Claudia Giambartolomei

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in https://www.linkedin.com/in/claudia-giambartolomei-77735820/

Education

Ph.D., Genomics of Complex Diseases
University College London *2010–2015*
Funded by the British Heart Foundation

Department of Genetics, Environment and Evolution
UCL Genetics Institute (UGI), London, U.K.
Supervisors: Primary: Dr. Vincent Plagnol; Secondary: Prof. Aroon Hingorani

- Analysis of the phenotypic consequences of genetic variations;
- Method development for integration of different sources of data to help in drug discovery;
- Developed a new method for colocalising genome-wide association signals with expression analysis in different tissues using summary statistics;
- Core analyst of the data from the UCL-London-School-Edinburgh-Bristol (UCLEB) consortium of highly phenotyped population-based prospective studies

M.Phil, Epidemiology
University of Cambridge *2008–2009*
Funded by Bill and Melinda Gates Foundation

Department of Public Health and Primary Care
Institute of Public Health, Sidney Sussex College, Cambridge, U.K.
Supervisor Professor Doug Easton

B.Sc., Biology
The George Washington University *2001–2005*
Partly funded by Presidential Achievement Scholarship

Columbian School of Arts and Sciences, Washington, D.C.
Summa cum Laude

High School Diploma, Classical Studies
Liceo Classico Linguistico Aristofane *1996–2001*
Rome, Italy

Score 98/100

Experience

Vocational

Senior Manager, Integrative Data Analysis Unit
Human Technopole Foundation *2023–present*
Health Data Science Centre, Milan, Italy

- Develop and manage a new state-of-the-art unit dedicated to the analysis of large volumes of multi-omic data from human populations generated by the Health Data Science Centre and from national and international collaborations;
- Coordinate a small team of doctoral and post-doctoral scientists with different background and expertise;
- Develop and apply new methods to perform integrative bioinformatics analysis and interpretation in relation to clinical endpoints, integrating a variety of processed molecular and clinical data types;
- Draft manuscripts and other reports for publication and help secure further funding for relevant projects;
- Help establish new projects and research consortia relating to research in the multi-omic field.

Consultant in statistical genetics analyses
Brigham and Women's Hospital, Harvard Medical School *2022–present*
Boston, MA, U.S.A.

Group Member of the Million Veteran Program Translational Genomics Applied to Drug Discovery

- Advise and construct pipelines using population-level data to evaluate which molecular features are most predictive of a successful drug target, specifically the QTL instruments characterization across tissues and cells, using publicly available dataset together with data from the Million Veteran Program.
- Work closely with colleagues to interpret findings, draft manuscripts and other reports for publication;
- Help establish new directions for target identification.

MINDED Researcher Marie Sklodowska-Curie Actions (H2020 COFUND)
Istituto Italiano di Tecnologia *2019–2022*
Non-coding RNAs and RNA-based Therapeutics, Genoa, Italy

- Study and characterize the role of non-coding mRNA expression and RNA binding proteins in neurodevelopmental disorders;
- Supervise students in population genetics methods and analyses.

Postdoctoral Fellow in Neurobehavioral Genetics (NIH T32NS048004)
Geffen School of Medicine at UCLA *2017–2019*
Pathology & Laboratory Medicine Department, Human Genetics Department, Los Angeles, U.S.A.

Supervisor: Dr. Pasaniuc Bogdan

- Method development to discover pathways from non-coding variants to diseases and complex traits.

Postdoctoral Fellow
Icahn School of Medicine at Mount Sinai *2015–2016*
Department of Genetics and Genomic Sciences and Department of Psychiatry, New York, U.S.A.

Supervisor: Dr. Panagiotis Roussos

- Develop strategies to integrate the different types of data available in the lab: genomic, expression, epigenetic, imaging, clinical outcomes, to identify possible mechanisms through which non-coding variants influence complex diseases;
- Supervise students in genetic association methods and analyses:

- Build pipelines to analyse data from ATAC-sequencing methods newly developed in the lab.

Research Assistant
London School of Hygiene and Tropical Medicine *2009–2010*
Non-Communicable Disease Epidemiology Unit, London U.K.

- Statistical genetics analyses.

Research Intern
GlaxoSmithKline *08/2009–10/2009*
Harlow Town, Essex, U.K.

- Analyses of Clinical Data (extraction of relevant information and statistical modelling using SAS), including data on drug adverse reaction, extracted from clinical trials ($>$ 90 trials). The main objective of the project was to compare four different equations used in clinical practice to determine kidney failure.
- Literature Review of clinical trials.

Cancer Research Training Award (CRTA) Fellow
National Cancer Institute (NCI) *2006–2008*
Division of Cancer Epidemiology and Genetics (DCEG), Rockville, M.D., U.S.A.

- Manage clinical and pedigree data in preparation for publication and update data information with occasional interviews when needed for the eight ongoing clinical protocols of Families at Increased Risk of Cancer;
- Assist in substantially modifying data-collection procedures to better monitor patient's visit;
- Liaison role between data management group and clinical team;
- Assist in reviews to be sent to the Institutional Review Board (IRB).

Research Experience for Undergraduates (REU) Intern
University of Florida *06/2004–08/2004*
Gainesville, FL, U.S.A.

- Funded by National Science Foundation;
- Conduct inorganic chemistry laboratory research in the Department of Chemistry, mentored by Dr. Michael Scott;
- Receive Intensive Nuclear Magnetic Resonance (NMR) training;
- Paper & poster presentation of research results, "A synthetic pathway to a novel proton sponge using a three-coordinated amine ligand", University of Florida, October 2004

Miscellaneous

Volunteer
European Red Cross *09/2005–12/2005*
Rome, Italy

Languages

English: native;
Italian: native.

Awards and Honors

European Union's Horizon 2020 under Marie Sklodowska-Curie Actions (grant#754490)	*2019–present*
Training grant in Neurobehavioral Genetics (NIH T32NS048004)	*2017–2018*
British Heart Foundation Ph.D. Studentship	*2010–2014*
Gates-Cambridge Scholar, Gates Cambridge Trust, Bill and Melinda Gates Foundation	*2008–2009*
Phi Beta Kappa, Member	*2004–present*
Presidential Achievement Scholarship	*2003–2005*
Dean's List of Honor Students	*2002–2004*
Golden Key International Honor Society, Member	*2002–2010*

Other Experience and Professional Memberships

- Affiliation and collaboration with Prof. Tartaglia at Istituto Italiano di Tecnologia, RNA Systems Biology, Genoa, Italy, to complete supervision of Ph.D. student Magda Arnal Segura and as collaborator in the project characterizing non-coding mRNA di eQTL and pQTL, 2023-present
- Million Veteran Program (MVP), member of working groups PheWAS Druggable Research Program, COVID-19 Research Program, MR CVD Proteomics Weekly, 2019-present
- Manuscript Reviewer: PLoS Genetics, PLoS One, The American Journal of Human Genetics, Genome Biology, Elsevier, Open Biology, NAR Genomics, Alzheimer's and Dementia, Genome Biology, Nature Communications, Bioinformatics
- Psychiatric Genomics Consortium, member 2015-2017
- CHARGE Consortium, Mendelian randomization and subclinical and coronary heart disease working groups, member 2017
- American Society of Human Genetics, member 2014-present

Teaching and Supervision

- Ph.D. Thesis Examiner, June 2023 University of Edinburgh
- Supervisor of Ph.D. (Magdalena Arnal Segura, Giorgio Bini), M.Sc. (Maddalena Pacelli) and postgraduate students (Alexandros Armaos), at IIT, 2019-2021
- Supervisor of undergraduate students, Bruins-In-Genomics (B.I.G.) Summer Research Program, July-August 2017; July-August 2018
- Supervisor of M.D./Ph.D. student in genetic assocation analyses, January-July 2016
- Statistics for Interpreting Genetic Data, GENEG005, Lecturer: Prof. David Balding, Dr. Vincent Plagnol, March 2014
- Computational Biology course, BIOL2015, Division of Biosciences, Lecturer: Dr. Max Reuter, October 6 2012/ October-December 2013

- UCL-LSHTM short class, Lecturer: Prof. David Balding, Prof. Vincent Plagnol, "Introduction to sequencing technologies, basic concept and file formats", September 12-13 2011/ September 12-13 2012 / September 2-4 2014

Publications

Most updated list in Google Scholar: https://scholar.google.com/citations?hl=enuser=quIST80AAAAJ

Published and In Press.....

2023 Schmidt, Amand F., Roshni Joshi, Maria Gordillo-Marañón, Fotios Drenos, Pimphen Charoen, Claudia Giambartolomei, Joshua C. Bis, Tom R. Gaunt, Alun D. Hughes, Deborah A. Lawlor, Andrew Wong, Jackie F. Price, Nishi Chaturvedi, Goya Wannamethee, Nora Franceschini, Mika Kivimaki, Aroon D. Hingorani, and Chris Finan (Jan. 2023). "Biomedical consequences of elevated cholesterol-containing lipoproteins and apolipoproteins on cardiovascular and non-cardiovascular outcomes." In: *Communications medicine* 3, p. 9. ISSN: 2730-664X. DOI: 10.1038/s43856-022-00234-0. epublish.

Role: Interpretation of findings.

2022 Arnal Segura, Magdalena, Giorgio Bini, Dietmar Fernandez Orth, Eleftherios Samaras, Maya Kassis, Fotis Aisopos, Jordi Rambla De Argila, George Paliouras, Peter Garrard, **Giambartolomei, C.**, and GG Tartaglia (2022). "Machine learning methods applied to genotyping data capture interactions between single nucleotide variants in late onset Alzheimer's disease." In: *Alzheimer's & dementia (Amsterdam, Netherlands)* 14, e12300. ISSN: 2352-8729. DOI: 10.1002/dad2.12300.

Castaneda, Andy B, Lauren E Petty, Markus Scholz, Rick Jansen, Stefan Weiss, Xiaoling Zhang, Katharina Schramm, Frank Beutner, Holger Kirsten, Ulf Schminke, Shih-Jen Hwang, Carola Marzi, Klodian Dhana, Adrie Seldenrijk, Knut Krohn, Georg Homuth, Petra Wolf, Marjolein J Peters, Marcus Dörr, Annette Peters, Joyce B J van Meurs, André G Uitterlinden, Maryam Kavousi, Daniel Levy, Christian Herder, Gerard van Grootheest, Melanie Waldenberger, Christa Meisinger, Wolfgang Rathmann, Joachim Thiery, Joseph Polak, Wolfgang Koenig, Jochen Seissler, Joshua C Bis, Nora Franceshini, **Giambartolomei, C.**, Cohorts for Heart, Aging Research in Genomic Epidemiology (CHARGE) Subclinical Working Group, Albert Hofman, Oscar H Franco, Brenda W J H Penninx, Holger Prokisch, Henry Völzke, Markus Loeffler, Christopher J O'Donnell, Jennifer E Below, Abbas Dehghan, and Paul S de Vries (Mar. 2022). "Associations of carotid intima media thickness with gene expression in whole blood and genetically predicted gene expression across 48 tissues." In: *Human molecular genetics* 31, pp. 1171–1182. ISSN: 1460-2083. DOI: 10.1093/hmg/ddab236.

Linder, Simon, Marlous Hoogstraat, Suzan Stelloo, Nils Eickhoff, Karianne Schuurman, Hilda de Barros, Maartje Alkemade, Elise M Bekers, Tesa M Severson, Joyce Sanders, Chia-Chi Flora Huang, Tunc Morova, Umut Berkay Altintas, Liesbeth Hoekman, Yongsoo Kim, Sylvan C Baca, Martin Sjöström, Anniek Zaalberg, Dorine C Hintzen, Jeroen de Jong, Roelof J C Kluin, Iris de Rink, **Giambartolomei, C.**, Ji-Heui Seo, Bogdan Pasaniuc, Maarten Altelaar, René H Medema, Felix Y Feng, Amina Zoubeidi, Matthew L Freedman, Lodewyk F A Wessels, Lisa M Butler, Nathan A Lack, Henk van der Poel, Andries M Bergman, and Wilbert Zwart (Sept. 2022). "Drug-Induced Epigenomic Plasticity Reprograms Circadian Rhythm Regulation to Drive

Prostate Cancer toward Androgen Independence." In: *Cancer discovery* 12, pp. 2074–2097. ISSN: 2159-8290. DOI: 10.1158/2159-8290.CD-21-0576.

Rasooly, Danielle, Gina M. Peloso, and Claudia Giambartolomei (Dec. 2022). "Bayesian Genetic Colocalization Test of Two Traits Using coloc." In: *Current protocols* 2, e627. ISSN: 2691-1299. DOI: 10.1002/cpz1.627. ppublish.

Role: supervision and writing.

2021 Baca, Sylvan C, David Y Takeda, Ji-Heui Seo, Justin Hwang, Sheng Yu Ku, Rand Arafeh, Taylor Arnoff, Supreet Agarwal, Connor Bell, Edward O'Connor, Xintao Qiu, Sarah Abou Alaiwi, Rosario I Corona, Marcos A S Fonseca, **Giambartolomei, C.**, Paloma Cejas, Klothilda Lim, Monica He, Anjali Sheahan, Amin Nassar, Jacob E Berchuck, Lisha Brown, Holly M Nguyen, Ilsa M Coleman, Arja Kaipainen, Navonil De Sarkar, Peter S Nelson, Colm Morrissey, Keegan Korthauer, Mark M Pomerantz, Leigh Ellis, Bogdan Pasaniuc, Kate Lawrenson, Kathleen Kelly, Amina Zoubeidi, William C Hahn, Himisha Beltran, Henry W Long, Myles Brown, Eva Corey, and Matthew L Freedman (Mar. 2021). "Reprogramming of the FOXA1 cistrome in treatment-emergent neuroendocrine prostate cancer." In: *Nature communications* 12, p. 1979. ISSN: 2041-1723. DOI: 10.1038/s41467-021-22139-7.

Role: HiChIP analysis.

Blokland, G.A.M and Schizophrenia Working Group of the Psychiatric Genomics Consortium et al. (Mar. 2021). "Sex-Dependent Shared and Nonshared Genetic Architecture Across Mood and Psychotic Disorders." In: *Biological psychiatry*. ISSN: 1873-2402. DOI: 10.1016/j.biopsych.2021.02.972.

Role: eQTL analysis.

Gaziano, L., **Giambartolomei, C.**, A. Pereira, A. Gaulton, D.C. Posner, S.A. Swanson, Y. Ho, S.K. Iyengar, N.M. Kosik, M. Vujkovic, D.R. Gagnon, A.P. Bento, I. Barrio-Hernandez, L. Rönnblom, N. Hagberg, C. Lundtoft, C. Langenberg, M. Pietzner, D. Valentine, S. Gustincich, G.G. Tartaglia, E. Allara, P. Surendran, S. Burgess, J.H. Zhao, J.E. Peters, B.P. Prins, E. Angelantonio, P. Devineni, Y. Shi, K.E. Lynch, S.L. DuVall, H. Garcon, L.O. Thomann, J.JJ Zhou, B.R. Gorman, J.E. Huffman, C.J. O'Donnell, P.S. Tsao, J.C Beckham, S. Pyarajan, S. Muralidhar, G.D. Huang, R. Ramoni, P. Beltrao, J. Danesh, A.M. Hung, KM. Chang, Y.V. Sun, J. Joseph, A.R. Leach, T.L. Edwards, K. Cho, J.M. Gaziano, A.S. Butterworth, J P Casas, and VA Million Veteran Program COVID-19 Science Initiative (Apr. 2021). "Actionable druggable genome-wide Mendelian randomization identifies repurposing opportunities for COVID-19." In: *Nature medicine* 27, pp. 668–676. ISSN: 1546-170X. DOI: 10.1038/s41591-021-01310-z.

Role: eQTL analyses and colocalization with GWAS.

Majumdar, Arunabha, **Giambartolomei, C.**, Na Cai, Tanushree Haldar, Tommer Schwarz, Michael Gandal, Jonathan Flint, and Bogdan Pasaniuc (May 2021). "Leveraging eQTLs to identify individual-level tissue of interest for a complex trait." In: *PLoS computational biology* 17, e1008915. ISSN: 1553-7358. DOI: 10.1371/journal.pcbi.1008915.

Role: participated in method development and interpretation of results and simulations.

Schmidt, Amand F, Nicholas B Hunt, Maria Gordillo-Marañón, Pimphen Charoen, Fotios Drenos, Mika Kivimaki, Deborah A Lawlor, **Giambartolomei, C.**, Olia Papacosta, Nishi Chaturvedi, Joshua C Bis, Christopher J O'Donnell, Goya Wannamethee, Andrew Wong, Jackie F Price, Alun D Hughes, Tom R Gaunt, Nora Franceschini, Dennis O Mook-Kanamori, Magdalena Zwierzyna, Reecha Sofat, Aroon D Hingorani, and Chris Finan (Sept. 2021). "Cholesteryl ester transfer protein (CETP) as a drug target for cardiovascular disease." In: *Nature communications* 12, p. 5640. ISSN: 2041-1723. DOI: 10.1038/s41467-021-25703-3.

Role: eQTL analyses and interpretation of results.

Watt, April C, Paloma Cejas, Molly J DeCristo, Otto Metzger-Filho, Enid Y N Lam, Xintao Qiu, Haley BrinJones, Nikolas Kesten, Rhiannon Coulson, Alba Font-Tello, Klothilda Lim, Raga Vadhi, Veerle W Daniels, Joan Montero, Len Taing, Clifford A Meyer, Omer Gilan, Charles C Bell, Keegan D Korthauer, **Giambartolomei, C.**, Bogdan Pasaniuc, Ji-Heui Seo, Matthew L Freedman, Cynthia Ma, Matthew J Ellis, Ian Krop, Eric Winer, Anthony Letai, Myles Brown, Mark A Dawson, Henry W Long, Jean J Zhao, and Shom Goel (Jan. 2021). "CDK4/6 inhibition reprograms the breast cancer enhancer landscape by stimulating AP-1 transcriptional activity." In: *Nature cancer* 2, pp. 34–48. ISSN: 2662-1347. DOI: 10.1038/s43018-020-00135-y.

Role: HiChIP analysis.

Giambartolomei, C., Ji-Heui Seo, Tommer Schwarz, Malika Kumar Freund, Ruth Dolly Johnson, Sandor Spisak, Sylvan C Baca, Alexander Gusev, Nicholas Mancuso, Bogdan Pasaniuc, and Matthew L Freedman (Dec. 2021b). "H3K27ac HiChIP in prostate cell lines identifies risk genes for prostate cancer susceptibility." In: *American journal of human genetics* 108, pp. 2284–2300. ISSN: 1537-6605. DOI: 10.1016/j.ajhg.2021.11.007.

2020 Coleman, J.R.I., H.A. Gaspar, J. Bryois, Bipolar Disorder Working Group of the Psychiatric Genomics Consortium, Major Depressive Disorder Working Group of the Psychiatric Genomics Consortium, and Gerome Breen (July 2020). "The Genetics of the Mood Disorder Spectrum: Genome-wide Association Analyses of More Than 185,000 Cases and 439,000 Controls." In: *Biological psychiatry* 88, pp. 169–184. ISSN: 1873-2402. DOI: 10.1016/j.biopsych.2019.10.015.

Pomerantz, Mark M, Xintao Qiu, Yanyun Zhu, David Y Takeda, Wenting Pan, Sylvan C Baca, Alexander Gusev, Keegan D Korthauer, Tesa M Severson, Gavin Ha, Srinivas R Viswanathan, Ji-Heui Seo, Holly M Nguyen, Baohui Zhang, Bogdan Pasaniuc, Claudia Giambartolomei, Sarah A Alaiwi, Connor A Bell, Edward P O'Connor, Matthew S Chabot, David R Stillman, Rosina Lis, Alba Font-Tello, Lewyn Li, Paloma Cejas, Andries M Bergman, Joyce Sanders, Henk G van der Poel, Simon A Gayther, Kate Lawrenson, Marcos A S Fonseca, Jessica Reddy, Rosario I Corona, Gleb Martovetsky, Brian Egan, Toni Choueiri, Leigh Ellis, Isla P Garraway, Gwo-Shu Mary Lee, Eva Corey, Henry W Long, Wilbert Zwart, and Matthew L Freedman (Aug. 2020). "Prostate cancer reactivates developmental epigenomic programs during metastatic progression." In: *Nature genetics* 52, pp. 790–799. ISSN: 1546-1718. DOI: 10.1038/s41588-020-0664-8.

2019 Andlauer, Till F M and Bipolar Disorder Working Group and Major Depressive Disorder Working Group of the Psychiatric Genomics Consortium et al. (Nov. 2019). "Bipolar multiplex families have an increased burden of common risk variants for psychiatric disorders." In: *Molecular psychiatry*. ISSN: 1476-5578. DOI: 10.1038/s41380-019-0558-2.

Drange, Ole Kristian and Psychiatric Genomics Consortium Bipolar Disorder Working Group at al. (2019). "Genetic Overlap Between Alzheimer's Disease and Bipolar Disorder Implicates the MARK2 and VAC14 Genes." In: *Frontiers in neuroscience* 13, p. 220. ISSN: 1662-4548. DOI: 10.3389/fnins.2019.00220.

Gill, Dipender, Christopher F Brewer, Grace Monori, David-Alexandre Tregouet, Nora Franceschini, **Giambartolomei, C.**, INVENT Consortium, Ioanna Tzoulaki, and Abbas Dehghan (Aug. 2019). "Effects of Genetically Determined Iron Status on Risk of Venous Thromboembolism and Carotid Atherosclerotic Disease: A Mendelian Randomization Study." In: *Journal of the American Heart Association* 8, e012994. ISSN: 2047-9980. DOI: 10.1161/JAHA.119.012994.

Role: statistical association analysis.

Hauberg, Mads E, John F Fullard, Lingxue Zhu, Ariella T Cohain, **Giambartolomei, C.**, Ruth Misir, Sarah Reach, Jessica S Johnson, Minghui Wang, Manuel Mattheisen, Anders Dupont Børglum, Bin Zhang, Solveig K Sieberts, Mette A Peters, Enrico Domenici, Eric E Schadt, Bernie Devlin, Pamela Sklar, Kathryn Roeder, Panos Roussos, and CommonMind Consortium (Nov. 2019). "Differential activity of transcribed enhancers in the prefrontal cortex of 537 cases with schizophrenia and controls." In: *Molecular psychiatry* 24, pp. 1685–1695. ISSN: 1476-5578. DOI: 10.1038/s41380-018-0059-8.

Role: statistical association analysis.

Stahl, Eli A et al. (May 2019). "Genome-wide association study identifies 30 loci associated with bipolar disorder." In: *Nature genetics* 51, pp. 793–803. ISSN: 1546-1718. DOI: 10.1038/s41588-019-0397-8.

Role: statistical association analysis.

2018 Bipolar Disorder and Schizophrenia Working Group of the Psychiatric Genomics Consortium (June 2018). "Genomic Dissection of Bipolar Disorder and Schizophrenia, Including 28 Subphenotypes." In: *Cell* 173, 1705–1715.e16. ISSN: 1097-4172. DOI: 10.1016/j.cell.2018.05.046.

Role: statistical association analysis.

Dobbyn, Amanda, Laura M Huckins, James Boocock, Laura G Sloofman, Benjamin S Glicksberg, **Giambartolomei, C.**, Gabriel E Hoffman, Thanneer M Perumal, Kiran Girdhar, Yan Jiang, Towfique Raj, Douglas M Ruderfer, Robin S Kramer, Dalila Pinto, CommonMind Consortium, Schahram Akbarian, Panos Roussos, Enrico Domenici, Bernie Devlin, Pamela Sklar, Eli A Stahl, and Solveig K Sieberts (June 2018). "Landscape of Conditional eQTL in Dorsolateral Prefrontal Cortex and Co-localization with Schizophrenia GWAS." In: *American journal of human genetics* 102, pp. 1169–1184. ISSN: 1537-6605. DOI: 10.1016/j.ajhg.2018.04.011.

Role: bioinformatics pipeline.

Giambartolomei, Claudia, Jimmy Zhenli Liu, Wen Zhang, Mads Hauberg, Huwenbo Shi, James Boocock, Joe Pickrell, Andrew E Jaffe, CommonMind Consortium, Bogdan Pasaniuc, and Panos Roussos (Aug. 2018). "A Bayesian framework for multiple trait colocalization from summary association statistics." In: *Bioinformatics (Oxford, England)* 34, pp. 2538–2545. ISSN: 1367-4811. DOI: 10.1093/bioinformatics/bty147.

Giambartolomei*, C. and Nora et al. Franceschini* (Dec. 2018). "GWAS and colocalization analyses implicate carotid intima-media thickness and carotid plaque loci in cardiovascular outcomes." In: *Nature communications* 9, p. 5141. ISSN: 2041-1723. DOI: 10.1038/s41467-018-07340-5.

Giambartolomei, C., Jimmy Zhenli Liu, Wen Zhang, Mads Hauberg, Huwenbo Shi, James Boocock, Joe Pickrell, Andrew E Jaffe, Bogdan Pasaniuc, Panos Roussos, et al. (2018). "A Bayesian Framework for Multiple Trait Colocalization from Summary Association Statistics". In: *Bioinformatics*, p. 155481. DOI: http://dx.doi.org/10.1093/bioinformatics/bty147.

Role: analysis and write-up.

2017 Fairoozy, Roaa Hani, J Cooper, J White, **Giambartolomei, C.**, L Folkersen, SG Wannamethee, BJ Jefferis, P Whincup, Y Ben-Shlomo, M Kumari, M Kivimaki, A Wong, R Hardy, D Kuh, TR Gaunt, JP Casas, S McLachlan, JF Price, A Hingorani, A Franco-Cereceda, T Grewal, AZ Kalea, SE Humphries, and UCLEB consortium (Apr. 2017). "Identifying low density lipoprotein cholesterol associated variants in the Annexin A2 (ANXA2) gene." In: *Atherosclerosis* 261, pp. 60–68. ISSN: 1879-1484. DOI: 10.1016/j.atherosclerosis.2017.04.010.

Role: analysis of eQTL data.

Fullard, John F, **Giambartolomei, C.**, Mads E Hauberg, Ke Xu, Georgios Voloudakis, Zhiping Shao, Christopher Bare, Joel T Dudley, Manuel Mattheisen, Nikolaos K Robakis, et al. (2017). "Open chromatin profiling of human postmortem brain infers functional roles for non-coding schizophrenia loci". In: *Human Molecular Genetics* 26.10, pp. 1942–1951.

Role: write-up.

Hauberg, Mads Engel, Wen Zhang, **Giambartolomei, C.**, Oscar Franzén, David L Morris, Timothy J Vyse, Arno Ruusalepp, Pamela Sklar, Eric E Schadt, Johan LM Björkegren, et al. (2017). "Large-Scale Identification of Common Trait and Disease Variants Affecting Gene Expression". In: *The American Journal of Human Genetics*.

Role: integrative analysis.

Soderquest, Katrina, Arnulf Hertweck, Claudia Giambartolomei, Stephen Henderson, Rami Mohamed, Rimma Goldberg, Esperanza Perucha, Lude Franke, Javier Herrero, Vincent Plagnol, Richard G Jenner, and Graham M Lord (Feb. 2017a). "Genetic variants alter T-bet binding and gene expression in mucosal inflammatory disease." In: *PLoS genetics* 13, e1006587. ISSN: 1553-7404. DOI: 10.1371/journal.pgen.1006587.

Soderquest, Katrina, Arnulf Hertweck, **Giambartolomei, C.**, Stephen Henderson, Rami Mohamed, Rimma Goldberg, Esperanza Perucha, Lude Franke, Javier Herrero, Vincent Plagnol, et al. (2017b). "Genetic variants alter T-bet binding and gene expression in mucosal inflammatory disease". In: *PLoS genetics* 13.2, e1006587.

Role: analysis of eQTL data.

2016 Franzén, Oscar, Raili Ermel, Ariella Cohain, Nicholas K Akers, Antonio Di Narzo, Husain A Talukdar, Hassan Foroughi-Asl, **Giambartolomei, C.**, John F Fullard, Katyayani Sukhavasi, Sulev Köks, Li-Ming Gan, Chiara Giannarelli, Jason C Kovacic, Christer Betsholtz, Bojan Losic, Tom Michoel, Ke Hao, Panos Roussos, Josefin Skogsberg, Arno Ruusalepp, Eric E Schadt, and Johan L M Björkegren (Aug. 2016). "Cardiometabolic risk loci share downstream cis- and trans-gene regulation across tissues and diseases." In: *Science (New York, N.Y.)* 353, pp. 827–830. ISSN: 1095-9203. DOI: 10.1126/science.aad6970.

Role: Bioinformatics and statistical analysis for integration of STARNET eQTL with GWAS risk loci and epigenome data.

Fullard, J.F., T. B Halene, **Giambartolomei, C.**, V. Haroutunian, S. Akbarian, and P. Roussos (Nov. 2016). "Understanding the genetic liability to schizophrenia through the neuroepigenome." In: *Schizophrenia research* 177, pp. 115–124. ISSN: 1573-2509. DOI: 10.1016/j.schres.2016.01.039.

Role: analysis and write-up.

McLachlan, S., **Giambartolomei, C.**, J. White, P. Charoen, Andrew Wong, Chris Finan, Jorgen Engmann, Tina Shah, Micha Hersch, Clara Podmore, Alana Cavadino, Barbara J Jefferis, Caroline E Dale, Elina Hypponen, Richard W Morris, Juan P Casas, Meena Kumari, Yoav Ben-Shlomo, Tom R Gaunt, Fotios Drenos, Claudia Langenberg, Diana Kuh, Mika Kivimaki, Rico Rueedi, Gerard Waeber, Aroon D Hingorani, Jacqueline F Price, Ann P Walker, and UCLEB Consortium (2016). "Replication and Characterization of Association between ABO SNPs and Red Blood Cell Traits by Meta-Analysis in Europeans." In: *PloS one* 11, e0156914. ISSN: 1932-6203. DOI: 10.1371/journal.pone.0156914.

Role: eQTL and integrative analyses.

White, Jon, Reecha Sofat, Gibran Hemani, Tina Shah, Jorgen Engmann, Caroline Dale, Sonia Shah, Felix A Kruger, **Giambartolomei, C.**, Daniel I Swerdlow, Tom Palmer, Stela McLachlan, Claudia Langenberg, Delilah Zabaneh, Ruth Lovering, Alana Cavadino, Barbara Jefferis, Chris Finan, Andrew Wong, Antoinette Amuzu, Ken Ong, Tom R Gaunt, Helen Warren, Teri-Louise Davies, Fotios Drenos, Jackie Cooper, Shah Ebrahim, Debbie A Lawlor, Philippa J Talmud, Steve E Humphries, Christine Power, Elina Hypponen, Marcus Richards, Rebecca Hardy, Diana Kuh, Nicholas Wareham, Yoav Ben-Shlomo, Ian N Day, Peter Whincup, Richard Morris, Mark W J Strachan, Jacqueline Price, Meena Kumari, Mika Kivimaki, Vincent Plagnol, John C Whittaker, International Consortium for Blood Pressure (ICBP), George Davey Smith, Frank Dudbridge, Juan P Casas, Michael V Holmes, Aroon D Hingorani, and UCLEB (University College London-London School of Hygiene & Tropical Medicine-Edinburgh-Bristol Consortium (Apr. 2016). "Plasma urate concentration and risk of coronary heart disease: a Mendelian randomisation analysis." In: *The lancet. Diabetes & endocrinology* 4, pp. 327–336. ISSN: 2213-8595. DOI: 10.1016/S2213-8587(15)00386-1.

Role: meta-analysis.

Zheng, Jie, Santiago Rodriguez, Charles Laurin, Denis Baird, Lea Trela-Larsen, Mesut A Erzurumluoglu, Yi Zheng, Jon White, **Giambartolomei, C.**, Delilah Zabaneh, Richard Morris, Meena Kumari, Juan P Casas, Aroon D Hingorani, UCLEB Consortium, David M Evans, Tom R Gaunt, and Ian N M Day (Sept. 2016). "HAPRAP: a haplotype-based iterative method for statistical fine mapping using GWAS summary statistics." In: *Bioinformatics (Oxford, England)*. ISSN: 1367-4811. DOI: 10.1093/bioinformatics/btw565.

Role: meta-analysis.

2015 Nueesch, E., C. Dale, T.M. Palmer, J. White, B.J. Keating, E.P. van Iperen, A. Goel, S. Padmanabhan, F. W. Asselbergs, E. P. I. C-Netherland Investigators, and Verschuren , ... **Giambartolomei, C**. et al. (2015). "Adult height, coronary heart disease and stroke: a multi-locus Mendelian randomization meta-analysis." In: *Int J Epidemiol*.

Role: meta-analysis.

Oldoni, F., J. Palmen, **Giambartolomei, C.**, P. Howard, F. Drenos, V. Plagnol, S. E. Humphries, P. J. Talmud, and A. J. P. Smith (2015). "Post-GWAS methodologies for localisation of functional non-coding variants: ANGPTL3." eng. In: *Atherosclerosis* 246, pp. 193–201. DOI: 10.1016/j.atherosclerosis.2015.12.009.

Role: analysis of eQTL data.

Swerdlow, Daniel I et al. (Jan. 2015). "HMG-coenzyme A reductase inhibition, type 2 diabetes, and bodyweight: evidence from genetic analysis and randomised trials." In: *Lancet (London, England)* 385, pp. 351–361. ISSN: 1474-547X. DOI: 10.1016/S0140-6736(14)61183-1.

Role: analysis of eQTL data and meta-analysis.

Talmud, Philippa J, Jackie A Cooper, Richard W Morris, Frank Dudbridge, Tina Shah, Jorgen Engmann, Caroline Dale, Jon White, Stela McLachlan, Delilah Zabaneh, Andrew Wong, Ken K Ong, Tom Gaunt, Michael V Holmes, Debbie A Lawlor, Marcus Richards, Rebecca Hardy, Diana Kuh, Nicholas Wareham, Claudia Langenberg, Yoav Ben-Shlomo, S Goya Wannamethee, Mark W J Strachan, Meena Kumari, John C Whittaker, Fotios Drenos, Mika Kivimaki, Aroon D Hingorani, Jacqueline F Price, Steve E Humphries, and UCLEB Consortium (May 2015). "Sixty-five common genetic variants and prediction of type 2 diabetes." In: *Diabetes* 64, pp. 1830–1840. ISSN: 1939-327X. DOI: 10.2337/db14-1504.

2014 Giambartolomei, Claudia, Damjan Vukcevic, Eric E Schadt, Lude Franke, Aroon D Hingorani, Chris Wallace, and Vincent Plagnol (May 2014). "Bayesian test for colocalisation between pairs of genetic association studies using summary statistics." In: *PLoS genetics* 10, e1004383. ISSN: 1553-7404. DOI: 10.1371/journal.pgen.1004383.

Gierach, G. L., Lui Hui, J. T. Loud, M. H. Greene, C. K. Chow, Li Lan, S. A. Prindiville, J. Eng-Wong, P. W. Soballe, **Giambartolomei, C.**, Phuong L. Mai, C. E. Galbo, K. Nichols, K. A. Calzone, Olufunmilayo I Olopade, M. H. Gail, and M. L. Giger (2014). "Relationships between Computer-extracted Mammographic Texture Pattern Features and BRCA1/2 Mutation Status: A Cross-Sectional Study". In: *Breast Cancer Research*.

Role: assisted in data collection procedures and management.

Holmes, M.V., F.W. Asselbergs, T.M. Palmer, F. Drenos, M.B. Lanktree, C.P. Nelson, C.E. Dale, S. Padmanabhan, C. Finan, D.I. Swerdlow, V. Tragante, E.P.A. van Iperen, S. Sivapalaratnam, S. Shah, C.C. Elbers, T. Shah, J. Engmann, and **Giambartolomei C.** et al. (2014). "Mendelian randomization of blood lipids for coronary heart disease." In: *Eur Heart J*.

Role: analysis of eQTL data.

2013 Lopes, L. R., A. Zekavati, P. Syrris, M. Hubank, **Giambartolomei, C.**, C. Dalageorgou, S. Jenkins, W. McKenna, Uk10k Consortium, V. Plagnol, and P. M. Elliott (2013). "Genetic complexity in hypertrophic cardiomyopathy revealed by high-throughput sequencing." In: *J Med Genet* 50.4, pp. 228–239.

Role: analysis of exome sequencing data.

Shah, T., J. Engmann, C. Dale, S. Shah, J. White, **Giambartolomei, C.**, and U. C. L. E. B Consortium (2013). "Population genomics of cardiometabolic traits: design of the University College London-London School of Hygiene and Tropical Medicine-Edinburgh-Bristol (UCLEB) Consortium." In: *PLoS One* 8.8, e71345.

Role: single study associations and meta-analyses of cardiometabolic traits.

Sovio, U., **Giambartolomei, C.**, S. Kinra, L. Bowen, F. Dudbridge, D. Nitsch, G.D. Smith, S. Ebrahim, and Y. Ben-Shlomo (2013). "Early and current socio-economic position and cardiometabolic risk factors in the Indian Migration Study." In: *Eur J Prev Cardiol* 20.5, pp. 844–853.

Role: analysis of eQTL data.

2012 Borges, J. D., V. Souza, **Giambartolomei, C.**, F. Dudbridge, W.S. Freire, S.A. Gregorio, P.P.Q. Torrez, M. Quiroga, P. Mayaud, C.S. Pannuti, and M.C. Nascimento (2012). "Transmission of human herpesvirus type 8 infection within families in american indigenous populations from the Brazilian Amazon." In: *J Infect Dis* 205.12, pp. 1869–1876.

Role: statistical analysis.

Gupta, V., D. G. Vinay, S. Rafiq, M. V. Kranthikumar, C. S. Janipalli, **Giambartolomei, C.**, D. M. Evans, K. R. Mani, M. N. Sandeep, A. E. Taylor, S. Kinra, R. M. Sullivan, L. Bowen, N. J. Timpson, G. D. Smith, F. Dudbridge, D. Prabhakaran, Y. Ben-Shlomo, K. S. Reddy, S. Ebrahim, G. R. Chandak, and Indian Migration Study Group (2012). "Association analysis of 31 common polymorphisms with type 2 diabetes and its related traits in Indian sib pairs." In: *Diabetologia* 55.2, pp. 349–357.

Role: preparatory analysis of genetic and clinical data.

Smith, Andrew J. P., P. Howard, S. Shah, P. Eriksson, S. Stender, **Giambartolomei, C.**, L. Folkersen, A. Tybjærg-Hansen, M. Kumari, J. Palmen, A.D. Hingorani, P. J. Talmud, and S.E. Humphries (2012). "Use of allele-specific FAIRE to determine functional regulatory polymorphism using large-scale genotyping arrays." In: *PLoS Genetics* 8.8, e1002908.

Role: integrative analysis of FAIRE data.

2011 Taylor, A. E., M. N. Sandeep, C. S. Janipalli, **Giambartolomei, C**., D. M. Evans, M. V. Kranthi Kumar, D. G. Vinay, P. Smitha, V. Gupta, M. Aruna, S. Kinra, R. M. Sullivan, L. Bowen, N. J. Timpson, G. Davey Smith, F. Dudbridge, D. Prabhakaran, Y. Ben-Shlomo, K. S. Reddy, S. Ebrahim, and G. R. Chandak (2011). "Associations of FTO and MC4R Variants with Obesity Traits in Indians and the Role of Rural/Urban Environment as a Possible Effect Modifier." In: *J Obes* 2011, p. 307542.

Role: preliminary analysis of genetic and clinical data.

2010 Gierach, G.L., J.T. Loud, C.K. Chow, S.A. Prindiville, J. Eng-Wong, P.W. Soballe, **Giambartolomei, C.**, P.L. Mai, C.E. Galbo, K. Nichols, K.A. Calzone, C. Vachon, M.H. Gail, and M.H. Greene (2010). "Mammographic density does not differ between unaffected BRCA1/2 mutation carriers and women at low-to-average risk of breast cancer." In: *Breast Cancer Res Treat* 123.1, pp. 245–255.

Role: assisted in data collection procedures and management.

2009 **Giambartolomei, C.**, C.M. Mueller, M.H. Greene, and L.A. Korde (2009). "A mini-review of familial ovarian germ cell tumors: an additional manifestation of the familial testicular germ cell tumor syndrome." In: *Cancer Epidemiol* 33.1, pp. 31–36.

Role: retrieved primary data, carried out the analysis, and wrote it as primary author.

Abstracts and Posters

- Alexandros Armaos, Ji-Heui Seo, Davide Cirillo, Francois Serra, Alfonso Valencia, Matthew L. Freedman, Claudia Giambartolomei, Gian Gaetano Tartaglia, "Reconstructing protein interactions at enhancer-promoter regions in prostate cancer " Abstract and poster, Virtual Meeting of the American Society of Human Genetics (ASHG), 2020
- Claudia Giambartolomei, Ji-Heui Seo, Nicholas Mancuso, Malika Kumar Freund, Xintao Qui, Alexander Gusev, Matthew Freedman, Bogdan Pasaniuc, "Gene prioritization using HiChiP and eQTL in prostate cancer" Abstract and poster, 68th Annual Meeting of the American Society of Human Genetics (ASHG), 2018
- Claudia Giambartolomei, Nicholas Mancuso, Huwenbo Shi, Ben Strober, Alexis Battle, Bogdan Pasaniuc, "Enhanced methods to investigate the role of trans-eQTL to complex traits." Abstract and poster, 67nd Annual Meeting of the American Society of Human Genetics (ASHG), 2017
- Claudia Giambartolomei, Nora Franceschini, Chris O' Donnell, JP Casas, Johan LM Bjorkegren, Bogdan Pasaniuc, "Multiple trait co-localizations to characterize CAD loci." Abstract and poster, CHARGE Boston conference, 2017
- L. Rocha Lopes, C. Giambartolomei, P. Syrris, C. O'Mahony, C. Dalageorgou, S. Jenkins, M. Hubank, W. Mc Kenna, V. Plagnol, P. Elliott, "High-throughput genotyping and phenotyping reveals new genetic determinants of clinical phenotype in hypertrophic cardiomyopathy", European Society of Cardiology, 2013
- C. Giambartolomei, H.-J. Westra, M. Kivimaki, M. Kumari, E. Schadt, L. Franke, A. Hingorani, V. Plagnol. "Co-localization of lipid biomarker associations with gene expression across human tissues." Abstract and poster, 62nd Annual Meeting of the American Society of Human Genetics (ASHG), 2012
- Andrew J. P. Smith, Fotios Drenos, Philip Howard, Claudia Giambartolomei, Philippa J. Talmud, Vincent Plagnol, Steve E. Humphries, "Chipping a hole-in-one from the FAIRE way: use of post-GWAS fine-mapping genotyping arrays for functional variant discovery", ASHG, 2012

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on 23/10/2023 at 14:03:37
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