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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**

2015

#### Refereed Journal Articles

Mungall, C.J., Washington, N. L., Nguyen-Xuan, J., Condit, C., Smedley, D., Köhler, S., Groza, T., Shefchek, K., Hochheiser, H., Robinson, P. N., Lewis, S. E., and Haendel, M. A. (2015). Use of Model Organism and Disease Databases to Support Matchmaking for Human Disease Gene Discovery. *Human mutation*, 36(10):979–84

Robinson, P. N., **Mungall, C. J.**, and Haendel, M. (2015). Capturing phenotypes for precision medicine. *Molecular Case Studies*, 1(1):a000372

Haendel, M. a., Vasilevsky, N., Brush, M., Hochheiser, H. S., Jacobsen, J., Oellrich, A., **Mungall, C. J.**, Washington, N., Köhler, S., Lewis, S. E., Robinson, P. N., and Smedley, D. (2015). Disease insights through cross-species phenotype comparisons. *Mammalian Genome* 

Groza, T., Köhler, S., Moldenhauer, D., Vasilevsky, N., Baynam, G., Zemojtel, T., Schriml, L., Kibbe, W., Schofield, P., Beck, T., Vasant, D., Brookes, A., Zankl, A., Washington, N., C.J. Mungall, Lewis, S., Haendel, M. a., Parkinson, H., and Robinson, P. (2015). The Human Phenotype Ontology: Semantic Unification of Common and Rare Disease. *The American Journal of Human Genetics*, 97:111–124

Lizio, M., Harshbarger, J., Shimoji, H., Severin, J., Kasukawa, T., Sahin, S., Abugessaisa, I., Fukuda, S., Hori, F., Ishikawa-Kato, S., **Mungall, C. J.**, Arner, E., Baillie, J. K., Bertin, N., Bono, H., de Hoon, M., Diehl, A. D., Dimont, E., Freeman, T. C., Fujieda, K., Hide, W., Kaliyaperumal, R., Katayama, T., Lassmann, T., Meehan, T. F., Nishikata, K., Ono, H., Rehli, M., Sandelin, A., Schultes, E. A., t Hoen, P. A., Tatum, Z., Thompson, M., Toyoda, T., Wright, D. W., Daub, C. O., Itoh, M., Carninci, P., Hayashizaki, Y., Forrest, A. R., and Kawaji, H. (2015). Gateways to the FANTOM5 promoter level mammalian expression atlas. *Genome Biology*, 16(1):22

Buske, O. J., Schiettecatte, F., Hutton, B., Dumitriu, S., Misyura, A., Huang, L., Hartley, T., Girdea, M., Sobreira, N., **Mungall, C.J.**, and Brudno, M. (2015). The Matchmaker Exchange API: Automating Patient Matching Through the Exchange of Structured Phenotypic and Genotypic Profiles. *Human mutation*, 36(10):922–7

Philippakis, A. A., Azzariti, D. R., Beltran, S., Brookes, A. J., Brownstein, C. A., Brudno, M., Brunner, H. G., Buske, O. J., Carey, K., Doll, C., Dumitriu, S., Dyke, S. O., den Dunnen, J. T., Firth, H. V., Gibbs, R. A., Girdea, M., Gonzalez, M., Haendel, M. A., Hamosh, A., Holm, I. A., Huang, L., Hurles, M. E., Hutton, B., Krier, J. B., Misyura, A., Mungall, C.J., Paschall, J., Paten, B., Robinson, P. N., Schiettecatte, F., Sobreira, N. L., Swaminathan, G. J., Taschner, P. E., Terry, S. F., Washington, N. L., Züchner, S., Boycott, K. M., and Rehm, H. L. (2015). The Matchmaker Exchange: A Platform for Rare Disease Gene Discovery. *Human Mutation*, 36(10):n/a-n/a

Deans, A. R., Lewis, S. E., Huala, E., Anzaldo, S. S., Ashburner, M., Balhoff, J. P., Blackburn, D. C., Blake, J. A., Burleigh, J. G., Chanet, B., Cooper, L. D., Courtot, M., Csösz, S., Cui, H., Dahdul, W., Das, S., Dececchi, T. A., Dettai, A., Diogo, R., Druzinsky, R. E., Dumontier, M., Franz, N. M., Friedrich, F., Gkoutos, G. V., Haendel, M., Harmon, L. J., Hayamizu, T. F., He, Y., Hines, H. M., Ibrahim, N., Jackson, L. M., Jaiswal, P., James-Zorn, C., Köhler, S., Lecointre, G., Lapp, H., Lawrence, C. J., Le Novère, N., Lundberg, J. G., Macklin, J., Mast, A. R., Midford, P. E., Mikó, I., Mungall, C. J., Oellrich, A., Osumi-Sutherland, D., Parkinson, H., Ramírez, M. J., Richter, S., Robinson, P. N., Ruttenberg, A., Schulz, K. S., Segerdell, E., Seltmann, K. C., Sharkey, M. J., Smith, A. D., Smith, B., Specht, C. D., Squires, R. B., Thacker, R. W., Thessen, A., Fernandez-Triana, J., Vihinen, M., Vize, P. D., Vogt, L., Wall, C. E., Walls, R. L., Westerfeld, M., Wharton, R. A., Wirkner, C. S., Woolley, J. B., Yoder, M. J., Zorn, A. M., and Mabee, P. (2015). Finding Our Way through Phenotypes. *PLoS Biology*, 13(1):e1002033

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

Dietze, H., Berardini, T. Z., Foulger, R. E., Hill, D. P., Lomax, J., Osumi-Sutherland, D., Roncaglia, P., and **Mungall, C. J.** (2014). TermGenie - A web application for pattern-based ontology class generation. *Journal of Biomedical Semantics*, 5(1):48

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., Koehler, 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., Furuhata, 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

Dahdul, W. M., Cui, H., Mabee, 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

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., Washingthon, 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

Forrest, A. 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., Francescatto, 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., Kaczkowski, 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., Shimoni, 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–

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

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
- 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
- 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

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., Laverty, T. R., **Mungall, C. J.**, Svirskas, 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* 

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

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

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)

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

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., Bayraktaroglir, L., Birney, E., Crosby, M. A., Kaminker, J. S., Matthews, B. B., Prochnik, S. E., Smithy, 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., Rinc?n-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., Lemaitre, B., Littleton, J. T., Morrison, D. K., Mungall, C. J., O'Farrell, P. H., Pickeral, O. K., Shue, C., Vosshall, L. B., Zhang, J., Zhao, Q., Zheng, X. H., and Lewis, S. (2000). Comparative genomics of the eukaryotes. Science, 287(5461):2204–2215

> 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**

1998

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

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

#### **Conference Proceedings**

- 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
- 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. 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 Rea-
	soning 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
- 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**

2014   Mungan, C. J. (2014). Formalization of Genome Interval Relations. <i>otorati</i>	2014	Mungall, C. J. (2014).	Formalization of Genome Interval Relation	as. $bioRxiv$
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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. *bioRxiv* 

#### Manuscripts in submission

Towards a computationally actionable phenotype data exchange (PDX). JAMIA. Role: middle author

## **Awards and Honors**

2013 Encyclopedia of Life Rubenstein Fellowship award. Role: collaborator

# Grants and Fellowships

Semantic LAMHDI (NIH) to develop a semantic infrastructure for linking human diseases to model systems. 2012-2016. Role: Site PI.

## **Invited Talks**

2015 Crossing the Species Divide - NIH Symposium: Linking Disease Model Phenotypes to Human Conditions, NIH, September 2015

2015	From Phenotype Ontologies to Phenotype Networks, Stanford, May 2015
2015	Describing samples using the Uberon anatomy ontology - Genomics Standards Workshop JGI, May 2015
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
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 corganizer. WHO International Training Course on Bioinformatics, FIOCRUZ, Rio de Janeiro, Brazil, May 21-June 15, 2001

# Service

2015	Working Group Member National Center for Ecological Analysis and Synthesis Ontology WG
2015	<b>Program Committee Member</b> 4th Workshop on Knowledge Discovery and Data Mining Meets Linked Open Data
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	<b>Program Committee Member</b> 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, Nature Methods, Bioinformatics, Journal of Biomedical Informatics, Journal of Biomedical Semantics, Peer/J, Gigascience, Database, BMC Bioinformatics, Nucleic Acids Research

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