Development of Animal Trait Ontology

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ANIMAL SCIENCES GROUP
WAGENINGEN UR

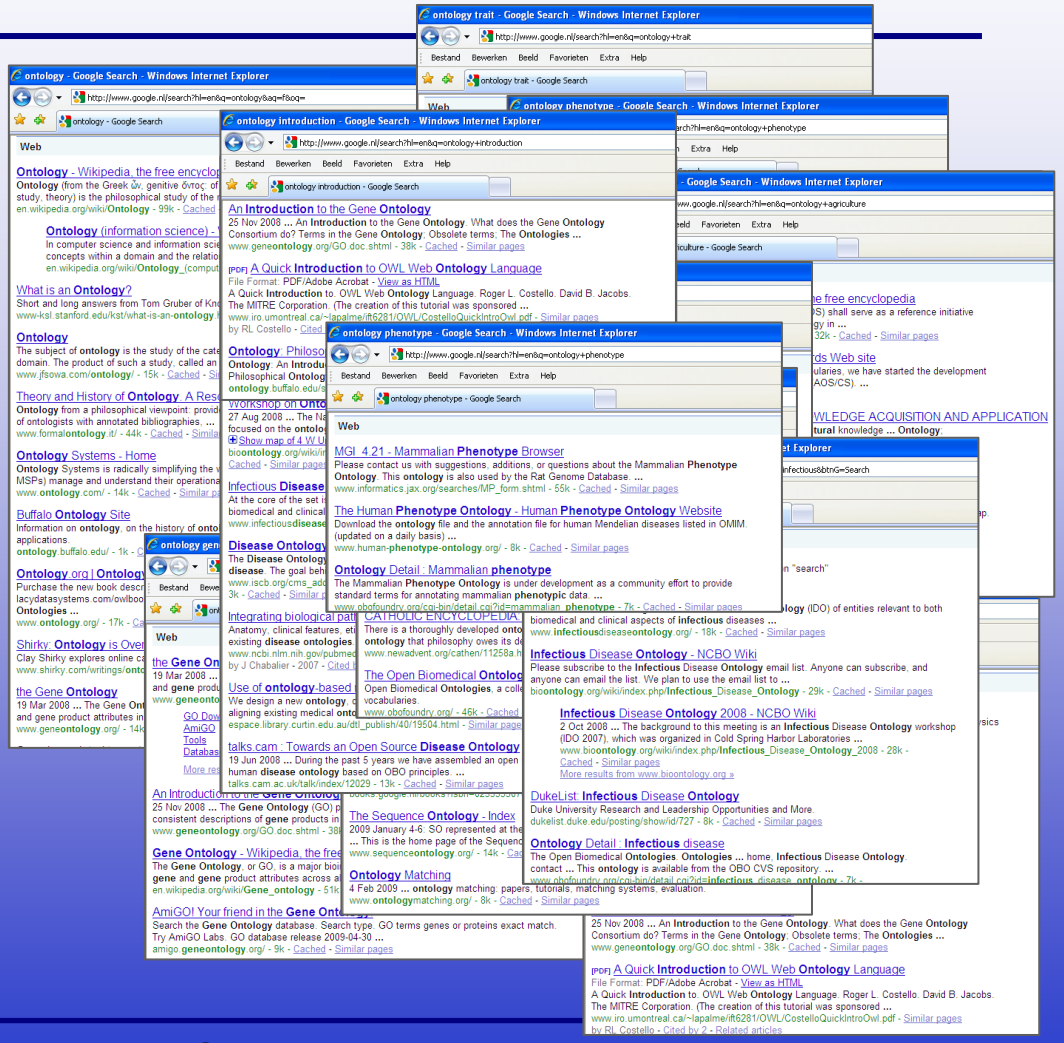
EADGENE – SABRE Industry Days
Rome – May 2009

Outline

- What is an ontology
- Animal Trait Ontology
  - REProduction Ontology (REPO)
  - Host-pathogen ontology for farm animals

What is an ontology?

Internet search for:
Ontology



What is an ontology?

Most quoted definition:

- An ontology is a description (like a formal specification of a program) of the concepts and relationships that can exist for an agent or a community of agents Gruber 1993

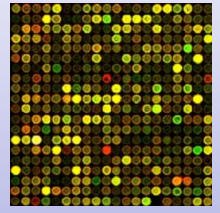
Simple definition:

- An ontology defines the terms used to describe and represent an area of knowledge w3c 2004
  - Ontologies are used by computer programs, database and people: Shared understanding of what particular information means.
  - Ontologies include computer-usable definitions of terminology in the domain and the relationships among them.
  - Ontologies encode knowledge in a domain and also knowledge that spans domains. In this way, they make that knowledge reusable.

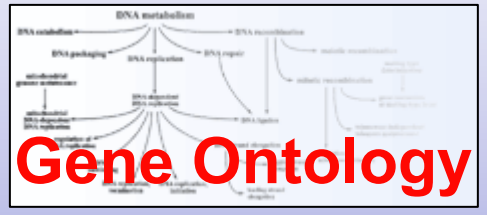
Ontologies are good for:

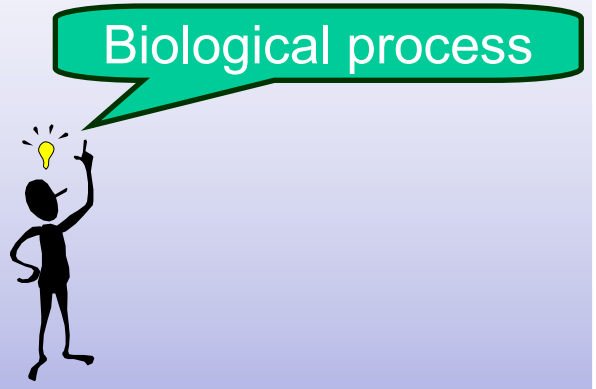
- Controlled vocabulary for science and industry
- Representation of biological knowledge, shared by computers and humans consistently and precisely curating data
- Knowledge source for biological applications
  - Decision support
  - Natural language-processing
  - Data integration
- Standardised descriptions for making inferences
- User search
- Terms for annotating experimental data (increasing number of biological databases using ontologies to annotate data content)

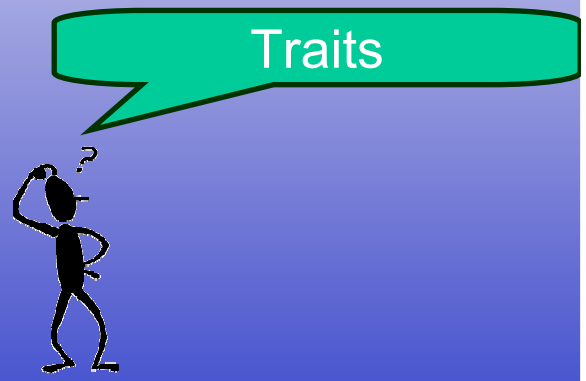
Ontology



Gene, QTL, SNP
expression (list)







Animal Trait Ontology

- A collaboration has started between the U.S. (NRSP-8 Animal Trait Ontology Project) and Europe (EADGENE and SABRE) with the objective of developing a unified animal trait ontology (ATO).

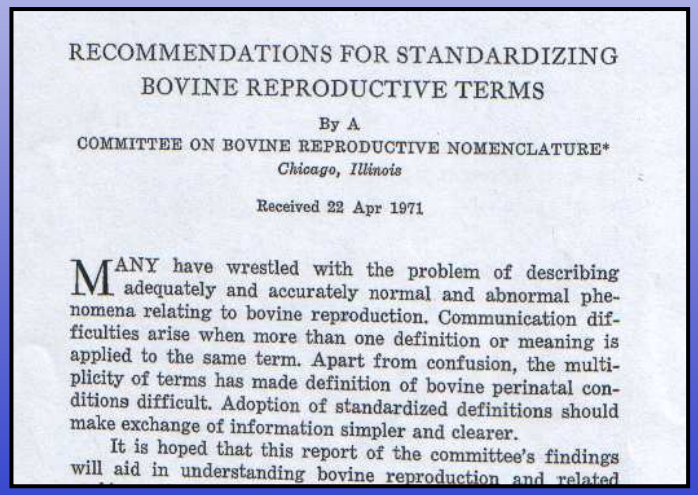
- Aim ATO

  Development of the Animal Trait Ontology (ATO) by including trait information for farm animals and aquaculture species and to create a database that will allow the linkage of the trait and other information (QTLs, SNPs, associations, …) within and across species incl. mouse, rat, human, etc.

  - scientific terminology ↔ industrial terminology

REProduction Ontology (REPO)

- Development of a phenotype ontology database concerning reproduction traits of interest for livestock production
  - An ontology for terms, definitions and relationships of female fertility for use by animal production science and –industry
- Specific sub-domain of REPO
  - female fertility in cattle



RECOMMENDATIONS FOR STANDARDIZING BOVINE REPRODUCTIVE TERMS

By A
COMMITTEE ON BOVINE REPRODUCTIVE NOMENCLATURE*
Chicago, Illinois

Received 22 Apr 1971

MANY have wrestled with the problem of describing adequately and accurately normal and abnormal phenomena relating to bovine reproduction. Communication difficulties arise when more than one definition or meaning is applied to the same term. Apart from confusion, the multiplicity of terms has made definition of bovine perinatal conditions difficult. Adoption of standardized definitions should make exchange of information simpler and clearer.

It is hoped that this report of the committee's findings will aid in understanding bovine reproduction and related

REProduction Ontology (REPO)

- Collection of terms concerning reproduction traits of interest for livestock production
  - Expert knowledge
  - Internet searches
  - Literature searches
- Development Thesaurus
  - Wikisaurus

    posted on the password protected SABRE Intranet for SABRE members
- Building ontology in Protege-owl

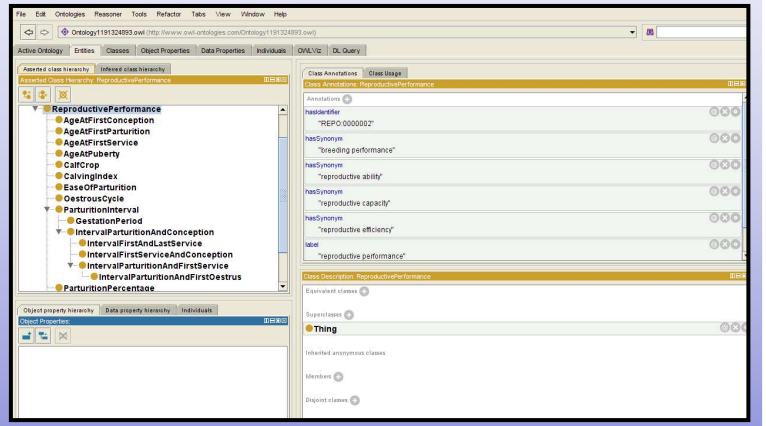
Screenshot wikisaurus

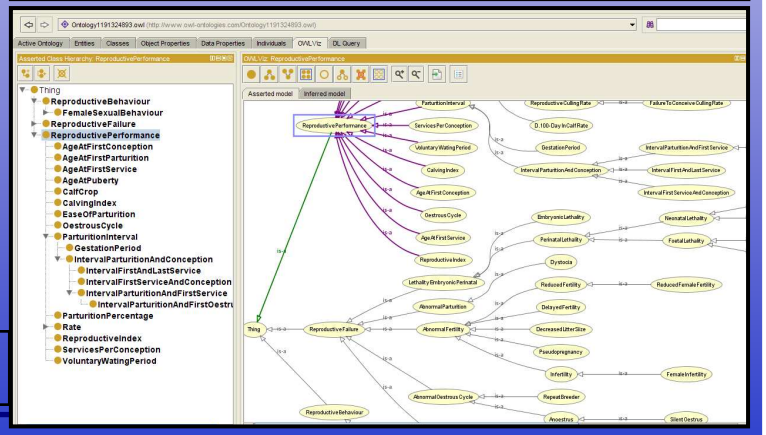
REProduction Ontology (REPO)

Subdomain: female fertility in cattle

The top level terms of REPO

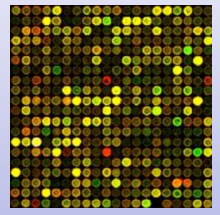
- Reproductive Performance
- Reproductive Behaviour
- Reproductive Failure





Screenshots protégé

Example



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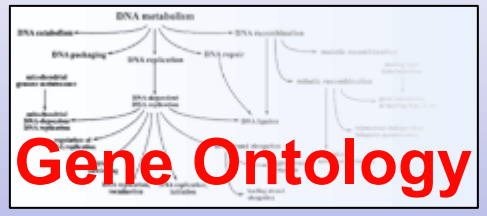
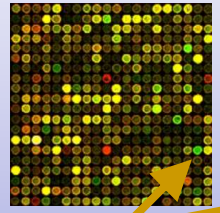


Biological process

Traits

Example



Gene **ESR1**

Biological process

Explorer

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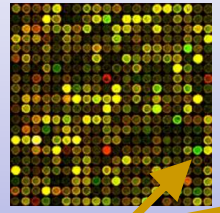
Detailed View

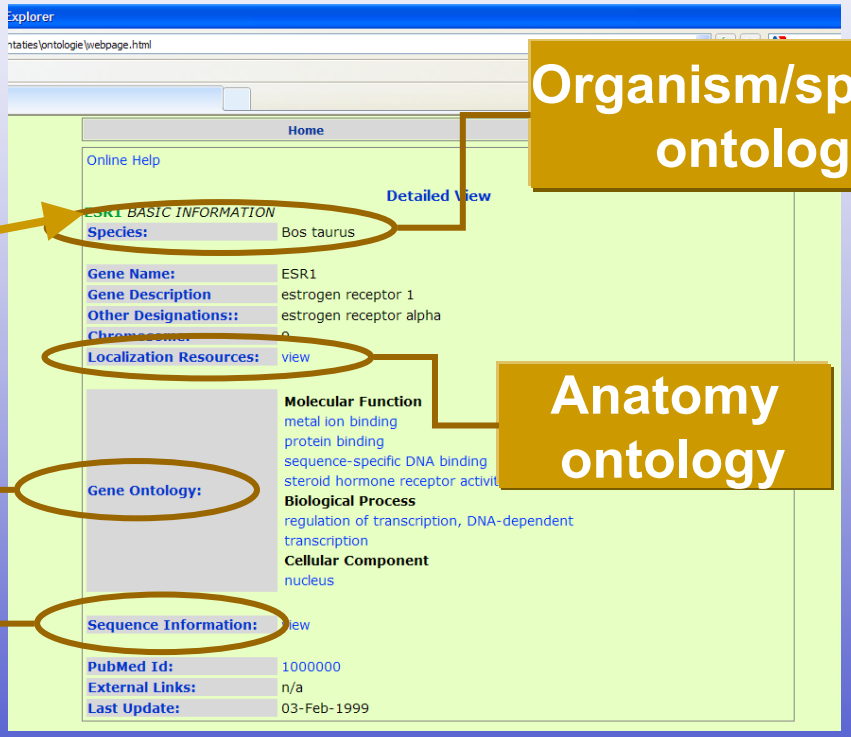
ESR1 *BASIC INFORMATION*

Species:	Bos taurus
Gene Name:	ESR1
Gene Description	estrogen receptor 1
Other Designations::	estrogen receptor alpha
Chromosome:	9
Localization Resources:	view
Gene Ontology:	**Molecular Function** metal ion binding protein binding sequence-specific DNA binding steroid hormone receptor activity **Biological Process** regulation of transcription, DNA-dependent transcription **Cellular Component** nucleus
Sequence Information:	view
PubMed Id:	1000000
External Links:	n/a
Last Update:	03-Feb-1999

Example



Gene **ESR1**



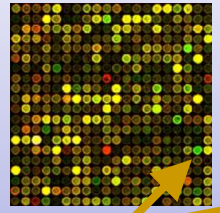
Organism/species ontology

Gene Ontology

Anatomy ontology

Sequence Ontology

Example



Gene **ESR1**

Traits

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Localization Resources:	view
Gene Ontology:	**Molecular Function** metal ion binding protein binding sequence-specific DNA binding steroid hormone receptor activity **Biological Process** regulation of transcription, DNA-dependent transcription **Cellular Component**
Animal Trait Ontology:	**Trait** infertility proceptive behaviour female sexual behaviour
Sequence Information:	view
PubMed Id:	1000000
External Links:	n/a
Last Update:	03-Feb-1999

Animal Trait Ontology:

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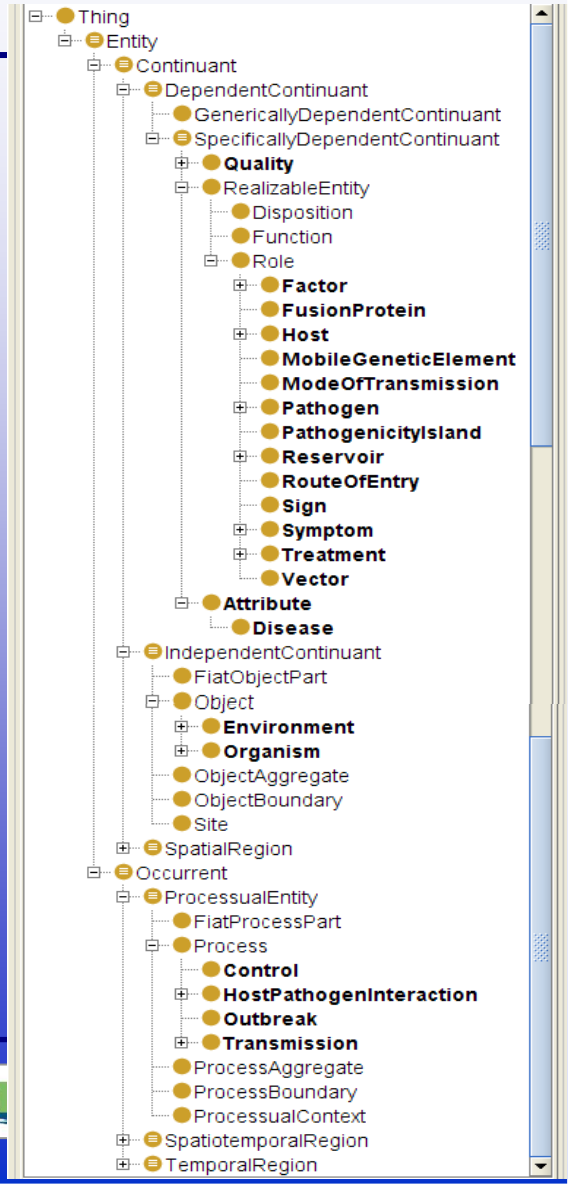
infe
pro
fem

REProduction Ontology (REPO)

http://www.animalgenome.org/cgi-bin/amigo/browse.cgi

Host-pathogen ontology for farm animals

- Development of a host-pathogen ontology for farm animals.
- Subdomain: salmonella – pig.
- Reuse: Reuse ontologies (or parts)
  - Infectious Disease Ontology (human)
  - Gemina
  - Transmission ontology
  - ……

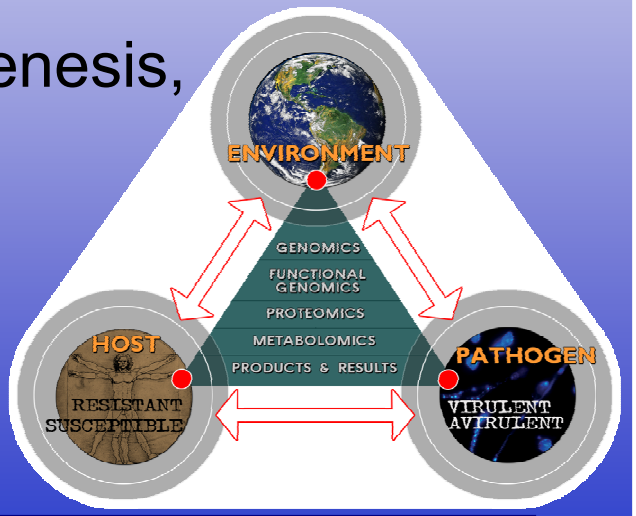


Host-pathogen ontology

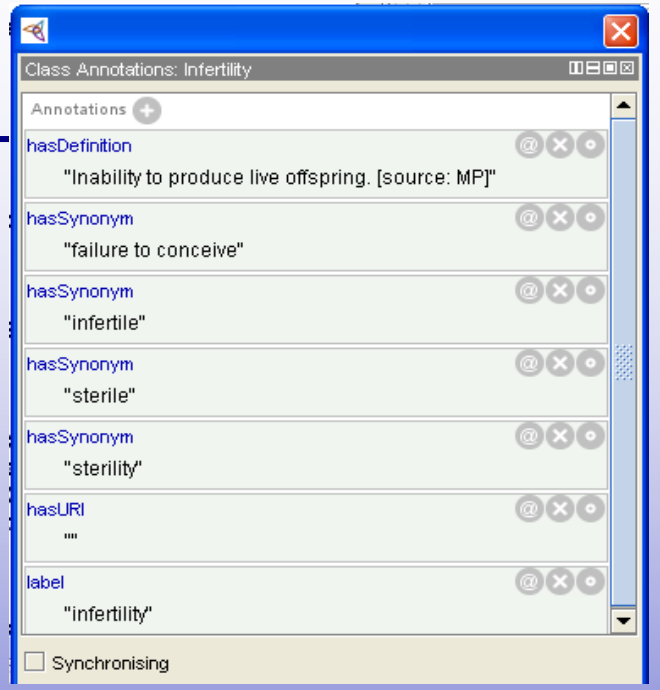
Scope

- **Scales:** molecules, cells, organisms, populations
- **Organisms:** host, pathogen, vector, model organisms, interactions between them
- **Domains:** biological, clinical care, health
- **Diseases:** etiology, nature of pathogenesis, signs, symptoms, treatments

Lindsay Cowell

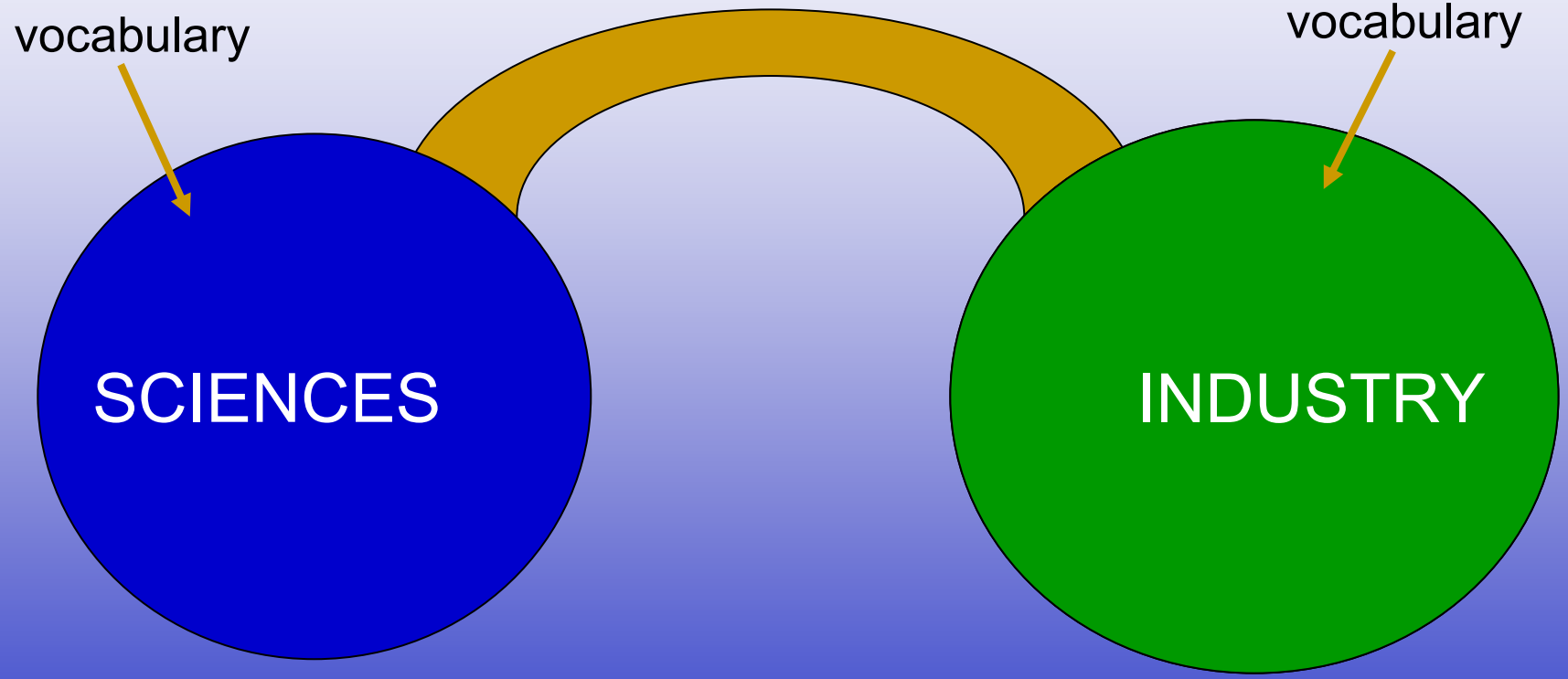


Ontology



- Terms
- Synonyms: They facilitate searching the ontology.
  - Different words that mean the same thing.
- Definitions: Each term should have a definition. A Definitions explains clearly what is meant by a particular term. A definition must have a reference to it's origin. (PubMed, database, website, the person that created it).
- Understanding relationships: Some type are: is_a, part_of and derived_from

Requirements



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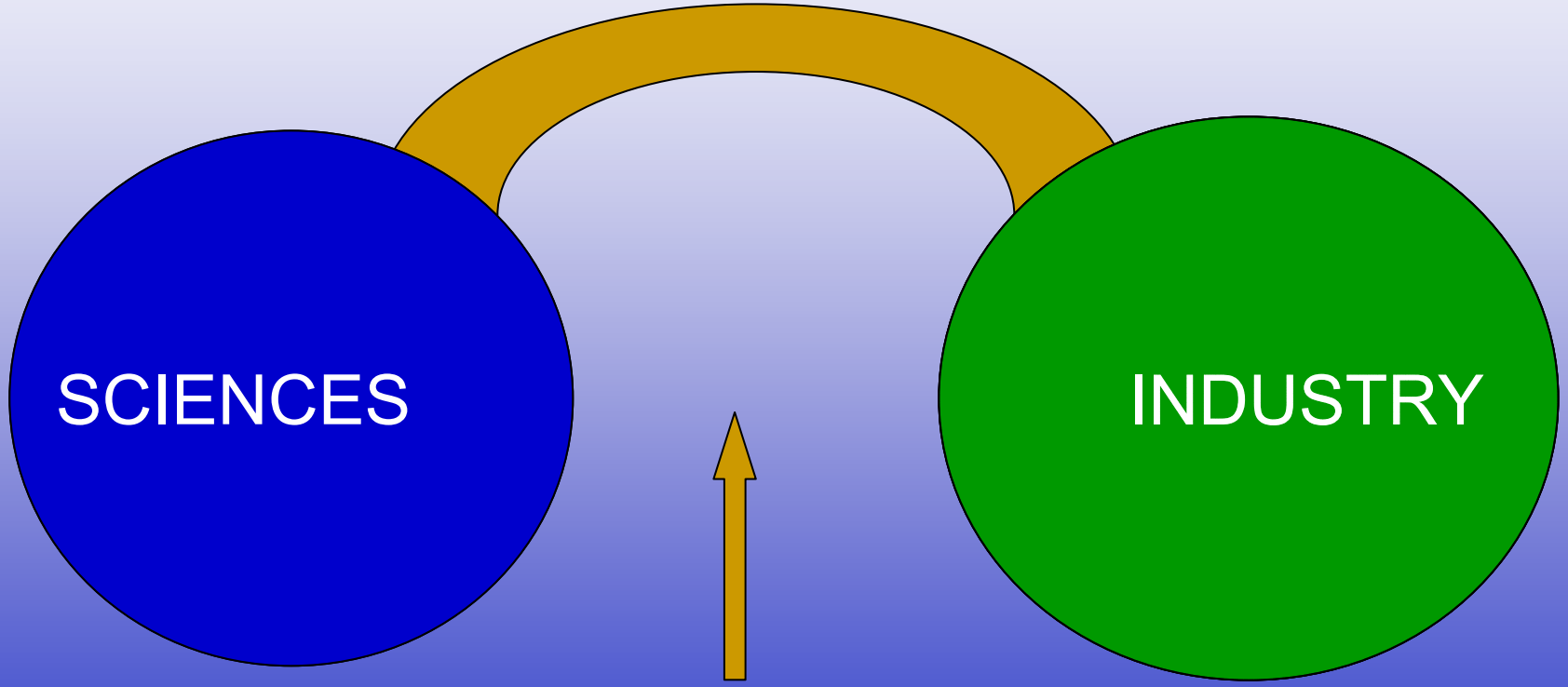

Better Animal Trait Ontology

Acknowledgements

- WP5
- WP6 and WP3
- KB