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Education

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

Employment

2015-Present **Scientist** Environmental Genomics and Systems Biology Division, LBNL
2006-2015 **Scientist** Genomics Division, LBNL
2001-2006 **Bioinformatics Specialist** Howard Hughes Medical Institute, UC Berkeley
1999-2001 **Bioinformatics Scientist** Life Sciences Division, Lawrence Berkeley National Laboratory
1994-1999 **Bioinformatician** Roslin Institute, Edinburgh, UK

Informatics Projects

Software

For additional software contributions see github.com/cmungall

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|--------------|---|--|
| OwlSim | Cross-species phenotype inference, search and matching tool Role: Main Developer | Website: owlsim.org |
| Exomiser | Functional annotation of variants in clinical genomes Role: Co-developer | Website: monarchinitiative.org/page/exomes |
| TermGenie | Web-based ontology term generation system Role: Manager | Website: termgenie.org |
| AmiGO | Gene functional annotation browser Role: Manager | Website: amigo.geneontology.org |
| Noctua | Functional annotation and network biology environment Role: Manager | Website: noctua.berkeleybop.org |
| Uberon | Multi-species organ system ontology Role: Lead Developer | Website: uberon.org |
| Phenopackets | Phenotype exchange format and tools Role: Designer | Website: github.com/cmungall/phenopacket-format |
| SciGraph | Graph database system Role: Manager | Website: github.com/SciGraph |
| Dipper | Python biomedical data extraction pipeline Role: Manager | Website: github.com/monarch-initiative/dipper |
| JBrowse | Javascript genome browser Role: Contributor | Website: jbrowse.org |
| OBO Edit | Ontology development environment Role: Manager | Website: oboedit.org |

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| Chado | Generic Model Organism Database schema and information system Role: Main Designer | Website: gmod.org/wiki/Chado |
| BioPerl | Perl bioinformatics library and software collection Role: Contributor | Website: bioperl.org |

Database and resources

For additional contributions, see publication list

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| Monarch | Genotype-phenotype information system for humans and model system Role: Architect, PI | Website: monarchinitiative.org |
| GO | Resource for functional annotation of genes in all species Role: Informatics Manager | Website: geneontology.org |
| Planteome | Database of plant genotypes and traits Role: co-PI | Website: planteome.org |
| Phenoscape | Evolutionary phenotype database Role: co-PI | Website: phenoscape.org |
| SDGIO | Official UN Environment Program Sustainable Development Goals Ontology Role: Consultant | Website: github.com/SDG-InterfaceOntology |
| OBO Library | Biomedical ontology repository and infrastructure Role: Co-founder and developer | Website: obofoundry.org |
| FANTOMO | Ontology for functional annotation of the mammalian genome Role: Developer | Website: github.com/cmungall/fantom5-ontology |
| BgeeDb | Database for gene expression evolution Role: Ontology Developer | Website: http://bgee.org/ |
| GloBI | Database and infrastructure for aggregating and analyzing species interactions Role: Consultant | Website: globalbioticinteractions.org |
| HPO | Human phenotype ontology | |

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| | Role: Developer | Website: human-phenotype-ontology.org |
| RO | Biological relationship type ontology Role: Lead Developer | Website: github.com/oborel |
| SO | Ontology of sequences and genome features Role: Contributor | Website: sequenceontology.org |
| FlyBase | Drosophila melanogaster database Role: Genome Re-annotation | Website: flybase.org |
| BDGP | Berkeley Drosophila Genome Project Role: Analysis Pipeline | Website: bdgp.org |
| ArkDb | Genome mapping database for farmed and other animal species Role: Designer | Website: thearkdb.org |

Publications

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|-----------------|--|------------|----|
| Google Scholar: | goo.gl/x2R5PC | h-index: | 54 |
| MyNCBI: | goo.gl/OFQ9k6 | i10-index: | 95 |

Refereed Journal Articles

- 2016 **Mungall, C.J.**, McMurry, J. A., Köhler, S., Balhoff, J. P., Borromeo, C., Brush, M., Carbon, S., Conlin, T., Dunn, N., Engelstad, M., Foster, E., Gourdine, J., Jacobsen, J. O., Keith, D., Laraway, B., Lewis, S. E., NguyenXuan, J., Shefchek, K., Vasilevsky, N., Yuan, Z., Washington, N., Hochheiser, H., Groza, T., Smedley, D., Robinson, P. N., and Haendel, M. A. (2016b). The Monarch Initiative: an integrative data and analytic platform connecting phenotypes to genotypes across species. *Nucleic Acids Research*, page gkw1128
- Köhler, S., Vasilevsky, N. A., Engelstad, M., Foster, E., McMurry, J., Aymé, S., Baynam, G., Bello, S. M., Boerkoel, C. F., Boycott, K. M., Brudno, M., Buske, O. J., Chinnery, P. F., Cipriani, V., Connell, L. E., Dawkins, H. J., DeMare, L. E., Devereau, A. D., deVries, B., Firth, H. V., Freson, K., Greene, D., Hamosh, A., Helbig, I., Hum, C., Jähn, J. A., James, R., Krause, R., Laulederkind, S. J., Lochmüller, H., Lyon, G. J., Ogishima, S., Olry, A., Ouwehand, W. H., Pontikos, N., Rath, A., Schaefer, F., Scott, R. H., Segal, M., Sergouniotis, P. I., Sever, R., Smith, C. L., Straub, V., Thompson, R., Turner, C., Turro, E., Veltman, M., Vulliamy, T., Yu, J., vonZiegenweidt, J., Zankl, A., Züchner, S., Zemojtel, T., Jacobsen, J., Groza, T., Smedley, D., **Mungall, C.J.**, Haendel, M., and Robinson, P. N. (2016). The Human Phenotype Ontology in 2017. *Nucleic Acids Research*, page gkw1039
- Diehl, A. D., Meehan, T. F., Bradford, Y. M., Brush, M. H., Dahdul, W. M., Dougall, D. S., He, Y., Osumi-Sutherland, D., Ruttenberg, A., Sarntivijai, S., Van Slyke, C. E., Vasilevsky, N. A., Haendel, M. A., Blake, J. A., and **Mungall, C.J.** (2016). The Cell Ontology 2016: enhanced content, modularization, and ontology interoperability. *Journal of Biomedical Semantics*, 7(1):44
- Buttigieg, P. L., Pafilis, E., Lewis, S. E., Schildhauer, M. P., Walls, R. L., and **Mungall, C. J.** (2016). The environment ontology in 2016: bridging domains with increased scope, semantic density, and interoperation. *Journal of Biomedical Semantics*, 7(1):57
- McMurry, J. A., Köhler, S., Washington, N. L., Balhoff, J. P., Borromeo, C., Brush, M., Carbon, S., Conlin, T., Dunn, N., Engelstad, M., Foster, E., Gourdine, J.-P., Jacobsen, J. O. B., Keith, D., Laraway, B., Xuan, J. N., Shefchek, K., Vasilevsky, N. A., Yuan, Z., Lewis, S. E., Hochheiser, H., Groza, T., Smedley, D., Robinson, P. N., **Mungall, C.J.**, and Haendel, M. A. (2016). Navigating the Phenotype Frontier: The Monarch Initiative. *Genetics*, 203(4):1491–5

Smedley, D., Schubach, M., Jacobsen, J., Köhler, S., Zemojtel, T., Spielmann, M., Jäger, M., Hochheiser, H., Washington, N., McMurtry, J., Haendel, M., **Mungall, C.J.**, Lewis, S., Groza, T., Valentini, G., and Robinson, P. (2016). A Whole-Genome Analysis Framework for Effective Identification of Pathogenic Regulatory Variants in Mendelian Disease. *The American Journal of Human Genetics*, 99(3):595–606

Hill, D. P., D'Eustachio, P., Berardini, T. Z., **Mungall, C.J.**, Renedo, N., and Blake, J. A. (2016). Modeling biochemical pathways in the gene ontology. *Database*, 2016:baw126

Lizio, M., Harshbarger, J., Abugessaisa, I., Noguchi, S., Kondo, A., Severin, J., **Mungall, C.J.**, Arenillas, D., Mathelier, A., Medvedeva, Y. A., Lennartsson, A., Drabløs, F., Ramilowski, J. A., Rackham, O., Gough, J., Andersson, R., Sandelin, A., Ienasescu, H., Ono, H., Bono, H., Hayashizaki, Y., Carninci, P., Forrest, A. R., Kasukawa, T., and Kawaji, H. (2016). Update of the FANTOM web resource: high resolution transcriptome of diverse cell types in mammals. *Nucleic Acids Research*, page gkw995

Ong, E., Xiang, Z., Zhao, B., Liu, Y., Lin, Y., Zheng, J., **Mungall, C.J.**, Courtot, M., Ruttenberg, A., and He, Y. (2016). Ontobee: A linked ontology data server to support ontology term dereferencing, linkage, query and integration. *Nucleic Acids Research*, page gkw918

Links, A. E., Draper, D., Lee, E., Guzman, J., Valivullah, Z., Maduro, V., Lebedev, V., Didenko, M., Tomlin, G., Brudno, M., Girdea, M., Dumitriu, S., Haendel, M. A., **Mungall, C.J.**, Smedley, D., Hochheiser, H., Arnold, A. M., Coessens, B., Verhoeven, S., Bone, W., Adams, D., Boerkoel, C. F., Gahl, W. A., and Sincan, M. (2016). Distributed Cognition and Process Management Enabling Individualized Translational Research: The NIH Undiagnosed Diseases Program Experience. *Frontiers in Medicine*, 3(October):1–9

Bolleman, J. T., **Mungall, C.J.**, Strozzi, F., Baran, J., Dumontier, M., Bonnal, R. J. P., Buels, R., Hoehndorf, R., Fujisawa, T., Katayama, T., and Cock, P. J. A. (2016). FALDO: a semantic standard for describing the location of nucleotide and protein feature annotation. *Journal of Biomedical Semantics*, 7(1):39

Druzinsky, R. E., Balhoff, J. P., Crompton, A. W., Done, J., German, R. Z., Haendel, M. A., Herrel, A., Herring, S. W., Lapp, H., Mabey, P. M., Muller, H.-M., **Mungall, C.J.**, Sternberg, P. W., Van Auken, K., Vinyard, C. J., Williams, S. H., and Wall, C. E. (2016). Muscle Logic: New Knowledge Resource for Anatomy Enables Comprehensive Searches of the Literature on the Feeding Muscles of Mammals. *PloS one*, 11(2):e0149102

2015

Dumontier, M., Gray, A. J., Marshall, M. S., Alexiev, V., Ansell, P., Bader, G., Baran, J., Bolleman, J. T., Callahan, A., Cruz-Toledo, J., Gaudet, P., Gombocz, E. A., Gonzalez-Beltran, A. N., Groth, P., Haendel, M., Ito, M., Jupp, S., Juty, N., Katayama, T., Kobayashi, N., Krishnaswami, K., Laibe, C., Le Novère, N., Lin, S., Malone, J., Miller, M., **Mungall, C.J.**, Rietveld, L., Wimalaratne, S. M., and Yamaguchi, A. (2016). The health care and life sciences community profile for dataset descriptions. *PeerJ*, 4:e2331

Xin, J., Mark, A., Afrasiabi, C., Tsueng, G., Juchler, M., Gopal, N., Stupp, G. S., Putman, T. E., Ainscough, B. J., Griffith, O. L., Torkamani, A., Whetzel, P. L., **Mungall, C.J.**, Mooney, S. D., Su, A. I., and Wu, C. (2016). High-performance web services for querying gene and variant annotation. *Genome Biology*, 17(1):91

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

Bone, W. P., Washington, N. L., Buske, O. J., Adams, D. R., Davis, J., Draper, D., Flynn, E. D., Girdea, M., Godfrey, R., Golas, G., Groden, C., Jacobsen, J., Köhler, S., Lee, E. M. J., Links, A. E., Markello, T. C., **Mungall, C. J.**, Nehrebecky, M., Robinson, P. N., Sincan, M., Soldatos, A. G., Tifft, C. J., Toro, C., Trang, H., Valkanas, E., Vasilevsky, N., Wahl, C., Wolfe, L. A., Boerkoel, C. F., Brudno, M., Haendel, M. A., Gahl, W. A., and Smedley, D. (2015). Computational evaluation of exome sequence data using human and model organism phenotypes improves diagnostic efficiency. *Genetics in medicine*

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*, 26(9-10)

Thessen, A. E., Bunker, D. E., Buttigieg, P. L., Cooper, L. D., Dahdul, W. M., Domisch, S., Franz, N. M., Jaiswal, P., Lawrence-Dill, C. J., Midford, P. E., **Mungall, C. J.**, Ramírez, M. J., Specht, C. D., Vogt, L., Vos, R. A., Walls, R. L., White, J. W., Zhang, G., Deans, A. R., Huala, E., Lewis, S. E., and Mabee, P. M. (2015). Emerging semantics to link phenotype and environment. *PeerJ*, 3:e1470

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., **Mungall, C.J.**, 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., Abuges-saisa, 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)

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., Charet, 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., Seltsmann, 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

- Arner, E., Daub, C. O., Vitting-Seerup, K., Andersson, R., Lilje, B., Drablos, F., Lennartsson, A., Ronnerblad, M., Hrydziuszko, O., Vitezic, M., Freeman, T. C., Alhendi, A., Arner, P., Axton, R., Baillie, J. K., Beckhouse, A., Bodega, B., Briggs, J., Brombacher, F., Davis, M., Detmar, M., Ehrlund, A., Endoh, M., Eslami, A., Fagiolini, M., Fairbairn, L., Faulkner, G. J., Ferrai, C., Fisher, M. E., Forrester, L., Goldowitz, D., Guler, R., Ha, T., Hara, M., Herlyn, M., Ikawa, T., Kai, C., Kawamoto, H., Khachigian, L., Klinken, P. S., Kojima, S., Koseki, H., Klein, S., Mejhert, N., Miyaguchi, K., Mizuno, Y., Morimoto, M., Morris, K. J., Mummery, C., Nakachi, Y., Ogishima, S., Okada-Hatakeyama, M., Okazaki, Y., Orlando, V., Ovchinnikov, D., Passier, R., Patrikakis, M., Pombo, A., Qin, X.-Y., Roy, S., Sato, H., Savvi, S., Saxena, A., Schwegmann, A., Sugiyama, D., Swoboda, R., Tanaka, H., Tomoiu, A., Winteringham, L. N., Wolvetang, E., Yanagi-Mizuochi, C., Yoneda, M., Zabierowski, S., Zhang, P., Abugessaisa, I., Bertin, N., Diehl, A. D., Fukuda, S., Furuno, M., Harshbarger, J., Hasegawa, A., Hori, F., Ishikawa-Kato, S., Ishizu, Y., Itoh, M., Kawashima, T., Kojima, M., Kondo, N., Lizio, M., Meehan, T. F., **Mungall, C.J.**, Murata, M., Nishiyori-Sueki, H., Sahin, S., Sato-Nagao, S., Severin, J., de Hoon, M. J., Kawai, J., Kasukawa, T., Lassmann, T., Suzuki, H., Kawaji, H., Summers, K. M., Wells, C., Hume, D. A., Forrest, A. R., Sandelin, A., Carninci, P., and Hayashizaki, Y. (2015). Transcribed enhancers lead waves of coordinated transcription in transitioning mammalian cells. *Science*, 347(6225):1010–1014
- 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
- 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., 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

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

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. (2013). The Human Phenotype Ontology project: linking molecular biology and disease through phenotype data. *Nucleic Acids Res.*, 42(D1):D966–D974

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. (2014). 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

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., Lewis, S., and Robinson, P. (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

2009

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

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

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

- 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., 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

Conference Proceedings

- 2016 Manda, P., **Mungall, C. J.**, Balhoff, J., Lapp, H., and Vision, T. (2016). Investigating the importance of anatomical homology for cross-species phenotype comparisons using semantic similarity. In *Pacific Symposium on Biocomputing 21*, pages 132–143. World Scientific Publishing Company
- 2014 **Mungall, C. J.**, Dietze, H., and Osumi-Sutherland, D. (2014). Use of OWL within the Gene Ontology. In Keet, M. and Tamma, V., editors, *Proceedings of the 11th International Workshop on OWL: Experiences and Directions (OWLED 2014)*, pages 25–36, Riva del Garda, Italy, October 17-18, 2014
- 2013 Brush, M. H., **Mungall, C.J.**, Washington, N., and Haendel, M. A. (2013). What’s in a Genotype ? An Ontological Characterization for Integration of Genetic Variation Data. In Dumontier, M., Hoehndorf, R., and Baker, C. J. O., editors, *Proceedings of the International Conference on Biomedical Ontology 2013, Montreal, Canada, July 7-12, 2013*, pages 105–108
- 2011 **Mungall, C.** (2011). POSH: The Prolog OWL Shell. In Dumontier, M. and Courtot, M., editors, *Proceedings of the 8th International Workshop on OWL: Experiences and Directions (OWLED2011), San Francisco, USA. June 5-6, 2011*
- 2009 **Mungall, C. J.** (2009). Experiences Using Logic Programming in Bioinformatics. In *Lecture notes in computer science*, volume Volume 564, pages 1–21. Springer
- Vassiliadis, V., Wielemaker, J., and **Mungall, C. J.** (2009). Processing OWL2 ontologies using Thea: An application of logic programming. In *6th OWL Experiences and Directions Workshop (OWLED 2009)*
- Gkoutos, G. V., **Mungall, C.J.**, Doelken, S., Ashburner, M., Lewis, S., Hancock, J., Schofield, P., Köhler, S., Robinson, P. N., Dolken, S., and Kohler, S. (2009). Entity/Quality-Based Logical Definitions for the Human Skeletal Phenome using PATO. In *Proceedings of the 31st Annual International Conference of the IEEE Engineering in Medicine and Biology Society (EMBC 2009)*, volume 2009, pages 7069–72
- 2008 Bada, M., **Mungall, C. J.**, and Hunter, L. (2008). A Call for an Abductive Reasoning Feature in OWL-Reasoning Tools toward Ontology Quality Control. In *5th OWL Experiences and Directions Workshop (OWLED 2008)*

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| 2007 | Mungall, C. J. , Gkoutos, G., Washington, N., and Lewis, S. (2007b). Representing Phenotypes in OWL. In Golbreich, C., Kalyanpur, A., and Parsia, B., editors, <i>Proceedings of the OWLED 2007 Workshop on OWL: Experience and Directions</i> , Innsbruck, Austria |
| 2003 | Ashburner, M., Mungall, C.J. , and Lewis, S. (2003). Ontologies for biologists: a community model for the annotation of genomic data. In <i>Cold Spring Harbor symposia on quantitative biology</i> , volume 68, pages 227–235 |

Pre-prints

All pre-prints are on bioRxiv.

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| 2017 | Köhler, S., Robinson, P., and Mungall, C.J. (2017). Opposite-of information improves similarity calculations in phenotype ontologies |
| 2016 | Mungall, C.J. , Koehler, S., Robinson, P., Holmes, I., and Haendel, M. (2016a). k-BOOM: A Bayesian approach to ontology structure inference, with applications in disease ontology construction |
| | Holmes, I. H. and Mungall, C.J. (2016). BioMake: a GNU Make-compatible utility for declarative workflow management |
| 2014 | Mungall, C. J. (2014). Formalization of Genome Interval Relations |

Awards and Honors

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| 2013 | Encyclopedia of Life Rubenstein Fellowship award. Role: collaborator |
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Awarded Proposals

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| 2016-2018 | NIH/NCATS OT3. co-PD. TransMed: A translational data integration machine for biomedical discovery. |
| 2016-2020 | NIH/OD. co-PD. The Monarch Initiative: Linking Diseases to Model Organism Resources. |
| 2016-2019 | NIH/BD2K. PI. An Intelligent Concept Agent for Assisting with the Application of Metadata |
| 2016-2017 | NIH/NHGRI. co-PI. The Alliance of Genomic Resources: towards integration of six model organism databases and the Gene Ontology Consortium. |

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| 2016-2017 | NIH/BD2K. co-PI. Supplement: Prefix-commons - BioCADDIE: Biomedical and healthCARE Data Discovery and Indexing Engine. |
| 2016-2017 | NIH/BD2K. co-PI. Supplement: BioCADDIE Harvester: Enhancing metadata using GitHub, YAML and Markdown. |
| 2015-2016 | NSF. co-PI. Ontology-enabled reasoning across phenotypes from evolution and model organisms. |
| 2015-2017 | NSF. co-PI. cROP: Common Reference Ontologies and Applications for Plant Biology. |
| 2012-2016 | NIH/OD. co-PI. Semantic LAMHDI: Linking Diseases to Model Organism Resources. |

Invited Talks

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| 2016 | Incorporating the exposome into machine intelligence methods in biomedical research - Sanford Imagenetics, Sioux Falls, September 2016 |
| 2016 | Panelist, Critical Assessment of Genome Interpretation, Open Challenges Conference, UCSF, March 2016 |
| 2016 | Panelist, Data Integration Challenges, NSF Phenotype Research Coordination Network Meeting, BioSphere2, February 2016 |
| 2015 | Computing on phenotypes across scale and species - Association for Molecular Pathology Annual Meeting, Austin, November 2015 |
| 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 |

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| 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

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| 2015 | Tutorial Organizer. Introduction to ontologies, international plant trait curation workshop, Corvallis, OR, 2015 |
| 2014 | Mentor. Harvey Mudd College, Industry Clinic Program |
| 2012 | Course organizer. Developing ontologies in Protege/OWL, Hinxton, UK, January 2012 |
| 2011 | Tutorial organizer. Developing ontologies in OBO and OWL, International Conference on Biomedical Ontologies, Buffalo, NY, 2011 |
| 2009 | Tutorial organizer. Developing ontologies in OBO and OWL, International Conference on Biomedical Ontologies, Buffalo, NY, 2009 |
| 2005 | Undergraduate guest lecturer. Introduction to the Gene Ontology, Bio-Engineering, UC Berkeley |
| 2001-2003 | Instructor Programming for biology. Cold Spring Harbor Laboratory |
| 2001 | Module Organizer. World Health Organization International Training Course on Bioinformatics, FIOCRUZ, Rio de Janeiro, Brazil, May 21-June 15, 2001 |

Thesis Committees

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| 2015 | Master thesis Committee Bryan Laraway, Department of Biomedical Informatics, Oregon Health and Sciences University |
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| 2013 | PhD Thesis Committee Sebastian Koehler, Department of Mathematics and Computer Science, Charite - Universitätsmedizin Berlin |
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Service

Program Committees

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| 2016 | Steering Committee 7th International Conference on Biological Ontology: Food, Nutrition, Health and Environment for the 9 billion, Corvalis, August 2016 |
| 2016 | Program Committee 8th International Conference on Neural Computation Theory and Applications |
| 2016 | Program Committee ECCB 2016 |
| 2016 | Program Committee Bio-Ontologies 2016 |
| 2016 | Program Committee PhenoDay 2016 |
| 2016 | Reviewer Pacific Symposium on Biocomputing 2016 |
| 2015 | Program Committee Member 4th Workshop on Knowledge Discovery and Data Mining Meets Linked Open Data |
| 2015 | Program Committee Member Resources Program, ISWC 2016 |
| 2015 | Program Committee Member Data Sets and Ontologies Program, ISWC 2015 |
| 2015 | Program Committee Member Bio-Ontologies |
| 2014 | Program Committee Member 10th Workshop on Constraint-Based Methods for Bioinformatics, September 8 2014, Lyon, France |
| 2014 | Reviewer AMIA 2014 |
| 2014 | Program Committee Member 6th Workshop on Formal Ontologies meet Industry |
| 2014-Present | Program Committee Member PhenoDay ISMB 2014-2016 |
| 2013 | Program Committee Member Declarative Logic Programming: Theory, Systems, and Applications |
| 2009-2013 | Program Committee Member Bio-Ontologies |

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| 2009-2013 | Program Committee Member Semantic Web Applications in the Life Sciences |
| 2009-2013 | Program Committee Member / Track Chair International Conference on Biomedical Ontologies |
| 2010 | Program Committee Member OWL: Experience and Directions |
| 2010-Present | Program Committee Member ISMB/ECCB |
| 2010 | Reviewer AMIA/TBI 2014 |
| 2007 | Program Committee Member Bio-Ontologies |
| 2007 | Program Committee Member OWL: Experience and Directions |
| 2007 | Program Committee Member ISMB/ECCB |

Journal Reviews

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| 2016 | Reviewing Editor eLife |
| 2016 | Reviewer International Journal of Approximate Reasoning |
| 2016 | Reviewer Systematic Biology |
| 2016 | Reviewer Nature Methods |
| 2016 | Reviewer Genome Research |
| 2016 | Reviewer Nucleic Acids Research |
| 2016 | Reviewer BioMed Research International |
| 2015 | Reviewer Nature Methods |
| 2014-Present | Reviewer PLoS ONE |
| 2014 | Reviewer Peer/J |
| 2014 | Reviewer Gigascience |
| 2014 | Reviewer International Journal of Human-Computer Studies |
| 2013 | Reviewer PLoS Genetics |

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| 2013- Present | Reviewer Journal of Biomedical Semantics |
| 2011- Present | Reviewer PLoS Computational Biology |
| 2011- Present | Reviewer Human Mutation |
| 2011- 2012 | Reviewer Journal of Biomedical Informatics |
| 2012- Present | Reviewer Nucleic Acids Research |
| 2012 | Reviewer International Journal on Semantic Web and Information Systems |
| 2011- Present | Reviewer Database |
| 2010- Present | Reviewer Bioinformatics |
| 2010 | Reviewer Genome Research |

Working Groups and Advisory Boards

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| 2016 | Scientific Advisory Board Member NSF Phylotranscriptomics for the Tree of Life |
| 2015 | Working Group Member National Center for Ecological Analysis and Synthesis Ontology WG |
| 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- Present | WG1 co-leader Transcription Factor TG Consortium |
| 2010, 2011 | Scientific Advisory Board Member Disease Ontology |
| 2004- Present | Coordinator Open Biological Ontologies Library |

Institutional Service

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| 2016 | Interview Committee Interviews for division head |
| 2016 | Working Group Member Neurosciences Interest Working Group |
| 2016 | Visioning Group Biosciences Strategic Plan |

Review Panels

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| 2017 | NIH Proposal Review Panel Genomics, Computational Biology and Technology (GCAT) study panel |
| 2017 | SIB Proposal Review Swiss Institute of Bioinformatics, competitive proposal review |
| 2014 | DOE Proposal Review Department of Energy, Biological and Environmental Research, SBIR/STTR Program |
| 2012 | DOE Proposal Review Department of Energy, Biological and Environmental Research, SBIR/STTR Program |

Last updated: March 2, 2017