# 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 1). Original publications characterizing to-date information is more readily 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 pure breeds is still incomplete (Figure research group post-publication. Upavailable to commercial genetic test providers offering screening for the discovered variant, and to dog registries maintaining records on DNA test results.

### 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.

## 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

- 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

### CONCLUSIONS

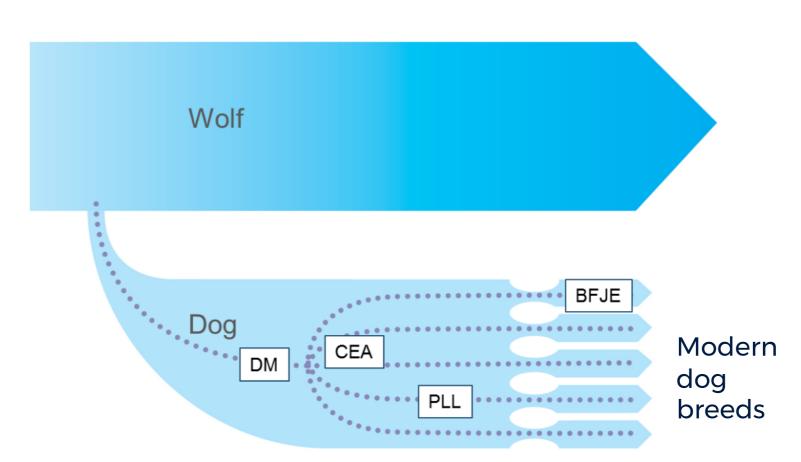
- Genetic test providers have access to a significant amount of data generated through commercial screening of disease variants across breeds
- Such information can be made available and amenable to updates online, providing current information on disease variant frequencies within and across breeds
- 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) Donner J, Anderson H, Davison S, Hughes AM, Bouirmane J, Lindqvist J, Lytle KM, Ganesan B, Ottka C, Ruotanen P, Kaukonen M, Forman OP, Fretwell N, Cole CA, Lohi H. Frequency and distribution of 152 genetic disease variants in over 100,000 mixed breed and purebred dogs. PLoS Genet 14(4): e1007361. 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.

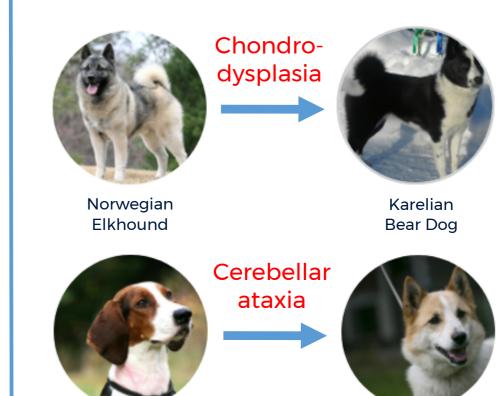
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#### **SHARED BREED ANCESTRY**



Some disease variants are ancient, common and widespread; others are recent and breed-specific

#### **RECENT CROSS-BREEDING**



Disease variants can transfer from breed to breed through intentional or inadvertent cross-breeding

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

#### PLOS ONE

**BEAGLE** 

Genetic Panel Screening of Nearly 100 Mutations Reveals New Insights into the Breed Distribution of Risk Variants for Canine Hereditary Disorders

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PLOS GENETICS

Finnish Hound

RESEARCH ARTICLE

Frequency and distribution of 152 genetic disease variants in over 100,000 mixed breed and purebred dogs

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

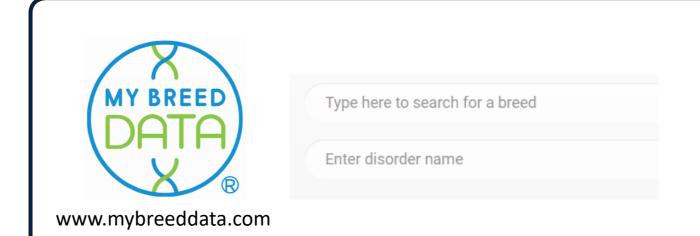


Figure 3. MyBreedData – a freely available online resource for canine disease variant genotype frequency and distribution information

#### **EXAMPLE: SEARCH BY BREED**

>100 INDIVIDUALS TESTED					
Туре		Disorder			
Neurological Disorders		Degenerative Myelopathy, (DM; SOD1A)		^	
MODE OF INHERITANCE Autosomal Recessive (Incomplete Penetrance)	SEVERITY Considerable		CARRIERS < 1%	GENETICALLY AT RISK < 1%	
Blood Disorders		Factor VII Deficiency			^
MODE OF INHERITANCE Autosomal Recessive	SEVERITY Mild		CARRIERS 35.77%	GENETICALLY AT RISK 6.20%	
Metabolic Disorders		Hypocatalasia or Acatalasemia			^
MODE OF INHERITANCE Autosomal Recessive	SEVERITY Mild		CARRIERS 6.93%	GENETICALLY AT RISK < 1%	
Metabolic Disorders		Intestinal Cobalamin Malabsorption or Imerslund-Gräsbeck Syndrome, (IGS); mutation originally found in Beagle			^
MODE OF INHERITANCE Autosomal Recessive	SEVERITY Moderate		CARRIERS 1.13%	GENETICALLY AT RISK < 1%	
Dermal Disorders		Musladin-Lueke syndrome, (MLS)			^
MODE OF INHERITANCE Autosomal Recessive	SEVERITY Moderate		CARRIERS 4.04%	GENETICALLY AT RISK < 1%	
Neurological Disorders		Neonatal Cerebellar Cortical Degeneration or Cerebellar Abiotrophy, (NCCD)			^
MODE OF INHERITANCE Autosomal Recessive	SEVERITY Severe		CARRIERS 3.65%	GENETICALLY AT RISK < 1%	
Skeletal Disorders	l Disorders Osteogenesis Imperfecta, (OI); mutation originally found in Beagle			^	
MODE OF INHERITANCE Autosomal Dominant	SEVERITY Severe		GENETICALLY AT RISK (HETEROZYGOTES) < 1%	GENETICALLY AT RISK (HOMOZYGOTES) < 1%	
Ocular Disorders		Primary Open Angle Glaucoma. (PO	AG): mutation originally found in Beagle		^

#### **EXAMPLE: SEARCH BY DISEASE VARIANT**

Breed	Genetically at risk (heterozygotes)	Genetically at risk (homozygo
Australian Cattle Dog	3.28%	< 1%
Australian Shepherd	33.46%	5.91%
Border Collie	< 1%	< 1%
Chinook	13.54%	< 1%
Collie	42.99%	28.04%
Collie Rough	45.85%	36.92%
Collie Smooth	40.38%	46.63%
Danish-Swedish Farmdog	< 1%	< 1%
East-Siberian Laika	1.45%	< 1%
German Shepherd Dog	1.88%	< 1%
German Shepherd Dog - Longhaired	< 1%	< 1%
McNab	Not available	Not available
Miniature American Shepherd	17.07%	2.44%
Mixed breed	1.80%	< 1%
Old English Sheepdog	7.77%	< 1%
Shetland Sheepdog	25.20%	7.32%
Silken Windhound	Not available	Not available
White Swiss Shepherd Dog	15.15%	< 1%
Windsprite (formerly Longhaired Whippet)	12.24%	< 1%

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



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