Plant and Trait Ontology Current Status

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Plant Ontologies for Agronomic Traits
EBI, UK
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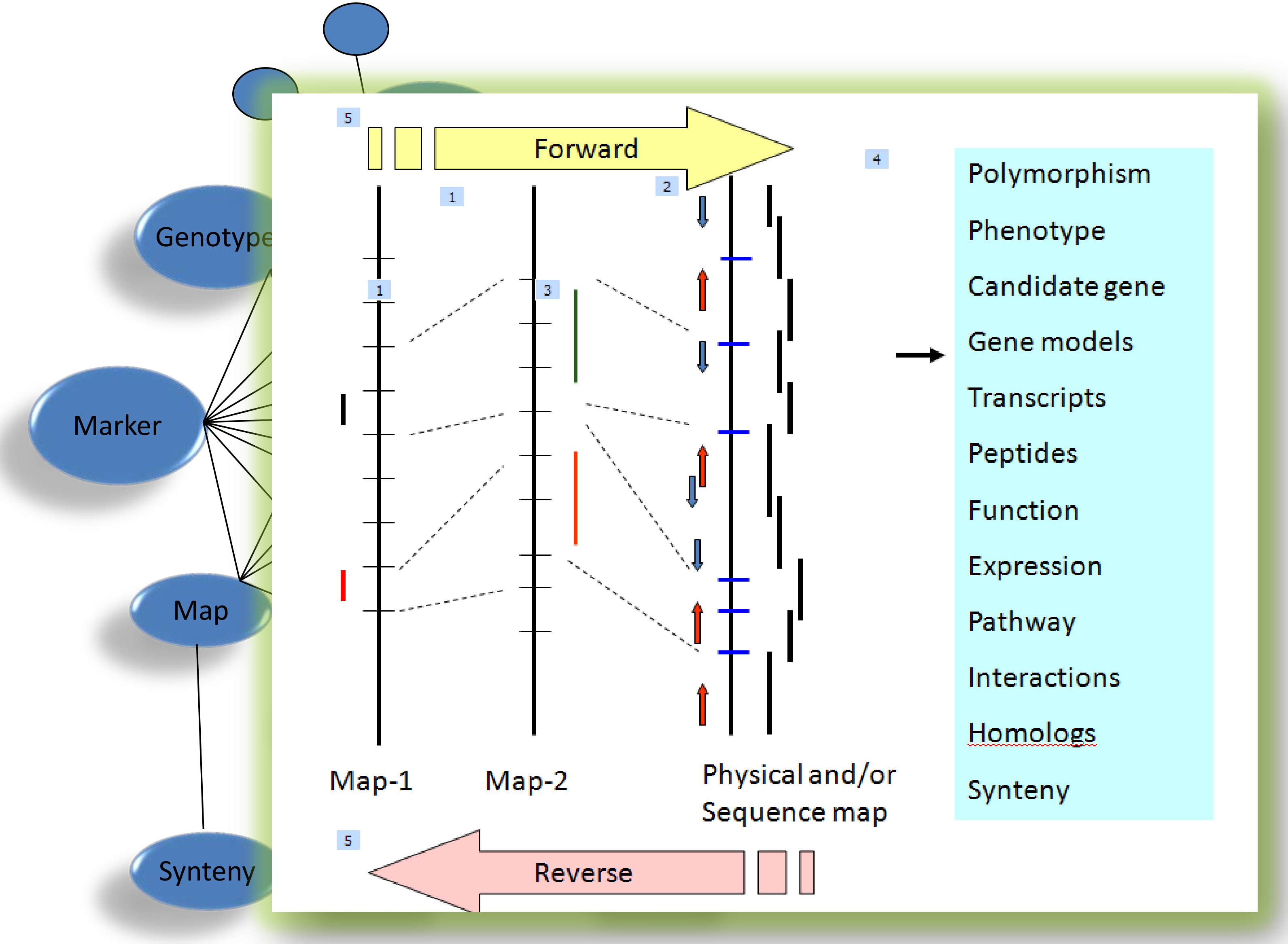
Comparative and Translational Genomics



Polymorphism

Phenotype

Candidate gene

Gene models

Transcripts

Peptides

Function

Expression

Pathway

Interactions

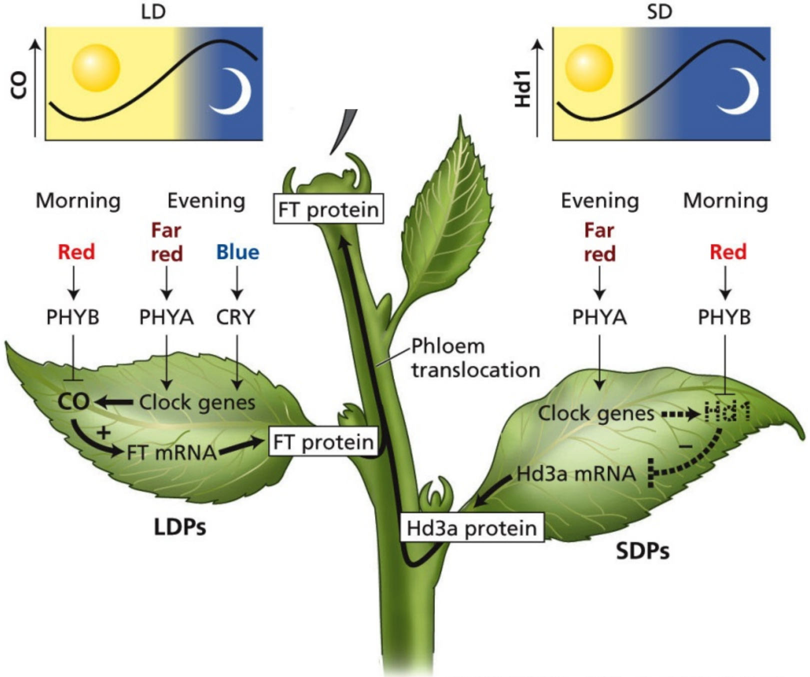
Homologs

Synteny

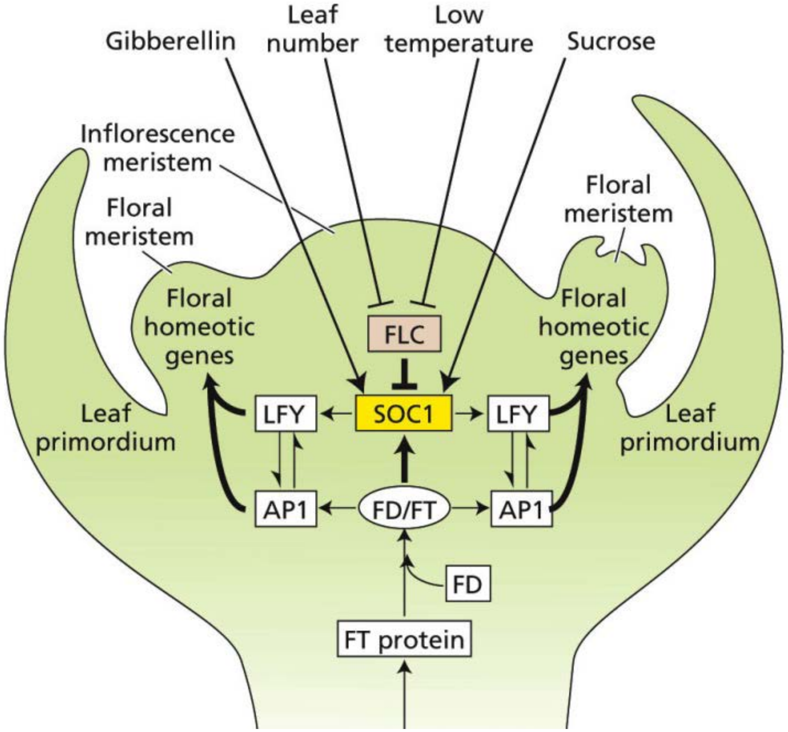
Photoperiod and the process of flowering

Arabidopsis
long-day
flowering

Rice
Short-day
flowering



PLANT PHYSIOLOGY, Fourth Edition, Figure 25.33 (Part 1) © 2006 Sinauer Associates, Inc.



GWAS and QTL studies

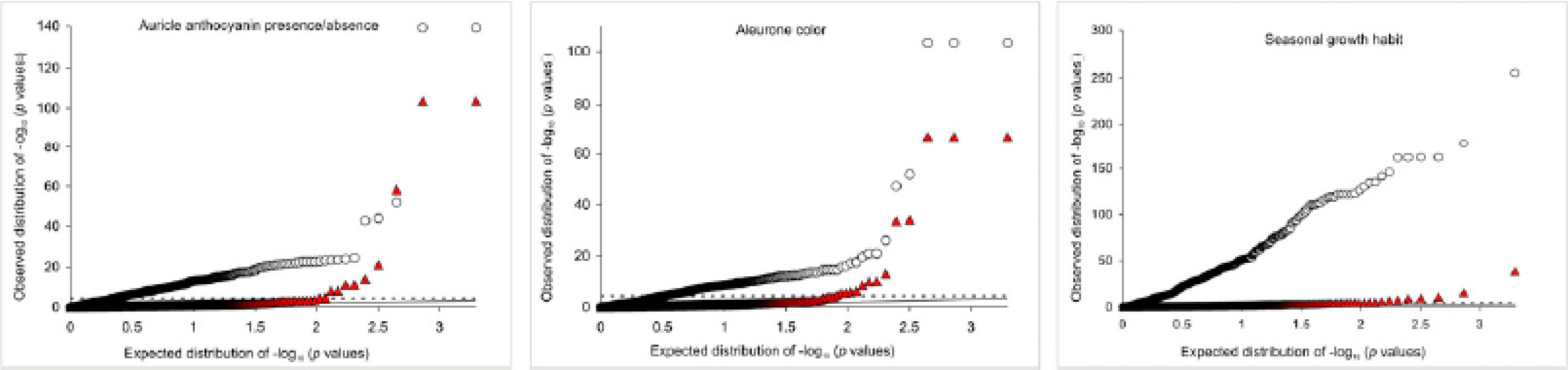
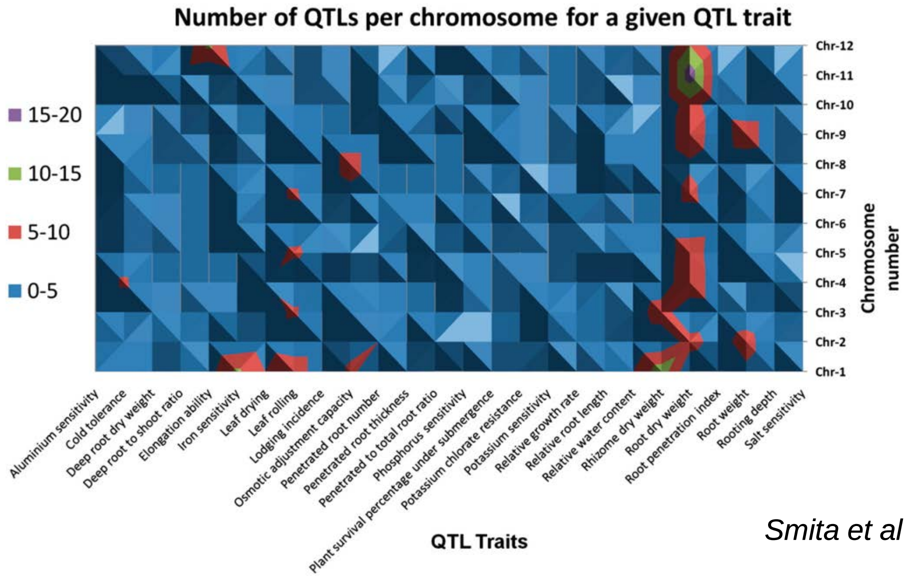


Fig. S4. Quantile–quantile plots for traits returning significant associations after GWA analysis. Expected vs. observed P values are plotted for naive (circles) and mixed model corrected (triangles) analyses. The $x = y$ line (solid) and Bonferroni corrected $P = 0.05$ significance thresholds (dashed line) are indicated.

Cockram et al (2010) PNAS



Smita et al (2011) doi: 10.1093/database/bar037

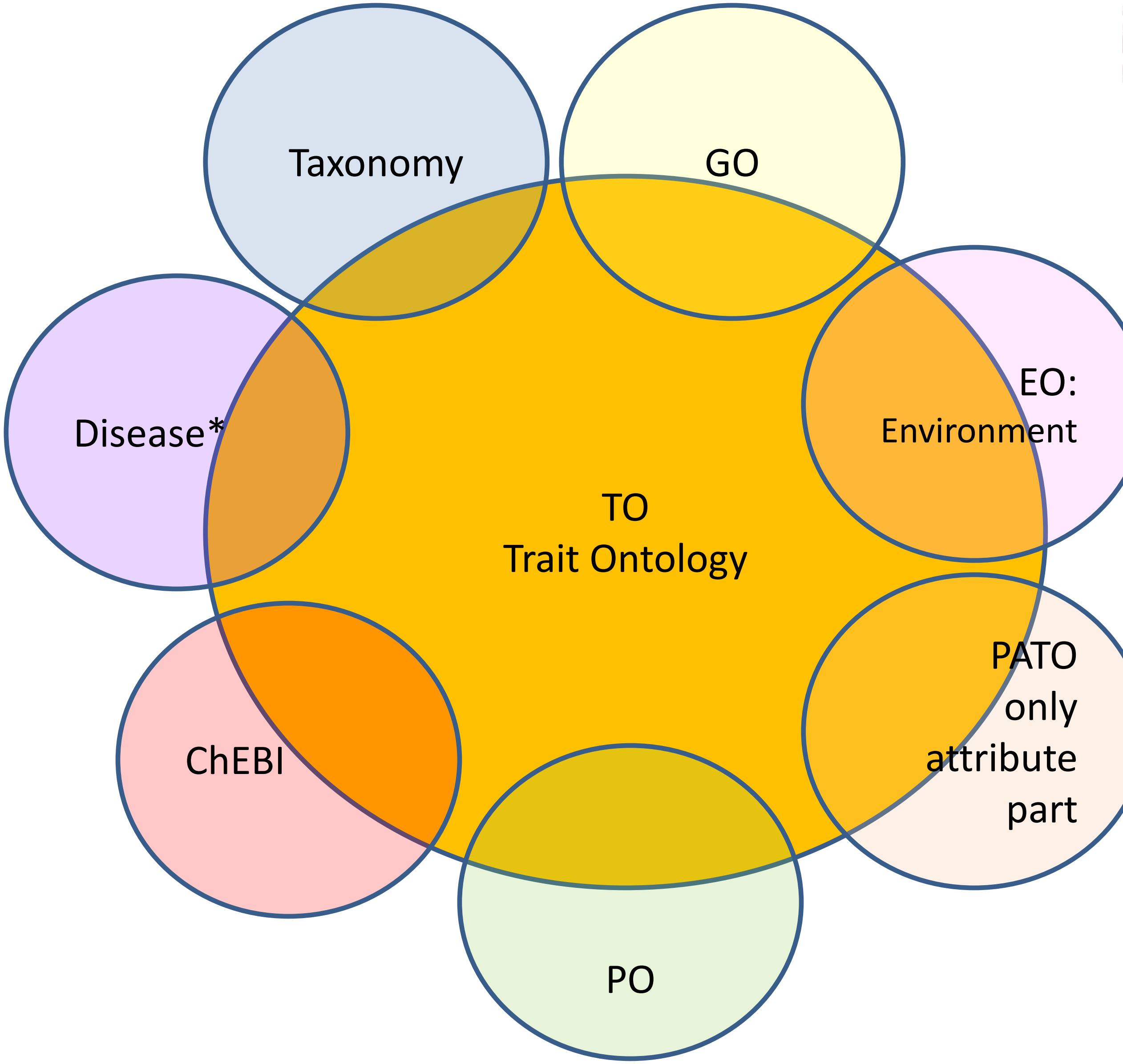
Trait vs Phenotype

- Entity+Attribute = Trait [**observable**]
  E.g. Leaf(PO) + color (PATO-A) = Leaf color(TO)

- [Entity+Attribute+Value] = Phenotype [**observed**]
  e.g. Leaf(PO) + color (PATO-A) + yellow (PATO-V) = Leaf color yellow [EAV model:old]

- [Entity+(Attribute+Value)] = Phenotype [**observed**]
  e.g. Leaf(PO) + color yellow (PATO-AV) = Leaf color yellow
  [EA model:NEW]

Traits are the visible markers from multiple dimensions



Taxonomy

GO

Disease*

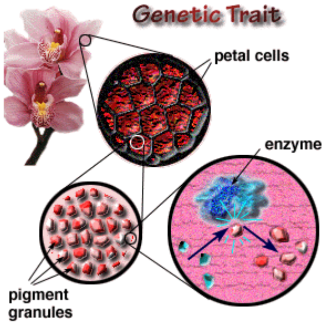
EO:
Environment

TO
Trait Ontology

PATO
only
attribute
part

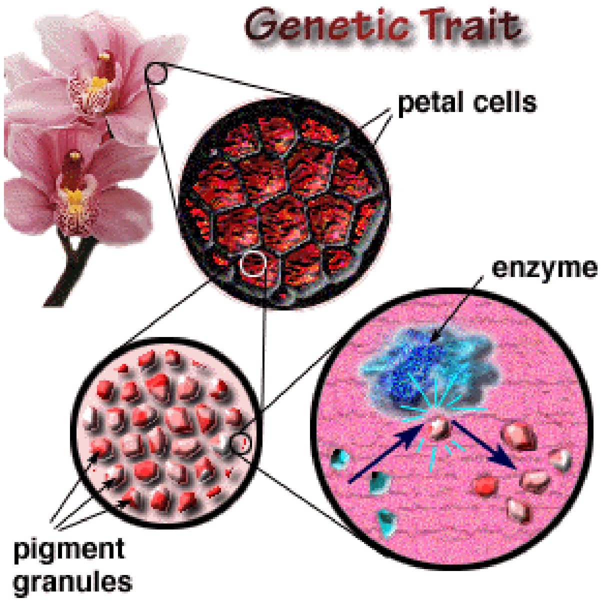
ChEBI

PO



Trait Ontology

- It’s an ontology of Plant traits (characteristics/observable)
- Precomposed trait terms
- Includes aspects of:
    - Anatomy trait
    - Growth and development (phenology) trait
    - Biochemical trait
    - Stress (abiotic and biotic stress) trait
    - Agronomic quality trait
    - Yield trait
    - Metabolomic trait
- Does not include phenotype term



Pre Composition Approach-1

```
[Term] id: TO:0000372 name: amylose to amylopectin ratio
namespace: plant_trait_ontology
intersection_of: PATO:0000025  !composition
intersection_of: towards CHEBI:28102  ! amylose
intersection_of: relative_to CHEBI:28057  ! amylopectin
```

```
[Term] id: TO:0000188 name: drought sensitivity
namespace: plant_trait_ontology
intersection_of: PATO:0000085  ! sensitivity
intersection_of: towards EO:0007404 ! drought environment
```

```
[Term] id: TO:0000227 name: root length
intersection_of: PATO:0000122  ! Length
intersection_of: inheres_in PO:0009005  ! root
```

Pre Composition Approach-1

Disease resistance phenotypes

[Term] id: TO:0000323 name: stem rot disease resistance

def: "Causal agent: Magnaporthe salvinii (Nakataea sigmoidea, Sclerotium oryzae), and Helminthosporium sigmoideum var. irregulare. Symptoms: dark lesions develop on the stems near the water line. Small, dark bodies (sclerotia) develop, weaken the stem and cause lodging."

*Easy to define if we had an orthogonal ontology of plant diseases and infectious agents. The definition pattern would be: A <TO:resistance> which *towards* <InfectiousAgent>*

Assay-specific terms

Example: root dry weight

It is unclear how to proceed with these. No action was taken for these.

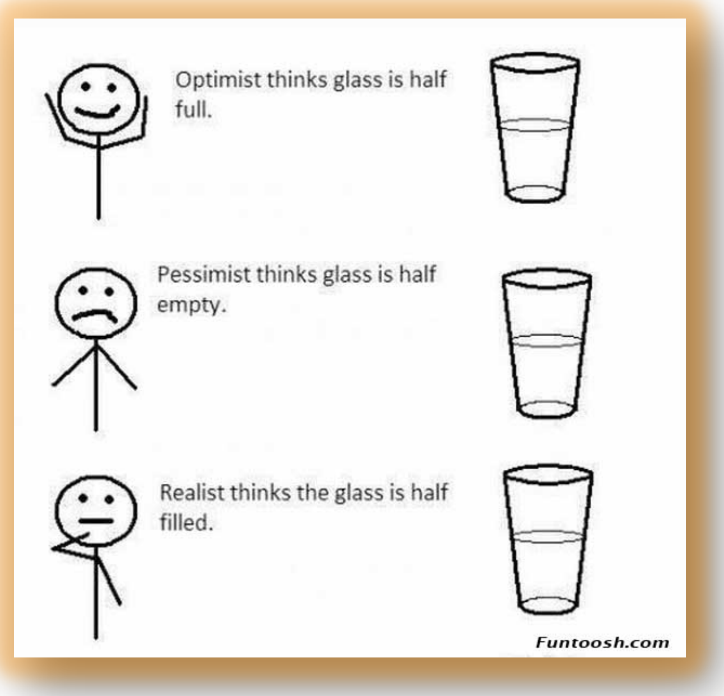
Conjunctive terms

Example: lemma and palea related traits

Example: lemma and palea pubescence

a pubescence which *inheres_in* the lemma and *inheres_in* the palea

Why TO Has No Observed Phenotype Values?



$$C = \begin{array}{c|cccccccccccccccc|}
 & A & B & C & D & E & F & G & H & I & J & K & L & M & N & O & P \\
A & 4 & 3 & 2 & 2 & 1 & 1 & 1 & 1 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 \\
B & 3 & 4 & 2 & 2 & 1 & 1 & 1 & 1 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 \\
C & 2 & 2 & 4 & 3 & 1 & 1 & 1 & 1 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 \\
D & 2 & 2 & 3 & 4 & 1 & 1 & 1 & 1 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 \\
E & 1 & 1 & 1 & 1 & 4 & 3 & 2 & 2 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 \\
F & 1 & 1 & 1 & 1 & 3 & 4 & 2 & 2 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 \\
G & 1 & 1 & 1 & 1 & 2 & 2 & 4 & 3 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 \\
H & 1 & 1 & 1 & 1 & 2 & 2 & 3 & 4 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 \\
I & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 4 & 3 & 2 & 2 & 1 & 1 & 1 & 1 \\
J & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 3 & 4 & 2 & 2 & 1 & 1 & 1 & 1 \\
K & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 2 & 2 & 4 & 3 & 1 & 1 & 1 & 1 \\
L & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 2 & 2 & 3 & 4 & 1 & 1 & 1 & 1 \\
M & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 1 & 1 & 1 & 1 & 4 & 3 & 2 & 2 \\
N & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 1 & 1 & 1 & 1 & 3 & 4 & 2 & 2 \\
O & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 1 & 1 & 1 & 1 & 2 & 2 & 4 & 3 \\
P & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 1 & 1 & 1 & 1 & 2 & 2 & 3 & 4
\end{array}.$$

Scale-1	Scale-2	Scale-3	Scale-4
0	Resistant/tolerant (R)	Susceptible (S)	R/S
2-3	Moderate R	Moderate S	-
5	Normal (no change)	Normal	Moderate
7-8	Moderate S	Moderate R	
9	S	R	S/R

Do we benefit from TO ?

- **Standard EQ Terms**

The standard PT term is a pre-coordinated EQ term; this can easily be defined as:

A <Q> *which* inheres_in an <E>

i.e. a quality carried by a bearer entity

- **Benefits**

– PATO definitions of qualities can be shared and reused

– The oboedit reasoner can keep TO in sync with PO/GO/ChEBI, Taxonomy, EO and PATO, and perform automatic DAG placement of terms

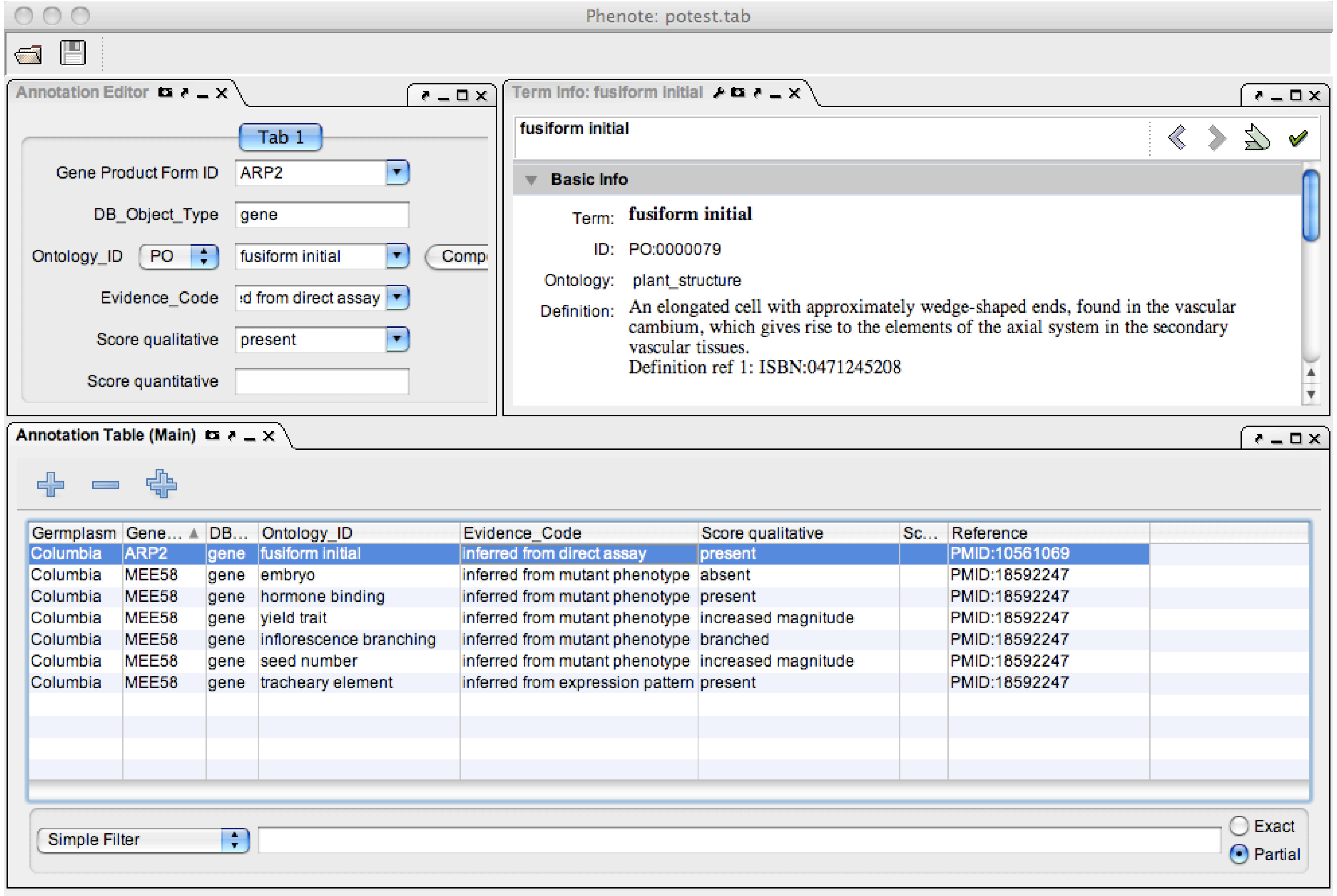
– PATO terms can be used to query TO-annotated phenotypes

– PO/GO/ChEBI, Taxonomy, EO terms can be used to query PT-annotated phenotypes

– Users/curators do not have to deal with post composition of terms with about 100K entity terms from different knowledge domains.

Phenote Annotation tool

With Two Configurations : Advanced and ‘Smart’



Gene Product Form ID: ARP2
DB_Object_Type: gene
Ontology_ID: PO fusiform initial
Evidence_Code: :d from direct assay
Score qualitative: present
Score quantitative:

fusiform initial

Basic Info

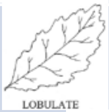
Term: **fusiform initial**
ID: PO:0000079
Ontology: plant_structure
Definition: An elongated cell with approximately wedge-shaped ends, found in the vascular cambium, which gives rise to the elements of the axial system in the secondary vascular tissues.
Definition ref 1: ISBN:0471245208

Germplasm	Gene... ▲	DB...	Ontology_ID	Evidence_Code	Score qualitative	Sc...	Reference
Columbia	ARP2	gene	fusiform initial	inferred from direct assay	present		PMID:10561069
Columbia	MEE58	gene	embryo	inferred from mutant phenotype	absent		PMID:18592247
Columbia	MEE58	gene	hormone binding	inferred from mutant phenotype	present		PMID:18592247
Columbia	MEE58	gene	yield trait	inferred from mutant phenotype	increased magnitude		PMID:18592247
Columbia	MEE58	gene	inflorescence branching	inferred from mutant phenotype	branched		PMID:18592247
Columbia	MEE58	gene	seed number	inferred from mutant phenotype	increased magnitude		PMID:18592247
Columbia	MEE58	gene	tracheary element	inferred from expression pattern	present		PMID:18592247

○ Exact
● Partial

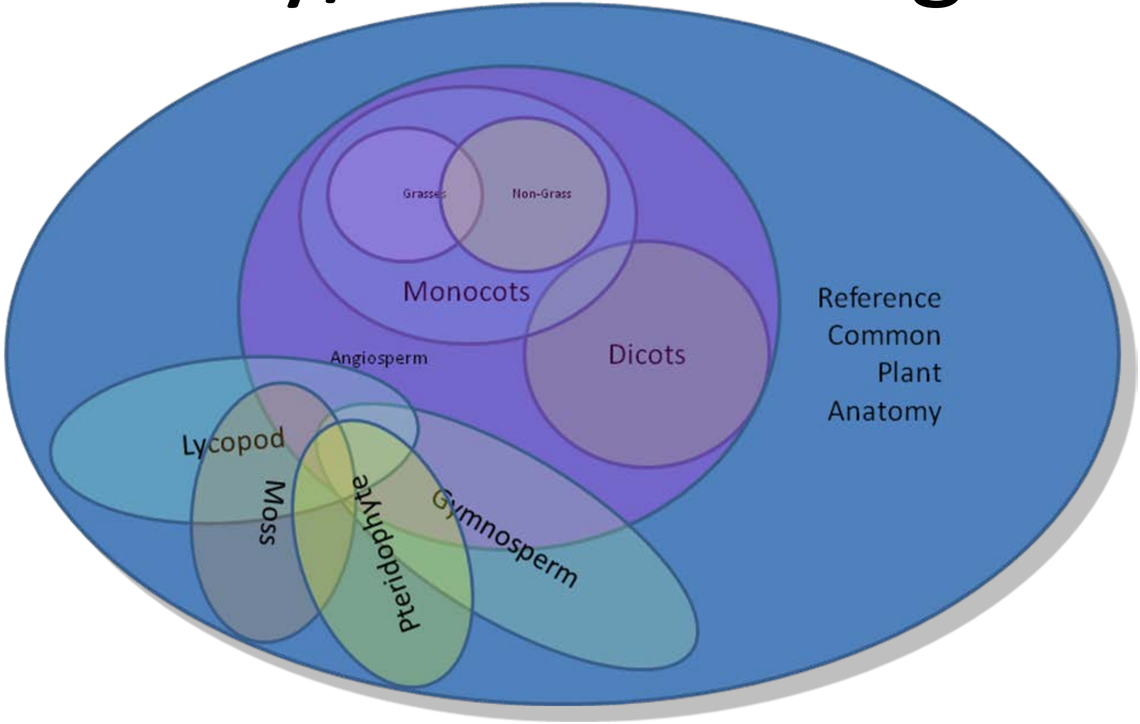
Custom Phenote tool (phenote.org) allows inputs by authors. Advanced configuration allows savvy users to build cross-products for phenotype annotations.

Creating Character Matrices

PO: Leaf					
TO: Leaf lamina shape (PO: Leaf lamina x PATO: shape)			**TO: Leaf lamina margin quality** (PO: Lamina margin x PATO: quality)		
Lyrate	☐		Lacerate	☐	
Linear	☐		Laciniate	☐	
Lanceolate	☐		Lobed	☐	
Hastate	☐		Lobulate	☒	
Deltoid	☒		Dentate	☐	

Sample: xxxxxxxxx
Species: xxxxxxxxx
Source: xxxxxxxxxx

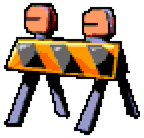
Do we need multiple Plant anatomy/trait ontologies?



Create various version and sub-versions of the PO that may cater to two aspects of the Ontology.

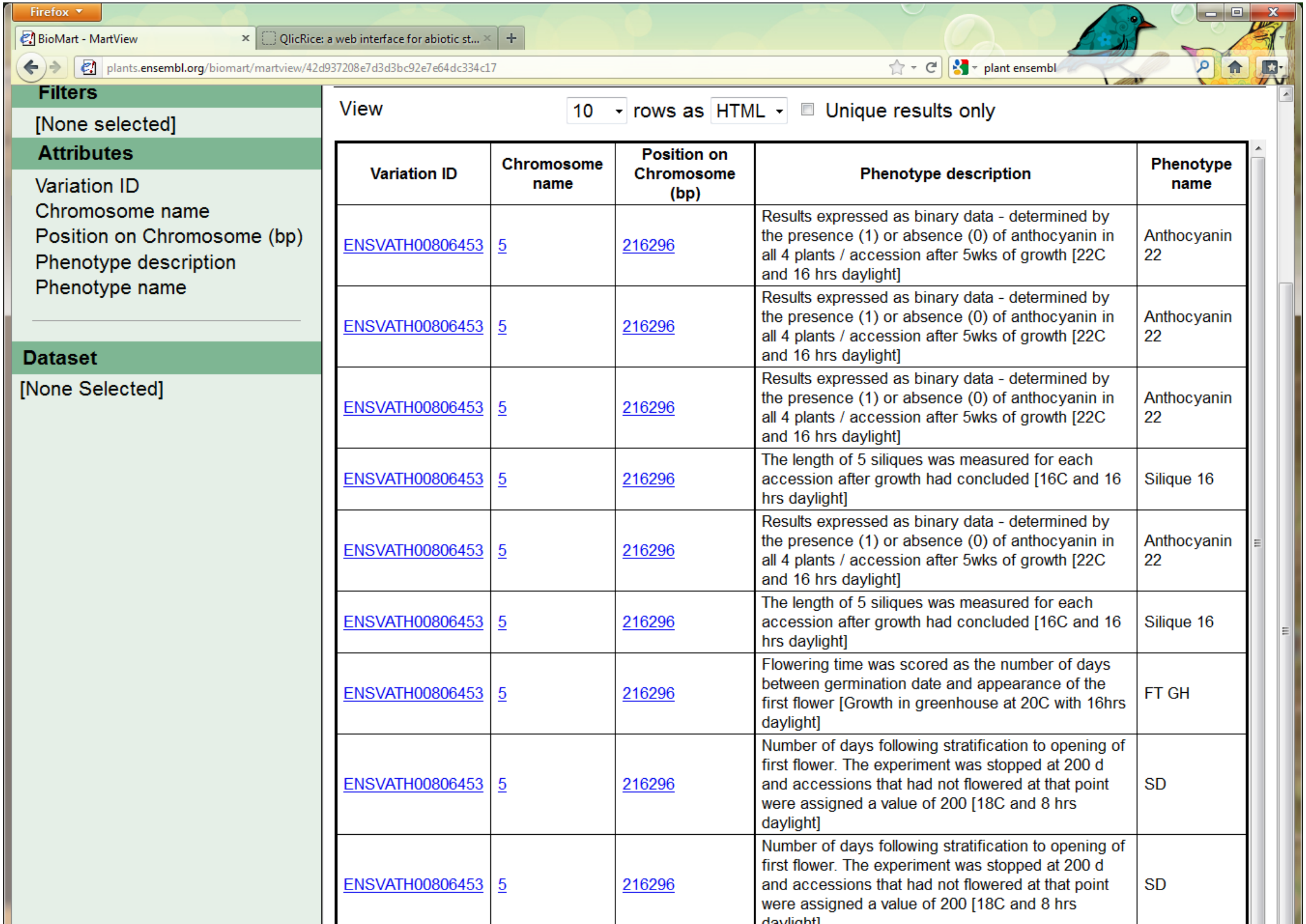
- Common Reference Ontology for Plants (CROP)
- Clade-specific Enriched Ontologies (CLEO)

Challenges



- Encourage species-specific vocabularies to use cross references to PO and TO terms
- Share annotations for robust comparison and hypothesis building
- Shortage of plant-specific phenotypic descriptors in PATO
- Add mapping files for clade oriented trait/phenotype glossaries
- Build consensus on metadata and minimal information guide

Capturing Phenotype Metadata



Filters

[None selected]

Attributes

Variation ID
Chromosome name
Position on Chromosome (bp)
Phenotype description
Phenotype name

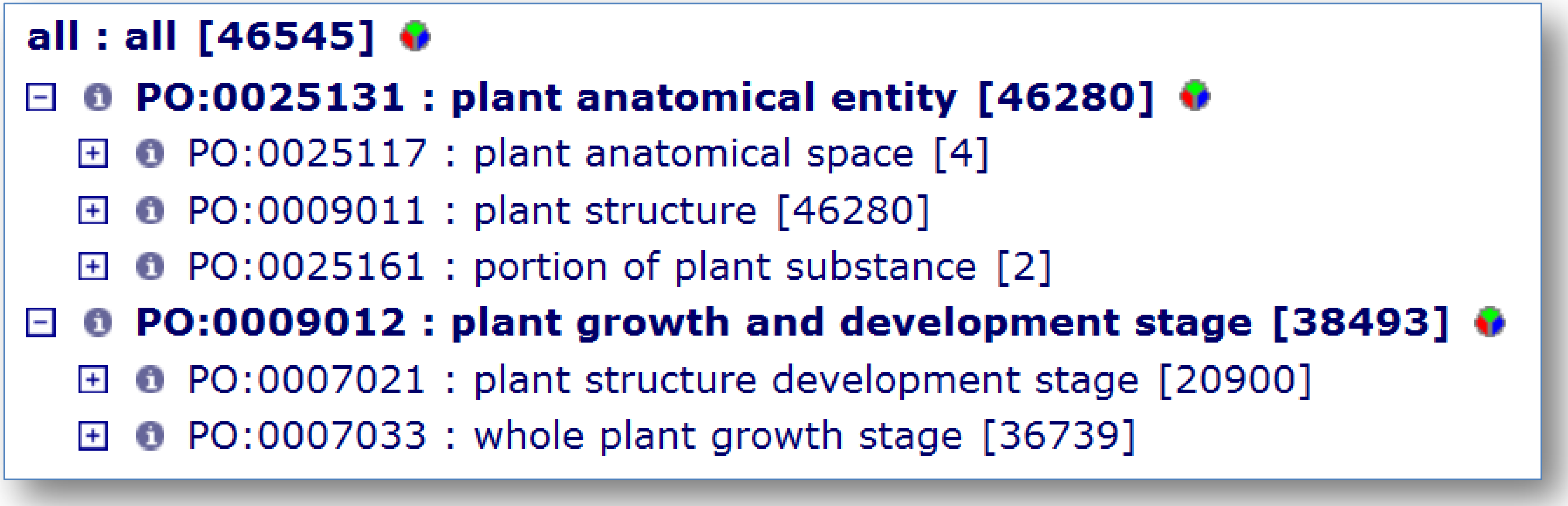
Dataset

[None Selected]

View 10 rows as HTML ☐ Unique results only

Variation ID	Chromosome name	Position on Chromosome (bp)	Phenotype description	Phenotype name
ENSVATH00806453	5	216296	Results expressed as binary data - determined by the presence (1) or absence (0) of anthocyanin in all 4 plants / accession after 5wks of growth [22C and 16 hrs daylight]	Anthocyanin 22
ENSVATH00806453	5	216296	Results expressed as binary data - determined by the presence (1) or absence (0) of anthocyanin in all 4 plants / accession after 5wks of growth [22C and 16 hrs daylight]	Anthocyanin 22
ENSVATH00806453	5	216296	Results expressed as binary data - determined by the presence (1) or absence (0) of anthocyanin in all 4 plants / accession after 5wks of growth [22C and 16 hrs daylight]	Anthocyanin 22
ENSVATH00806453	5	216296	The length of 5 siliques was measured for each accession after growth had concluded [16C and 16 hrs daylight]	Silique 16
ENSVATH00806453	5	216296	Results expressed as binary data - determined by the presence (1) or absence (0) of anthocyanin in all 4 plants / accession after 5wks of growth [22C and 16 hrs daylight]	Anthocyanin 22
ENSVATH00806453	5	216296	The length of 5 siliques was measured for each accession after growth had concluded [16C and 16 hrs daylight]	Silique 16
ENSVATH00806453	5	216296	Flowering time was scored as the number of days between germination date and appearance of the first flower [Growth in greenhouse at 20C with 16hrs daylight]	FT GH
ENSVATH00806453	5	216296	Number of days following stratification to opening of first flower. The experiment was stopped at 200 d and accessions that had not flowered at that point were assigned a value of 200 [18C and 8 hrs daylight]	SD
ENSVATH00806453	5	216296	Number of days following stratification to opening of first flower. The experiment was stopped at 200 d and accessions that had not flowered at that point were assigned a value of 200 [18C and 8 hrs daylight]	SD

Plant Ontology



all : all [46545]
- ⊟ **PO:0025131 : plant anatomical entity [46280]**
  - ⊞ PO:0025117 : plant anatomical space [4]
  - ⊞ PO:0009011 : plant structure [46280]
  - ⊞ PO:0025161 : portion of plant substance [2]
- ⊟ **PO:0009012 : plant growth and development stage [38493]**
  - ⊞ PO:0007021 : plant structure development stage [20900]
  - ⊞ PO:0007033 : whole plant growth stage [36739]

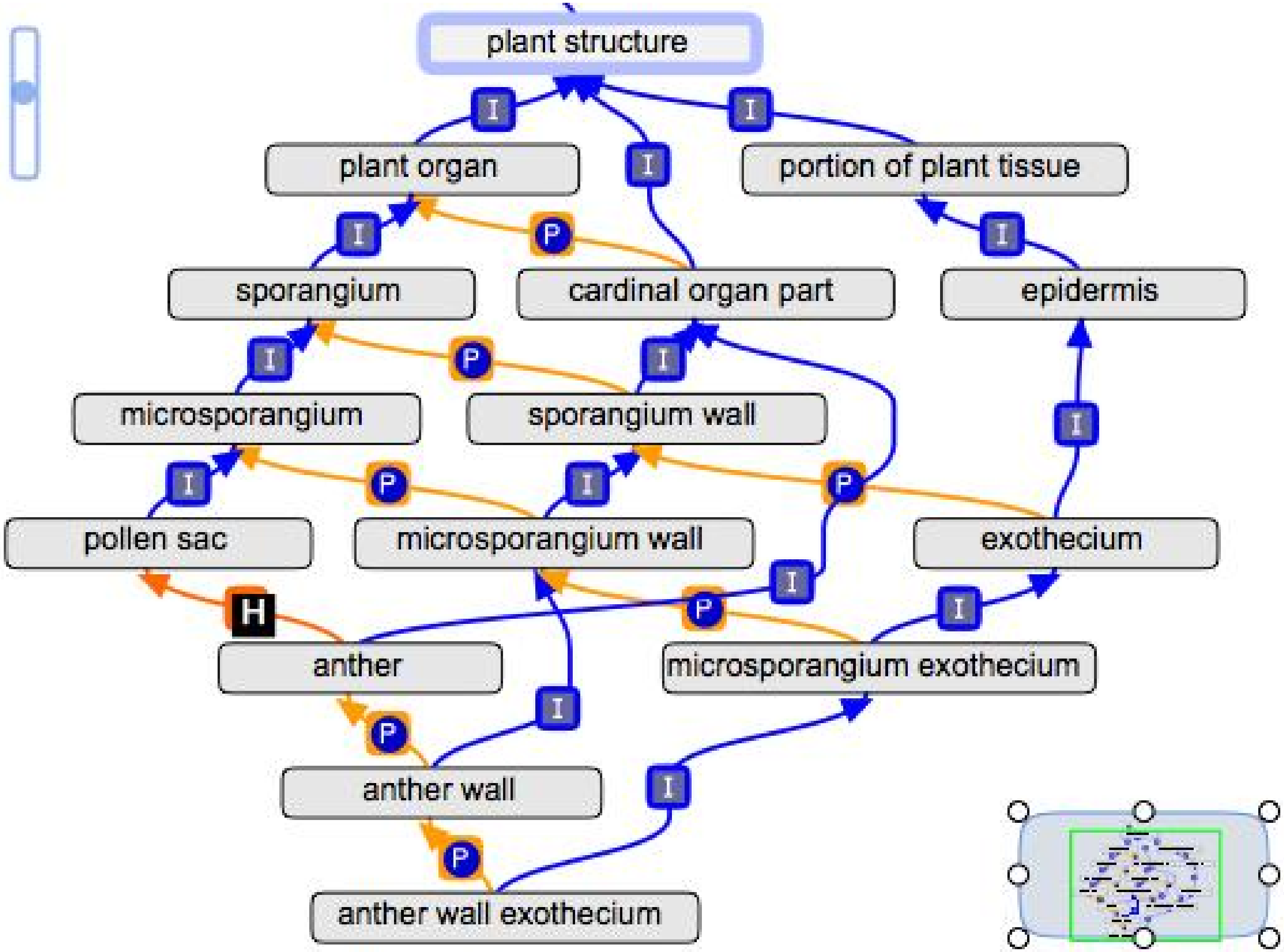
- Has two classes for **Plant Anatomical entities** and **Growth and Development**
- Every term has definition
- Every term has is_a relationship as mandated to have single inheritance (some terms may have multiple for enriched biological reasoning)
- Use multiple relationship types
  - is_a, part_of, has_part, adjacent_to, develops_from, derived_from, participates_in

Vision for Plant Ontology:

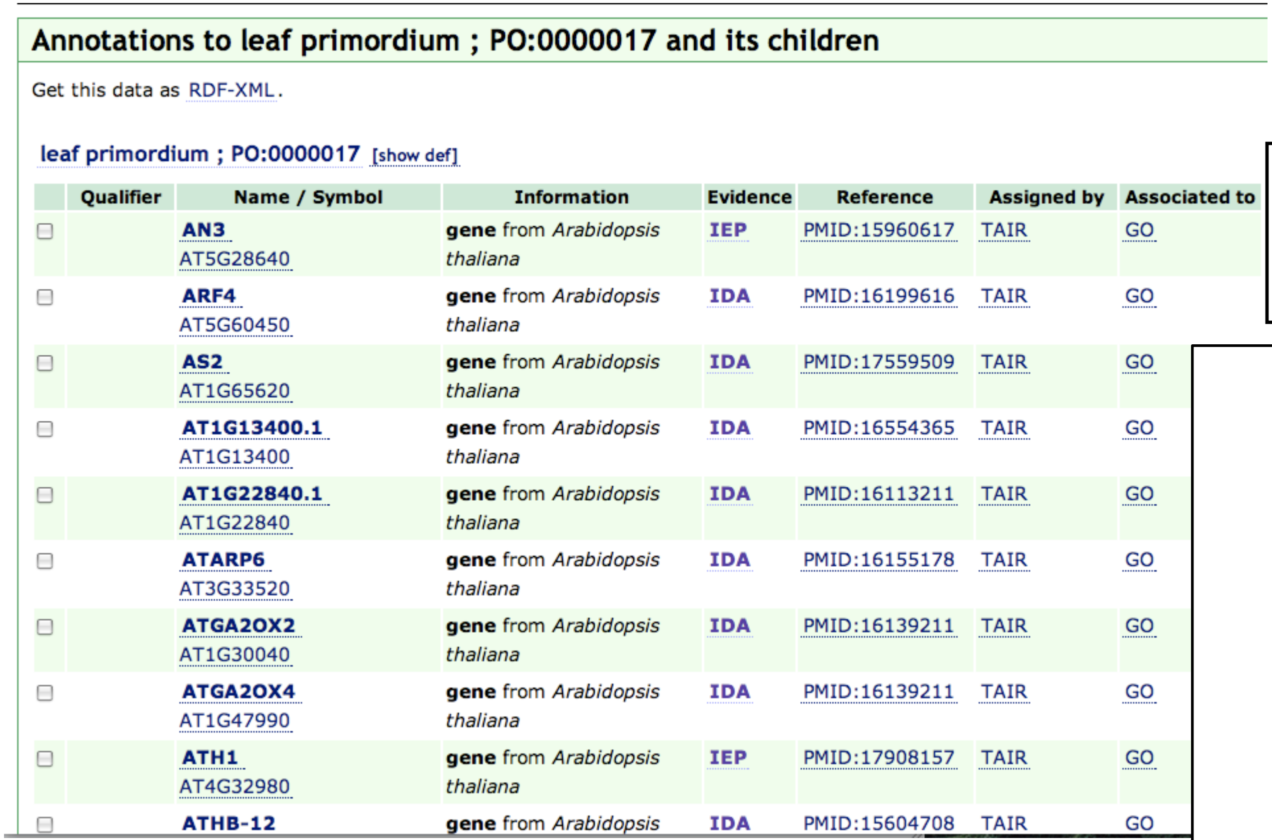
- Encompass **all** plants
- Facilitate consistency in:
    - Annotation of comparative genomics data
    - Cross-database queries
- Develop PO as a reference ontology for plants:
    - Provide mappings to other ontologies in use by plant databases
- Create cross-products to other ontologies such as PATO, to describe phenotypic characters

Source: Laurel Cooper

An example from PO Anatomical Entity



The Plant Ontology facilitates comparative plant genomics, developmental biology and systematics



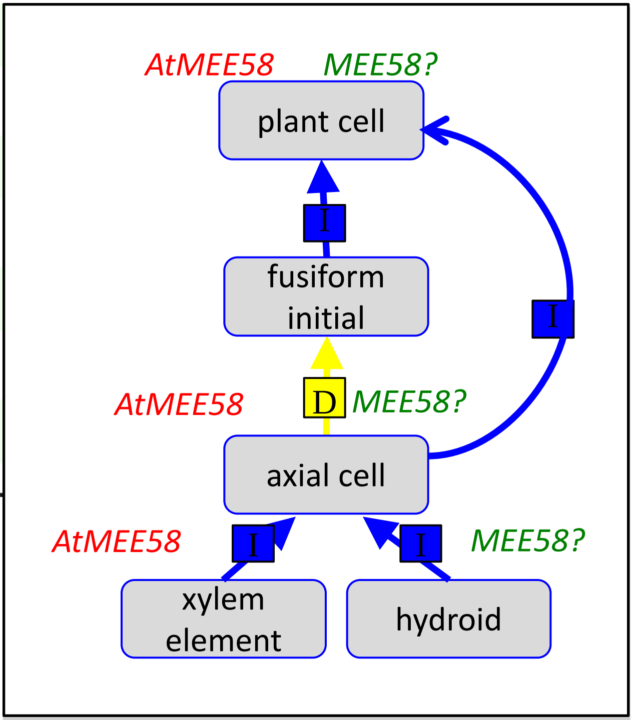
Annotations to leaf primordium ; PO:0000017 and its children

Get this data as RDF-XML.

leaf primordium ; PO:0000017 [show def]

	Qualifier	Name / Symbol	Information	Evidence	Reference	Assigned by	Associated to
☐		**AN3** AT5G28640	**gene** from *Arabidopsis thaliana*	IEP	PMID:15960617	TAIR	GO
☐		**ARF4** AT5G60450	**gene** from *Arabidopsis thaliana*	IDA	PMID:16199616	TAIR	GO
☐		**AS2** AT1G65620	**gene** from *Arabidopsis thaliana*	IDA	PMID:17559509	TAIR	GO
☐		**AT1G13400.1** AT1G13400	**gene** from *Arabidopsis thaliana*	IDA	PMID:16554365	TAIR	GO
☐		**AT1G22840.1** AT1G22840	**gene** from *Arabidopsis thaliana*	IDA	PMID:16113211	TAIR	GO
☐		**ATARP6** AT3G33520	**gene** from *Arabidopsis thaliana*	IDA	PMID:16155178	TAIR	GO
☐		**ATGA2OX2** AT1G30040	**gene** from *Arabidopsis thaliana*	IDA	PMID:16139211	TAIR	GO
☐		**ATGA2OX4** AT1G47990	**gene** from *Arabidopsis thaliana*	IDA	PMID:16139211	TAIR	GO
☐		**ATH1** AT4G32980	**gene** from *Arabidopsis thaliana*	IEP	PMID:17908157	TAIR	GO
☐		**ATHB-12**	**gene** from *Arabidopsis*	IDA	PMID:15604708	TAIR	GO

Cross-taxa comparisons:



>500,000 associations for >1300 terms

Source: Laurel Cooper

Plant Ontology
Taxon Relevance, Synonyms and Xrefs

subsetdef: Angiosperm "Term for angiosperms"

subsetdef: Arabidopsis "Term used for Arabidopsis"

subsetdef: Bryophytes "Term used for mosses, liverworts, and/or hornworts"

subsetdef: Citrus "Term used for citrus"

subsetdef: Gymnosperms "Term used for gymnosperms"

subsetdef: Maize "Term used for maize"

subsetdef: Musa "Terms used for banana"

subsetdef: Poaceae "Term used for grasses"

subsetdef: Potato "Term used for potato“

subsetdef: Solanaceae "Term used for solanaceous plants“

subsetdef: Pteridophytes "Term used for ferns and allies"

subsetdef: reference "reference plant structure term"

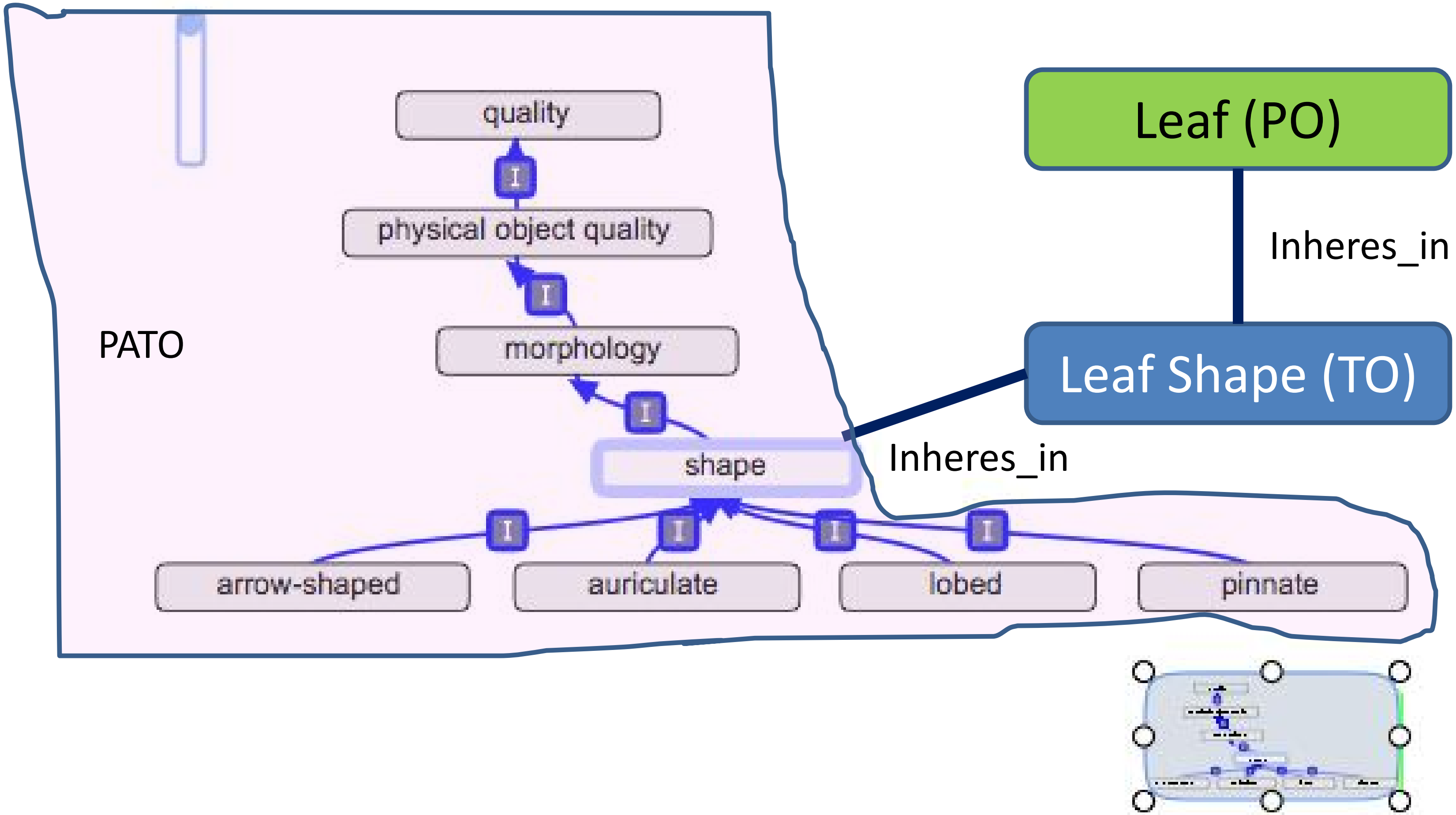
subsetdef: Rice "Term used for rice"

subsetdef: Tomato "Term used for tomato"

subsetdef: TraitNet "Plant Functional Traits“

```
synonymtypedef: Japanese "Japanese synonym" EXACT
synonymtypedef: Plural "Plural" EXACT
synonymtypedef: Spanish "Spanish synonym" EXACT
treat-xrefs-as-is_a: CARO
dbxrf: CL:Cell_type | GO:cellular_component | APweb
```

Examples of shape qualities that are relevant to leaves



PATO

Leaf (PO)

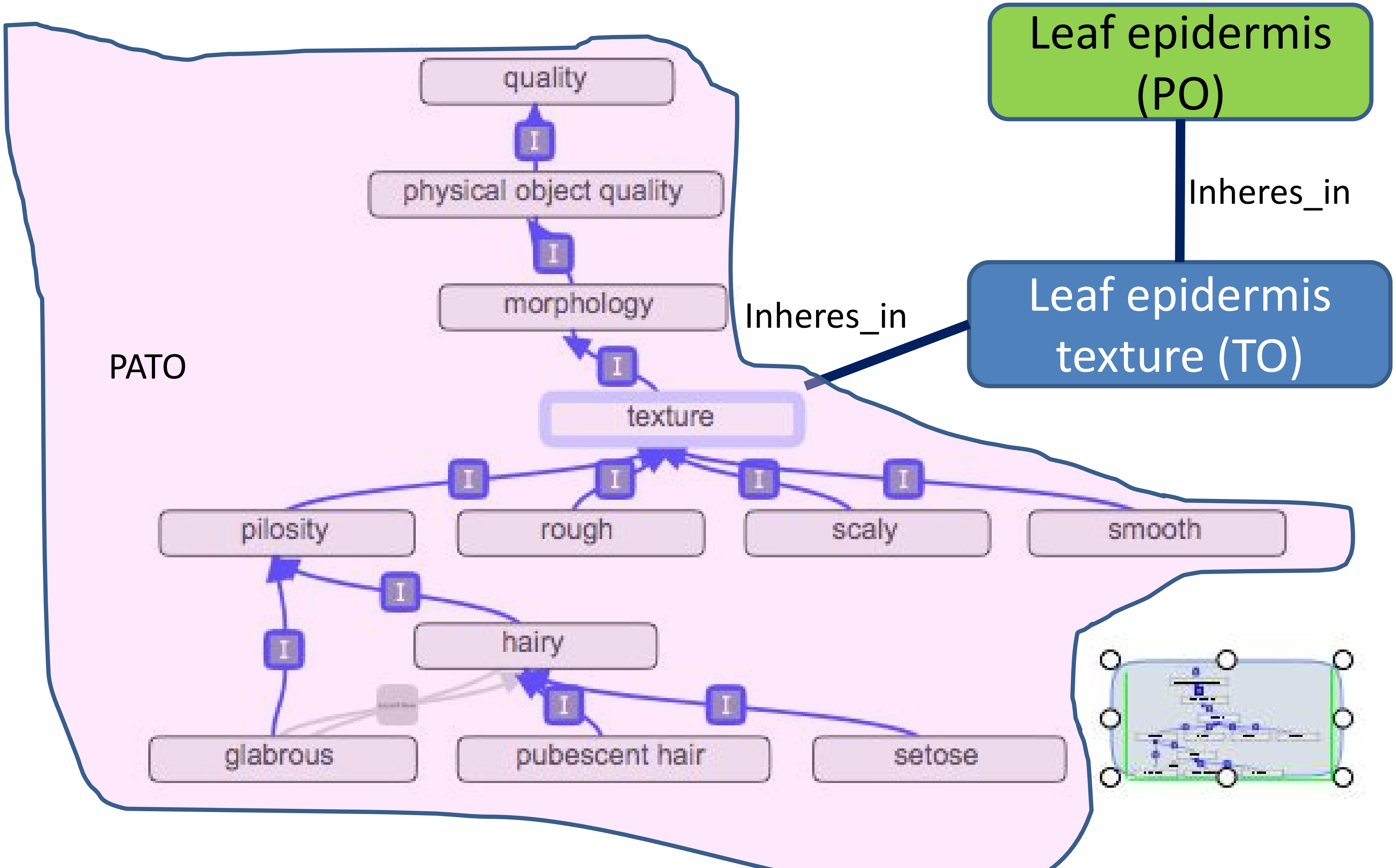
Inheres_in

Leaf Shape (TO)

Inheres_in

Source: Ramona Walls

Examples of textures that are relevant to leaves



PATO

Leaf epidermis (PO)

Inheres_in

Inheres_in

Leaf epidermis texture (TO)

Source: Ramona Walls