SPECIAL REPORT
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The brave new world of clinical veterinary genetics

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ABSTRACT

The creation of purebred breeds of domestic animals has been accompanied by inbreeding and an increased incidence of simple genetic diseases within these breeds and animals. The veterinary profession has largely been a spectator of recent developments in companion animal genetics. A review of the current state of veterinary genetics for the small animal practitioner is provided that includes an overview of commercial DNA service providers as well as a review of information sources useful for clinical veterinary genetics.

RÉSUMÉ

Le meilleur des mondes de la génétique vétérinaire clinique

La création de races pures d'animaux domestiques s'est accompagnée de consanguinité et d'une augmentation de la fréquence de maladies génétiques simples parmi ces races et ces animaux. La profession vétérinaire a été plutôt spectatrice des développements récents en matière de génétique des animaux de compagnie. Une revue de l'état actuel en génétique vétérinaire destinée aux praticiens dans le domaine des animaux de compagnie est fournie, comprenant une vue d'ensemble des fournisseurs commerciaux de services d'ADN ainsi qu'une recension des sources d'informations utiles pour la génétique vétérinaire clinique.

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The creation of purebred breeds of domestic animals has been accompanied by genetic bottlenecks, inbreeding, and an increased incidence of simple genetic diseases. Constantly developing technologies have contributed to our understanding of these genetic diseases, and include traditional DNA sequencing, polymerase chain reaction (PCR) amplification of DNA and more recently developed techniques such as DNA microarrays and high-volume DNA sequencing methods. Traditionally, university-based not-for-profit DNA service labs, associated with academic research labs, have offered DNA diagnostic tests to both veterinarians and animal breeders. More recently, these service labs have been replaced by for-profit companies offering microarray-based DNA profiling tests for domestic animals, using a direct-to-consumer business model, common in the human sector. Initially, these for-profit companies targeted the dog-owning public, offering genetic analysis of dog breeds. In addition, they are now targeting animal breeders and veterinarians by including trait and genetic disease markers on their DNA microarrays. Another trend is for smaller startup DNA diagnostic companies to be bought and incorporated into large multinational food and pharmaceutical companies. These multinational companies are now in stiff competition for market share of the lucrative direct-to-consumer DNA testing of dogs and cats. Indeed, DNA microarray profiling services can provide information on > 250 disease-causing DNA mutations for dogs and > 50 for cats, in addition to trait and breed

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analyses. However, the fact that only a fraction of these mutations is of relevance for a particular breed has led to the description of this business model and as technologies in search of a marketplace.

EDUCATIONAL AND SERVICE PROVIDERS

The veterinary profession has largely been a spectator to these developments in companion animal genetics. This being so, the profession needs access to reliable non-commercial information regarding genetic diseases of domestic animals, plus knowledge of animal genetics.

The following is a snapshot view of a rapidly evolving area and is not intended to be exhaustive. The names of companies do not represent endorsements and are presented only as examples, with web addresses included for convenience. Descriptions represent the author's opinion. Unless noted, websites are in English only.

Labgenvet (https://labgenvet.ca/)

Labgenvet is an academic resource originating from the Faculty of Veterinary Medicine, University of Montreal that provides a web-based source of academic and clinical information concerning domestic animal genetic diseases. Labgenvet was created primarily for Quebec and Canadian clinical veterinarians, with international relevance. The website contains searchable databases of simple genetic diseases for dogs, cats, cattle, and horses. For dogs and cats, the disease database can be searched and regrouped by breed, by gene, by disease name, and by disease system. Disease pages are provided that present genetic, hereditary, and clinical information relevant to the veterinary clinician. Unique to the Labgenvet website is a systematic presentation of genetic disease mutation frequencies listed by individual dog and cat breed. Links to the scientific literature for genetic diseases are provided. The website functions in both English and French.

OTHER NOT-FOR-PROFIT EDUCATIONAL AND INFORMATIONAL RESOURCES

Online Mendelian Inheritance in Animals (OMIA) (https://www.omia.org/home/)

OMIA, affiliated with the University of Sydney (Australia), is the omnibus academic database and resource for simple genetic inheritance and diseases of multiple animal species, including domestic animals. It is current, curated, and has search functions that work well for genes and individual diseases, but it is not designed for breed compilations of diseases or for frequencies of disease mutations by breed. Functional links to the scientific literature are provided.

DogWellNet (https://dogwellnet.com/)

DogWellNet is the website for The International Partnership for Dogs (IPFD), with a mandate to enhance dog health, well-being, and welfare. An information hub for multiple aspects of canine health and welfare, it provides a database for simple genetic diseases in the dog that can be searched by disease or by breed, with a qualitative assessment of the relevance of a disease for a particular breed. The scientific literature is cited without links, but links are provided to appropriate OMIA citations. The website functions in multiple languages.

NOT-FOR-PROFIT SERVICE RESOURCES

Veterinary Genetics Laboratory (VGL), University of California, Davis (UC Davis) (https://vgl.ucdavis.edu/)

As a not-for-profit, self-supporting unit of UC Davis, the VGL is one of the original university-based DNA service labs that still provides DNA testing for multiple species of domestic and wild animals. Mutation testing is *via* traditional DNA methods, thereby facilitating rapid results. The VGL website provides search functions by both genetic disease and breed. Disease pages are provided that include clinical information interspersed with sample collection, pricing, and turnaround information. Information on mutation frequency within a breed is not provided. Functional links to the scientific literature are provided.

PennGen, University of Pennsylvania (https://www.vet.upenn.edu/research/academic-departments/clinical-sciences-advanced-medicine/research-labs-centers/penngen/penngen-tests/genetic-tests)

The PennGen laboratory of the University of Pennsylvania is another university-based DNA testing lab that is still operational and provides DNA tests for disease mutations for the dog and cat. Testing for genetic disease mutations is by traditional DNA methods. The PennGen website provides search functions by disease and by breed. Disease pages are cursory and no information of mutation frequency within a breed is provided. Scientific literature citations are not provided.

FOR PROFIT (COMMERCIAL) SERVICE RESOURCES

Wisdom Panel (Mars)
(https://www.wisdompanel.com/en-us)

Wisdom Panel was introduced as a dog DNA testing company by the privately held multinational food company Mars in 2007, as a part of its veterinary and pet care division. Initially the business model involved veterinarians submitting blood samples; however, this proved cumbersome and a direct-to-consumer model with buccal swabs was adapted. In 2009, DNA tests were initially designed for breed determination and targeting mixed-breed dog owners. To further target dog breeders and veterinarians, microarray tests were expanded to include genetic disease mutations and trait analysis. The Wisdom Panel website has a database of canine genetic diseases presented as an alphabetic listing without search functions, but with links to individual disease pages; these pages have paragraphs for breeders and for veterinarians. No mutation breed frequencies are given. Links to the scientific literature are cursory. In 2021, Wisdom Panel expanded its product line to include DNA testing for cats.

Embark (https://embarkvet.com/)

Embark came out of academic research labs at the Cornell University School of Veterinary Medicine. It was launched as a public company in 2016, with the goal of providing dog owners, breeders, and veterinarians with microarray DNA profiling diagnostic services for canine ancestry, trait and genetic disease risk analysis. The Embark website provides access to a data bank of canine genetic diseases that can be searched by name or breed. Breed-relevant genetic diseases can be retrieved, although breed specific frequencies of disease mutations are not provided. Diseases can be regrouped by medical category, but this cannot be further grouped by breed. Disease pages are designed for the consuming public and include links for the purchase of products. Cursory links to the scientific literature are provided. Embark has now partnered with dog nutrition companies (Purina, Wellness, others) and provides a portal for the purchase of canine nutrition products.

Paw Print Genetics (Neogen)
(https://www.pawprintgenetics.com/)

Paw Print Genetics is a for-profit company that specializes in DNA testing for disease mutations and traits in dogs. A division of Genetic Veterinary Sciences, it was founded in 2012 and in 2021 was purchased by Neogen, a multinational food safety and animal diagnostic company. As a company, Paw Print Genetics has maintained a mandate for providing an educational axis to complement its service function. The Paw Print Genetics web site provides a database for canine genetic diseases that is searchable by disease name and by breed. Disease pages are provided and disease relevance for a particular breed is given. Breed-relevant mutation frequencies can be provided on a single-disease basis. Links to the scientific literature are provided.

My CatScan (Neogen)
(https://www.mycatscan.com/)

My CatScan is a for-profit company that specializes in DNA testing for disease mutations and traits in cats. Like Paw Print Genetics, it is a division of Genetic Veterinary Sciences and is now a part of the Neogen portfolio of animal diagnostic services. The My CatScan website provides a database for cat simple genetic diseases that is mixed with physical traits and can be searched by breed. Disease pages can be accessed, and relevant diseases are listed, but breed relevant mutation frequencies are not provided. Links to the scientific literature are given.

Basepaws (Zoetis) (https://basepaws.com/)

Basepaws started in 2017 as a DNA diagnostic company for genetic analysis of cats. In 2022, Basepaws was acquired by Zoetis, and in 2023 its product line was extended to include DNA tests for dogs. The Basepaws website provides access to databases for both dog and cat genetic diseases. Search functions are on disease and gene name but not on breed. Disease pages are provided with a cursory clinical signs section. Breeds at higher risk for a disease are currently not well-documented and no indication of breed-specific mutation frequencies is given. Citations to the scientific literature are provided, with clickable links for feline diseases.

Betagene (University of Laval)
(https://betagene.ca/)

Betagene is a private animal DNA diagnostic company that operates out of the Department of Animal Sciences, University Laval, in Quebec. Betagene provides rapid conventional DNA services to animal breeders, owners, and veterinarians, particularly in Quebec but also in the rest of Canada. The Betagene website provides a database for genetic diseases of the dog, cat, and horse that is catered to user's needs. Some breed specificity is provided, but mutation frequencies are not given. The scientific literature as well as OMIA listings for diseases are given but are not linked. The Betagene website functions in English and French.