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Education

2011 **Ph.D.** Bioinformatics, University of Edinburgh
1993 **BSc.** Artificial Intelligence and Computer Science, University of Edinburgh

Employment

2006-Present **Staff Scientist** Genomics Division, Lawrence Berkeley National Laboratory
2001-2006 **Bioinformatics specialist** Howard Hughes Medical Institute, UC Berkeley
1999-2006 **Bioinformatician** Life Sciences Division, Lawrence Berkeley National Laboratory
1994-1999 **Bioinformatician** Roslin Institute, Edinburgh, UK

Publications

Refereed Journal Articles

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|------|---|
| 2014 | Kohler, S., Doelken, S. C., Mungall, C. J. , Bauer, S., Firth, H. V., Bailleul-Forestier, I., Black, G. C. M., Brown, D. L., Brudno, M., Campbell, J., FitzPatrick, D. R., Eppig, J. T., Jackson, A. P., Freson, K., Girdea, M., Helbig, I., Hurst, J. A., Jahn, J., Jackson, L. G., Kelly, A. M., Ledbetter, D. H., Mansour, S., Martin, C. L., Moss, C., Mumford, A., Ouwehand, W. H., Park, S.-M., Riggs, E. R., Scott, R. H., Sisodiya, S., Vooren, S. V., Wapner, R. J., Wilkie, A. O. M., Wright, C. F., Vulto-van Silfhout, A. T., de Leeuw, N., de Vries, B. B. A., Washington, N. L., Smith, C. L., Westerfield, M., Schofield, P., Ruef, B. J., Gkoutos, G. V., Haendel, M., Smedley, D., Lewis, S. E., and Robinson, P. N. (2014). The Human Phenotype Ontology project: linking molecular biology and disease through phenotype data. <i>Nucleic Acids Res.</i> , 42(D1):D966–D974 |
| 2013 | Smedley, D., Oellrich, A., Köhler, S., Ruef, B., Westerfield, M., Robinson, P., Lewis, S., and Mungall, C. (2013). PhenoDigm: analyzing curated annotations to associate animal models with human diseases. <i>Database: the journal of biological databases and curation</i> , 2013: bat025 |
| | Köhler, S., Doelken, S. C., Ruef, B. J., Bauer, S., Washington, N., Westerfield, M., Gkoutos, G., Schofield, P., Smedley, D., Lewis, S. E., Robinson, P. N., and Mungall, C. J. (2013). Construction and accessibility of a cross-species phenotype ontology along with gene annotations for biomedical research. <i>F1000Research</i> , 2(30) |

Robinson, P., Kohler, S., Oellrich, A., Wang, K., **Mungall, C.**, Lewis, S. E., Washington, N., Bauer, S., Seelow, D. S., Krawitz, P., Gilissen, C., Haendel, M., and Smedley, D. (2013). Improved exome prioritization of disease genes through cross species phenotype comparison. *Genome Research*, Epub ahead of print

Meehan, T., Vasilevsky, N., **Mungall, C.**, Dougall, D., Haendel, M., Blake, J., and Diehl, A. (2013). Ontology based molecular signatures for immune cell types via gene expression analysis. *BMC Bioinformatics*, 14(1):263

Buttigieg, P. L., Morrison, N., Smith, B., **Mungall, C. J.**, and Lewis, S. E. (2013). The environment ontology: contextualising biological and biomedical entities. *Journal of Biomedical Semantics*, 4(1):43

Maynard, S. M., **Mungall, C.J.**, Lewis, S. E., Imam, F. T., and Martone, M. E. (2013). A knowledge based approach to matching human neurodegenerative disease and animal models. *Frontiers in neuroinformatics*, 7:7

Roncaglia, P., Martone, M. E., Hill, D. P., Berardini, T. Z., Foulger, R. E., Imam, F. T., Drabkin, H., **Mungall, C.**, and Lomax, J. (2013). The Gene Ontology (GO) Cellular Component Ontology: integration with SAO (Subcellular Anatomy Ontology) and other recent developments. *Journal of Biomedical Semantics*, 4(1):20

Hill, D. P., Adams, N., Bada, M., Batchelor, C., Berardini, T. Z., Dietze, H., Drabkin, H. J., Ennis, M., Foulger, R. E., Harris, M. A., Hastings, J., Kale, N. S., de Matos, P., **Mungall, C. J.**, Owen, G., Roncaglia, P., Steinbeck, C., Turner, S., and Lomax, J. (2013). Dovetailing biology and chemistry: integrating the Gene Ontology with the ChEBI chemical ontology. *BMC genomics*, 14(1):513

Doelken, S. C., Köhler, S., **Mungall, C. J.**, Gkoutos, G. V., Ruef, B. J., Smith, C., Smedley, D., Bauer, S., Klopocki, E., Schofield, P. N., et al. (2013). Phenotypic overlap in the contribution of individual genes to CNV pathogenicity revealed by cross-species computational analysis of single-gene mutations in humans, mice and zebrafish. *Disease models & mechanisms*, 6(2):358–372

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Blake, J., Dolan, M., Drabkin, H., Hill, D., Ni, L., Sitnikov, D., Bridges, S., Burgess, S., Buza, T., McCarthy, F., Peddinti, D., Pillai, L., Carbon, S., Dietze, H., Ireland, A., Lewis, S., **Mungall, C.J.**, et al. (2013). Gene ontology annotations and resources. *Nucleic Acids Research*, 41(D1):D530–D535

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Osumi-Sutherland, D., Reeve, S., **Mungall, C. J.**, Neuhaus, F., Ruttenberg, A., Jefferis, G. S. X. E., and Armstrong, J. D. (2012). A strategy for building neuroanatomy ontologies. *Bioinformatics (Oxford, England)*, pages 1–10

Walls, R. L., Athreya, B., Cooper, L., Elser, J., Gandolfo, M. A., Jaiswal, P., **Mungall, C. J.**, Preece, J., Rensing, S., Smith, B., and Stevenson, D. W. (2012). Ontologies as integrative tools for plant science. *American journal of botany*, pages ajb.1200222–

Chen, C.-K., **Mungall, C. J.**, Gkoutos, G. V., Doelken, S. C., Köhler, S., Ruef, B. J., Smith, C., Westerfield, M., Robinson, P. N., Lewis, S. E., Schofield, P. N., and Smedley, D. (2012). MouseFinder: Candidate disease genes from mouse phenotype data. *Human mutation*, 33(5):858–66

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Meehan, T., Masci, A. M., Abdulla, A., Cowell, L., Blake, J., **Mungall, C. J.**, and Diehl, A. (2011). Logical Development of the Cell Ontology. *BMC Bioinformatics*, 12(1):6

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- 2010 Deegan, J., Dimmer, E., and **Mungall, C. J.** (2010). Formalization of taxon-based constraints to detect inconsistencies in annotation and ontology development. *BMC bioinformatics*, 11(1):530
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- 2009 **Mungall, C. J.**, Gkoutos, G., Smith, C., Haendel, M., Lewis, S., and Ashburner, M. (2010). Integrating phenotype ontologies across multiple species. *Genome Biology*, 11(1):R2
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- Hancock, J., Mallon, A.-M., Beck, T., Gkoutos, G., **Mungall, C. J.**, and Schofield, P. (2009). Mouse, man, and meaning: bridging the semantics of mouse phenotype and human disease. *Mammalian Genome*
- Diehl, A. D., Augustine, A. D., Blake, J. A., Cowell, L. G., Gold, E. S., Gondré-Lewis, T. A., Masci, A. M., Meehan, T. F., Morel, P. A., Group, N. C. O. W., Nijnik, A., Peters, B., Pulendran, B., Scheuerman, R. H., Yao, Q. A., Zand, M. S., and **Mungall, C. J.** (2010). Hematopoietic Cell Types: Prototype for a Revised Cell Ontology. *Journal of Biomedical Informatics*, Epub ahead

- 2008 Yandell, M., Moore, B., Salas, F., **Mungall, C. J.**, MacBride, A., White, C., and Reese, M. G. (2008). Genome-wide analysis of human disease alleles reveals that their locations are correlated in paralogous proteins. *PLoS Computational Biology*, 4:e1000218
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- 2007 **Mungall, C. J.**, Emmert, D. B., and Consortium, T. F. (2007a). A Chado case study: an ontology-based modular schema for representing genome-associated biological information. *Bioinformatics*, 23(13):i337–346
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- 2004 **Mungall, C. J.** (2004). Obol: Integrating Language and Meaning in Bio-Ontologies. *Comparative and Functional Genomics*, 5(7):509–520
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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. 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

Letters to the Editor

- 2013 Druzinsky, R., Mungall, C. J., Haendel, M., Lapp, H., and Mabee, P. (2013). What is an anatomy ontology? Technical report

Conference Proceedings

- 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*
- Mungall, C.**, Torniai, C., Diehl, A., Haendel, M., Ireland, A., Manzoor, S., Meehan, T., and Osumi-Sutherland, D. (2011). Modularization for the Cell Ontology
- 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)*

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| 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, <i>Proceedings of the OWLED 2007 Workshop on OWL: Experience and Directions</i> , 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 <i>Cold Spring Harbor symposia on quantitative biology</i> , volume 68, pages 227–235 |

Pre-prints

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| 2014 | Bolleman, J., Mungall, C. J. , Strozzi, F., Baran, J., Dumontier, M., Bonnal, R. J., Buels, R., Hoehndorf, R., Fujisawa, T., Katayama10, T., et al. (2014). Faldo: A semantic standard for describing the location of nucleotide and protein feature annotation. <i>bioRxiv</i> |
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Manuscripts in submission

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| 2014 | <p>An atlas of active enhancers across human cell types and tissues. Accepted. Role: middle author</p> <p>A promoter level mammalian expression atlas. Accepted. Role: middle author</p> <p>The Porifera Ontology (PORO). Submitted to Journal of Biomedical Semantics. Role: senior author</p> <p>Uberon in 2013. Submitted to Journal of Biomedical Semantics. Role: senior author</p> <p>'Deletions of Chromosomal Regulatory Boundaries are Associated with Congenital Disease. Submitted to Genome Biology. Role: middle author</p> <p>The influence of disease groups on gene candidate predictions from model organism phenotypes. Accepted Journal of Biomedical Semantics. Role: middle author</p> <p>The Biological Spatial Ontology. Accepted by Journal of Biomedical Semantics. Role: middle author</p> <p>The Cell Line Ontology (CLO). Provisionally accepted by Journal of Biomedical Semantics. Role: middle author</p> |
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Awards and Honors

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| 2013 | Encyclopedia of Life Rubenstein Fellowship award. Role: collaborator |
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Grants and Fellowships

Semantic LAMHDI to develop a semantic infrastructure for linking human diseases to model systems. 2012-2016. Role: Co-investigator.

Invited Talks

2013	Uberon : an integrative multi-species ontology - European Bioinformatics Institute Industry Workshop, Hinxton Genome Campus, April 2013
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

- 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
- 2009: **Tutorial co-organizer**. Developing ontologies in OBO and OWL, International Conference on Biomedical Ontologies, Buffalo, NY, 2009
- 2001, 2002, 2003: **Teaching assistant** Programming for biology. Cold Spring Harbor Laboratory
- 2001: **Module Organizer**. Module organizer. WHO International Training Course on Bioinformatics, FIOCRUZ, Rio de Janeiro, Brazil, May 21-June 15, 2001

Service

2014- Present	WG1 co-leader Transcription Factor TG Consortium
2014	Program Committee Member, 6th Workshop on Formal Ontologies meet Industry
2014	Program Committee Member, PhenoDay
2012, 2014	SBIR/STTR Reviewer , Department of Energy
2004- Present	Open Biological Ontologies Library Coordinator
2009- 2013	Program Committee Member, Bio-Ontologies
2011- 2013	Program Committee Member / Track Chair, International Conference on Biomedical Ontologies
2011- 2013	Program Committee Member, Semantic Web Applications in the Life Sciences
2011	Program Committee Member, OWL: Experience and Directions
2010- 2012	Program Committee Member, ISMB/EECB
2010, 2011	Scientific Advisory Board Member, Disease Ontology
2005- Present	Reviewer PLoS Genetics, PLoS Computational Biology, PLoS ONE, Bioinformatics, Journal of Biomedical Semantics, Database, BMC Bioinformatics, Nucleic Acids Research

Last updated: March 19, 2014