
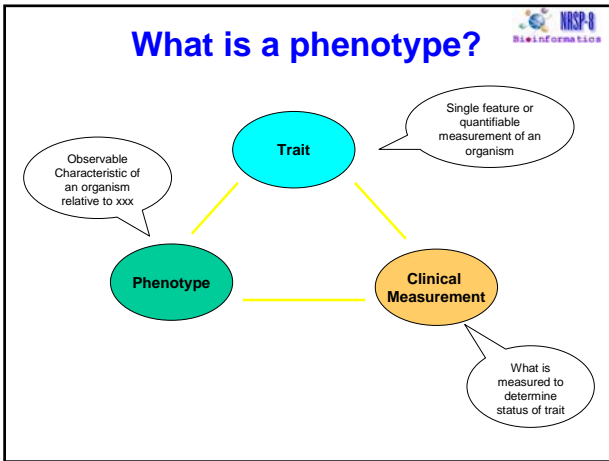
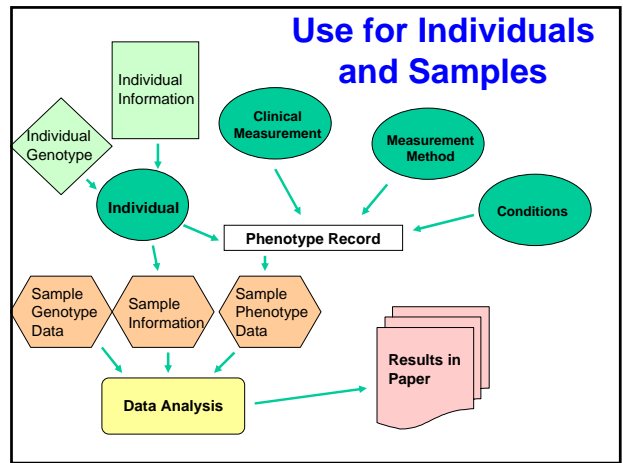

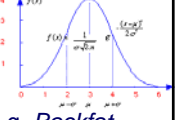




Global perspectives on animal trait ontology

J.M. Reecy
 Iowa State University





- 
- 
- Trait measurement variations, e.g. *Backfat*
 - Backfat Thickness (Average Backfat) by ruler
 - Backfat Thickness (Average Backfat) by ultrasound
 - Backfat Thickness (Average Backfat) by Fat-O-Meater
 - Backfat at First Rib (First Rib Backfat)
 - Backfat at First Rib (Measured at 14 Weeks of Age)
 - Backfat at First Rib (Measured at 26 Weeks of Age)
 - Backfat at Last Rib (Measured at 14 Weeks of Age)
 - Backfat at Last Rib (Measured at 26 Weeks of Age)
 - Backfat Thickness at Last Rib
 - Backfat at Shoulder
 - Backfat at Tenth Rib
 - Backfat between 3th and 4th Rib
 - Backfat between 6th and 7th Rib
 - Backfat Depth at Max. Mus. Depth
 - Backfat at Last Lumbar
 - Backfat weight (dissected total weight)
- by methods
- by time
- by locations







Why trait ontology?


Everyone can tell the difference between Apples and Oranges



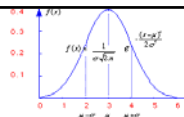
But can you tell **McIntosh** from **Cortland**?




- 
- 
- ## What is an ontology?
- Webster's Dictionary: a branch of metaphysics concerned with the nature of relations of being
 - Dictionary.com: the hierarchical structuring of knowledge about things by subcategorizing them according to their essential (or at least relevant and/or cognitive) qualities
 - Gruber: a specification of a conceptualization



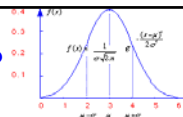
What an ontology is NOT




- Not a collection of facts rising from a specific situation
 - Provides all semantic entities to describe that situation
- Not a model of an application domain
 - List of all building blocks with their valid modes of combination
- Not a database schema (categories/data types)
 - describes relations between entities in the real world
- Not a knowledge base
 - A particular knowledge base about concepts and their relations
- Not a taxonomy (superclass and subclass relations)
 - Open to many types of relations
- Not a vocabulary or dictionary
 - Describes the hierarchy and relations of each term
- Not a semantic net (use of words)
 - Nature of the entity referenced by the word



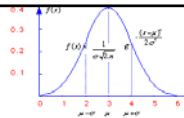
Why use an Ontology?




- To share common understanding of the structure of information among people or software agents
- To enable reuse of domain knowledge
- To make domain assumptions explicit
- To separate domain knowledge from the operational knowledge
- To analyze domain knowledge



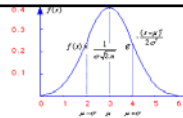
AIMS



- Development of the Animal Trait Ontology (ATO) by including trait information for the pig, chicken, cow, and other farm animals
- Create a database that will allow the linkage of the trait information for each species stored in the ontology to its QTL information
 - Mouse, Rat, Human, etc.



Classification of Traits



- Mammalian Phenotype Ontology (MGI website) not suitable for production traits.
- Define a 3 level Ontology based on the different publications :
 - ↳ **Body traits**
 - ↳ Behaviour
 - ↳ Immune response
 - ↳ Infectious disease
 - ↳ Metabolic disease
 - ↳ Reproduction
 - ↳ Carcass traits
 - ↳ Fat
 - ↳ Growth traits
 - ↳ Meat quality
 - ↳ Metabolic traits
 - ↳ Muscular system
 - ↳ Nervous system
 - ↳ Organs
 - ↳ Plumage
 - ↳ Skeletal system
 - ↳ % Abdominal fat weight
 - ↳ Abdominal fat weight
 - ↳ Abdominal fat weight / BW
 - ↳ Abdominal fat width
 - ↳ Fat distribution
 - ↳ Skin fat weight
 - ↳ Skin fatness
- Interactions between traits or traits belonging to different group : Thigh meat / bone ratio; meat color adjusted for body weight...

Kindly supplied by Wilfrid Curre

Iowa State University

Collaboration is Needed

Example: Gene Ontology Consortium



Iowa State University

Challenges

- Knowledge Integration
- Management of the Ontology
 - Concurrence Management
 - Consistency Maintenance
 - Privilege Management
 - History Maintenance
 - Scalability
- Providing the community with what it needs!
 - Once size does not fit all

Iowa State University

Collaboration

An animal trait ontology that involves multiple research groups across the world

Swine, Horse, Cattle, Chicken

Each group works on an ontology module for a particular species (according to the group's best expertise)

13

Iowa State University

Future Human/Animal Trait Ontology

Case: A Case: B Case: C

Disease X X X

Phenotype Y Y Y1 Y2

Trait Z Z1 Z2 Z

14

Iowa State University

ATO Example

Disease	Phenotype(s)	Trait
1. Diabetes	Decreased insulin level, Increased insulin level	Insulin concentration
2. Cardiomyopathy	Increased blood vessel dilation, Decreased blood vessel dilation	Blood vessel diameter
3. Cancer	Decreased tumor incidence, Increased tumor incidence, Decrease tumor growth/size, Increased tumor growth/size Late onset, Early onset	Tumor number Tumor diameter Tumor onset

15

Iowa State University

Phenotype And Trait Ontology (PATO)

- Phenotypes may be described in many different dimensions, e.g.
 - the biochemical ('alcohol dehydrogenase null')
 - the cellular ('cell division arrested at metaphase'),
 - the anatomical ('eye absent')
 - the behavioural ('hyperactive').
 - etc.
- In whatever dimension and granularity, however, there is a commonality and the great majority of, if not all, phenotypic descriptions can be decomposed into two parts
 - An **entity** that is affected. This entity may be an enzyme, an anatomical structure or a complex biological process.
 - The **qualities** of that entity.

16

Iowa State University

Phenotypic Character

Phenotypic Character entity + quality

(mouse body weight) (mouse anatomy: body + PATO: weight)

(eye colour) (Drosophila anatomy: eye + PATO: colour)

(glucose concentration) (ChEBI: glucose + PATO: concentration)

increased size hepatocellular carcinoma hepatocellular carcinoma (MPATH:357) has_quality increased size (PATO:0000586)

17

18

Iowa State University

Advantages

Providing these definitions in obo format has the following advantages:

- PATO definitions of qualities can be shared and reused
- The obo edit reasoner can keep pre-composed terms in sync with the various phenotype ontology they reside and PATO, and perform automatic DAG placement of terms
- PATO terms can be used to query pre-composed annotated phenotypes
- Pre-composed terms can be used to query annotated phenotypes (eg "halogen sensitivity" can be used as a query, even though this might not be pre-composed yet)

19

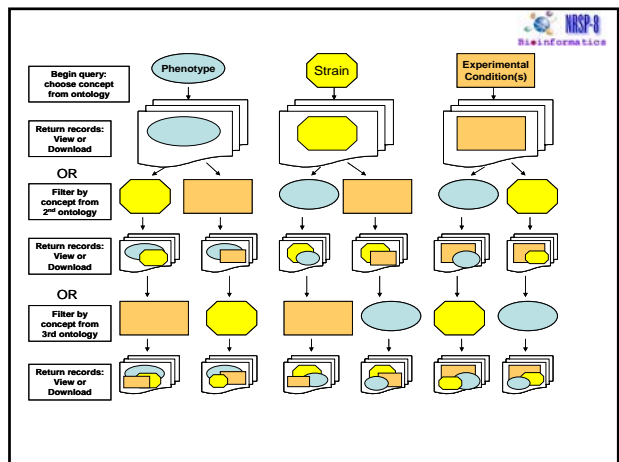
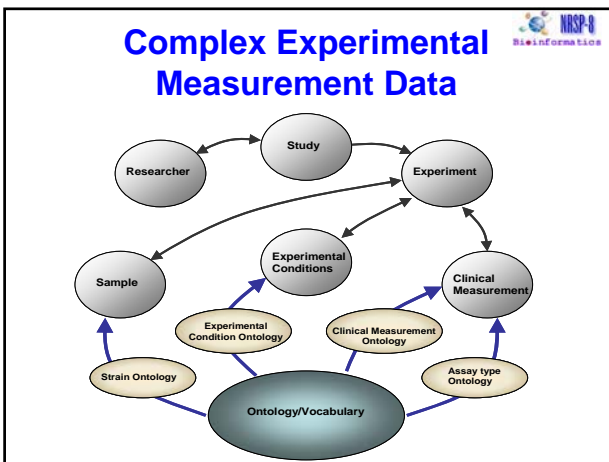
Iowa State University

Future ATO

The diagram illustrates the integration of PATO (Phenotype And Trait Ontology) into three different cases (A, B, and C) across three levels: Disease, Phenotype, and Trait.

- Case A:** Disease X is linked to Phenotype Y, which is linked to Trait Z.
- Case B:** Disease X is linked to Phenotype Y, which is linked to Trait Z1 and Z2.
- Case C:** Disease X is linked to Phenotype Y1 and Y2, which are both linked to Trait Z.

20



Future Development

- **Pig, Cattle, Chicken**
 - All QTL traits
 - All phenotypes
- **Integrate with Rat and Mouse databases**
- **Decompose Trait terms into PATO**
- **Develop ontology query tools**
- **Link information across databases**

NIRSP-B
Bioinformatics

Acknowledgements

Iowa State University

- Sue Lamont
- Max Rothschild
- Chris Tuggle
- Zhiliang Hu
- Eric Fritz
- Laron Hughes
- Jie Bao
- Neeraj Kaul
- Vasant Honavar

Collaborators

- Mary Shimoyama
- Anne Kwitek
- Mindy Dwindell
- EADGENE

NIRSP-B
Bioinformatics