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## Education

2011 **Ph.D.** Bioinformatics, University of Edinburgh

1993 **BSc.** Artificial Intelligence and Computer Science, University of Edinburgh

## **Employment**

2019-Present Staff Scientist, Molecular Ecosystems Biology Dept., Environmental Genomics and

Systems Biology Division, LBNL

2018-Present Dept. Head, Molecular Ecosystems Biology Dept., Environmental Genomics and Systems

Biology Division, LBNL

2015-2018 **Research Scientist**, Environmental Genomics and Systems Biology Division, LBNL

2006-2015 **Research Scientist**, Genomics Division, LBNL

2001-2006 **Bioinformatics Specialist**, Howard Hughes Medical Institute, UC Berkeley

1999-2001 **Bioinformatics Scientist**, Life Sciences Division, Lawrence Berkeley National

Laboratory

1994-1999 **Bioinformatician**, Roslin Institute, Edinburgh, UK

#### **Publications**

Google Scholar: goo.gl/x2R5PC h-index: 64 MyNCBI: goo.gl/OFQ9k6 i10-index: 120

#### **Refereed Journal Articles**

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- 2001 Hu, J., **Mungall, C. J.**, Law, A., Papworth, R., Nelson, J. P., Brown, A., Simpson, I., Leckie, S., Burt, D. W., Hillyard, A. L., and Archibald, A. L. (2001). The ARKdb: genome databases for farmed and other animals. Nucleic Acids Res, 29(1):106–110
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- 1998 Hu, J., **Mungall, C. J.**, Nicholson, D., and Archibald, A. L. (1998). Design and implementation of a CORBA-based genome mapping system prototype. Bioinformatics, 14(2):112–120

### **Book Chapters**

2007 Haendel, M. A., Neuhaus, F., Osumi-Sutherland, D., Mabee, P. M., Mejino, J. L. J., Mungall, C. J., and Smith, B. (2007). CARO - The Common Anatomy Reference Ontology. In Anatomy Ontologies for Bioinformatics, Principles and Practice, volume Albert Burger, Duncan Davidson and Richard Baldock (Eds.). Springer

### **Published Conference Proceedings**

- 2016 Manda, P., Mungall, C. J., Balhoff, J., Lapp, H., and Vision, T. (2016). Investigating the importance of anatomical homology for cross-species phenotype comparisons using semantic similarity. In Pacific Symposium on Biocomputing 21, pages 132–143. World Scientific Publishing Company
- 2014 Mungall, C. J., Dietze, H., and Osumi-Sutherland, D. (2014). Use of OWL within the Gene Ontology. In Keet, M. and Tamma, V., editors, Proceedings of the 11th International Workshop on OWL: Experiences and Directions (OWLED 2014), pages 25–36, Riva del Garda, Italy, October 17-18, 2014
- 2013 Brush, M. H., **Mungall, C.J.**, Washington, N., and Haendel, M. A. (2013). What's in a Genotype? An Ontological Characterization for Integration of Genetic Variation Data. In Dumontier, M.,

- Hoehndorf, R., and Baker, C. J. O., editors, Proceedings of the International Conference on Biomedical Ontology 2013, Montreal, Canada, July 7-12, 2013, pages 105–108
- 2011 Mungall, C. (2011). POSH: The Prolog OWL Shell. In Dumontier, M. and Courtot, M., editors, Proceedings of the 8th International Workshop on OWL: Experiences and Directions (OWLED2011), San Francisco, USA. June 5-6, 2011
- 2009 Mungall, C. J. (2009). Experiences Using Logic Programming in Bioinformatics. In Lecture notes in computer science, volume Volume 564, pages 1–21. Springer
  - Vassiliadis, V., Wielemaker, J., and **Mungall, C. J.** (2009). Processing OWL2 ontologies using Thea: An application of logic programming. In 6th OWL Experiences and Directions Workshop (OWLED 2009)
  - Gkoutos, G. V., **Mungall, C.J.**, Doelken, S., Ashburner, M., Lewis, S., Hancock, J., Schofield, P., Köhler, S., Robinson, P. N., Dolken, S., and Kohler, S. (2009). Entity/Quality-Based Logical Definitions for the Human Skeletal Phenome using PATO. In Proceedings of the 31st Annual International Conference of the IEEE Engineering in Medicine and Biology Society (EMBC 2009), volume 2009, pages 7069–72
- 2008 Bada, M., **Mungall, C. J.**, and Hunter, L. (2008). A Call for an Abductive Reasoning Feature in OWL-Reasoning Tools toward Ontology Quality Control. In 5th OWL Experiences and Directions Workshop (OWLED 2008)
- 2007 Mungall, C. J., Gkoutos, G., Washington, N., and Lewis, S. (2007b). Representing Phenotypes in OWL. In Golbreich, C., Kalyanpur, A., and Parsia, B., editors, Proceedings of the OWLED 2007 Workshop on OWL: Experience and Directions, Innsbruck, Austria
- 2003 Ashburner, M., **Mungall, C.J.**, and Lewis, S. (2003). Ontologies for biologists: a community model for the annotation of genomic data. In Cold Spring Harbor symposia on quantitative biology, volume 68, pages 227–235

#### **Pre-prints**

All pre-prints are on bioRxiv.

2019 Mungall, C.J., Koehler, S., Robinson, P., Holmes, I., and Haendel, M. (2019). k-BOOM: A Bayesian approach to ontology structure inference, with applications in disease ontology construction. <a href="https://www.biorxiv.org/content/10.1101/048843v3">https://www.biorxiv.org/content/10.1101/048843v3</a>

Zhang XA, Yates A, Vasilevsky N, Gourdine JP, Carmody LC, Danis D, Joachimiak MP, Ravanmehr V, Pfaff ER, Champion J, Robasky K, Xu H, Fecho K, Walton NA, Zhu R, Ramsdill J, **Mungall C,** Kohler S, Haendel MA, McDonald C, Vreeman DJ, Peden DB, Chute CG, Robinson PN. Semantic Integration of Clinical Laboratory Tests from Electronic Health Records for Deep Phenotyping and Biomarker Discovery. bioRxiv. 2019 p. 519231. <a href="https://www.biorxiv.org/content/10.1101/519231v1.abstract">https://www.biorxiv.org/content/10.1101/519231v1.abstract</a>

Zhang XA, Yates A, Vasilevsky N, Gourdine JP, Carmody LC, Danis D, Joachimiak MP, Ravanmehr V, Pfaff ER, Champion J, Robasky K, Xu H, Fecho K, Walton NA, Zhu R, Ramsdill J, **Mungall C**, Kohler S, Haendel MA, McDonald C, Vreeman DJ, Peden DB, Chute CG, Robinson PN. Semantic Integration of Clinical Laboratory Tests from Electronic Health Records for Deep

Phenotyping and Biomarker Discovery. bioRxiv. 2019 p. 519231. https://www.biorxiv.org/content/10.1101/519231v1.abstract

2017 Köhler, S., Robinson, P., and **Mungall, C.J.** (2017a). Opposite-of information improves similarity calculations in phenotype ontologies. <a href="https://www.biorxiv.org/content/10.1101/108977v1">https://www.biorxiv.org/content/10.1101/108977v1</a>

**Mungall, C.J.** and Holmes, I. (2017). WTFgenes: What's The Function of these genes? Static sites for model-based gene set analysis. <a href="https://www.biorxiv.org/content/10.1101/114785v1">https://www.biorxiv.org/content/10.1101/114785v1</a>

2014 **Mungall, C. J.** (2014a). Formalization of Genome Interval Relations. https://www.biorxiv.org/content/10.1101/006650v1

**Mungall, C.J.,** Dietze, H., and Osumi-Sutherland, D. (2014). Use of OWL within the Gene Ontology. <a href="https://www.biorxiv.org/content/10.1101/010090v1">https://www.biorxiv.org/content/10.1101/010090v1</a>

### **Awards and Honors**

2017 International Society for Biocuration, Exceptional Contributions to Biocuration Award

# **Awarded Proposals**

U01 CA239108-01 Robinson (PI), Mungall (PI), Oprea (PI)

02/20/19-02/19/21

NIH—Office of Strategic Coordination – The Common Fund

Illuminating the Druggable Genome by Knowledge Graphs

Prioritizing novel drug targets is a slow, high-risk process. This proposal leverages two knowledge sources: the Monarch Initiative, which aggregates gene-disease-phenotype knowledge, and the Illuminating the Druggable Genome (IDG) project knowledgebase, which collects information on drug characteristics.

Role: PI

R24 OD011883 Haendel (PI), Mungall (PI), Robinson (PI)

09/01/12-05/31/20

Oregon State University / NIH—Office of the Director

The Monarch Initiative: Linking Diseases to Model Organism Resources

We are developing a community resource that integrates genotype-phenotype data from numerous model organisms to support translational medicine and improved disease diagnostics and mechanism discovery. Role: PI

OT3 TR002019 Chute (PI), Haendel (PI), Mungall (PI)

09/23/16-08/31/19

Johns Hopkins University / NIH NCATS

TransMed: A translational data integration machine for biomedical discovery

This collaboration integrates multiple types of existing data sources, including objective signs and symptoms of disease, drug effects, and intervening types of biological data relevant to understanding pathophysiology.

Role: PI

U01 HG009453-01 Mungall (PI)

09/23/16-08/31/19

15

NIH NHGRI

An Intelligent Concept Agent (INCA) for Assisting with the Application of Metadata

The goal of this project is to develop an agent capable of learning and adapting, as well as interacting with researchers to extend a core set of concepts. Role: PI

OT3 HL142479 Ahalt (PI)

09/30/17 - 03/31/19

UNC Chapel Hill / NHLBI)

NIH DataSTAGE

This project will perform high impact, cutting-edge scientific and computing activities to support an NIH Data Commons. We will develop a pilot system to demonstrate the feasibility of a vision we have created for allowing researchers to share data and perform computation on the cloud.

Role: Co-Investigator

U41 HG02273 Thomas (PI)

03/01/12-02/28/22

USC / NIH/NHGRI

The Gene Ontology Consortium

The major goal is to provide a complete and integrated picture of what every single gene in a human being does, thus allowing us to better understand the genetic and cellular workings of human health and disease. Role: Co-Investigator

R24 ES028518 Peters (PI), Mungall (PI)

07/01/17-06/30/20

La Jolla Institute For Allergy and Immunology / NIH

Services to support the OBO foundry standards

This project provides catalytic support to migrate the technical services of the Open Biomedical Ontologies (OBO) Foundry which is an established community resource that promotes and facilitates the development, application, harmonization, and sharing of special digital objects known as 'ontologies', to modern cloud-based computing. Role: PI

R01 3U41HG001315-23S1 Cherry (PI)

05/01/18-04/30/22

Stanford Univ. (NIH/NIGMS)

GO Enrichment in the Cloud

This project will create a web-based system for biomedical researchers to perform analyses of their data using the Gene Ontology system. It will serve as a demonstrator project for a high impact, cutting-edge scientific and computing activities to support an NIH Data Commons, potentially changing the way scientists work with the increasing levels of data being generated by a variety of new techniques. Role: Co-Investigator

### **Invited Talks**

2019 Panelist, CGIAR webinar on reference ontologies for agriculture, <a href="https://youtu.be/mpfUaGn-l00">https://youtu.be/mpfUaGn-l00</a>

2018 Online seminar, "GO Causal Activity Models" (<a href="https://vimeo.com/307092087">https://vimeo.com/307092087</a>), BioOntologies Seminar Series, Iowa State University, December 2018

- Gene Ontology Program at Berkeley Lab Triennial Biosciences Review, LBNL, January 2018
- 2017 Ontology Learning for Biosciences, Energy, and the Environment Environmental Knowledgebase Workshop Berkeley Institute for Data Science, January 2017
- 2016 Incorporating the exposome into machine intelligence methods in biomedical research Sanford Imagenetics, Sioux Falls, September 2016
  - Panelist, Critical Assessment of Genome Interpretation, Open Challenges Conference, UCSF, March 2016
  - Panelist, Data Integration Challenges, NSF Phenotype Research Coordination Network Meeting, BioSphere2, February 2016
- 2015 Computing on phenotypes across scale and species Association for Molecular Pathology Annual Meeting, Austin, November 2015
  - Crossing the Species Divide NIH Symposium: Linking Disease Model Phenotypes to Human Conditions, NIH, September 2015
  - From Phenotype Ontologies to Phenotype Networks, Stanford, May 2015
  - Describing samples using the Uberon anatomy ontology Genomics Standards Workshop JGI, May 2015
  - Towards Common Peer Based Standards Development NIH BD2K Data Standards Workshop, Bethesda, February 2015
- 2014 Computing on the environment NIEHS Workshop, NC State, September 2014
- 2013 Uberon : an integrative multi-species ontology European Bioinformatics Institute Industry Workshop, Hinxton Genome Campus, April 2013
  - Mapping Phenotype Ontologies for diabetes and obesity European Bioinformatics Institute, Hinxton Genome Campus, April 2013
- 2012 Helping Machines to Help Us (Keynote) Rocky Bioinformatics Summit, November 2012
- 2011 The Environment Ontology, Environmental Protection Agency offices, San Francisco, March 2011
- 2009 Logic Programming in Bioinformatics (Keynote) International Conference on Logic Programming, Pasadena, July 2009
- 2007 Overview of the Open Biomedical Ontologies Foundry Clinical Trial Ontology Workshop, NIH, Bethesda, May 2007
- 2006 Ontologies for Evo-Devo, National Evolutionary Synthesis Center, Nov 2006

# **Teaching Experience**

- 2018 Guest lecturer, Health Informatics Graduate Program, UC Davis, CA
- 2017 Tutorial Co-organizer. OBO Tutorial. International Conference on Biomedical Ontology, Newcastle upon Tyne, UK
- 2017 Tutorial Co-organizer, 2017 Berkeley GO Editors Workshop
- 2015 Tutorial Organizer, Introduction to ontologies, International Plant Trait Curation workshop, Corvallis, OR
- 2014 Mentor. Harvey Mudd College, Industry Clinic Program
- 2012 Course organizer. Developing ontologies in Protege/OWL, Hinxton, UK, January 2012

- 2011 Tutorial organizer. Developing ontologies in OBO and OWL, International Conference on Biomedical Ontologies, Buffalo, NY
- 2009 Tutorial organizer. Developing ontologies in OBO and OWL, International Conference on Biomedical Ontologies, Buffalo, NY
- 2005 Undergraduate guest lecturer, Introduction to the Gene Ontology, BioEngineering, UC Berkeley
- 2001-2003 Instructor, Programming for biology. Cold Spring Harbor Laboratory
- 2001 Module Organizer. World Health Organization International Training Course on Bioinformatics, FIOCRUZ, Rio de Janeiro, Brazil, May 21-June 15, 2001

#### **Thesis Committees**

- 2015 Master thesis Committee: Bryan Laraway, Department of Biomedical Informatics, Oregon Health and Sciences University
- 2013 Ph.D. Thesis Committee: Sebastian Koehler, Department of Mathematics and Computer Science, Charite Universitatsmedizin Berlin

#### Service

### **Program Committees**

- 2019 Steering Committee, U.S. Semantic Technologies Symposium 2019
- 2018-present Program co-chair, International Conference on Biological Ontology, Corvallis, OR
- 2019 Co-organizer, Phenotype Ontologies Traversing All The Organisms (POTATO) workshop, International Conference on Biological Ontology, Corvallis, OR
  - Scientific Committee, Gene Ontology Workshop, Montreal, Canada, October 2018
- 2017 Review Committee, Internet of Food Conference, Davis, CA, November 2017
- 2016 Steering Committee, 7th International Conference on Biological Ontology: Food, Nutrition, Health and Environment for the 9 billion, Corvallis, OR, August 2016
  - Program Committee, 8th International Conference on Neural Computation Theory and Applications
  - Program Committee, ECCB 2016
  - Program Committee, Bio-Ontologies 2016
  - Reviewer, Pacific Symposium on Biocomputing 2016
- 2015 Program Committee, 4th Workshop on Knowledge Discovery and Data Mining Meets Linked Open Data
  - Program Committee, Resources Program, ISWC 2016
  - Program Committee, Data Sets and Ontologies Program, ISWC 2015
  - Program Committee, Bio-Ontologies
- 2014 Program Committee, 10th Workshop on Constraint-Based Methods for Bioinformatics, September 8, 2014, Lyon, France
  - Reviewer, AMIA 2014
  - Program Committee, 6th Workshop on Formal Ontologies meet Industry
  - Co-organizer Uberon workshop, Biocuration 2014, Toronto, Canada.
- 2014-2016 Program Committee, PhenoDay ISMB
- 2013 Program Committee, Declarative Logic Programming: Theory, Systems, and Applications

2009-2013 Program Committee, Bio-Ontologies

Program Committee, Semantic Web Applications in the Life Sciences

Program Committee / Track Chair, International Conference on Biomedical Ontologies

2010-2016 Program Committee, ISMB/ECCB

2010 Program Committee, OWL: Experience and Directions

Reviewer. AMIA/TBI 2014

2007 Program Committee, Bio-Ontologies

Program Committee, OWL: Experience and Directions

Program Committee, ISMB/ECCB

### **Journal Reviews**

ACS Synthetic Biology

**BMC** Bioinformatics

**BioMed Research International** 

**Bioinformatics** 

Cancer Research

Database

Environmental Health Perspectives

Genome Research

GigaScience

**Human Mutation** 

International Journal of Approximate Reasoning

International Journal of Human-Computer Studies

International Journal on Semantic Web and Information Systems

Journal of Biomedical Informatics

Journal of Biomedical Semantics

Journal of Inherited Metabolic Diseases

Nature Methods

Nucleic Acids Research

Open Biology

PLoS Computational Biology

**PLoS Genetics** 

PLoS ONE

Peer.J

Review Editor eLife

Systematic Biology

## **Working Groups and Advisory Boards**

2016 Scientific Advisory Board Member, NSF Phyloreferences for the Tree of Life

Subcommittee member, International Federation of Associations of Anatomists (IFAA)

2015 National Center for Ecological Analysis and Synthesis Ontology Working Group

2014-Present Global Alliance for Genomics and Health, Clinical Working Group

International Rare Diseases Consortium, Ontologies and rare disease prioritization

Working Group

Working Group 1, co-leader Transcription Factor TG Consortium

2010, 2011 Scientific Advisory Board Member Disease Ontology

2004-Present Coordinator Open Biological Ontologies Library

## **Institutional Service**

2018 Department Head, Molecular Ecosystems Biology2016 Interview Committee, Interviews for division head

Working Group Member, Neurosciences Interest Working Group

Visioning Group, Biosciences Strategic Plan

### **Review Panels**

2019	NIH BDMA ad-hoc panel member
2018	NLM ad-hoc reviewer, March 2018
2017	NIH ad-hoc Proposal Review Panel member, Genomics, Computational Biology and Technology (GCAT) study panel
	NCI CBIIT Project Review
	SIB Proposal Review Swiss Institute of Bioinformatics, competitive proposal review
2014	DOE Proposal Review Department of Energy, Biological and Environmental Research, SBIR/STTR Program
2012	DOE Proposal Review Department of Energy, Biological and Environmental Research, SBIR/STTR Program