http://www.cavalierhealth.org/health_clinics.htm. A list of available genotypic tests is included in the proceedings of the 4th Tufts Canine and Feline Breeding & Genetics Conference: www.vin.com/tufts/2009.

The Orthopedic Foundation for Animals maintains registries for testable genetic disorders in dogs on their searchable website: www.offa.org/search.html. For many disorders, genetic test results for individual breeding dogs are available on-line. When obtaining a dog for breeding purposes, you can type in the parent's name or registration number (from any registry; AKC, CKC, UKC, Sally's Registry, etc.), and if test information is available the dog's webpage will come up. This is Facebook for dogs, with their own web pages and health test information. Each page also includes health test information on a dog's parents, offspring, full-siblings, and half-siblings, which allows breadth of pedigree analysis. If a dog's

SHAANKATA'S TEQUILA TEASER
CHIC

Registration:	SN89876701	**Sire:**	CJ199702
Breed:	PARTI-COLOR COCKER SPANIEL	**Dam:**	EQ323261
Sex:	F	***Titles:**	CH
Color:	BLACK & WHITE	**CHIC #:**	27495
Birthdate:	Jan 9 2000	**Addtl. Reg. #**	KA640960
DNA Profile:			

OFA Number	Registry	Report Date	Age	Final Conclusion
CS-CA19/15F/C-PI	CARDIAC	Sep 18 2001	15	NORMAL - CARDIOLOGIST
CS-PA129/24F-PI	PATELLA	Feb 19 2002	24	NORMAL
CS-7428E31F-PI	HIPS	Sep 12 2002	31	EXCELLENT
CS-LP74/31F-PI	LEGG-CALVE-PERTHES	Jun 23 2005	31	NORMAL
CS-2682	CERF	Oct 17 2005	68	TESTED: 00,03,04,05
CS-TH29/74F-PI	THYROID	Apr 27 2006	74	NORMAL

Sire/Dam	Registration	Birthdate	Sex	Relation	CARDIAC	CERF	GDC HIPS	HIPS	THYROID
NICKLEODEON'S DEVIL MAY DARE	SN28739801	May 18 1994	M	Sire	CS-CA6/54M/C-T			CS-4509G24M-T	
KATAXIN SHAANKATA TEQUILA MITE CHIC	EQ323261	Aug 4 1995	F	Dam		CS-2393	OFA11060H24GN	CS-5045G24F-T	CS-TH6/84F-PI

Offspring	Registration	Birthdate	Sex	CARDIAC	CERF	PATELLA	THYROID
SHAANKATA KATAXIN GUILTYAS SIN	PC926097	Feb 14 2004	F	CS-CA47/17F/C-PI	CS-3611	CS-PA256/17F/P-PI	CS-TH69/25F-PI
SHAANKATA'S PEPERMINT PIE	PW982176	Nov 16 2004	F		CS-3612		CS-TH70/15F-PI

Half Siblings(Sire)	Registration	Birthdate	Sex	Relation	CARDIAC	ELBOW	CERF	GDC HIPS	HIPS	LEGG-CALVE-PERTHES
CLASSIC'S LADY SINGS THE BLUES	SN43821301	Apr 6 1997	F	Half(Sire)	CS-CA5/19F/C-T		CS-2143			
NICKLEODEON CLASSIC JAZZ CHIC	SN55252301	Aug 18 1997	M	Half(Sire)			CS-3602		CS-7641G66M-PI	CS-LP620/66M-PI
CLASSIC'S MYSTICAL IMAGE		Mar 10 2000	M	Half(Sire)			CS-3148	GDC20672H18EN		
RENDITION FINAL DESTINATION	SN81275203	Oct 5 2000	M	Half(Sire)					CS-7629G28M-NOPI	CS-LP606/28M-NOPI
NICKLEODEAN LIVIN LA VIDA LOCA	KY713425	Dec 20 2000	M	Half(Sire)		CS-EL137M50-NOPI			CS-8770G50M-NOPI	CS-LP1724/50M-NOPI
CHRIS-DI'S DEVILISH ENCHANTMENT	SN87204801	Aug 19 2001	M	Half(Sire)					CS-7948G24M-PI	CS-LP925/24M-PI
GUNTHERS AMERICAN HERO CHIC	SN91623203	Jul 4 2002	F	Half(Sire)			CS-3729		CS-8385E24F-PI	CS-LP1366/24F-PI

information does not come up on the OFA website, you should also check the Canine Eye Registry Foundation (CERF) website for eye examination results: www.vmdb.org/verify.html.

If test results are not available for the parents of a dog that you are looking to purchase on the web-based registries, ask the owner if the breeder provided them with verification of each of the required genetic test results on the parents; I.e., a copy of the official test results from the testing agencies. If no verified test

results are available, then the puppy was not bred by a health conscious breeder. There is no expectation of genetic health. When you perform genetic testing prior to breeding, the results may be a surprise.

As most of you are commercial breeders, you will not receive feedback on the health status or genetic test results of the puppies that you sell. The only way to know the genetic disease risk of your breeding stock is through their test results, and the test results of their parents and relatives.

Occasionally a genetic disease just appears without warning. However, most common genetic diseases are predictable. Using valid breeding strategies, these genetic diseases are for the most part preventable. Simple Mendelian (one gene pair) disorders with a direct genetic test are absolutely preventable. With direct genetic tests for recessive disorders, carriers can be bred to normal testing mates. This prevents affected offspring from being produced. The long-term goal is to have normal testing breeding stock. With quality carrier breeding stock, you can prevent affected offspring, and replace quality carriers with normal testing offspring for your next generation of breeding stock.

Dogs affected with an autosomal dominant disease should be selected against, as half of their offspring will be affected with the genetic disorder. With polygenic or complexly inherited disorders, knowledge of the test results or affected status of the close relatives allows the breeder to recognize the risk of producing the genetic disorder. Breadth of pedigree normalcy (test results of the siblings of prospective breeding dogs) gives a better indication of the genetic load of normal genes that the prospective breeding dog may carry. Breadth and depth of pedigree normalcy can be visualized in vertical pedigrees on the dog's page of the OFA website.

Figure 3: OFA Vertical Depth & Breadth of Pedigree

Complexly inherited disease and diseases without direct tests for carriers cannot absolutely be prevented. No one wants to produce genetic disease, and we can all empathize when it does occur. If you are doing genetic testing and plan matings accordingly, you are doing your best to be health conscious and fulfilling your breeder responsibility.

Everyone loves their breed, and their own breeding stock. The more genetic tests that are developed, the greater chance there is of identifying an undesirable gene in your dog. Conscientious breeders understand that negative test results limit their breeding options. With direct gene tests, you can use

carriers when bred to normal testing mates. For disorders without direct gene tests, you may have to choose a normal relative, as opposed to one you were planning on using in the next generation. Matings should be planned that prevent or minimize the risk of producing genetic disease.

When your prospective breeding stock has a carrier or affected test result, please release this information to the OFA or other listing registry. (You have to mail CERF results in to release negative eye examination results.) If negative test results are not made available, then other breeders will not be able to ascertain the disease risk of their own breeding stock to make informed breeding decisions. An OFA webpage of all normal individuals is great to see, but of little value if those with negative results are not listed. As opposed to the stigma that used to be attached to the *appearance* of genetic disease, the stigma now rests on those that hide the occurrence of genetic disease. Dealing with genetic disorders is a community effort.

When making breeding decisions, you can search the OFA, CERF, CHIC or other websites for genetic test results on prospective mates. The OFA search page is extremely flexible and useful on search requirements. If test results are not available on dogs that have already been bred, then it must be assumed that they are affected or carriers – otherwise the results would be available.

When selling a puppy, please provide your new owners with full documentation of the health test results (copies of official test results) on the parents. It is not enough to say that the testing was done. If testing was done then you have the paperwork, and it should be provided. It must be impressed upon the public that health consciousness is one of the most important considerations when getting a puppy.

Health guarantees that provide for replacement of puppies with genetic defects are not a replacement for health testing. Such a guarantee is of little value, as no one wants to part with their family member once the emotional bonds have been made. This is not a toaster. Breeders need to fulfill their ethical responsibility and obligation of health testing.

The frequency of genetic diseases can be significantly decreased, if not eliminated by valid testing and breeding selection in purposely bred dogs. We are now at the point where the tools are available, and the information is well established. It is time to put an end to the excuse of ignorance of the breeder in their role and responsibility to improve the genetic health of dogs.