

# Enabling a Cross-Species Disease Research Ecosystem

*Melissa Haendel*  
*Oregon Health & Science University*

NIH Symposium: Linking Disease Model  
Phenotypes to Human Conditions

2015-09-10



@monarchinit



@ontowonka

# Connecting all the things that relate to disease research

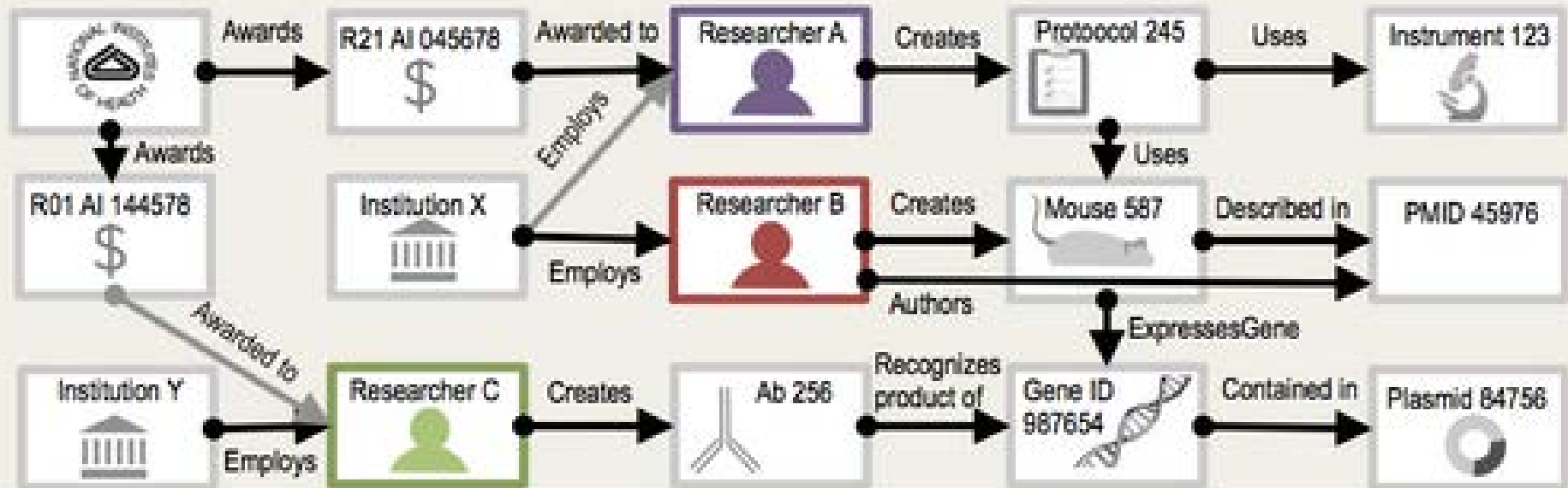


Image by Julie McMurry 2012

=> we need to uniquely reference these entities and their connections

# Uniquely Identifying people

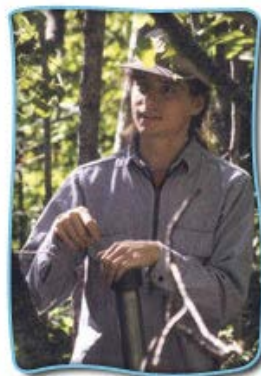


"campbell, jl" and ecology



Scholar

About 2,070 results (0.09 sec)



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Create alert

## [Post-wildfire logging hinders regeneration and increases fire risk](#)

[DC Donato](#), [JB Fontaine](#), [JL Campbell](#), [WD Robinson](#)... - *Science*, 2006 - sciencemag.org

... However, our data indicate that delay was responsible for ~10% of the woody fuel present after logging. ← JK Agee, *Fire Ecology of Pacific Northwest Forests* (Island Press, Washington, DC, 1993). ... More in Collections. **Ecology**. Related Jobs from ScienceCareers. ...

Cited by 149 Related articles All 66 versions Cite

[PDF] from [humboldt.edu](#)

## [Winter in northeastern North America: a critical period for ecological processes](#)

[JL Campbell](#), [MJ Mitchell](#), [PM Groffman](#)... - *Frontiers in Ecology* ..., 2005 - Eco Soc America

Ecological research during winter has historically been a low priority in northeastern North America, an oversight that stems from the commonly accepted notion that there is little biological activity when temperatures drop below freezing. However, recent research has ...

Cited by 95 Related articles All 10 versions Cite

[PDF] from [esf.edu](#)

## [Can fuel-reduction treatments really increase forest carbon storage in the western US by reducing future fire emissions?](#)

[JL Campbell](#), [ME Harmon](#)... - *Frontiers in Ecology and* ..., 2011 - Eco Soc America

It has been suggested that thinning trees and other fuel-reduction practices aimed at reducing the probability of high-severity forest fire are consistent with efforts to keep carbon (C) sequestered in terrestrial pools, and that such practices should therefore be rewarded ...

Cited by 17 Related articles All 5 versions Cite

[PDF]

## [Mixed-severity fire regimes: lessons and hypotheses from the Klamath-Siskiyou Ecoregion](#)

[JE Halofsky](#), [DC Donato](#), [DE Hibbs](#), [JL Campbell](#)... - *Ecosphere*, 2011 - Eco Soc America

... Fire regimes—the characteristic pattern and effects of wildland fire in a given area—are major drivers of the **ecology** of many vegetation types and are widely applied as a guiding framework for management of fire-prone ecosystems globally (Bond and van Wilgen 1996 ...

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[PDF] from

## [Intake, digestibility, and passage of a commercially designed diet by two \*Propithecus\* species](#)

[JL Campbell](#), [JH Eisemann](#), [KE Glander](#)... - *American journal of* ..., 1999 - Wiley Online Library

... **Campbell**, **JL**, Eisemann, JH, Glander, KE and Crissey, SD (1999), Intake, digestibility, and passage of a commercially designed diet by two propithecus ... Though both animals have a similar feeding **ecology**, the captive status of *P. verreauxi* is considered more stable than that of ...

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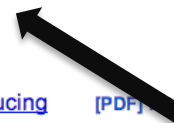
[PDF] from [fs.fed.us](#)

## [Dissolved organic nitrogen budgets for upland, forested ecosystems in New England](#)

[JL Campbell](#), [JW Hornbeck](#), [WH McDowell](#), [DC Buso](#)... - *Biogeochemistry*, 2000 - Springer

Page 1. *Biogeochemistry* 49: 123–142, 2000. © 2000 Kluwer Academic Publishers. Printed in the Netherlands. Dissolved organic nitrogen budgets for upland, forested ecosystems in New England JOHN L. CAMPBELL1, JAMES ...

Cited by 155 Related articles All 17 versions Cite



So... we can identify people, but how identifiable are models in the published literature?

# An experiment in reproducibility

84 Journals

248 papers

Domains:  
Immunology  
Cell biology  
Neuroscience  
Developmental  
biology  
General biology

Impact factors:  
High  
Medium  
Low

Reporting Guidelines:  
Stringent  
Satisfactory  
Loose

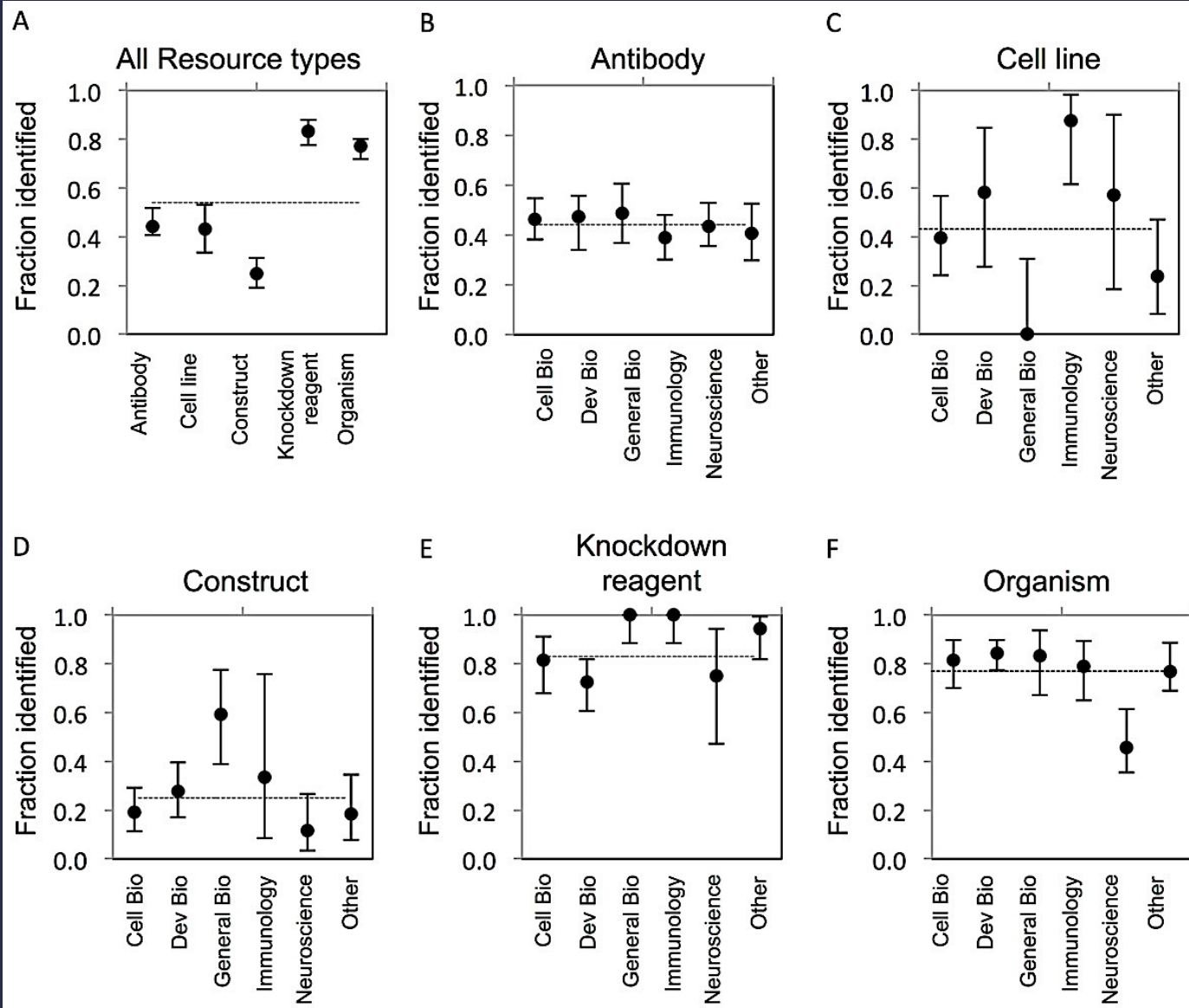
707 antibodies

104 cell lines

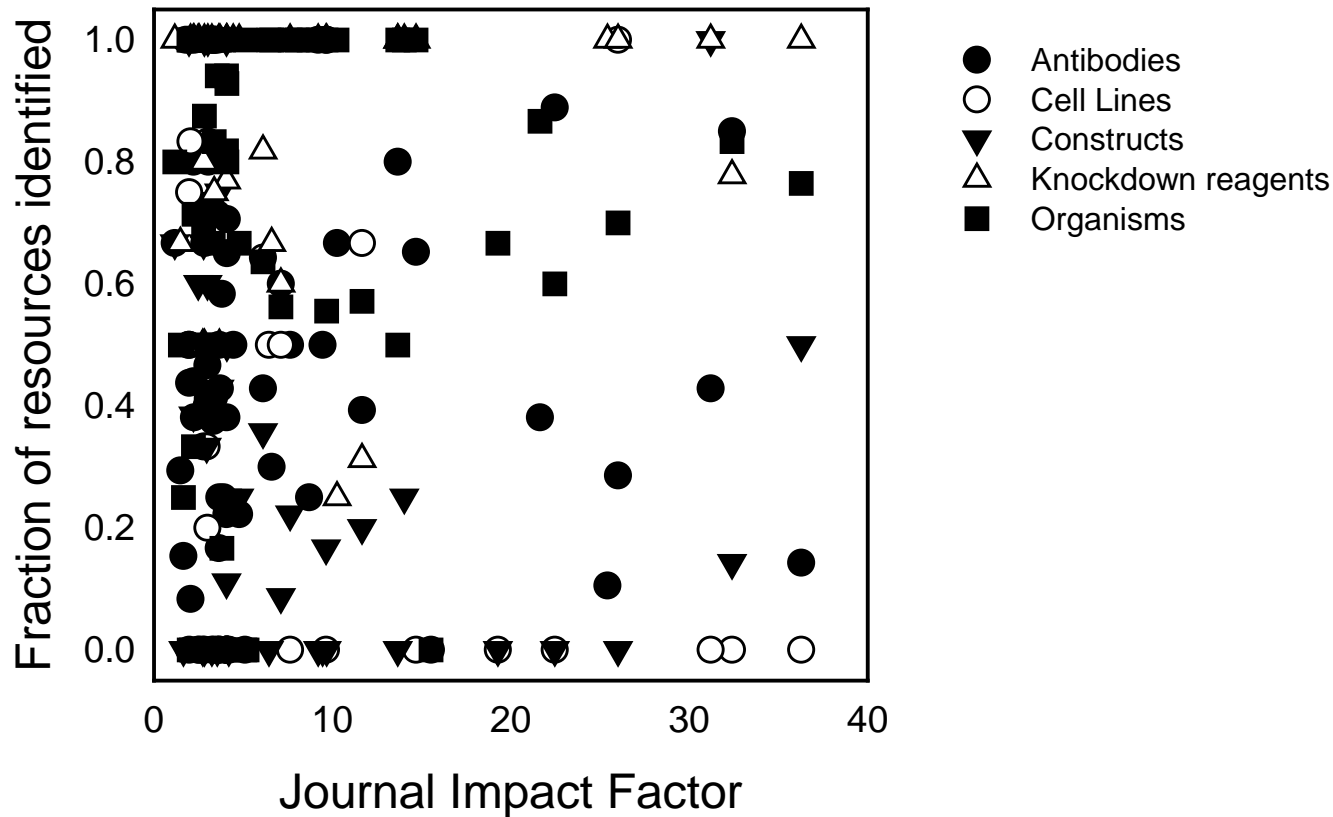
258 constructs

437 model  
organisms

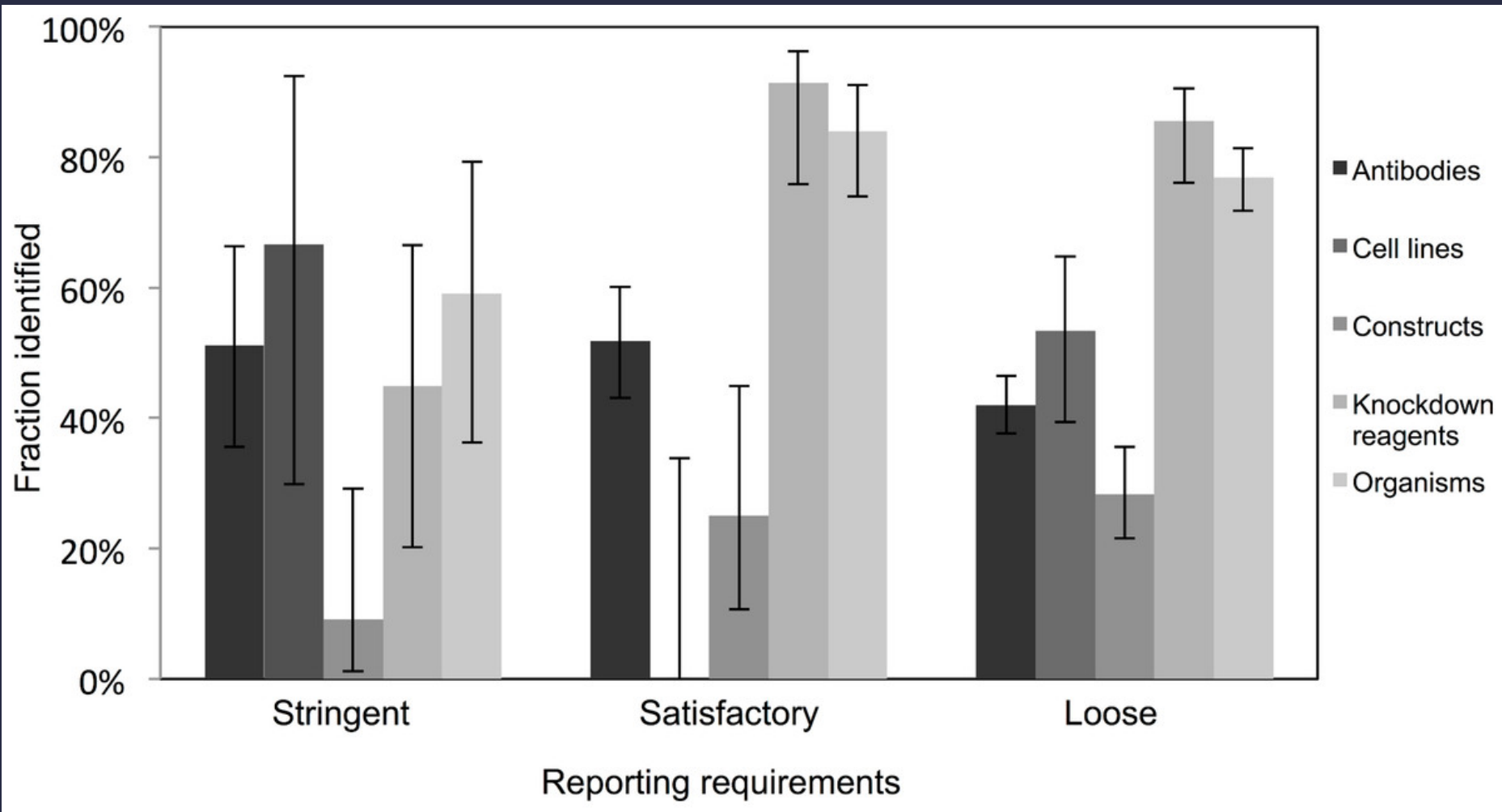
210 knockdown  
reagents



Only ~50% of resources were identifiable



**No correlation between impact factor and resource identification**



**Resources are not more identifiable in journals with stricter reporting requirements**



# Darn, that is a lot of lost mice

44,557 alleles in MGI

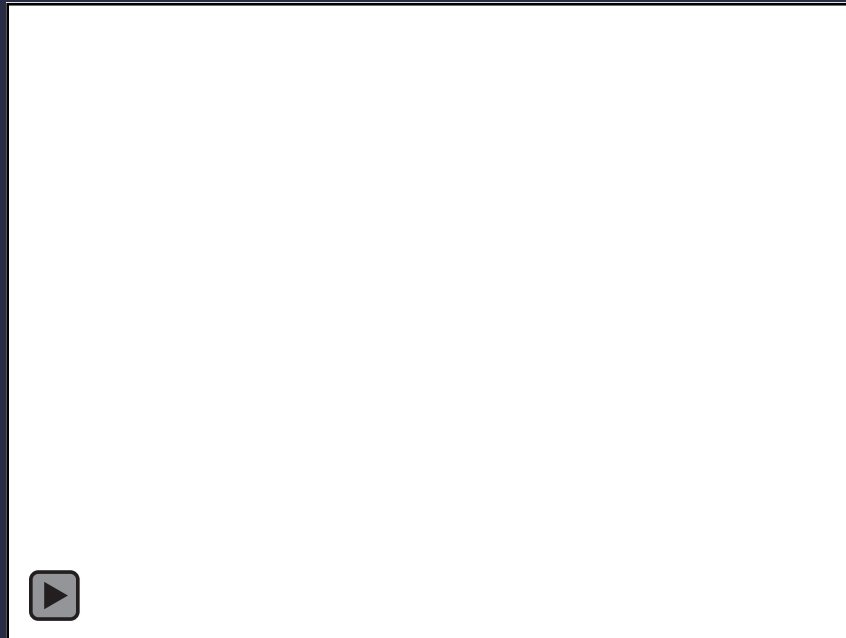
@21,946 missing alleles

11,154 Genes with mutant alleles

@5,494 missing genes?

55,388 Genotypes with phenotypes

@27,281 missing genotypes?





# Resource Identification Initiative



## FORCE11

The Future of Research Communications and e-Scholarship

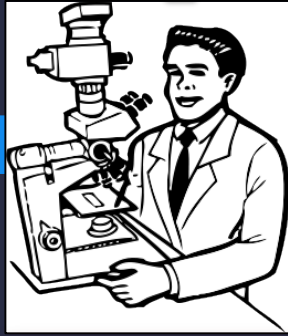


Numerous endorsers <https://www.force11.org/RII/SignUp>

Implementation of the new standard for authors

<http://biosharing.org/bsg-000532>

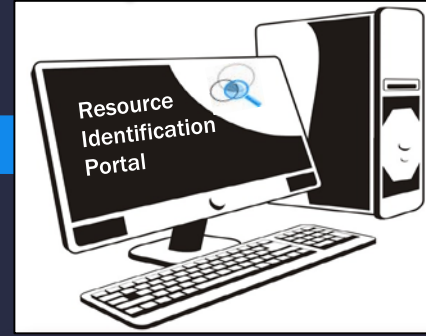
[http://www.force11.org/Resource\\_Identification\\_Initiative](http://www.force11.org/Resource_Identification_Initiative)



1. Researcher submits a manuscript for publication



2. Editor or Publisher asks for inclusion of RRID



3. Author goes to Resource Identification Portal to locate RRID



4. RRID is included in Methods section and as Keyword

**Sample citation:**

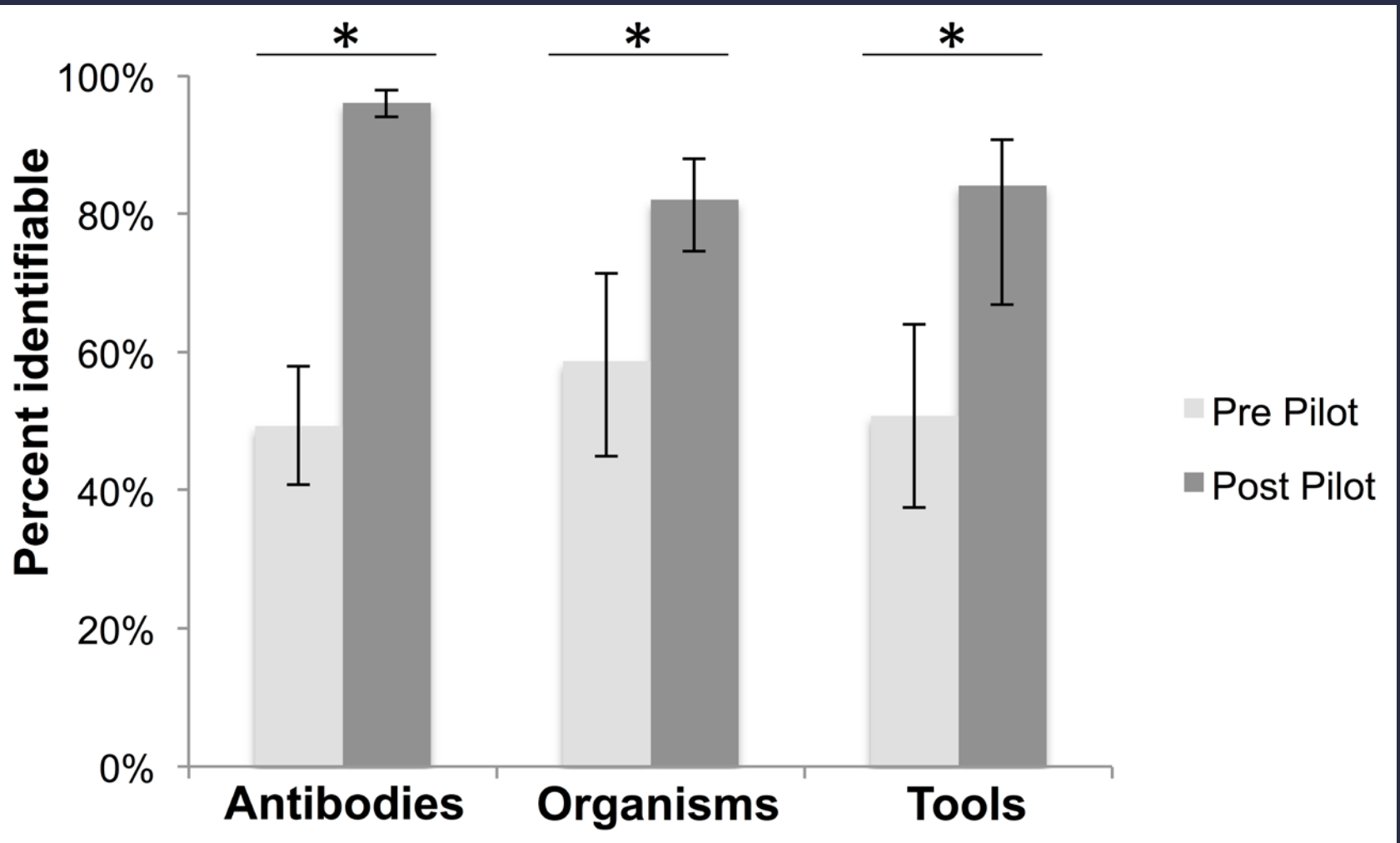
Source Database: MGI

Genetic Background: involves: 129P2/OlaHsd \* C3H/He

Affected Genes: Pax2; Variant Alleles: tm1Mbu

RRID:MGI:3586676

# Post-pilot identification



How can we enable genotype-phenotype data capture AND interoperability with the literature?

# Any-species G2P curation tool

**BRCA demo**

Store into a file

**Edit collaboratively, group sharing** ★★★★★

**Uniquely identify a model or disease, Check organism/genotype nomenclature**

**Choose terms from any selected phenotype ontology**

**Check annotation sufficiency**

**Provide evidence**

**Add another evidence**

**Add**

Disease	Phenotype	Age of Onset	Evidence	Reference	Description	Actions
DOID:3773 (third ventricle chordoid glioma)	FBbt:00000499PHENOTYPE (Drosophila dorsal mouthpart muscle 2 phenotype)	HP:0003596 (Middle age onset)	["ECO:0000006 (experimental evidence)","ECO:0000152 (in vitro recombinant protein transcription reconstitution assay evidence)"]	ref1	test description 2	Edit Delete
DOID:3773 (third ventricle chordoid glioma)	FBbt:00110314PHENOTYPE (Drosophila histaminergic HP4 neuron phenotype)	FBbt:00000499PHENOTYPE (Drosophila dorsal mouthpart muscle 2 phenotype)	["ECO:0000006 (experimental evidence)"]	ref1	test description	Edit Delete

**Familiar spreadsheet and export format**



<http://create.monarchinitiative.org/>

Based on standalone Phenote tool:  
[Phenote.org](http://Phenote.org)

# Case Reports and recording phenotypes at publication time



COLD SPRING HARBOR  
Molecular Case Studies



*A journal of precision medicine*

HOME | AUTHOR INFO | SUBJECT COVERAGE | SUBMIT A MANUSCRIPT

[Submit a manuscript](#)

[Author information](#)

CSH *Molecular Case Studies* is an open-access, peer-reviewed, international journal in the field of precision medicine. Articles in the journal present genomic and molecular

The journal publishes:

- **Research Reports** presenting detailed case studies of individuals and small cohorts
- **Research Articles** describing more extensive work using larger cohorts and/or functional analyses
- **Follow-up Reports** linked to previous observations

EDITOR-IN-CHIEF: Elaine Mardis

DEPUTY EDITOR: Ralph DeBerardinis

EDITORS: Stylianos Antonarakis  
Steven Jones  
Stephen Kingsmore  
Heidi Rehm  
Lillian Siu

EDITORIAL BOARD:

=> New phenotype data sharing recommendations to assist in quality genotype-2-phenotype data capture

# G2P integration with the literature



## PubMed Browser

Human Phenotype Ontology

Q Search

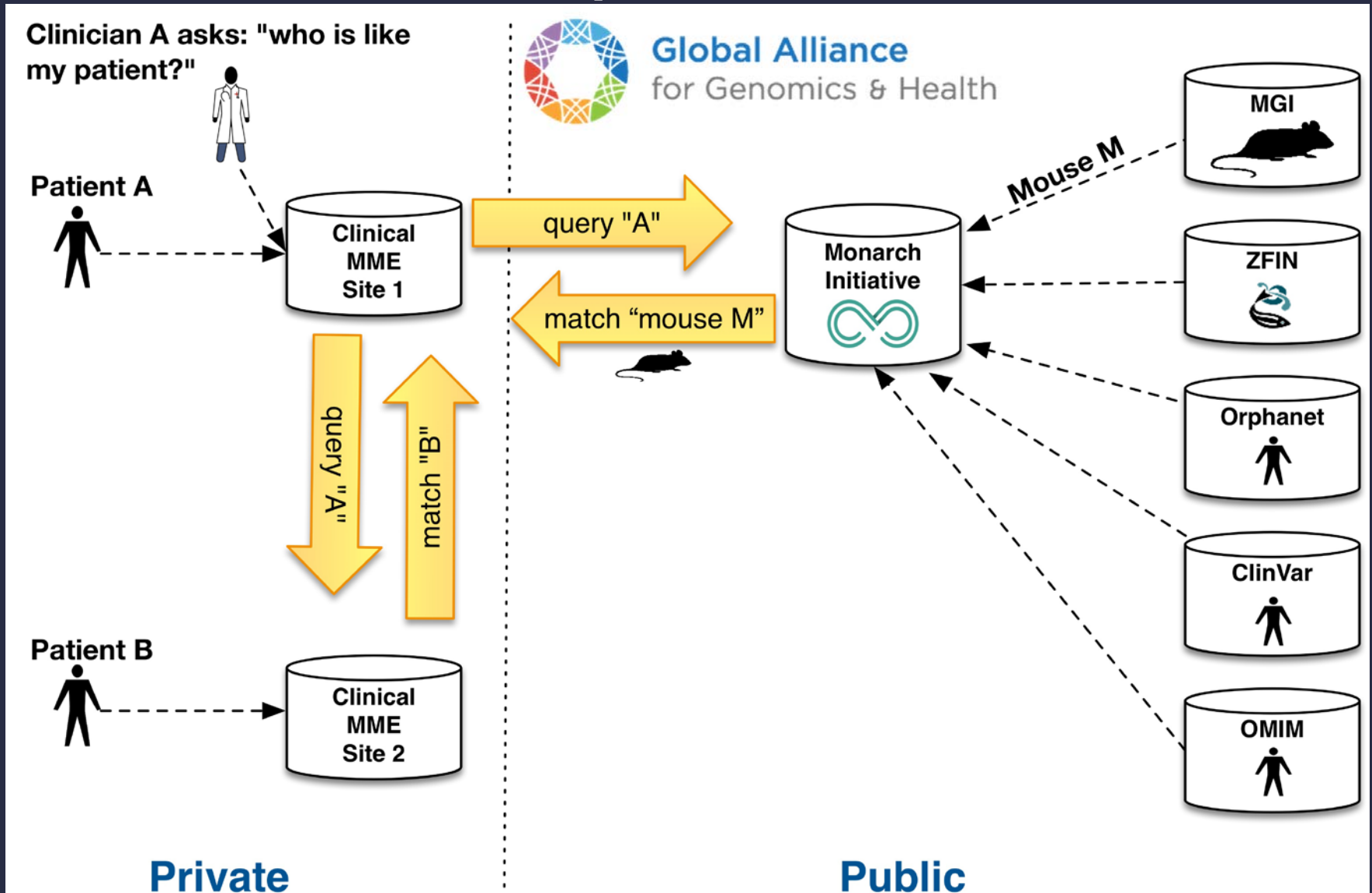
Not sure what to do? Take the tour

<http://pubmed-browser.human-phenotype-ontology.org/>

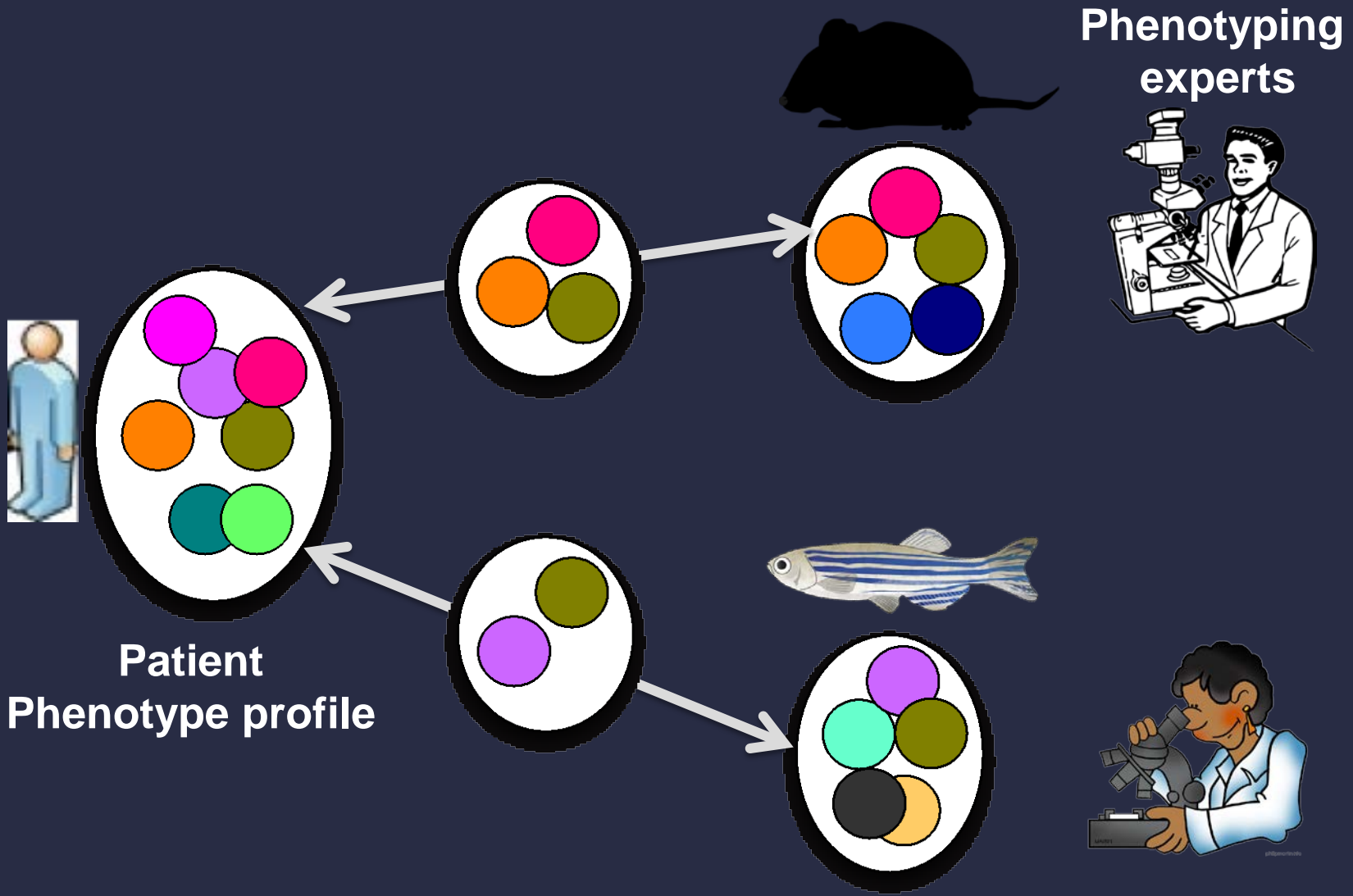
Currently searches the literature for combinations of human phenotype terms, using synonyms and HPO graph  
=> Need to link identified model organisms and model phenotypes



# Using and sharing G2P data across species



# Finding collaborators for functional validation



# Acknowledgments

Lots and lots of people, but mostly:

**Nicole Vasilevsky – Resource Identification**

**Maryann Martone – Force11/RRID pilot**

**Chris Mungall and Nicole Washington – G2P curation tool**

**Tudor Groza – PubMed Browser**

**Kent Shefchek – GA4GH**

**Peter Robinson – HPO, among other things!**

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NIH Office of Director: 1R24OD011883

HHSN268201300036C, HHSN268201400093P