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Veterinary genetic counseling service: a preliminary report

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Abstract

Data generated from an online veterinary genetics counseling service from November 1, 2018 to October 31, 2019 were reviewed. Data were tabulated regarding the individual submitting the inquiry and relationship to the pet, species, breed, and medical issue(s) thought to be familial. Inquiries were assigned to 1 or more of the following: risk assessment and heritability, pre-breeding testing and pedigree review, and information regarding a specific mutation or minor health concern. This study demonstrated that clients are desirous for information that could help promote health in canine companions. Furthermore, there is an interest and need for additional genetic resources for both veterinarians and owners.

Keywords: Canine, genetic counseling, heritability, genetic testing

Introduction

Improvement of offspring genetic health begins with an understanding of the selection tools that are available and the ethical responsibilities to use these tools in assisting with breeding decisions.[1] Discovery of genetic markers of disease in companion animals and the advent of readily available at-home testing kits increased testing for genetic disease in companion animals.[2] Dog's breed identification testing is performed to fulfill owner's interest and curiosity. However, knowing that their pet carries a genetic mutation for a specific medical issue might have important clinical implications for the future care of the pet. Although the ability to estimate an individual animal's risk of genetic disease has become increasingly available,[3,4] accessible resources to navigate test interpretation and clinical implications of the findings might be inadequate, leaving owners and veterinarians with many unanswered questions.[5,6] Veterinarians, especially those working in general practice or having an interest in theriogenology, are often the first line of contact for evaluation of an animal suspected for a familial disease or a genetic problem. Genetic diseases are of significant concern for veterinarians and their patients.

A genetic online veterinary counseling service was created to provide a forum so that the complex nature of genetic disease could be better navigated and discussed. To the authors' knowledge, this is the first database of its kind providing a free, readily available counseling service. Services are provided by a university veterinary genetics laboratory in conjunction with the theriogenology services of 2 veterinary colleges. Goals are to provide information regarding familial diseases, specific mutations, and genetic test interpretation to veterinarians, pet breeders and owners.

Objective of this paper was to review 1 year data generated from this service and provide a summary of the types of questions and areas of concerns that veterinarians and owners submitted regarding familial diseases.

Materials and methods

Consultations were provided by a team including a veterinarian with a PhD in genetics, a veterinarian who had completed a medical genetics residency, and 2 theriogenology residents in combined residency/medical genetics programs. The service was promoted both through its location on the genetic testing website (https://cvm.ncsu.edu/nc-state-vet-hospital/small-animal/genetics/) and through the American Kennel Club-Canine Health Foundation.

Consultation requests from November 1, 2018 until October 31, 2019 were gathered and tabulated. Information on the individual submitting the inquiry and their relationship to the pet was obtained. The term breeder was defined as any person who owns or plans to own an animal intended for breeding, or has an animal already used to produce progeny. Pet owner was defined as any person who owns or plans to own an animal not intended for breeding purposes. A veterinarian acted on behalf a client where a valid veterinary-client-patient relationship existed. Patient demographics including species, breed, and medical issue(s) recognized as familial were collected. Each inquiry was assigned to 1 or more of the following: risk assessment and heritability estimates, pre-breeding testing, pedigree record review, information regarding a specific mutation (either to determine presence or have results analyzed), and minor

health concern (including coat color, breed make-up, and age).

Results

Thirty-five submissions (Table 1) were received and evaluated through the online genetic testing portal during the 12-month interval. Over half (63 %; 22/35) of the submissions were from pet breeders with 2 of those indicating breed club affiliation with the intent to distribute knowledge to their respective breed club. Of the remaining submissions, 20% (7/35) were pet owners with questions regarding an individual animal and 17% (6/35) were veterinarians submitting on behalf of a client with questions regarding an individual animal, unrelated to breeding. Although the service was provided for all species, all submissions were for dogs. Inquires represented dogs in 23 American Kennel Club recognized breeds, and 4 mixed breed, 2 non-American Kennel Club breeds, and 1 foundation stock service breed.

Overall, 75% (26/35) of the inquiries were related to breeding an animal and the impact of a familial trait on the breed overall and 25% (9/35) were inquiries regarding the impact of a trait on an individual pet animal not intended for breeding. Inquiries regarding risk assessment and heritability were most common (17 inquiries), followed by pre-breeding testing and pedigree record review (9), information regarding a specific mutation (either to determine presence or have results analyzed (6), and of minor health concern (6 [including coat color, breed make-up, and age]). Distribution of inquiries is summarized (Figure).

Genetic consultations are categorized by organ system (Table 2). Largest number of inquiries included questions regarding neurologic, musculoskeletal, and ophthalmic diseases or disorders. Although kinked tails and umbilical hernias are seemingly relatively benign issues, kinked tails could be indicative of other spinal abnormalities and could be disqualifications in conformation showing for certain breeds. Similarly, umbilical hernias

Table 1. Number of inquiries for each breed and topic of inquiry

Breeds represented	Number of inquiries	Topic of inquiry
Labrador Retriever	4	Ocular certification; hepatitis with copper accumulation; hemangiosarcoma; coat color
Mixed breed	4	Coat color; age testing; breed identification & dilated cardiomyopathy; deafness
Australian Shepherd	2	Breed screening pre-breeding & coat color; degenerative myelopathy
Golden Retriever	2	Cataracts
American Bully	1	Breed screening pre-breeding
Beagle	1	Degenerative myelopathy
Bearded Collie	1	Coat color
Black Russian Terrier	1	Epilepsy
Boston Terrier	1	Deafness
Cairn Terrier	1	Breed screening pre-breeding
Cavalier King Charles Spaniel	1	Midline defect (cleft palate), kinked tail, umbilical hernia & hip dysplasia
Chinese Shar-Pei	1	Persistent right aortic arch & megaesophagus
Clumber Spaniel	1	Midline defect (bifid nose) & umbilical hernia
Doberman Pinscher	1	Dilated cardiomyopathy & Von Willebrand's Disease
Giant Schnauzer	1	Hypothyroidism
Great Dane	1	Polycythemia Vera
Labradoodle	1	Hypoadrenocorticism
Leon Berger	1	Hip dysplasia
Miniature Schnauzer	1	Breed screening pre-breeding
Norwich Terrier	1	Norwich Terrier
Pembroke Welsh Corgi	1	Degenerative myelopathy & cystic endometrial hyperplasia-pyometra complex
Portuguese Water Dog	1	GM1-gangliosidosis
GM1-gangliosidosis	1	Breed screening pre-breeding
Standard Poodle	1	Keratoconjunctivitis sicca & hypoadrenocorticism
Vizsla	1	Cataracts
Whippet	1	Lung lobe torsion, skin allergy & hemangiosarcoma

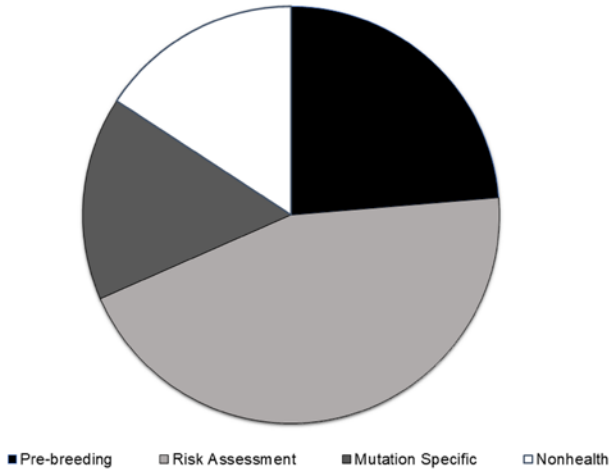

Figure. Distribution of inquires

might have a hereditary basis and could have larger implications regarding a dam's ability to carry a litter to term successfully.

Discussion

The majority of inquiries to the genetics consultation service were from individuals self-identified as dog breeders; however, almost 40% of the inquiries came from dog owners or veterinarians requesting information regarding an individual pet indicating increased interest in familial disease beyond that needed for breeding purposes. Although inquiries were made most commonly regarding purebred dogs, mixed breed dogs were also represented. There were zero inquiries regarding familial health issues in cats or other species. This could simply be a reflection of promotion of the program through the American Kennel Club-Canine Health Foundation. However, it could also be indicative of a lesser understanding or interest in familial disease in the cat. Promotion of this tool to cat breeders and cat owners might be beneficial to draw attention to feline familial health concerns.

There were 4 inquiries regarding mixed breed dogs indicating an interest in genetics and familial disease, even in mixed breed dog populations. The most common disease or disorder inquiries appeared to parallel the commonality of diseases that occur across canine population and included: cataracts, degenerative myelopathy, deafness, endocrine disease, cancer (hemangiosarcoma), and dilated cardiomyopathy.[8,9] However, some inquiries were in diseases to be familial (lung lobe torsion) or without a well-known breed predilection for an issue. This might be associated with an increased interest and curiosity regarding genetics as a cause of medical issues even in situations where it is not clearly a familial issue.

Questions regarding risk assessment, heritability and pre-breeding screening were most common. However, questions regarding genetic test interpretation were also frequent and illustrate the important need for increased availability and promotion for additional guidance in this area.

Table 2. Genetic consultations categorized by organ system (number within parenthesis indicates total consults)

Organ System	Disease, Disorder or Condition of Concern
Neurologic	Degenerative myelopathy[3] Deafness[2] GM1-gangliosidosis Epilepsy
Ophthalmic	Cataracts[3] Keratoconjunctivitis sicca
Digestive	Hepatic fibrosis Megaesophagus Hepatitis with copper accumulation
Respiratory	Lung lobe torsion
Urogenital / Reproductive	Cystic renal dysplasia Ectopic ureter Cystic endometrial hyperplasia-pyometra complex
Endocrine	Hypoadrenocorticism[2] Hypothyroidism
Musculoskeletal	Midline defects[2] Umbilical hernia Kinked tail Hip dysplasia
Cardiovascular	Dilated cardiomyopathy[2] Persistent right aortic arch
Integumentary	Skin Allergies
Other	Cancer risk – hemangiosarcoma[2] Von Willebrand's Disease Polycythemia Vera

This study demonstrated the interest and need for additional genetic resources for veterinarians and owners who have access to genetic tests. Careful use of genetic information (refer 'selected genetic resources for veterinarians and clients') might help to prevent or decrease disease occurrence in companion animals. However, the impact of genetic testing on animal health is likely dependent on the owner and veterinarian understanding the genetic information obtained from the test and its possible applications.[10] As these records imply, clients are interested in information that could help promote the familial health of their canine companions. As concerted efforts are made in implementing canine genetic counseling recommendations, a shift in the health of dogs might be incrementally improved with both time and knowledge.[1]

Conflict of interest

None to report.

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Selected genetic resources for veterinarians and clients		
Orthopedic Foundation for Animals/ Canine Health Information Center	Database, statistics by breed, disease overview, DNA tests, research, health screening clinic search	https://www.ofa.org/
Canine Health Foundation	Review of the current state of genetic testing in dogs, a living resource	https://www.akcchf.org/educational-resources/library/articles/review-of-the-current-state.html
The International Partnership for Dogs (in collaboration with CHF, WASAVA and others)	Harmonization of genetic testing for dogs by breed, disease or test, labs	https://dogwellnet.com/ctp/
UC Davis Veterinary Genetics Laboratory	DNA testing	https://vgl.ucdavis.edu/
UPEI CIDD Database	Canine inherited disorders database	https://cidd.discoveryspace.ca/index.html
UPenn PennGen	DNA testing	https://www.vet.upenn.edu/research/academic-departments/clinical-sciences-advanced-medicine/research-labs-centers/penngen

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