

Christopher J Mungall

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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 **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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|------|---|
| 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 |
| | 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. <i>Genome Research</i> |
| | 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. (2013). The Human Phenotype Ontology project: linking molecular biology and disease through phenotype data. <i>Nucleic Acids Res.</i> , pages gkt1026– |

- 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
- 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
- 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. *F1000Research*, 2
- 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
- Egaña Aranguren, M., Fernández-Breis, J. T., Antezana, E., **Mungall, C.J.**, Rodríguez González, A., and Wilkinson, M. D. (2013). OPPL-Galaxy, a Galaxy tool for enhancing ontology exploitation as part of bioinformatics workflows. *Journal of biomedical semantics*, 4(1):2
- Cooper, L., Walls, R. L., Elser, J., Gandolfo, M. a., Stevenson, D. W., Smith, B., Preece, J., Athreya, B., **Mungall, C. J.**, Rensing, S., Hiss, M., Lang, D., Reski, R., Berardini, T. Z., Li, D., Huala, E., Schaeffer, M., Menda, N., Arnaud, E., Shrestha, R., Yamazaki, Y., and Jaiswal, P. (2013). The plant ontology as a tool for comparative plant anatomy and genomic analyses. *Plant & cell physiology*, 54(2):e1
- 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

- 2012 **Mungall, C. J.**, Torniai, C., Gkoutos, G. V., Lewis, S. E., and Haendel, M. A. (2012). Uberon, an integrative multi-species anatomy ontology. *Genome Biology*, 13(1):R5
- Thomas, P. D., Wood, V., **Mungall, C. J.**, Lewis, S. E., and Blake, J. A. (2012). On the Use of Gene Ontology Annotations to Assess Functional Similarity among Orthologs and Paralogs: A Short Report. *PLoS computational biology*, 8(2):e1002386
- 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
- 2011 **Mungall, C. J.**, Batchelor, C., and Eilbeck, K. (2011b). Evolution of the Sequence Ontology terms and relationships. *Journal of Biomedical Informatics*, 44(1):87–93
- 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
- Tirmizi, S., Aitken, S., Moreira, D., **Mungall, C. J.**, Sequeda, J., Shah, N., and Miranker, D. (2011). Mapping between the OBO and OWL ontology languages. *Journal of Biomedical Semantics*, 2(Suppl 1):S3
- Kohler, S., Bauer, S., **Mungall, C. J.**, Carletti, G., Smith, C. L., Schofield, P., Gkoutos, G. V., and Robinson, P. N. (2011). Improving ontologies by automatic reasoning and evaluation of logical definitions. *BMC Bioinformatics*, 12(1):418
- Hoehndorf, R., Batchelor, C., Bittner, T., Dumontier, M., Eilbeck, K., Knight, R., **Mungall, C.J.**, Richardson, J. S., Stombaugh, J., Westhof, E., Zirbel, C. L., and Leontis, N. B. (2011). The RNA Ontology (RNAO): An ontology for integrating RNA sequence and structure data. *Applied Ontology*, 6(1):53–89
- 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

- Mungall, C. J.**, Bada, M., Berardini, T. Z., Deegan, J., Ireland, A., Harris, M. A., Hill, D. P., and Lomax, J. (2011a). Cross-product extensions of the Gene Ontology. *Journal of Biomedical Informatics*, 44(1):80–86
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- Alterovitz, G., Xiang, M., Hill, D. P., Lomax, J., Liu, J., Cherkassky, M., Dreyfuss, J., **Mungall, C.J.**, Harris, M. A., Dolan, M. E., Blake, J. A., and Ramoni, M. F. (2010). Ontology engineering. *Nature Biotechnology*, 28(2):128–130
- 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
- Skinner, M. E., Uzilov, A. V., Stein, L. D., **Mungall, C. J.**, and Holmes, I. H. (2009). JBrowse: A next-generation genome browser. *Genome Research*
- 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*
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- Masci, A., Arighi, C., Diehl, A., Lieberman, A., **Mungall, C.**, Scheuermann, R., Smith, B., and Cowell, L. (2009). An improved ontological representation of dendritic cells as a paradigm for all cell types. *BMC bioinformatics*, 10(1):70
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- 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
- Smith, C. D., Shu, S., **Mungall, C. J.**, and Karpen, G. H. (2007b). The Release 5.1 annotation of *Drosophila melanogaster* heterochromatin. *Science*, 316:1586–1591
- Smith, B., Ashburner, M., Rosse, C., Bard, J., Bug, W., Ceusters, W., Goldberg, L. J., Eilbeck, K., Ireland, A., **Mungall, C. J.**, Consortium, T. O. B. I., Leontis, N., Rocca-Serra, P., Ruttenberg, A., Sansone, S.-A., Scheuermann, R. H., Shah, N., Whetzel, P. L., and Lewis, S. (2007a). The OBO Foundry: coordinated evolution of ontologies to support biomedical data integration. *Nat Biotechnol*, 25(11):1251–1255
- 2006 Yandell, M., **Mungall, C. J.**, Smith, C., Prochnik, S., Kaminker, J., Hartzell, G., Lewis, S., and Rubin, G. M. (2006). Large-scale trends in the evolution of gene structures within 11 animal genomes. *PLoS Computational Biology*, 2(3):e15
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- 2005 Smith, B., Ceusters, W., Kohler, J., Kumar, A., Lomax, J., **Mungall, C. J.**, Neuhaus, F., Rector, A., and Rosse, C. (2005). Relations in Biomedical Ontologies. *Genome Biology*, 6(5)
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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

2002

Harris, M. A., Clark, J., Ireland, A., Lomax, J., Ashburner, M., Foulger, R., Eilbeck, K., Lewis, S., Marshall, B., **Mungall, C. J.**, Richter, J., Rubin, G. M., Blake, J. A., Bult, C., Dolan, M., Drabkin, H., Eppig, J. T., Hill, D. P., Ni, L., Ringwald, M., Balakrishnan, R., Cherry, J. M., Christie, K. R., Costanzo, M. C., Dwight, S. S., Engel, S., Fisk, D. G., Hirschman, J. E., Hong, E. L., Nash, R. S., Sethuraman, A., Theesfeld, C. L., Botstein, D., Dolinski, K., Feierbach, B., Berardini, T., Mundodi, S., Rhee, S. Y., Apweiler, R., Barrell, D., Camon, E., Dimmer, E., Lee, V., Chisholm, R., Gaudet, P., Kibbe, W., Kishore, R., Schwarz, E. M., Sternberg, P., Gwinn, M., Hannick, L., Wortman, J., Berriman, M., Wood, V., de la Cruz, N., Tonellato, P., Jaiswal, P., Seigfried, T., White, R., and Consortium, G. O. (2004). The Gene Ontology (GO) database and informatics resource. *Nucleic Acids Res*, 32(Database issue):D258—D261

Mungall, C. J., Misra, S., Berman, B. P., Carlson, J., Frise, E., Harris, N., Marshall, B., Shu, S., Kaminker, J. S., Prochnik, S. E., Smith, C. D., Smith, E., Tupy, J. L., Wiel, C., Rubin, G. M., and Lewis, S. E. (2002). An integrated computational pipeline and database to support whole-genome sequence annotation. *Genome Biol*, 3(12):RESEARCH0081

Stein, L. D., **Mungall, C. J.**, Shu, S., Caudy, M., Mangone, M., Day, A., Nickerson, E., Stajich, J. E., Harris, T. W., Arva, A., and Lewis, S. (2002). The generic genome browser: a building block for a model organism system database. *Genome Res*, 12(10):1599–1610

Stajich, J. E., Block, D., Boulez, K., Brenner, S. E., Chervitz, S. A., Dagdigian, C., Fuellen, G., Gilbert, J. G., Korf, I., Lapp, H., Lehvaslaiho, H., Matsalla, C., **Mungall, C. J.**, Osborne, B. I., Pocock, M. R., Schattner, P., Senger, M., Stein, L. D., Stupka, E., Wilkinson, M. D., and Birney, E. (2002). The Bioperl toolkit: Perl modules for the life sciences. *Genome Res*, 12(10):1611–1618

Misra, S., Crosby, M. A., **Mungall, C. J.**, Matthews, B. B., Campbell, K. S., Hradecky, P., Huang, Y., Kaminker, J. S., Millburn, G. H., Prochnik, S. E., Smith, C. D., Tupy, J. L., Whitfied, E. J., Bayraktaroglu, L., Berman, B. P., Bettencourt, B. R., Celniker, S. E., de Grey, A. D. N. J., Drysdale, R. A., Harris, N. L., Richter, J., Russo, S., Schroeder, A. J., Shu, S. Q., Stapleton, M., Yamada, C., Ashburner, M., Gelbart, W. M., Rubin, G. M., and Lewis, S. E. (2002). Annotation of the *Drosophila melanogaster* euchromatic genome: a systematic review. *Genome Biol*, 3(12)

Lewis, S. E., Searle, S. M., Harris, N., Gibson, M., Lyer, V., Richter, J., Wiel, C., Bayraktaroglu, L., Birney, E., Crosby, M. A., Kaminker, J. S., Matthews, B. B., Prochnik, S. E., Smith, C. D., Tupy, J. L., Rubin, G. M., Misra, S., **Mungall, C. J.**, and Clamp, M. E. (2002). Apollo: a sequence annotation editor. *Genome Biol*, 3(12):81–82

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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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- 2000 Rubin, G. M., Yandell, M. D., Wortman, J. R., Miklos, G. L. G., Nelson, C. R., Hariharan, I. K., Fortini, M. E., Li, P. W., Apweiler, R., Fleischmann, W., Cherry, J. M., Henikoff, S., Skupski, M. P., Misra, S., Ashburner, M., Birney, E., Boguski, M. S., Brody, T., Brokstein, P., Celniker, S. E., Chervitz, S. A., Coates, D., Cravchik, A., Gabrielian, A., Galle, R. F., Gelbart, W. M., George, R. A., Goldstein, L. S., Gong, F., Guan, P., Harris, N. L., Hay, B. A., Hoskins, R. A., Li, J., Li, Z., Hynes, R. O., Jones, S. J., Kuehl, P. M., Lemaitre, B., Littleton, J. T., Morrison, D. K., **Mungall, C. J.**, O’Farrell, P. H., Pickeral, O. K., Shue, C., Vossell, L. B., Zhang, J., Zhao, Q., Zheng, X. H., and Lewis, S. (2000). Comparative genomics of the eukaryotes. *Science*, 287(5461):2204–2215
- 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

Other Articles

- 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 |

Manuscripts in submission

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| 2013 | <p>An atlas of active enhancers across human cell types and tissues. In press. Nature. Role: middle author</p> <p>A promoter level mammalian expression atlas. Submitted to Nature. 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>The Cell Ontology in 2013. Submitted to Journal of Biomedical Semantics. Role: senior author</p> <p>The influence of disease groups on gene candidate predictions from model organism phenotypes. Submitted to Journal of Biomedical Semantics. Role: middle author</p> <p>The Environment Ontology. Accepted by Journal of Biomedical Semantics pending revisions. Role: middle author</p> <p>The Biological Spatial Ontology. Submitted to 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**. Taught perl scripting module as part of 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

2012-Present: **SBIR/STTR Reviewer**, Department of Energy

2004-Present : **Open Biological Ontologies Library Coordinator**

2011, 2012, 2013: **Program Committee Member, International Conference on Biomedical Ontology**

2011, 2012, 2013: **Program Committee Member, Semantic Web Applications in the Life Sciences**

2011: **Program Committee Member, OWL: Experience and Directions**

2011: **Program Committee Member, ISMB/EECB**

2010, 2011: **Scientific Advisory Board Member, Disease Ontology**

2005-Present : **Reviewer** PLoS Genetics, PLoS Computational Biology, Bioinformatics, Journal of Biomedical Semantics, Database, BMC Bioinformatics, Nucleic Acids Research

Last updated: December 3, 2013