Phenoscape: use cases and anatomy ontology requirements for linking evolutionary and model organism phenotypes

Wasila Dahdul, James Balhoff, Hilmar Lapp, Peter Midford, Todd Vision, Monte Westerfield, Paula Mabee

ICBO 2011 Anatomy ontology workshop
Phenoscape Knowledgebase

- Brings together data from two domains
- Search for candidate genes and candidate taxa

http://kb.phenoscape.org
Candidate genes

Did Mola lose its caudal fin because of changes in regulation of yap1?

genes: smc3, tll1, yap1

Caudal fin, absent

Jiang et al., 2009
Phenoscape

- Collaboration:
  - Mabee (lead PI, USD)
  - Westerfield (ZFIN, UO)
  - Vision and Lapp (NESCent, UNC)
  - Sereno (U Chicago)
  - Blackburn (CAS)
  - Blake (MGI)
  - Zorn (Xenbase, CCRF)

- Expanding to vertebrates

- Expanding reasoning (homology, phenoblast)

- Fin/limb use case

- Synchronization requirements
Vertebrate ontologies

Different strategies for referencing external terms
Synchronization by cross referencing terms

Synchronization Tool
• Plugin for Obo-Edit
• Further development planned
Synchronization by importing terms

- Strategy used by multispecies ontologies
- Better support needed in Obo-Edit
Improved tools for synchronization needed

• Synchronization Tool
• Imports/MIREOT in Obo-edit
  • roundtripping file with import statement
  • color code terms from different ontologies
  • saving to separate ontologies
• Reasoning to assist in synchronization
• Communication of ontology changes
  • GO diff tool
• Community development of shared tools
  • PhenotypeRCN travel support  (http://phenotypercn.org)
Acknowledgements

• Phenoscape team

• Chris Mungall

• Many contributors to data, character curation, ontology development and ideas (see: http://kb.phenoscape.org/contributors/)

• National Science Foundation (DBI 0641025, DBI 1062542, DBI 1062404)

• National Evolutionary Synthesis Center (NSF EF-0423641)