

Classifying Animal Breeds with the Vertebrate Breed Ontology (VBO)

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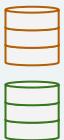


Monarch Initiative integrates disparate data and creates comparative tools to support diagnosis and treatments



Disparate Data

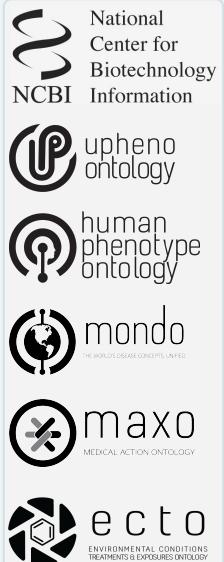
Human



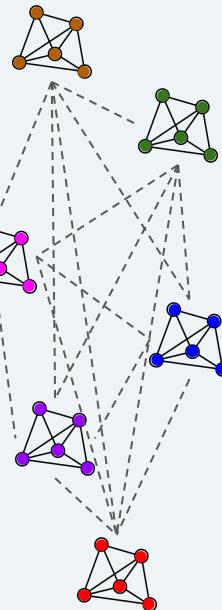
Animal models



Data integration (use of ontologies)

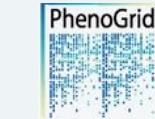


Unified data model



Tools

Phenotype comparison



Variant prioritization



Disease Diagnosis

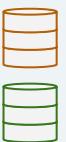
Treatments discovery

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



Disparate Data

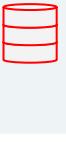
Human



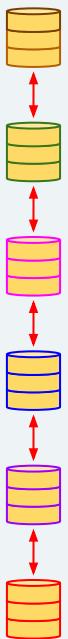
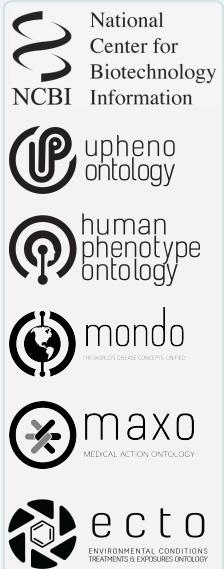
Animal models



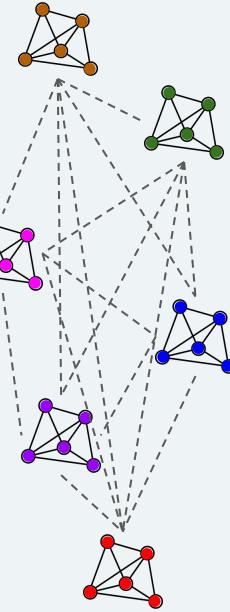
Non-human animals



Data integration (use of ontologies)

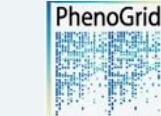


Unified data model



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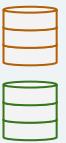


Integrating non-human animal/veterinary data would improve diagnosis for non-human animals (as well as human)



Disparate Data

Human



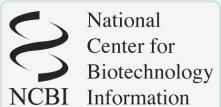
Animal models



Non-human animals



Data integration (use of ontologies)



National Center for Biotechnology Information



upheno ontology



human phenotype ontology



mondo
THE WORLD'S DISEASE CONCEPTS UNIFIED



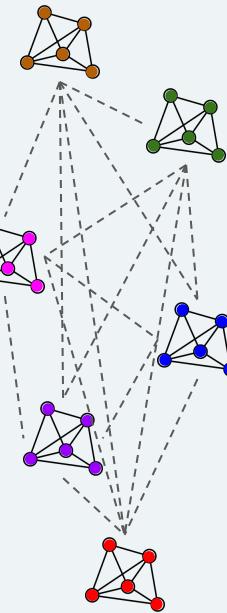
maxo
MEDICAL ACTION ONTOLOGY



ecto
ENVIRONMENTAL CONDITIONS TREATMENTS & EXPOSURES ONTOLOGY

Vertebrate
Breed
Ontology

Unified data model



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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

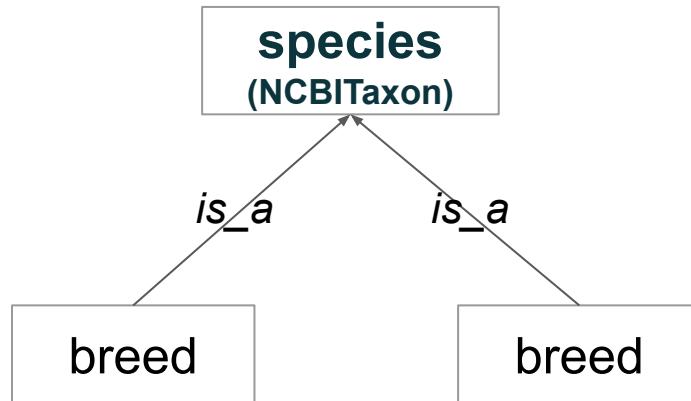
- that **share specific characteristics** (such as traits, behavior, etc) that **distinguish it from other organisms of the same species**, and/or
- which cultural or geographical separation has led to the general acceptance of its separate identity.

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”

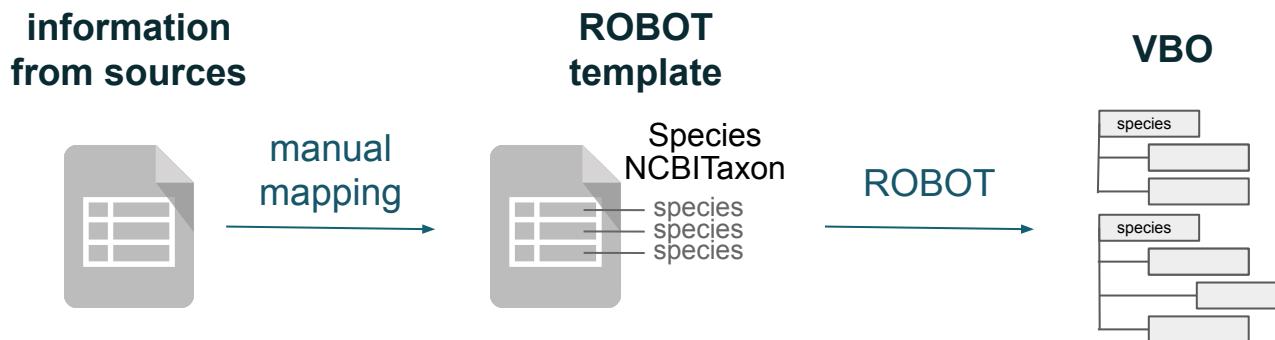


VBO was created using ODK and ROBOT tools.

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**



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 Camel	Dog	Llama	Vicuña
Alpaca	Dromedary	Muscovy duck	Yak (domestic)
American Bison	Duck (domestic)	Nandu	North American deer mouse
Ass	Emu	Ostrich	Domestic yak
Bactrian camel	Goat	Pig	Bighorn sheep
Buffalo	Goose (domestic)	Pigeon	Swallow
Cassowary	Guanaco	Quail	Partridge
Cattle	Guinea fowl	Rabbit	Duck(domestic) Muscovy duck
Chicken	Guinea pig	Sheep	Peacock
Deer	Horse	Turkey	Pheasant

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

- **Dog breeds (ongoing)**



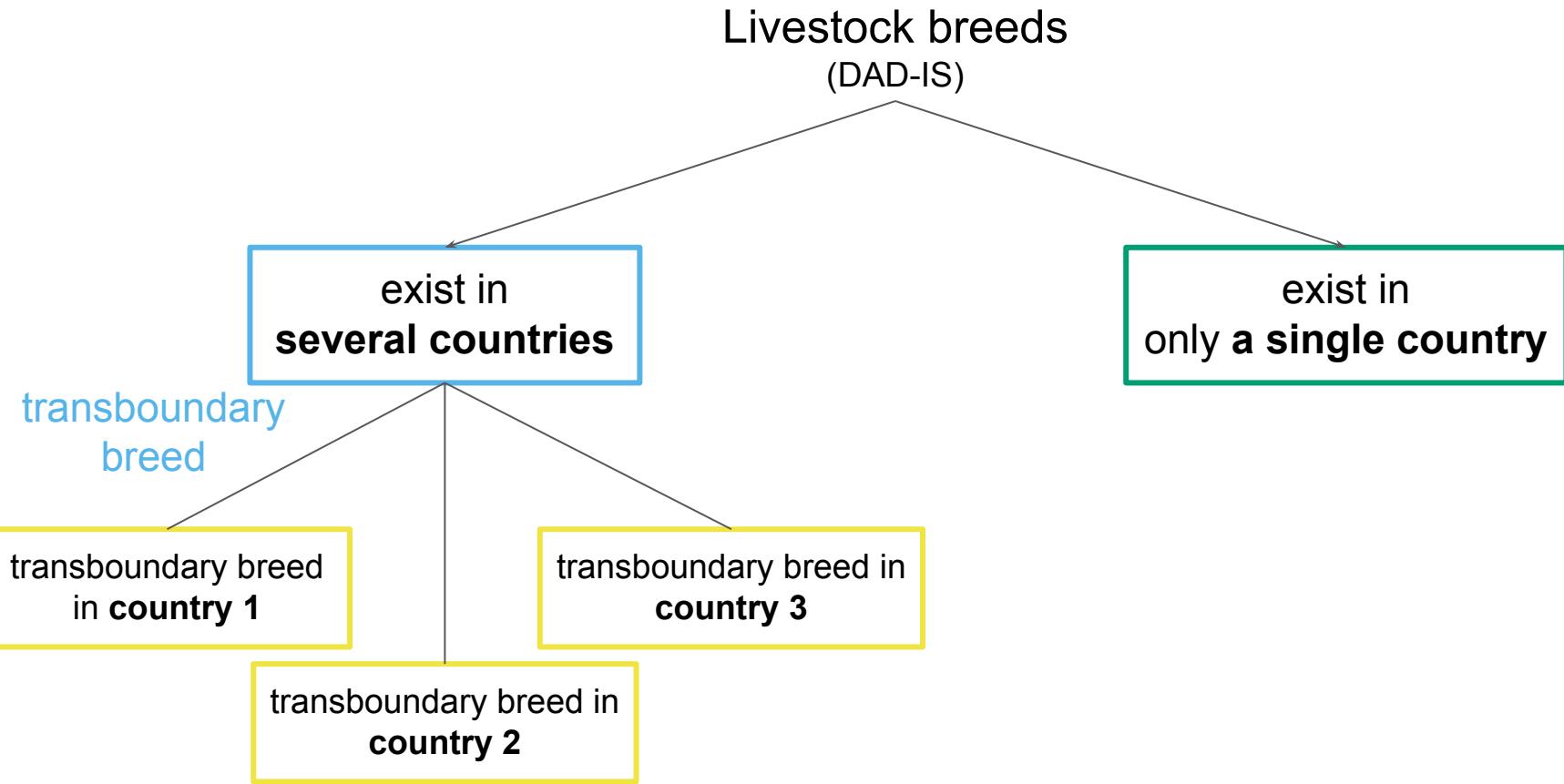
<https://github.com/monarch-initiative/vertebrate-breed-ontology>

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



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)

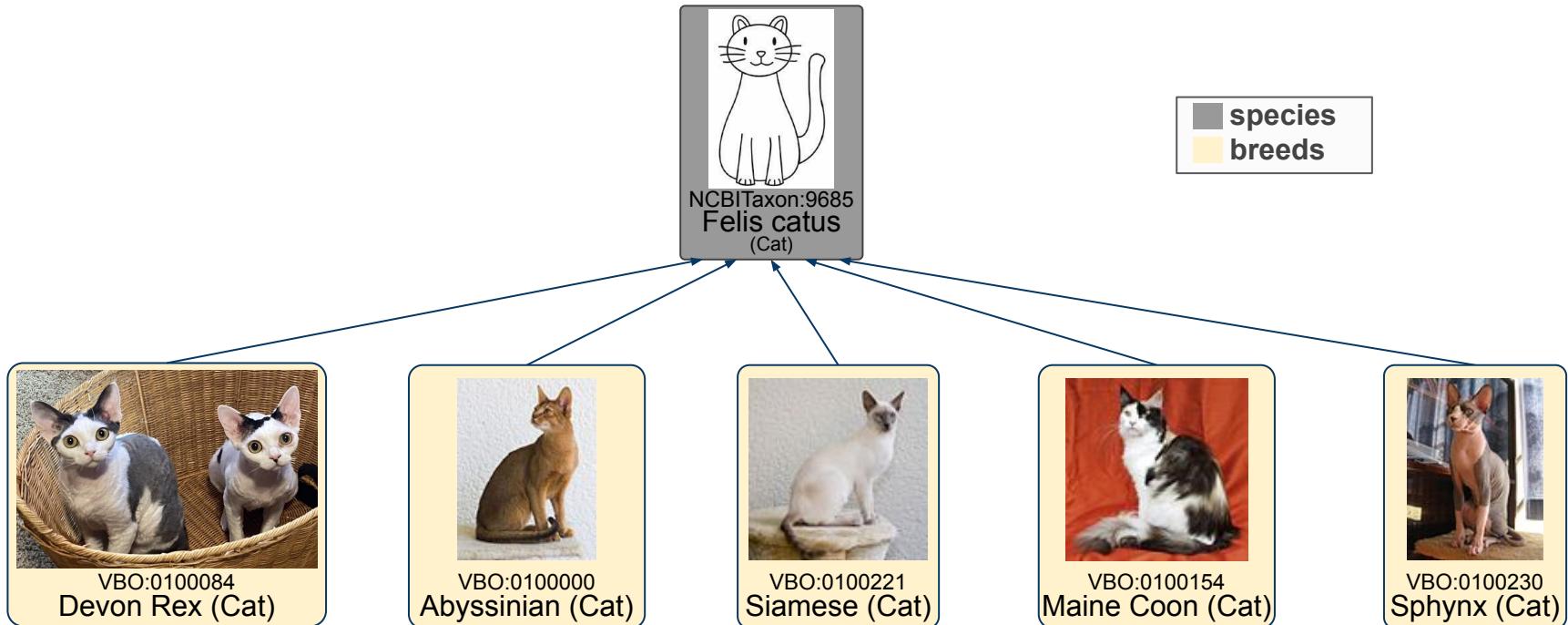
Experts:

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

Literature:

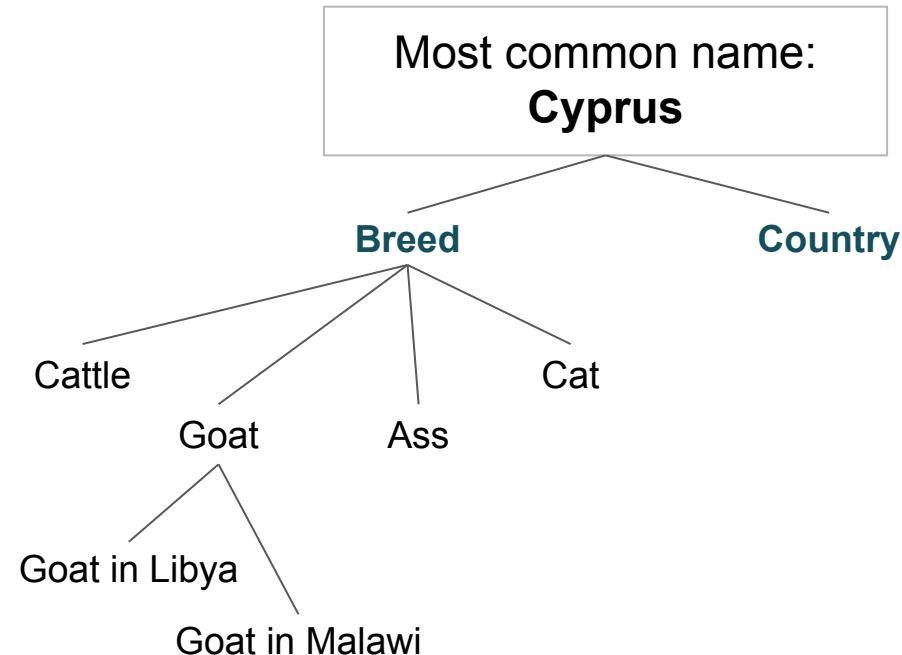
- Online Mendelian Inheritance in Animals (OMIA) (<https://www.omia.org/home/>)

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/>

most common name

sources of the information

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

Annotations Usage

Annotations: Fehér erdélyi kopasznakú, Hungary (Chicken)

Annotations +

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

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

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

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The screenshot shows a detailed view of a VBO term's annotations. The term is identified by a yellow circle icon and the text "Fehér erdélyi kopasznakú, Hungary (Chicken) — VBO:0007282". The interface includes tabs for "Annotations" and "Usage", with "Annotations" currently selected.

The annotations listed are:

- rdfs:label** [type: xsd:string]
Fehér erdélyi kopasznakú, Hungary (Chicken)
- 'has exact synonym'** [type: xsd:string]
Fehér erdélyi kopasznakú
 - 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>

Three red arrows point to the "has exact synonym" annotations, highlighting the "most common name" type and the two source URLs.

VBO terms metadata

- most common name and synonyms
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The image shows two separate cards from a VBO (Veterinary Breeds Observatory) database, each representing a different chicken breed with its metadata.

Card 1: Fehér erdélyi kopasznakú, Hungary (Chicken) — VBO:0007282

Description: Fehér erdélyi kopasznakú, Hungary (Chicken)

Equivalent To +

SubClass Of +

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

wikidata:Q28

Card 2: Jersey Giant, Ireland (Chicken) — VBO:0006071

Description: Jersey Giant, Ireland (Chicken)

Equivalent To +

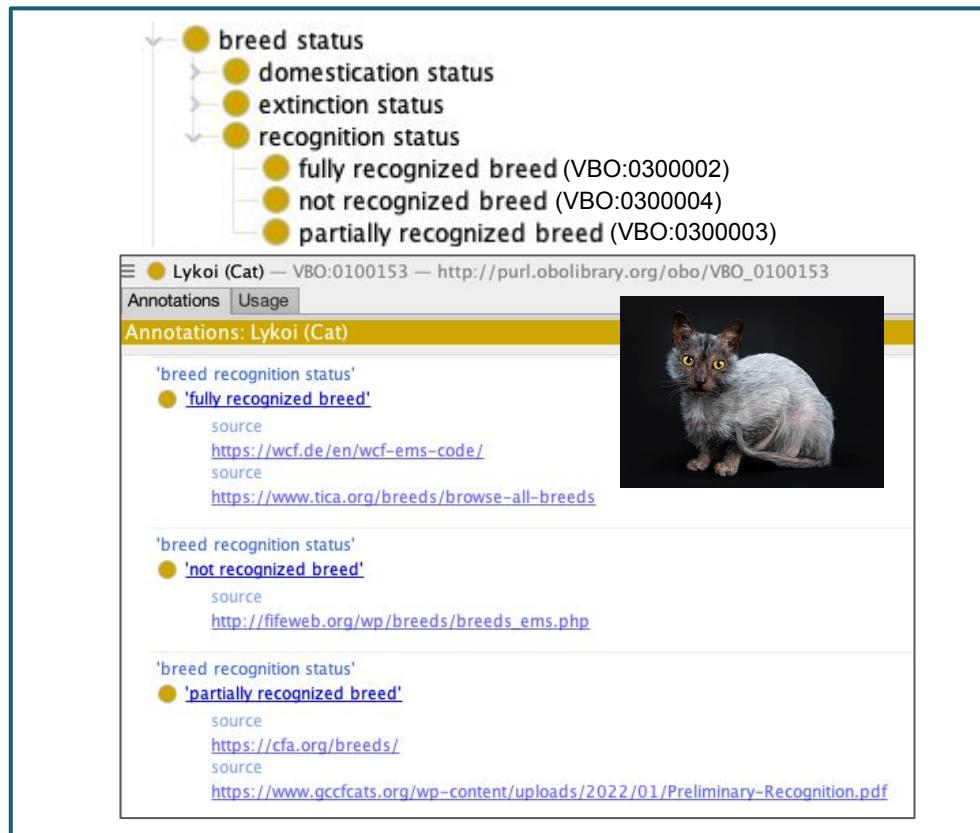
SubClass Of +

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

wikidata:Q22890

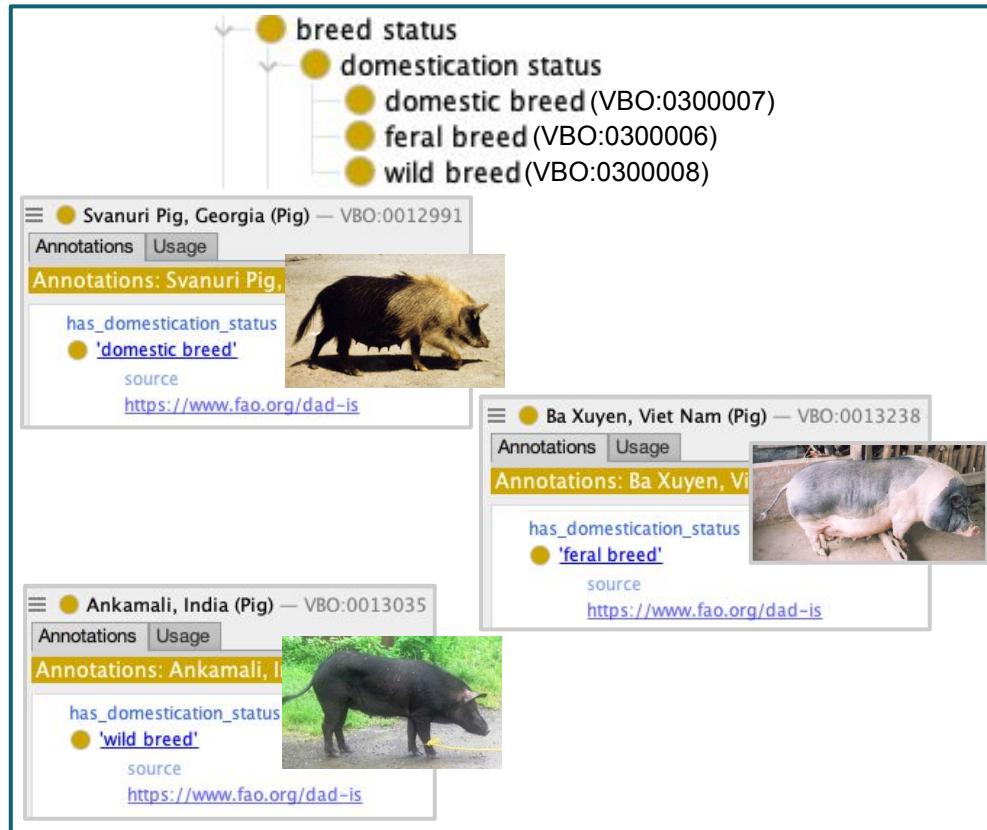
VBO terms metadata

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VBO terms metadata

- most common name and synonyms
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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
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- 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
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 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
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- **description of origin**
- source of information

A screenshot of a web-based metadata editor interface. At the top, there is a header with a yellow circular icon, the text "Aberdeen Angus, Austria (Cattle) — VBO:0002144 — http://purl.ob", and two tabs: "Annotations" (which is selected) and "Usage". Below the header, a yellow bar displays the title "Annotations: Aberdeen Angus, Austria (Cattle)". The main content area shows a field labeled "'description of origin'" with the type "xsd:string". The description text 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 this, there is a "source" section with a link: "<https://www.fao.org/dad-is>". On the right side of the annotations bar, there are three small circular icons with symbols: an '@' symbol, a 'X' symbol, and a 'O' symbol.

VBO terms metadata

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Abyssinian (Cat) — VBO:0100000 — http://purl.obolibrary.org/obo/VBO_0100000

Annotations Usage

Annotations: Abyssinian (Cat)

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

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

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

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

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

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

dcterms:source
<https://www.tica.org/breeds/browse-all-breeds>

dcterms:source
https://www.worldcatcongress.org/wp/cat_breed_comp_aby.php



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
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Summary

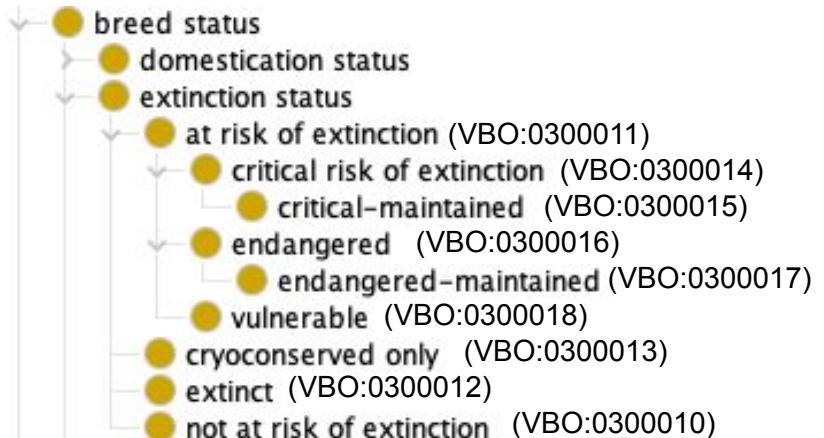
- VBO has been accepted into the OBO Foundry
<https://obofoundry.org/ontology/vbo.html>
- VBO is available in ontology browsers
 - OLS: <https://www.ebi.ac.uk/ols/ontologies/vbo>
 - Ontobee: <https://ontobee.org/ontology/VBO>
- VBO is still in early development, and improvements are in the works.
<https://github.com/monarch-initiative/vertebrate-breed-ontology>

Future work : additional breed information

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

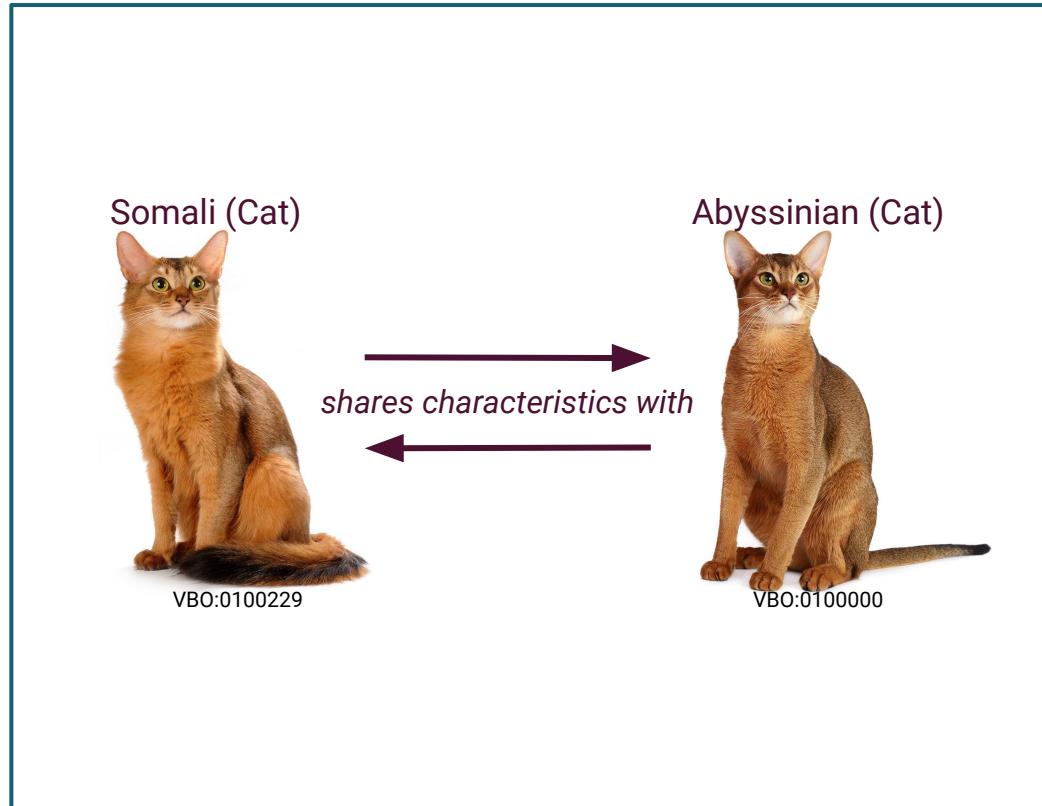
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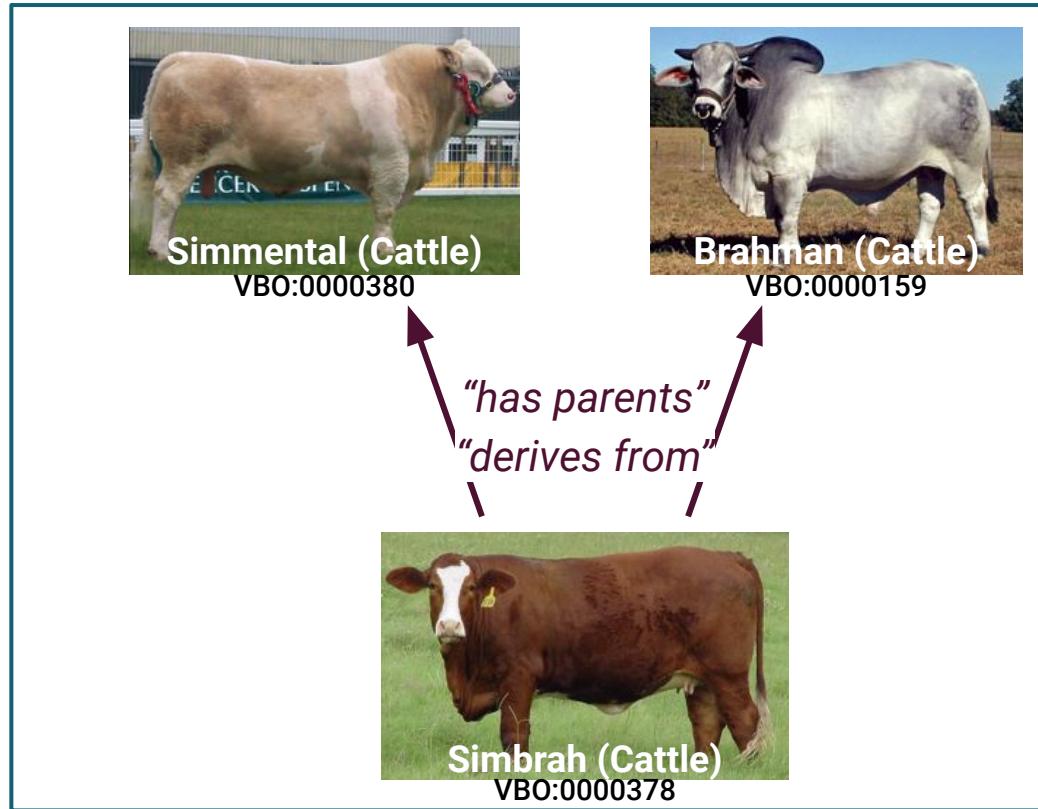
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Short tail trait has been associated with:				
HES7 $c.5T>C$ (homozygous)	100% prevalence	60-100% prevalence	60-100% prevalence	10-25% prevalence
T Box 4 variants identified (heterozygous)	0	0	0	1-10% prevalence
Short tail trait has been associated with:				
HES7 $c.5T>C$ (homozygous)	1-10% prevalence	1-10% prevalence	0	0
T Box 4 variants identified (heterozygous)	25-50% prevalence	25-50% prevalence	25-50% prevalence	10-25% prevalence

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.

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- 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>

VBO Team

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<https://omia.org/>

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(DAD-IS team)*

Gregoire Leroy

Roswitha Baumung

Iowa State University (LBO team)

Zhi-Liang Hu



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



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