MYBREEDDATA – A CANINE DISEASE VARIANT GENOTYPE FREQUENCY AND DISTRIBUTION DATABASE

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INTRODUCTION

More than 300 genetic disease variants have been identified and characterized in dogs to date. However, knowledge on their distribution and prevalence across pure breeds is still incomplete (Figure 1). Original publications characterizing a novel Mendelian disease variant typically report carrier frequency estimates based on a subset of dogs representing the studied breed, and feature limited genotyping of the novel discovered variant across other breeds. In most cases, no updates on population carrier frequency changes over time are offered by the discovery research group post-publication. Up-to-date information is more readily available to commercial genetic test providers offering screening for the discovered variant, and to dog registries maintaining records on DNA test results.

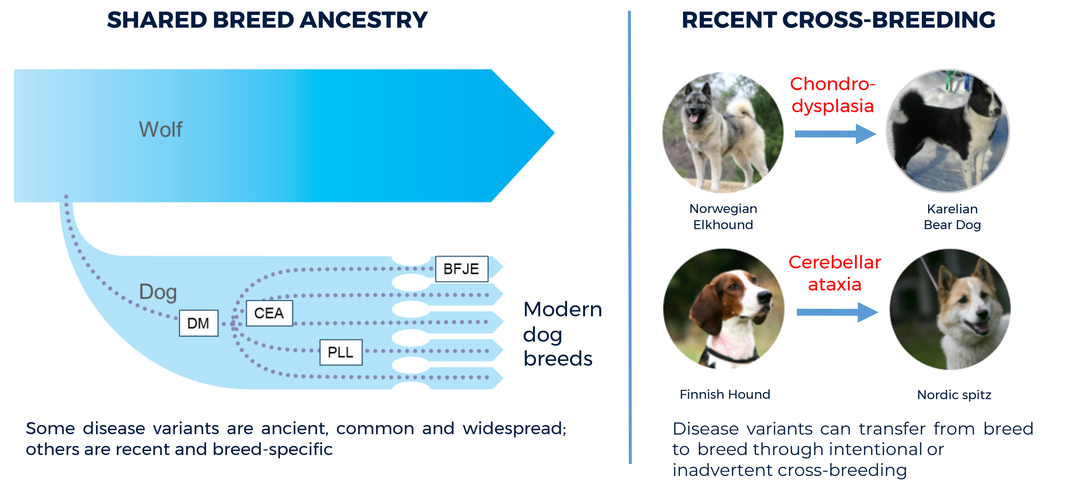


Figure 1. Defining with certainty which genetic mutations are present in any given breed is a near impossible task. Shared breed ancestry and recent cross-breeding represent potential explanations for mutation distribution and transfer between breeds. DM = Degenerative myelopathy ; CEA = Collie Eye Anomaly ; PLL = Primary Lens Luxation ; BFJE = Benign Familial Juvenile Epilepsy of Lagotto Romagnolos

AIM

To develop a freely available online portal providing current information on the frequency and distribution of canine inherited disease variants.

To ease access to big data relevant for breed health research, breeding selections and veterinary care.

MATERIALS

Genoscoper Laboratories and Wisdom Health are world leading canine genetic testing service providers, and contributing partners of the International Partnership for Dogs (IPFD) and its Harmonization of Genetic Testing in Dogs (HGTD) initiative. Through comprehensive commercial genetic screening of more than **1,000,000 dogs** to date, we have gained extensive insight into canine breed ancestry and disease heritage. Two recent peer-reviewed scientific publications (Figure 2) analyzing more than **83,000 mixed breed dogs and 18,000 purebred dogs** representing **330 different breeds** laid the foundation for our disease variant genotype database.

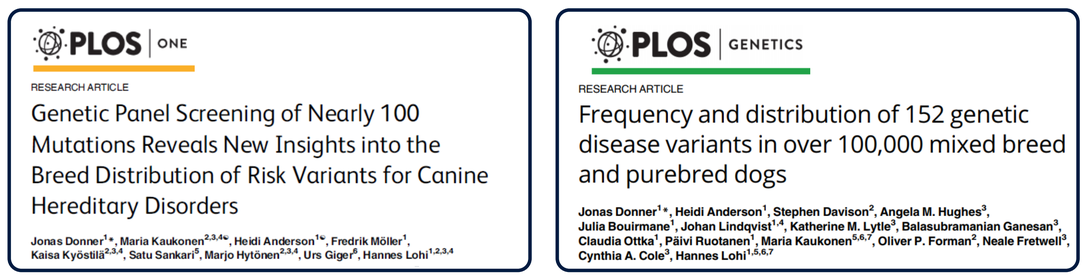


Figure 2. Recent relevant peer-reviewed publications from Genoscoper Laboratories, Wisdom Health and collaborators.

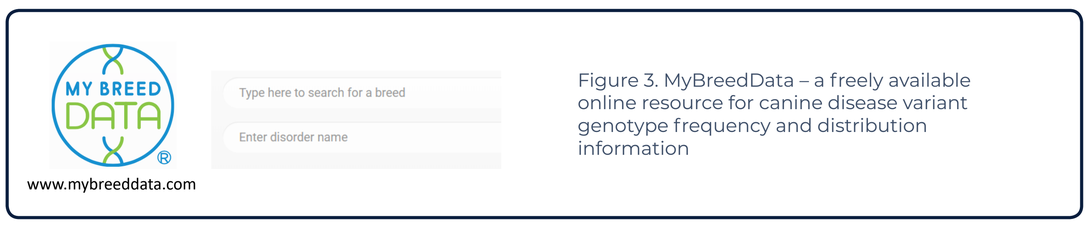


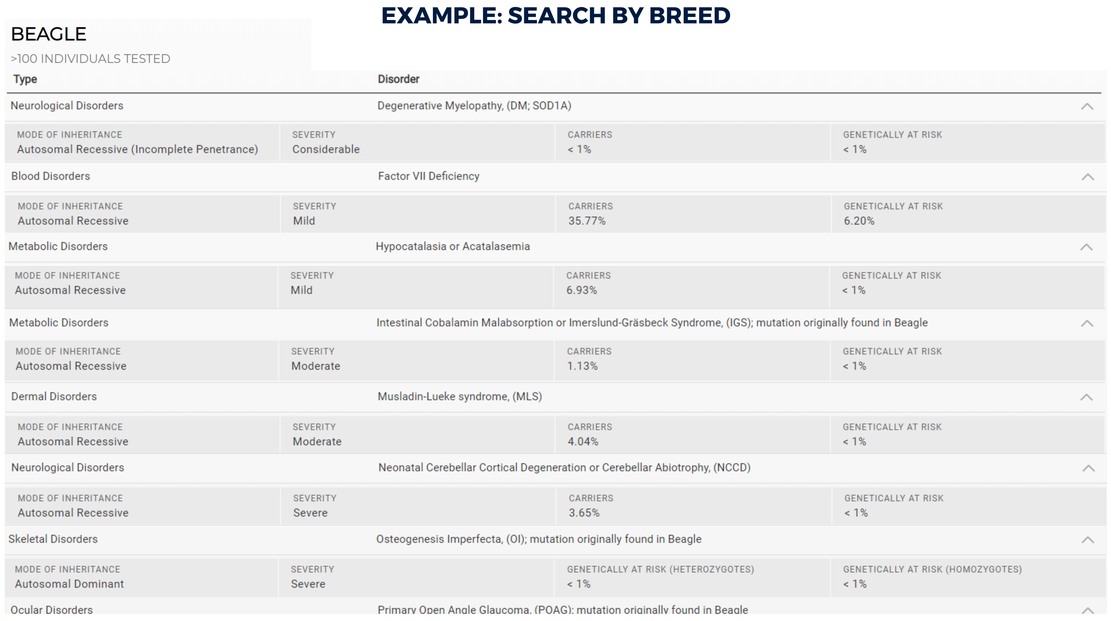
Figure 3. MyBreedData – a freely available online resource for canine disease variant genotype frequency and distribution information

OUTCOME

We developed an online portal and named it **MyBreedData** (**www.mybreeddata.com** ; Figure 3). The portal provides access to genetic disease variant genotype frequency information based on the aforementioned in house research projects and continuously ongoing commercial panel screening. The database is updated to include new analyzed dogs weekly. It features functionalities (Figure 4) for

1) **Searching by breed**; with the output being a listing of known breed-relevant Mendelian disease variants and their "carrier" and "genetically at risk" genotype frequencies

2) Searching by disease variant; with the output being a listing of all breeds known to carry the disease variant along with "carrier" and "genetically at risk" genotype frequencies by breed



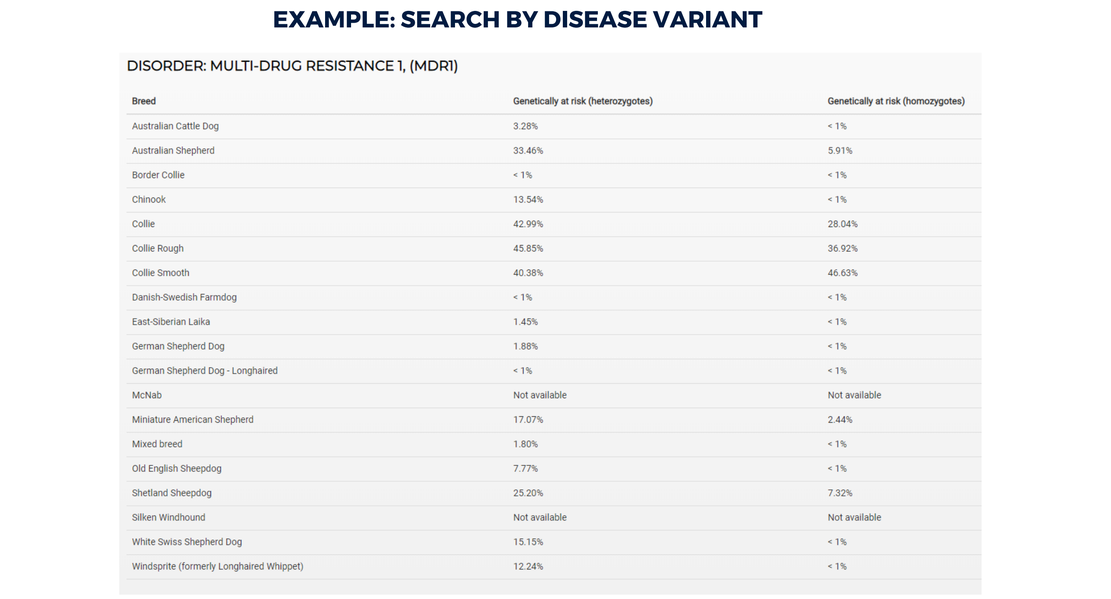


Figure 4. MyBreedData features the option of searching for disease variant frequencies either by breed or by disease variant.

CONCLUSIONS

- Genetic test providers have access to a significant amount of data generated through commercial screening of disease variants across breeds
- Such information can be made available and amenable to updates online, providing current information on disease variant frequencies within and across breeds
- Accessible disease variant frequency and distribution information could be leveraged to guide breed DNA testing priorities, breeding selections, breed health research and health committee work, veterinary education and pet care

References

MyBreedData - Canine Disease Variant Genotype Frequency and Distribution Database (https://www.mybreeddata.com)

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Donner J, Kaukonen M, Anderson H, Möller F, Kyöstilä K, Sankari S, Hytönen M, Giger U, Lohi H. Genetic panel screening of nearly 100 mutations reveals new insights into the breed distribution of risk variants for canine hereditary disorders. PLoS ONE 11(8): e0161005.

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