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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

- 2014 Huntley, R. P., Harris, M. A., Alam-Faruque, Y., Blake, J. A., Carbon, S., Dietze, H., Dimmer, E. C., Foulger, R. E., Hill, D. P., Khodiyar, V. K., Lock, A., Lomax, J., Lovering, R. C., Mutowo-Meullenet, P., Sawford, T., Van Auken, K., Wood, V., and **Mungall, C. J.** (2014). A method for increasing expressivity of Gene Ontology annotations using a compositional approach. *BMC Bioinformatics*, 15(1):155
- Haendel, M. A., Balhoff, J. P., Bastian, F. B., Blackburn, D. C., Blake, J. A., Bradford, Y., Comte, A., Dahdul, W. M., Dececchi, T. A., Druzinsky, R. E., Hayamizu, T. F., Ibrahim, N., Lewis, S. E., Mabee, P. M., Niknejad, A., Robinson-Rechavi, M., Sereno, P. C., and **Mungall, C. J.** (2014). Unification of multi-species vertebrate anatomy ontologies for comparative biology in Uberon. *Journal of Biomedical Semantics*, 5(1):21
- Poelen, J. H., Simons, J. D., and **Mungall, C. J.** (2014). Global Biotic Interactions: An open infrastructure to share and analyze species-interaction datasets. *Ecological Informatics*, 24:148–159
- Thacker, R. W., Díaz, M. C., Kerner, A., Vignes-Lebbe, R., Segerdell, E., Haendel, M. A., and **Mungall, C. J.** (2014). The Porifera Ontology (PORO): enhancing sponge systematics with an anatomy ontology. *Journal of Biomedical Semantics*, 5(1):39

Ibn-Salem, J., Köhler, S., Love, M. I., Chung, H.-R., Huang, N., Hurles, M. E., Haendel, M., Washington, N. L., Smedley, D., **Mungall, C. J.**, Lewis, S. E., Ott, C.-E., Bauer, S., Schofield, P. N., Mundlos, S., Spielmann, M., and Robinson, P. N. (2014). Deletions of chromosomal regulatory boundaries are associated with congenital disease. *Genome Biology*, 15(9):423

Oellrich, A., Köhler, S., Washington, N., **Mungall, C.J.**, Lewis, S., Haendel, M., Robinson, P. N., and Smedley, D. (2014). The influence of disease categories on gene candidate predictions from model organism phenotypes. *Journal of Biomedical Semantics*, 5(Suppl 1):S4

Andersson, R., Gebhard, C., Miguel-Escalada, I., Hoof, I., Bornholdt, J., Boyd, M., Chen, Y., Zhao, X., Schmidl, C., Suzuki, T., Ntini, E., Arner, E., Valen, E., Li, K., Schwarzfischer, L., Glatz, D., Raithel, J., Lilje, B., Rapin, N., Bagger, F. O., Jørgensen, M., Andersen, P. R., Bertin, N., Rackham, O., Burroughs, A. M., Baillie, J. K., Ishizu, Y., Shimizu, Y., Furuhashi, E., Maeda, S., Negishi, Y., **Mungall, C.J.**, Meehan, T. F., Lassmann, T., Itoh, M., Kawaji, H., Kondo, N., Kawai, J., Lennartsson, A., Daub, C. O., Heutink, P., Hume, D. A., Jensen, T. H., Suzuki, H., Hayashizaki, Y., Müller, F., Consortium, T. F., Forrest, A. R. R., Carninci, P., Rehli, M., and Sandelin, A. (2014). An atlas of active enhancers across human cell types and tissues. *Nature*, 507(7493):455–461

Köhler, 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. (2014a). The Human Phenotype Ontology project: linking molecular biology and disease through phenotype data. *Nucleic Acids Res.*, 42(D1):D966–D974

Dahdul, W. M., Cui, H., Mabey, P. M., **Mungall, C. J.**, Osumi-Sutherland, D., Walls, R. L., and Haendel, M. A. (2014). Nose to tail, roots to shoots: spatial descriptors for phenotypic diversity in the Biological Spatial Ontology. *Journal of Biomedical Semantics*, 5(1):34

Alam-Faruque, Y., Hill, D. P., Dimmer, E. C., Harris, M. A., Foulger, R. E., Tweedie, S., Attrill, H., Howe, D. G., Thomas, S. R., Davidson, D., Woolf, A. S., Blake, J. A., **Mungall, C. J.**, O'Donovan, C., Apweiler, R., and Huntley, R. P. (2014). Representing Kidney Development Using the Gene Ontology. *PloS one*, 9(6):e99864

Forrest, A. R. R., Kawaji, H., Rehli, M., Kenneth Baillie, J., de Hoon, M. J. L., Haberle, V., Lassmann, T., Kulakovskiy, I. V., Lizio, M., Itoh, M., Andersson, R., **Mungall, C. J.**, Meehan, T. F., Schmeier, S., Bertin, N., Jørgensen, M., Dimont, E., Arner, E., Schmidl, C., Schaefer, U., Medvedeva, Y. A., Plessy, C., Vitezic, M., Severin, J., Semple, C. A., Ishizu, Y., Young, R. S., Francescato, M., Alam, I., Albanese, D., Altschuler, G. M., Arakawa, T., Archer, J. A. C., Arner, P., Babina, M., Rennie, S., Balwierz, P. J., Beckhouse, A. G., Pradhan-Bhatt, S., Blake, J. A., Blumenthal, A., Bodega, B., Bonetti, A., Briggs, J., Brombacher, F., Maxwell Burroughs, A., Califano, A., Cannistraci, C. V., Carbajo, D., Chen, Y., Chierici, M., Ciani, Y., Clevers, H. C., Dalla, E., Davis, C. A., Detmar, M., Diehl, A. D., Dohi, T., Drabløs, F., Edge, A. S. B., Edinger, M., Ekwall, K., Endoh, M., Enomoto, H., Fagiolini, M., Fairbairn, L., Fang, H., Farach-Carson, M. C., Faulkner, G. J., Favorov, A. V., Fisher, M. E., Frith, M. C., Fujita, R., Fukuda, S., Furlanello, C., Furuno, M., Furusawa, J.-i., Geijtenbeek, T. B., Gibson, A. P., Gingeras, T., Goldowitz, D., Gough, J., Guhl, S., Guler, R., Gustincich, S., Ha, T. J., Hamaguchi, M., Hara, M., Harbers, M., Harshbarger, J., Hasegawa, A., Hasegawa, Y., Hashimoto, T., Herlyn, M., Hitchens, K. J., Ho Sui, S. J., Hofmann, O. M., Hoof, I., Hori, F., Huminiecki, L., Iida, K., Ikawa, T., Jankovic, B. R., Jia, H., Joshi, A., Jurman, G., Kaczowski, B., Kai, C., Kaida, K., Kaiho, A., Kajiyama, K., Kanamori-Katayama, M., Kasianov, A. S., Kasukawa, T., Katayama, S., Kato, S., Kawaguchi, S., Kawamoto, H., Kawamura, Y. I., Kawashima, T., Kempfle, J. S., Kenna, T. J., Kere, J., Khachigian, L. M., Kitamura, T., Peter Klinken, S., Knox, A. J., Kojima, M., Kojima, S., Kondo, N., Koseki, H., Koyasu, S., Krampitz, S., Kubosaki, A., Kwon, A. T., Laros, J. F. J., Lee, W., Lennartsson, A., Li, K., Lilje, B., Lipovich, L., Mackay-sim, A., Manabe, R.-i., Mar, J. C., Marchand, B., Mathelier, A., Mejhert, N., Meynert, A., Mizuno, Y., de Lima Morais, D. A., Morikawa, H., Morimoto, M., Moro, K., Motakis, E., Motohashi, H., Mummery, C. L., Murata, M., Nagao-Sato, S., Nakachi, Y., Nakahara, F., Nakamura, T., Nakamura, Y., Nakazato, K., van Nimwegen, E., Ninomiya, N., Nishiyori, H., Noma, S., Nozaki, T., Ogishima, S., Ohkura, N., Ohmiya, H., Ohno, H., Ohshima, M., Okada-Hatakeyama, M., Okazaki, Y., Orlando, V., Ovchinnikov, D. A., Pain, A., Passier, R., Patrikakis, M., Persson, H., Piazza, S., Prendergast, J. G. D., Rackham, O. J. L., Ramilowski, J. A., Rashid, M., Ravasi, T., Rizzu, P., Roncador, M., Roy, S., Rye, M. B., Saijyo, E., Sajantila, A., Saka, A., Sakaguchi, S., Sakai, M., Sato, H., Satoh, H., Savvi, S., Saxena, A., Schneider, C., Schultes, E. A., Schulze-Tanzil, G. G., Schwegmann, A., Sengstag, T., Sheng, G., Shimoji, H., Shimon, Y., Shin, J. W., Simon, C., Sugiyama, D., Sugiyama, T., Suzuki, M., Suzuki, N., Swoboda, R. K., t Hoen, P. A. C., Tagami, M., Takahashi, N., Takai, J., Tanaka, H., Tatsukawa, H., Tatum, Z., Thompson, M., Toyoda, H., Toyoda, T., Valen, E., van de Wetering, M., van den Berg, L. M., Verardo, R., Vijayan, D., Vorontsov, I. E., Wasserman, W. W., Watanabe, S., Wells, C. A., Winteringham, L. N., Wolvetang, E., Wood, E. J., Yamaguchi, Y., Yamamoto, M., Yoneda, M., Yonekura, Y., Yoshida, S., Zabierowski, S. E., Zhang, P. G., Zhao, X., Zucchelli, S., Summers, K. M., Suzuki, H., Daub, C. O., Kawai, J., Heutink, P., Hide, W., Freeman, T. C., Lenhard, B., Bajic, V. B., Taylor, M. S., Makeev, V. J., Sandelin, A., Hume, D. A., Carninci, P., and Hayashizaki, Y. (2014). A promoter-level mammalian expression atlas. *Nature*, 507(7493):462–470

- Kohler, S., Schoeneberg, U., Czeschik, J. C., Doelken, S. C., Hehir-kwa, J. Y., Ibn-Salem, J., **Mungall, C. J.**, Smedley, D., Haendel, M. A., and Robinson, P. N. (2014b). Clinical interpretation of CNVs with cross-species phenotype data. *J. Med. Genet.*, pages 1–7
- Chibucos, M. C., **Mungall, C. J.**, Balakrishnan, R., Christie, K. R., Huntley, R. P., White, O., Blake, J. A., Lewis, S. E., and Giglio, M. (2014). Standardized description of scientific evidence using the Evidence Ontology (ECO). *Database*, 2014(0):1–11
- Sarntivijai, S., Lin, Y., Xiang, Z., Meehan, T. F., Diehl, A. D., Vempati, U. D., Schürer, S. C., Pang, C., Malone, J., Parkinson, H., Liu, Y., Takatsuki, T., Saijo, K., Masuya, H., Nakamura, Y., Brush, M. H., Haendel, M. A., Zheng, J., Stoeckert, C. J., Peters, B., **Mungall, C.J.**, Carey, T. E., States, D. J., Athey, B. D., and He, Y. (2014). CLO: The cell line ontology. *Journal of Biomedical Semantics*, 5(1):37
- Kibbe, W. A., Arze, C., Felix, V., Mitraka, E., Bolton, E., Fu, G., **Mungall, C. J.**, Binder, J. X., Malone, J., Vasant, D., Parkinson, H., and Schriml, L. M. (2014). Disease Ontology 2015 update: an expanded and updated database of human diseases for linking biomedical knowledge through disease data. *Nucleic acids research*, pages gku1011–
- 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. *Database: the journal of biological databases and curation*, 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. *F1000Research*, 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
- 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
- Antezana, E., Venkatesan, A., **Mungall, C. J.**, Mironov, V., and Kuiper, M. (2010). ONTO-ToolKit: enabling bio-ontology engineering via Galaxy. *BMC Bioinformatics*, 11(Suppl 12):S8
- 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*
- Washington, N. L., Haendel, M. A., **Mungall, C. J.**, Ashburner, M., Westerfield, M., and Lewis, S. E. (2009). Linking Human Diseases to Animal Models using Ontology-based Phenotype Annotation. *PLoS Biology*, 7(11)
- Schober, D., Smith, B., Lewis, S., Kusnierczyk, W., Lomax, J., **Mungall, C. J.**, Taylor, C., Rocca-Serra, P., and Sansone, S.-A. (2009). Survey-based naming conventions for use in OBO Foundry ontology development. *BMC Bioinformatics*, 10(1):125
- 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
- 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
- Pfeiffer, B. D., Jenett, A., Hammonds, A. S., Ngo, T.-T. B., Misra, S., Murphy, C., Scully, A., Carlson, J. W., Wan, K. H., Lavery, T. R., **Mungall, C. J.**, Svirska, R., Kadonaga, J. T., Doe, C. Q., Eisen, M. B., Celniker, S. E., and Rubin, G. M. (2008). Tools for neuroanatomy and neurogenetics in Drosophila. *Proceedings of the National Academy of Sciences of the United States of America*, 105:9715–9720
- Carbon, S., Ireland, A., **Mungall, C. J.**, Shu, S., Marshall, B., Lewis, S., the AmiGO Hub, and the Web Presence Working Group (2008). AmiGO: online access to ontology and annotation data. *Bioinformatics*

- 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
- Rubin, D. L., Lewis, S. E., **Mungall, C. J.**, Misra, S., Westerfield, M., Ashburner, M., Sim, I., Chute, C. G., Solbrig, H., Storey, M.-A., Smith, B., Day-Richter, J., Noy, N. F., and Musen, M. A. (2006). National Center for Biomedical Ontology: advancing biomedicine through structured organization of scientific knowledge. *OMICS: A Journal of Integrative Biology*, 10(2):185–198
- 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)
- Eilbeck, K., Lewis, S. E., **Mungall, C. J.**, Yandell, M. D., Stein, L. D., Durbin, R., and Ashburner, M. (2005). The Sequence Ontology: a tool for the unification of genome annotations. *Genome Biology*, 6(5)
- 2004 **Mungall, C. J.** (2004). Obol: Integrating Language and Meaning in Bio-Ontologies. *Comparative and Functional Genomics*, 5(7):509–520
- 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

- 2002 **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., Whitfield, 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
- Gardner, M. J., Hall, N., Fung, E., White, O., Berriman, M., Hyman, R. W., Carlton, J. M., Pain, A., Nelson, K. E., Bowman, S., Paulsen, I. T., James, K., Eisen, J. A., Rutherford, K., Salzberg, S. L., Craig, A., Kyes, S., Chan, M. S., Nene, V., Shallom, S. J., Suh, B., Peterson, J., Angiuoli, S., Pertea, M., Allen, J., Selengut, J., Haft, D., Mather, M. W., Vaidya, A. B., Martin, D. M., Fairlamb, A. H., Fraunholz, M. J., Roos, D. S., Ralph, S. A., McFadden, G. I., Cummings, L. M., Subramanian, G. M., **Mungall, C. J.**, Venter, J. C., Carucci, D. J., Hoffman, S. L., Newbold, C., Davis, R. W., Fraser, C. M., and Barrell, B. (2002). Genome sequence of the human malaria parasite *Plasmodium falciparum*. *Nature*, 419(6906):498–511

- Bergman, C. M., Pfeiffer, B. D., Rincón-Limas, D. E., Hoskins, R. A., Gnirke, A., **Mungall, C. J.**, Wang, A. M., Kronmiller, B., Pacleb, J., Park, S., Stapleton, M., Wan, K., George, R. A., de Jong, P. J., Botas, J., Rubin, G. M., and Celniker, S. E. (2002). Assessing the impact of comparative genomic sequence data on the functional annotation of the *Drosophila* genome. *Genome Biol*, 3(12):RESEARCH0086
- 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
- Ashburner, M., Ball, C. A., Blake, J. A., Butler, H., Cherry, J. M., Corradi, J., Dolinski, K., Eppig, J. T., Harris, M., Hill, D. P., Lewis, S., Marshall, B., **Mungall, C. J.**, Reiser, L., Rhee, S., Richardson, J. E., Richter, J., Ringwald, M., Rubin, G. M., Sherlock, G., and Yoon, J. (2001). Creating the gene ontology resource: design and implementation. *Genome Res*, 11(8):1425–1433
- 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., Lemaître, B., Littleton, J. T., Morrison, D. K., **Mungall, C. J.**, O’Farrell, P. H., Pickeral, O. K., Shue, C., Vossball, 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

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

- 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*
- 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)*
- 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

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| 2014 | Mungall, C. J. (2014). Formalization of Genome Interval Relations. <i>bioRxiv</i> |
| 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> |

Manuscripts in submission

- | | |
|------|---|
| 2014 | Clinical Interpretation of Copy-Number Variants with Cross-Species Phenotype Data. Accepted by Journal of Medical Genetics. Role: middle author |
| | Gateways to the FANTOM5 promoter level mammalian expression atlas. Accepted by Genome Biology. Role: middle author |
| | Faldo: A semantic standard for describing the location of nucleotide and protein feature annotation. Submitted to JBMS. Role: middle author |
| | Towards a computationally actionable phenotype data exchange (PDX). JAMIA. Role: middle author |

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

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| 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 |
| 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	Working Group Member Global Alliance for Genomics and Health, Clinical Working Group
2014-Present	Working Group Member International Rare Diseases Consortium, Ontologies and rare disease prioritization WG
2014	Program Committee Member 10th Workshop on Constraint-Based Methods for Bioinformatics, September 8 2014, Lyon, France
2014-Present	WG1 co-leader Transcription Factor TG Consortium

2014- Present	Member Orion Bionetworks Knowledge Engineering Working Group
2014	Program Committee Member 10th WCB- Workshop on Constraint-Based Methods for Bioinformatics at CP 2014 (Lyon, France, 8th Sept. 2014)
2014	Program Committee Member, 6th Workshop on Formal Ontologies meet Industry
2014	Program Committee Member, PhenoDay ISMB 2014
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- Present	Program Committee Member, ISMB/ECCB
2010, 2011	Scientific Advisory Board Member, Disease Ontology
2005- Present	Reviewer PLoS Genetics, PLoS Computational Biology, PLoS ONE, Bioinformatics, Journal of Biomedical Semantics, Peer/J, Gigascience, Database, BMC Bioinformatics, Nucleic Acids Research

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