



University of Colorado
Anschutz Medical Campus



THE UNIVERSITY OF
SYDNEY

Classifying Animal Breeds with the Vertebrate Breed Ontology (VBO)

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International Conference on Biomedical Ontology
September 2022



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INITIATIVE

Monarch Initiative integrates disparate data and creates comparative tools to support diagnosis and treatments

Disparate Data

Human



GENE



VARIANT



PHENOTYPE



DISEASE



TREATMENT



EXPOSURE

Animal models



Data integration (use of ontologies)



National
Center for
Biotechnology
Information



upheno
ontology



human
phenotype
ontology



mondo
THE MONDO MONDO MONDO MONDO



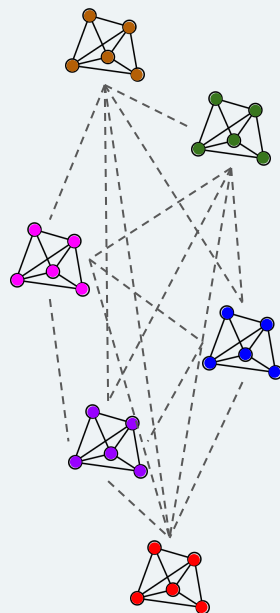
maxo
MEDICAL ACTION ONTOLOGY



ecto
ENVIRONMENTAL CONDITIONS
TREATMENTS & EXPOSURES ONTOLOGY



Unified data model

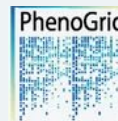


Tools



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Phenotype comparison



Variant prioritization



Disease Diagnosis

Treatments discovery



Integrating non-human animal/veterinary data would improve diagnosis for non-human animals (as well as human)



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DISEASE



TREATMENT

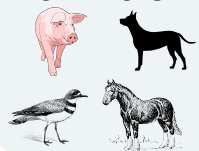


EXPOSURE

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Non-human
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NCBI

National
Center for
Biotechnology
Information



UPheno
ontology

upheno
ontology



human
phenotype
ontology

human
phenotype
ontology



mondo

mondo



maxo

maxo



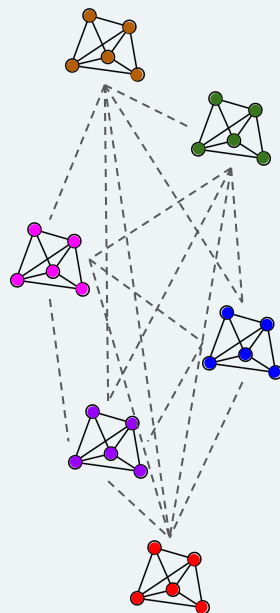
ecto

ecto

ENVIRONMENTAL CONDITIONS
TREATMENTS & EXPOSURES ONTOLOGY

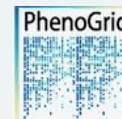


Unified data model



Tools

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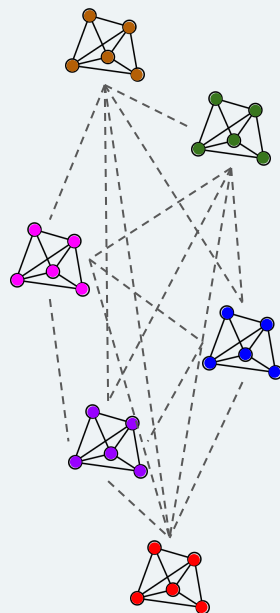


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ENVIRONMENTAL CONDITIONS
TREATMENTS & EXPOSURES ONTOLOGY

Vertebrate
Breed
Ontology
VBO

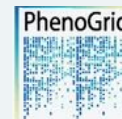


Unified data model



Tools

Phenotype comparison



Variant prioritization



Disease Diagnosis



Treatments discovery



Vertebrate Breed Ontology (VBO)

- Source for animal breeds to be used for data standardization and integration in databases (e.g. OMIA) and veterinary electronic health records
- Scope: vertebrate animals
 - Livestock
 - Companion animals (cats, dogs)
 - Laboratory animals
- VBO was created at the end of 2021 (still in early development stage)

VBO “breed” definition

A breed is a group of animals

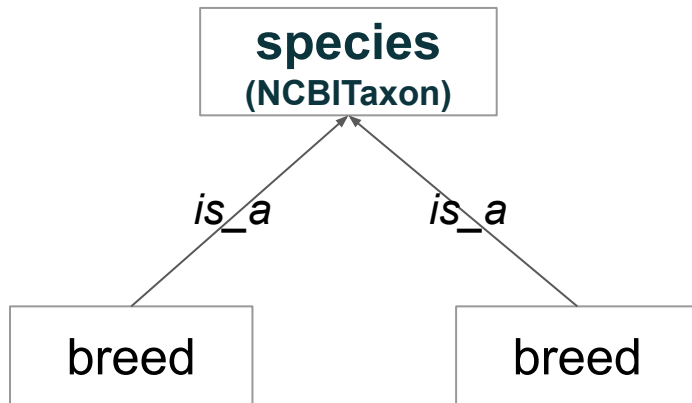
- that **share specific characteristics** (such as traits, behavior, etc) that **distinguish it from other organisms of the same species**, and/or
- which cultural or geographical separation has led to the general acceptance of its separate identity.

Breeds included in VBO have been characterized, defined, and determined **by international breed organizations, communities, experts, and/or have been reported in the literature.**

NCBITaxon as the top-level classification of VBO

Definition: A breed is a group of animals that share specific characteristics (such as traits, behavior, etc) that **distinguish it from other organisms of the same species**, and/or which cultural or geographical separation has led to the general acceptance of its separate identity.

→ a breed is a subclass of a “species”



VBO was created using ODK and ROBOT tools.

VBO was built using the **Ontology Development Kit (ODK)** which

- automatically checks for logical errors
- releases new ontology versions

VBO was created and is maintained via **ROBOT templates**

information
from sources



manual
mapping



ROBOT
template

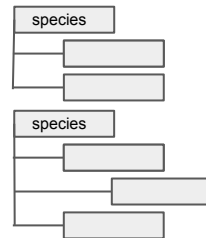


Species
NCBITaxon
species
species
species

ROBOT



VBO



VBO is managed and openly available on GitHub:

<https://github.com/monarch-initiative/vertebrate-breed-ontology>

VBO currently includes livestock and cats breeds

- **Livestock breeds** : 1st version available

Dromedary Bactrian Camel
Alpaca
American Bison
Ass
Bactrian camel
Buffalo
Cassowary
Cattle
Chicken
Deer

Dog
Dromedary
Duck (domestic)
Emu
Goat
Goose (domestic)
Guanaco
Guinea fowl
Guinea pig
Horse

Llama
Muscovy duck
Nandu
Ostrich
Pig
Pigeon
Quail
Rabbit
Sheep
Turkey

Vicuña
Yak (domestic)
North American deer mouse
Domestic yak
Bighorn sheep
Swallow
Partridge
Duck(domestic) Muscovy duck
Peacock
Pheasant

- **Cat breeds** : 1st version available

- **Dog breeds** (*ongoing*)



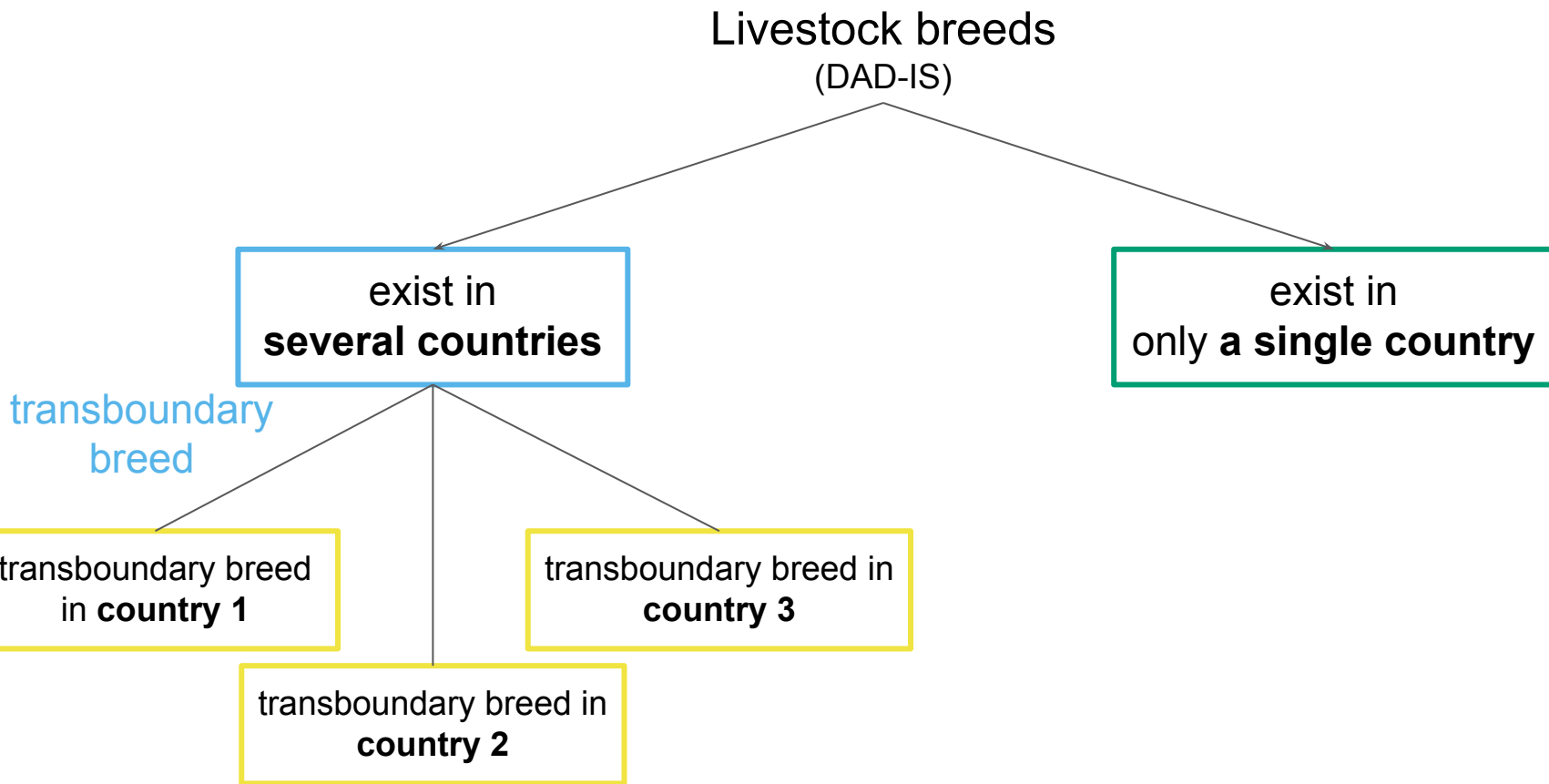
<https://github.com/monarch-initiative/vertebrate-breed-ontology>

DAD-IS is the VBO Source for Livestock Breeds

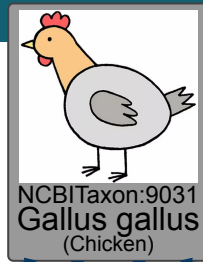
Domestic Animal Diversity Information (DAD-IS) (<https://www.fao.org/dad-is/en/>)

- maintained and developed by the Food and Agriculture Organization of the United Nations (FAO)
- exhaustive list maintained by national coordinators from 182 countries
 - 37 species; >8,800 breeds
- aims to list all breeds in all countries

Livestock Breeds in DAD-IS are identified based on their geographical location



VBO classification: livestock breeds example



VBO:0000547

Jersey Giant (Chicken)



VBO:0007282

Fehér erdélyi kopasznyakú,
Hungary (Chicken)



VBO:0007340

Danki, India (Chicken)



VBO:0000463

Brahma (Chicken)



VBO:0006071

Jersey Giant, Ireland
(Chicken)



VBO:0006068

Jersey Giant,
Canada (Chicken)



VBO:0005526

Brahma, Croatia
(Chicken)



VBO:0005523

Brahma, Australia
(Chicken)

Cat Breeds information in VBO comes from many sources.

International breed organizations:

- The Cat Fanciers' Association (CFA) (<https://cfa.org/>)
- Fédération Internationale Féline (FIFe) (<http://fifeweb.org/index.php>)
- Governing Council of the Cat Fancy (GCCF) (<https://www.gccfcats.org/>)
- Rare and Exotic Feline Registry (REFR) (<https://www.rareexoticfelineregistry.com/>)
- The International Cat Association (TICA) (<https://www.tica.org/>)
- World Cat Congress (WCC) (<https://www.worldcatcongress.org/index.php>)
- World Cat Federation (WCF) (<https://wcf.de/en/>)

Community:

- Wikipedia, List of cat breeds (Wiki) (https://en.wikipedia.org/wiki/List_of_cat_breeds#Breeds)

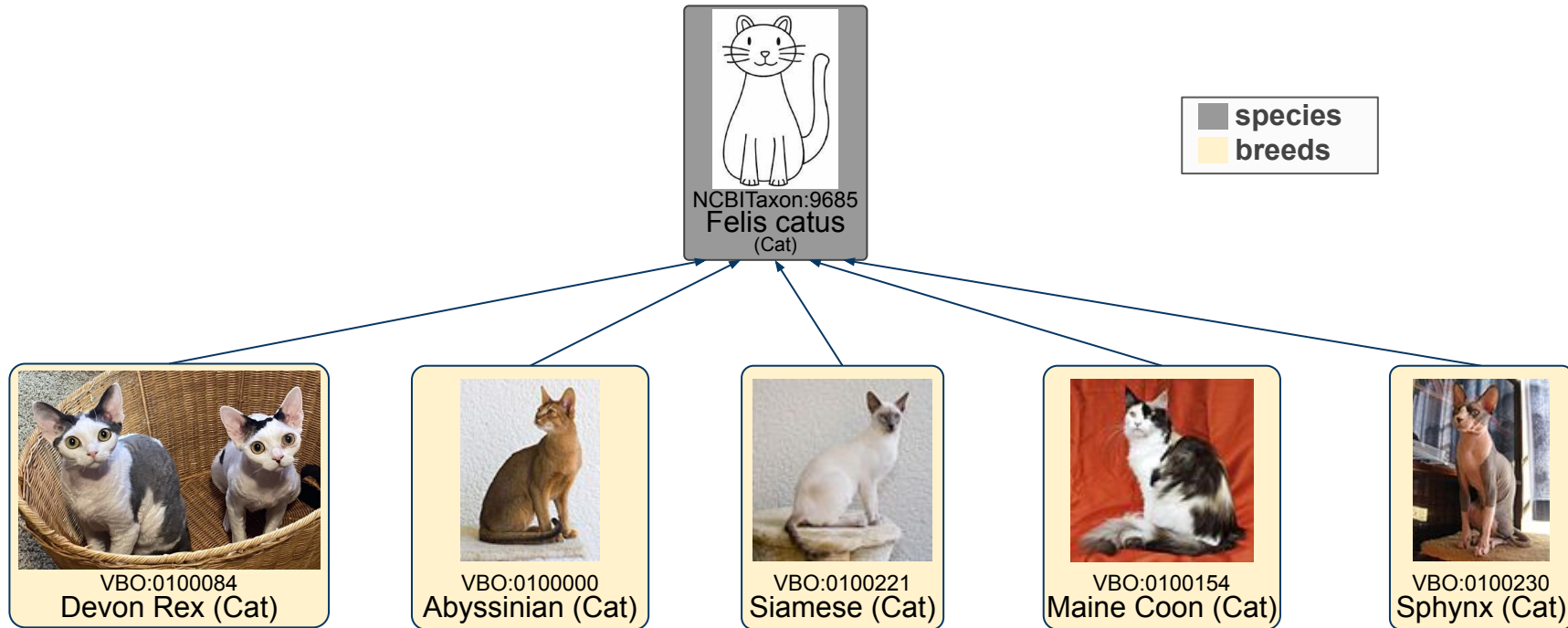
Experts:

- Professor Leslie Lyons, Feline Genetics and Comparative Medicine Laboratory (<http://felinegenetics.missouri.edu>)

Literature:

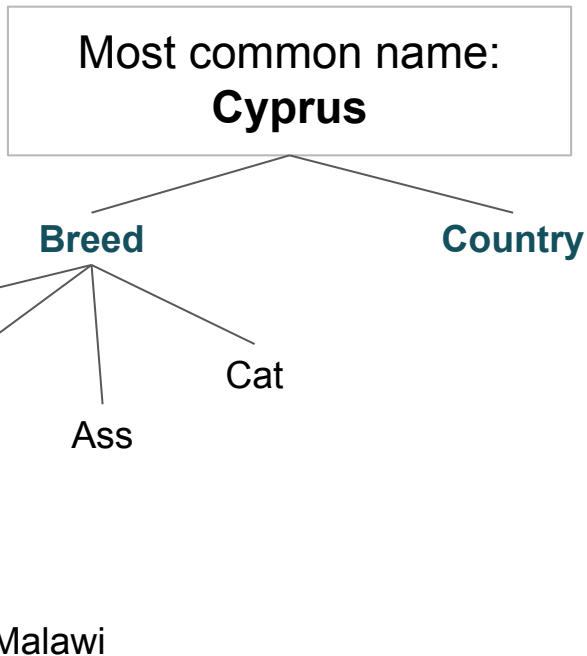
- Online Mendelian Inheritance in Animals (OMIA) (<https://www.omia.org/home/>)

VBO classification: cat breeds example



Insuring label uniqueness: Species and Country are included in the VBO term label

Issue:



Solution:

VBO term labels: **Most common name (Species)**

Eg: 'Cyprus (Ass)' (VBO:0000046)

Eg: 'Cyprus (Cat)' (VBO:0100081)

Eg: 'Cyprus (Goat)' (VBO:0000754)

For breeds identified based on the country where they are located (eg breeds from DAD-IS), VBO term labels:

Most common name, Country (Species)

Eg: 'Cyprus, Libya (Goat)' (VBO:0008930)

Eg: 'Cyprus, Malawi (Goat)' (VBO:0008931)

Metadata: most common names and other synonyms

☰ ● Siberian (Cat) — VBO:0100223 — http://purl.obolibrary.org/obo/VBO_0100223

Annotations Usage

Annotations: Siberian (Cat)

Annotations +

rdfs:label [type: xsd:string]
Siberian (Cat)

'has exact synonym' [type: xsd:string]
Siberian Cat

source
<https://wcf.de/en/wcf-ems-code/>

'has exact synonym' [type: xsd:string]
Siberian

has_synonym_type
■ 'most common name'

source
http://fifeweb.org/wp/breeds/breeds_prf_stn.php

source
<https://cfa.org/breeds/>

source
https://en.wikipedia.org/wiki/List_of_cat_breeds#Breeds

source
<https://wcf.de/en/wcf-ems-code/>

source
<https://www.gccfcats.org/getting-a-cat/choosing/cat-breeds/>

source
<https://www.rareexoticfelineregistry.com/breed-recognition/>

source

most common name

sources of the
information

☰ ● Fehér erdélyi kopasznnyakú, Hungary (Chicken) — VBO:0007282

Annotations Usage

Annotations: Fehér erdélyi kopasznnyakú, Hungary (Chicken)

Annotations +

rdfs:label [type: xsd:string]
Fehér erdélyi kopasznnyakú, Hungary (Chicken)

'has exact synonym' [type: xsd:string]
Fehér erdélyi kopasznnyakú

has_synonym_type
■ 'most common name'

source
<https://www.fao.org/dad-is>

'has exact synonym' [type: xsd:string]
White Transylvanian Naked Neck

source
<https://www.fao.org/dad-is>

VBO terms metadata

- most common name and synonyms
- country of existence
- breed recognition status
- breed domestication status
- breed codes
- reference of the breed to other databases
- description of origin
- source of information

VBO terms metadata

- **most common name and synonyms**
- country of existence
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The screenshot displays the VBO (Vocabulary Browser) interface for the term 'Fehér erdélyi kopasznnyakú, Hungary (Chicken)' with VBO ID 0007282. The interface includes tabs for 'Annotations' and 'Usage'. The 'Annotations' tab is active, showing a list of annotations for the term. The first annotation is 'rdfs:label' [type: xsd:string] with the value 'Fehér erdélyi kopasznnyakú, Hungary (Chicken)'. The second annotation is 'has exact synonym' [type: xsd:string] with the value 'Fehér erdélyi kopasznnyakú'. The third annotation is 'has_synonym_type' with the value 'most common name'. The fourth annotation is 'source' with the value 'https://www.fao.org/dad-is'. The fifth annotation is 'has exact synonym' [type: xsd:string] with the value 'White Transylvanian Naked Neck'. The sixth annotation is 'source' with the value 'https://www.fao.org/dad-is'. Red arrows point to the 'has exact synonym' values and the 'most common name' value.

Fehér erdélyi kopasznnyakú, Hungary (Chicken) — VBO:0007282

Annotations Usage

Annotations: Fehér erdélyi kopasznnyakú, Hungary (Chicken)

Annotations +

rdfs:label [type: xsd:string]
Fehér erdélyi kopasznnyakú, Hungary (Chicken)

'has exact synonym' [type: xsd:string]
Fehér erdélyi kopasznnyakú

has_synonym_type
most common name

source
<https://www.fao.org/dad-is>

'has exact synonym' [type: xsd:string]
White Transylvanian Naked Neck

source
<https://www.fao.org/dad-is>

VBO terms metadata

- most common name and synonyms
- **country of existence**
- breed recognition status
- breed domestication status
- breed codes
- reference of the breed to other databases
- description of origin
- source of information

☰ ● Fehér erdélyi kopasznnyakú, Hungary (Chicken) — VBO:0007282

Description: Fehér erdélyi kopasznnyakú, Hungary (Chicken)

Equivalent To +

SubClass Of +

- 'Gallus gallus'
- 'located in' value Hungary

wikidata:Q28

☰ ● Jersey Giant, Ireland (Chicken) — VBO:0006071

Description: Jersey Giant, Ireland (Chicken)

Equivalent To +

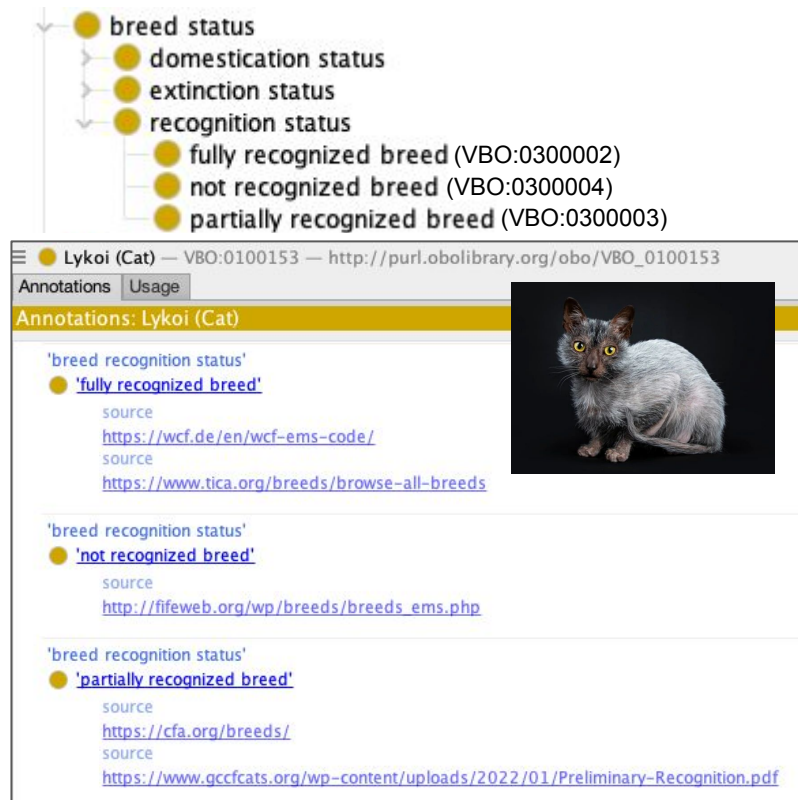
SubClass Of +

- 'Jersey Giant (Chicken)'
- 'located in' value Ireland

wikidata:Q22890

VBO terms metadata

- most common name and synonyms
- country of existence
- **breed recognition status**
- breed domestication status
- breed codes
- reference of the breed to other databases
- description of origin
- source of information



The screenshot displays the VBO interface for the term 'Lykoi (Cat)' (VBO:0100153). The top section shows a tree view of breed status terms:

- breed status
 - domestication status
 - extinction status
 - recognition status
 - fully recognized breed (VBO:0300002)
 - not recognized breed (VBO:0300004)
 - partially recognized breed (VBO:0300003)

The main section shows the 'Annotations: Lykoi (Cat)' for the term 'Lykoi (Cat)' (VBO:0100153). The annotations are:

- 'breed recognition status'
 - 'fully recognized breed'
 - source: <https://wcf.de/en/wcf-ems-code/>
 - source: <https://www.tica.org/breeds/browse-all-breeds>
- 'breed recognition status'
 - 'not recognized breed'
 - source: http://fifeweb.org/wp/breeds/breeds_ems.php
- 'breed recognition status'
 - 'partially recognized breed'
 - source: <https://cfa.org/breeds/>
 - source: <https://www.gccfcats.org/wp-content/uploads/2022/01/Preliminary-Recognition.pdf>

A small image of a Lykoi cat is shown on the right side of the annotations section.

VBO terms metadata

- most common name and synonyms
- country of existence
- breed recognition status
- **breed domestication status**
- breed codes
- reference of the breed to other databases
- description of origin
- source of information

breed status

- domestication status
 - domestic breed (VBO:0300007)
 - feral breed (VBO:0300006)
 - wild breed (VBO:0300008)

Svanuri Pig, Georgia (Pig) — VBO:0012991

Annotations Usage

Annotations: Svanuri Pig,

has_domestication_status

- 'domestic breed'

source

<https://www.fao.org/dad-is>

Ba Xuyen, Viet Nam (Pig) — VBO:0013238

Annotations Usage

Annotations: Ba Xuyen, Vi

has_domestication_status

- 'feral breed'

source

<https://www.fao.org/dad-is>

Ankamali, India (Pig) — VBO:0013035

Annotations Usage




Annotations: Ankamali, I

has_domestication_status

- 'wild breed'

source

<https://www.fao.org/dad-is>



Photos credit: http://www.agrowebcee.net/fileadmin/content/aw-gruzia/files/Svaneti_pig.pdf ; https://www.researchgate.net/figure/fig2_228095003 ; https://www.researchgate.net/figure/Figure-1-Andaman-Desi-pig-Andaman-wild-pig-Nicobari-pig-Andaman-Desi-pig-and-pure-and_fig1_273667616

VBO terms metadata

- most common name and synonyms
- country of existence
- breed recognition status
- breed domestication status
- **breed codes**
- reference of the breed to other databases
- description of origin
- source of information

Siberian (Cat) — VBO:0100223 — http://purl.obolibrary.org/obo/VBO_0100223

Annotations Usage

Annotations: Siberian (Cat)

'breed code' [type: xsd:string]

SB

source

<https://www.tica.org/phocadownload/sb.pdf>

'breed code' [type: xsd:string]

SIB

source

http://fifeweb.org/wp/breeds/breeds_prf_stn.php

source


<https://wcf.de/en/wcf-ems-code/>

source

https://www.gccfcats.org/wp-content/uploads/2022/04/SOP.final_February2022.pdf

source

<https://www.rareexoticfelineregistry.com/breed-recognition/>



VBO terms metadata

- most common name and synonyms
- country of existence
- breed recognition status
- breed domestication status
- breed codes
- **reference of the breed to other databases**
- description of origin
- source of information

The image shows two screenshots of the VBO (Vocabulary Browser) interface. Each screenshot displays a term's metadata, including its name, VBO ID, and a list of annotations. The first screenshot is for 'American Bobtail (Cat)' with VBO ID VBO:0100003. The second screenshot is for 'Alai, Kyrgyzstan (Sheep)' with VBO ID VBO:0016189. Both screenshots show a 'database_cross_reference' annotation with the value 'OMIA:893' for the cat and 'OMIA:216' for the sheep.

American Bobtail (Cat) — VBO:0100003

Annotations Usage

Annotations: American Bobtail (Cat)

database_cross_reference [type: xsd:string]
OMIA:893

Alai, Kyrgyzstan (Sheep) — VBO:0016189

Annotations Usage

Annotations: Alai, Kyrgyzstan (Sheep)

database_cross_reference [type: xsd:string]
OMIA:216

VBO terms metadata

- most common name and synonyms
- country of existence
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- breed domestication status
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- reference of the breed to other databases
- **description of origin**
- source of information



The screenshot shows a web browser window with the title "Aberdeen Angus, Austria (Cattle) — VBO:0002144 — http://purl.ob". Below the title bar, there are two tabs: "Annotations" (selected) and "Usage". The main content area has a yellow header bar that reads "Annotations: Aberdeen Angus, Austria (Cattle)". Below this, there is a section titled "'description of origin' [type: xsd:string]" with a yellow background. The text in this section reads: "The Aberdeen Angus has its origin in the Northeast of Scotland. In North America it was selected for a bigger frame and higher daily gain. German Angus resulted from a combination cross between Aberdeen Angus and German dual purpose breeds (Black pied, Gelbvieh, Simmental)". Below the text, there is a "source" label and a URL: <https://www.fao.org/dad-is>. The browser window also shows standard navigation buttons (back, forward, home, etc.) and a search bar.

VBO terms metadata

- most common name and synonyms
- country of existence
- breed recognition status
- breed domestication status
- breed codes
- reference of the breed to other databases
- description of origin
- **source of information**

A screenshot of a web browser displaying the VBO (Vocabulary Browser) page for the term "Abyssinian (Cat)". The browser address bar shows the URL: http://purl.obolibrary.org/obo/VBO_0100000. The page has two tabs: "Annotations" (selected) and "Usage". The main content area is titled "Annotations: Abyssinian (Cat)" and lists several "dcterms:source" entries, each with a corresponding URL:

- <https://cfa.org/breeds/>
- <https://wcf.de/en/wcf-ems-code/>
- <https://www.gccfcats.org/getting-a-cat/choosing/cat-breeds/>
- <https://www.rareexoticfelineregistry.com/breed-recognition/>
- https://en.wikipedia.org/wiki/List_of_cat_breeds#Breeds
- http://fifeweb.org/wp/breeds/breeds_prf_stn.php
- <https://www.tica.org/breeds/browse-all-breeds>
- https://www.worldcatcongress.org/wp/cat_breed_comp_abv.php

To the right of the list is a photograph of three Abyssinian cats sitting on a dark surface. The photo is credited to "© Chang".

Summary

- Vertebrate Breed Ontology (VBO) is an ontology created to serve as a single computable resource for vertebrate breed names.
- VBO is created based on information from international organizations, communities, and experts which have defined and/or generally accepted a group of animals as a “breed”.
- VBO currently includes livestock and cat breeds.
- VBO terms include information related to
 - most common name and synonyms
 - country of existence
 - breed recognition status
 - breed domestication status
 - breed codes
 - reference of the breed to other databases
 - description of origin
 - source of information

Summary

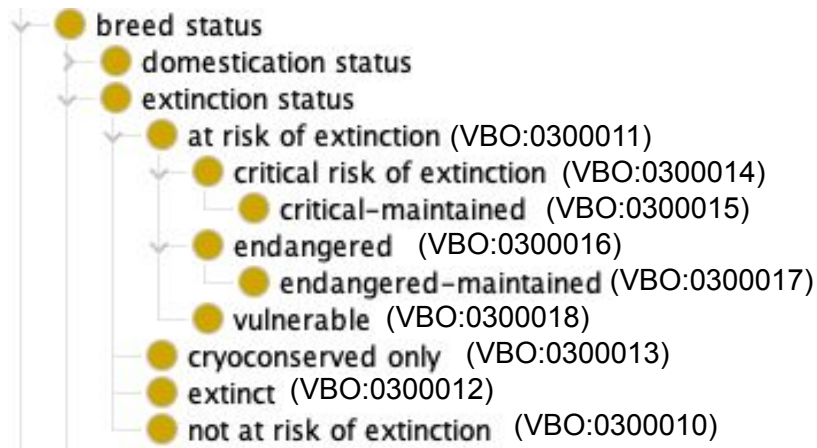
- VBO has been accepted into the OBO Foundry
<https://obofoundry.org/ontology/vbo.html>
- VBO is available in ontology browsers
 - OLS: <https://www.ebi.ac.uk/ols/ontologies/vbo>
 - Ontobee: <https://ontobee.org/ontology/VBO>
- VBO is still in early development, and improvements are in the works.
<https://github.com/monarch-initiative/vertebrate-breed-ontology>

Future work : additional breed information

- country of origin
- extinction status
- *“shares characteristics with”*
- breed parentage / pedigree
- genetic information of breeds

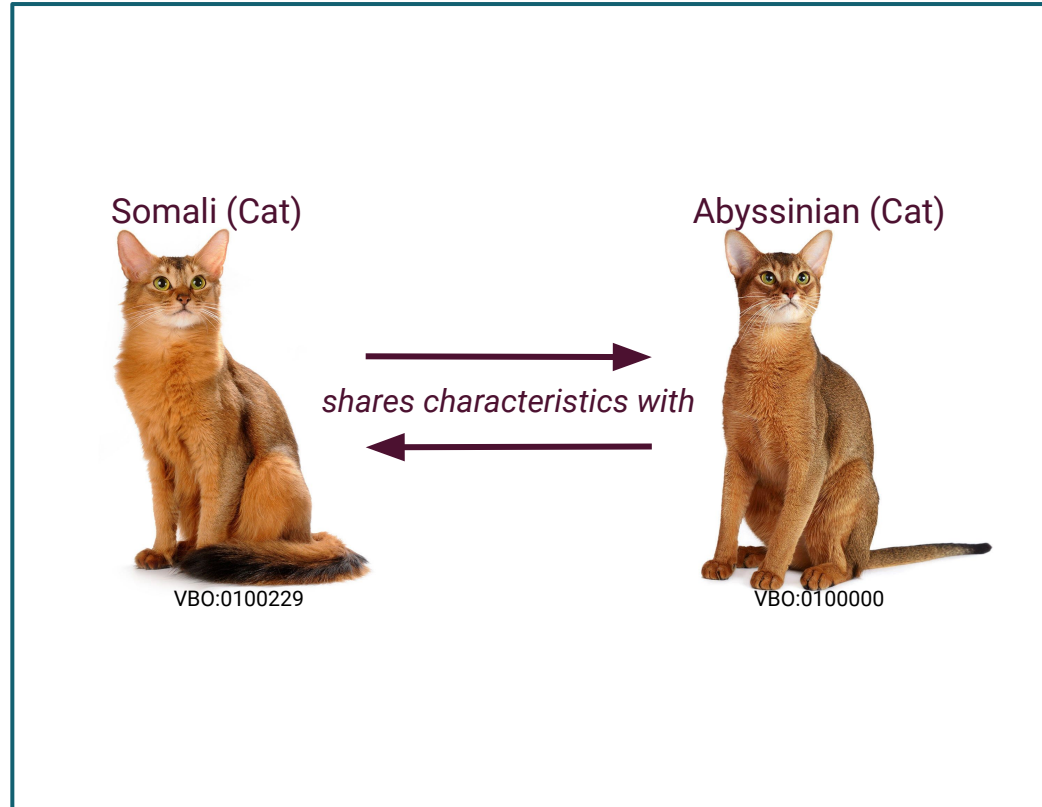
Future work : additional breed information

- country of origin
- **extinction status**
- *"shares characteristics with"*
- breed parentage / pedigree
- genetic information of breeds



Future work : additional breed information

- country of origin
- extinction status
- ***“shares characteristics with”***
- breed parentage / pedigree
- genetic information of breeds



Future work : additional breed information

- country of origin
- extinction status
- *"shares characteristics with"*
- **breed parentage / pedigree**
- genetic information of breeds



VBO:0000380



VBO:0000159



VBO:0000378

"has parents"
"derives from"

Future work : additional breed information

- country of origin
- extinction status
- “shares characteristics with”
- breed parentage / pedigree
- **genetic information of breeds**

Short tail trait has been associated with:	 Japanese Bobtail (Cat) VBO:0100128	 Kurilian Bobtail (Cat) VBO:0100146	 Mekong Bobtail (Cat) VBO:0100161	 Toybob (Cat) VBO:0100244
	HES7 c.5T>C (homozygous)	100% prevalence	60-100% prevalence	60-100% prevalence
	T Box 4 variants identified (heterozygous)	0	0	0
Short tail trait has been associated with:	 Cymric (Cat) VBO:0100080	 Manx (Cat) VBO:0100156	 Highlander (Cat) VBO:0100114	 American Bobtail (Cat) VBO:0100003
	HES7 c.5T>C (homozygous)	1-10% prevalence	1-10% prevalence	0
	T Box 4 variants identified (heterozygous)	25-50% prevalence	25-50% prevalence	25-50% prevalence

Anderson H, Davison S, Lytle KM, Honkanen L, Freyer J, Mathlin J, et al. (2022) Genetic epidemiology of blood type, disease and trait variants, and genome-wide genetic diversity in over 11,000 domestic cats. PLoS Genet 18(6): e1009804. <https://doi.org/10.1371/journal.pgen.1009804>

Photo credits: <https://www.wisdompanel.com/en-us/cat-breeds> ; <https://cattime.com/cat-breeds/mekong-bobtail#/slide/1>

Future work : expand sources and species coverage

- **New Sources:**

- **Livestock Breed Ontology (LBO)** (<https://www.animalgenome.org/bioinfo/projects/lbo/>)
 - short term plan: x-ref between VBO and LBO
 - long term plan: merge VBO & LBO
- **Veterinary Nomenclature (VeNom) Breed Codes** (<https://venomcoding.org/>)
 - Breed codes for cat, dog, equids, rabbit

- **New Breed Species:**

- **Dog breeds** (*ongoing*)
 - List all sources available
 - Investigate available data
- **Lab animals**
 - animals bred for research purposes

Future work: community and consortium

- Reach out to (potential) **VBO users**
 - Databases (e.g. OMIA)
 - Veterinary electronic health records
 - Breeders
 - ...
- Investigate **needs and requirements** from the VBO community and users
 - new annotations
 - “mixed breeds”, “unknown breeds”,...
 - new classification
 - “classification based on breed purpose”
e.g breeds used for milk/meat production, transportation, etc.
 - “classification based on breed type”
e.g draft horses, warmblood horses, sight hounds, herding dogs, etc.

Summary

- Vertebrate Breed Ontology (VBO) is an ontology created to serve as a single computable resource for vertebrate breed names.
- VBO is created based on information from international organizations, communities, and experts which have defined and/or generally accepted a group of animals as a “breed”.
- VBO currently includes livestock and cat breeds.
- VBO terms include metadata with breed information
- VBO has been accepted into the OBO Foundry, and is still in early development.



<https://github.com/monarch-initiative/vertebrate-breed-ontology>

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