

Carlo Berzuini

curriculum

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Key Areas of Interest

Carlo Berzuini develops and uses causal inference, machine learning and statistical methods in the study of genetic/molecular mechanisms of cardiovascular, renal, inflammatory and other types of disease. He enhances classical epidemiological methods for integrating evidence from multiple data types (observational, experimental, human phenotypic, genomic, epigenomic, bioinformatic, cell biology, clinical knowledge) within a proper (Bayesian or frequentist) causal/predictive inference scheme. He has experience in the analysis of large datasets, such as UK Biobank, UKBEC, QTE, pedigree banks and others.

Bio

Carlo Berzuini is currently holding a Research Chair in Biostatistics at the University of Manchester, United Kingdom, since 2011.

Previously, he has been Professor of Biostatistics (1983-2008) at the University of Pavia, Italy, and he has then moved to UK Academia, where he has worked in the capacity of Research Project PI at the Faculty of Mathematics of the University of Cambridge (2009-2011), United Kingdom, and won an Isaac Newton Institute Award for research in Causal Inference.

He has also worked at Cambridge's Medical Research Council Biostatistics Unit, where he fruitfully collaborated with D.Clayton, D.Cox, A.P. Dawid, F. Dudbridge, W.Gilks and D.Spiegelhalter, and where he established a team of statistical geneticists involved in the EU-funded pre-2007 Bloodomics Consortium. He has been scientific advisor at the Department of Gastroenterology of the University of Cambridge, plus member of the Cambridge Inflammatory Bowel Disease Research Group and of the International Inflammatory Bowel Disease Genetics

Consortium. His peer reviewing activity for scientific journals is documented in Publons. In addition, he has been statistical reviewer for such research funding bodies as MRC and NIHR.

See also <https://orcid.org/0000-0001-6056-0489>.

Carlo Berzuini has taught courses of Modelling of Complexly Structured Data and Causal Inference in numerous academic venues, notably Cambridge, Manchester, Pavia, Leiden, and Aarhus.

Carlo Berzuini has been at the forefront of landmark methodological developments in Bayesian modelling and inference, as a pioneer of particle-based Markov chain Monte Carlo (MCMC) methods for real time inference, and as the originator of the first (1996) practical method for a joint analysis of longitudinal and event history data, with medical applications. He has been an early developer of Bayesian MCMC approaches in disease monitoring, medical and epidemiological prediction, and of structural biology. He has a deep understanding of data science foundations. He is a leading expert in Statistical Genomics and Causal Inference, with notable contributions in Causative Interaction, Mediation and Mendelian Randomization.

In collaboration with neuroscientists of the universities of Manchester and Princeton, he is responsible for publishing on the journal *Cognition* the first example of a statistical framework for studying inter-individual differences in the relationship between expectation and pain perception, and he is now extending this to the study of autism.

In 2009 he organized in Cambridge the first major international conference on Causal Inference, jointly with L. Bernardinelli and A.P. Dawid. He lead-edited a landmark and successful book on Statistical Causality, for Wiley. He has a strong record of interdisciplinary research in a wide spectrum of substantive areas, and in 2014 his collaboration with Cambridge Abcodia Ltd for developing a biomarker-based approach to early diagnosis of pancreatic cancer earned him a Horizon 2020 nomination.

He counts about 140 peer-reviewed papers, including interdisciplinary publications on such journals as Nature, Nature Genetics, Nature Communications, Journal of the American College of Cardiology, Circulation, Cognition, Journal of Alzheimer's Disease, Multiple Sclerosis Journal, Gastroenterology, Diabetes, Cancer Research, British Journal of Cancer, European Journal of Neuroscience, Journal of Alzheimer's Disease, Cytometry and Journal of Molecular Biology. His more theoretical work has appeared on Biometrika, Statistics in Medicine, the Journal of the American Statistical Association (Theory and Methods), the Journals of the Royal Statistical Society, Series A, B and C, Bioinformatics, Genetic Epidemiology, Biostatistics, IEEE Transactions in Biomedical Engineering and IEEE Transactions in Pattern Analysis and Machine Intelligence, among others. He has participated with Principal Investigator capacity in European Research Projects.

In 2007-2013 he was proponent and chair of the Royal Statistical Society Study Group in Bioinformatics, and Member of the Royal Statistical Society Coun-

cil. Besides tons of undergraduate teaching, he has given courses in Bayesian modelling and causal inference all over Europe, for master and post doctoral students, for researchers in statistics and medical researchers, and is regularly giving invited lectures in top scientific meetings. He is the author of publicly available R and **Stan** software.

Employment

- University of Manchester: Manchester, United Kingdom, from 2011 to present. Employed as Research Professor in Biostatistics, at the Centre for Biostatistics. Institute of Population Health. Jean McFarlane Building, Oxford Road. Manchester, M13 9PL.
- University of Cambridge: Cambridge, United Kingdom, From 2008 to 2011. Position funded by the Isaac Newton Award I won for research in Causal Inference in Medicine, to be carried out at the Faculty of Mathematical Sciences (Statistical Laboratory)
- University of Pavia: Pavia, Italy. From 1983 to 2008, employed as Professor of Biostatistics, at the Department of Computer and Systems Science.
- University of Pavia: Pavia, Italy. From 1974 to 1983. Employed as Researcher in Artificial Intelligence and Statistics with teaching capacity, at the Department of Computer and Systems Science.

Education and qualifications

- University of Pavia: Pavia, Italy, from 1 September 1969 to 1974. Doctor in Computers and Systems Science (equivalent to current PhD title)

Invited positions

- Doctorate in Medical and Genomic Statistics of the University of Pavia: Pavia, from 2013 to present. In the capacity of Teacher and Faculty Member
- Master Course in Genetic and Molecular Epidemiology of the University of Pavia: Pavia, from 2010 to 2012, in the capacity of Teacher and Faculty member
- University of Cambridge: Cambridge, United Kingdom. From 2004 to 2010. Scientific Advisor at the Department of Gastroenterology.
- Biostatistics Unit of the Medical Research Council: Cambridge, United Kingdom. From 2001 to 2008. Invited to act as Project Leader (EU funded BLOODOMICS Project)(see 2006 MRC Biostatistics Unit QQR Review Committee Report).

Membership and service

- International Inflammatory Bowel Disease Genetics Consortium: London, United Kingdom. From 2004. Member, expert in Statistical Genetics and Statistics
- Publons (Clarivate Analytics): Philadelphia, PA, United States, from 1990 to present: Recognized reviewer, with a record of countless peer reviews for top statistical, bioinformatic and scientific journals, most recently for Nature Communications, Biometrika, Biostatistics, Statistics in Medicine, International Journal of Epidemiology, Journal of the American Heart Association and Statistica Sinica.
- Uncertainty in Artificial Intelligence International Conference Scientific Committee: Cambridge, from 2012 to 2018: Member of Scientific Committee
- Artificial Intelligence and Statistics Conference Scientific Committee: Cambridge, from 2014 to 2015: Member of Scientific Committee
- One-day Workshop on "The Statistical Contributions of A. Philip Dawid: Causal Inference, Graphical Models and Prediction": Cambridge, 30 April 2014. Organizer (Faculty of Mathematical Sciences, University of Cambridge)
- Royal Statistics Society Bioinformatics Study Group: London and Cambridge, From 2007 to 2013, Proponent and Chair
- Cambridge Inflammatory Bowel Disease Research Group: Cambridge, from 2004 to 2010: Member, statistician (Department of Gastroenterology, University of Cambridge)
- Causal Inference International Conference: Cambridge. 2009. Organizer (Statistical Laboratory, Faculty of Mathematical Sciences) jointly with L. Bernardinelli and A.P. Dawid.
- Royal Statistical Society: London, GB2006 to 2008: RSS Council Member

Funding

- Co-applicant in the proposed ERC project Infarctomics, jointly with Dr. Diego Ardissino, Director of Cardiology Department in the Parma Hospital, Italy, and member of the Italian Research Consortium Amici della Cardiologia European Research Council (Parma) 2019 to present. Grant
- Participant in an application for a Manchester University BHF Centre of Excellence in Integrative Genomics of Cardiovascular Disease British Heart Foundation (Manchester) 2019 to present—Award

- Functional Mechanisms Underlying Genomewide Associations with Chronic Kidney Disease and its Defining Traits Kidney Research UK (Manchester) 2018 to 2020—Grant GRANT NUMBER: R122698
- Translating Signals from Genome-Wide Association Studies into Biological Mechanisms of Hypertension - Expression Quantitative Trait Locus Analysis in the Human Kidney. British Heart Foundation (Manchester) 2017 to present—Grant GRANT NUMBER: R121157
- Methods for Integrated analysis of multiple Omics datasets (MIMOMICS). European Commission (Leiden) 2013 to 2018. GRANT NUMBER: FP7-HEALTH-2012-INNOVATION
- Contract for the development of a statistical method and a system for "Early Dynamic Detection of Pacreatic Adenocarcinoma", with ABCODIA Ltd (London). 2012 to 2015
- Award from the Isaac Newton Trust for research in statistical methods for causal inference at the Centre for Mathematical Science of Cambridge University. 2011 to 2012
- Genetic Variation, Disease Prediction and Causation. Funded by Research Council (London) 2009 to 2012. GRANT NUMBER: G0802320

Sample of post-1993 Works (45 out of 140)

- Long-Term Outcomes After Early-Onset Myocardial Infarction Journal of the American College of Cardiology JACC (in press) 2019
- Uncovering novel genetic mechanisms of hypertension Nature (submitted for publication) jointly with Maciej Tomaszewski et al, 2019
- Acid Sensing Ion Channel 2: a new potential player in the pathophysiology of multiple sclerosis European Journal of Neuroscience, 2018
- Molecular insights into genome-wide association studies of chronic kidney disease-defining traits. Nature communications, 2018. PMID: 30467309 DOI: 10.1038/s41467-018-07260-4
- Sensitivity to pain expectations: A Bayesian model of individual differences. Cognition, 2018 PMID: 30243037 DOI: 10.1016/j.cognition.2018.08.022
- Mendelian randomisation analysis of clustered causal effects of body mass on cardiometabolic biomarkers. BMC bioinformatics 2018-07, PMID: 30066639 PMC: PMC6069804. DOI: 10.1186/s12859-018-2178-2
- Women's Pregnancy Life History and Alzheimer's Risk: Can Immunoregulation Explain the Link? American journal of Alzheimer's disease and other dementias 2018l PMID: 30060670. DOI:10.1177/1533317518786447

- Bayesian Mendelian Randomization for incomplete pedigree data, and the characterisation of Multiple Sclerosis proteins. ArXiv (submitted for publication to an Epidemiology Journal) 2018.
- Investigating multiple sclerosis genetic susceptibility on the founder population of east-central Sardinia via association and linkage analysis of immune-related loci. Multiple sclerosis (Houndmills, Basingstoke, England) 2017, PMID: 28933650. DOI: 10.1177/1352458517732841
- Ovarian Cancer Follow-up: A Preliminary Comparison of 2 Approaches. International journal of gynecological cancer : official journal of the International Gynecological Cancer Society 2017, PMID: 28002208.
- Systematic analysis of circulating soluble angiogenesis-associated proteins in ICON7 identifies Tie2 as a biomarker of vascular progression on bevacizumab. British journal of cancer 2016, PMID: 27351218.
- Stochastic mechanistic interaction, Biometrika 2016, DOI: 10.1093/biomet/asv072.
- Causality: Statistical Perspectives and Applications by Carlo Berzuini, Philip Dawid, and Luisa Bernardinelli, Wiley Series in Probability and Statistics 2012, book, DOI: 10.1002/9781119945710
- The combination of circulating Ang1 and Tie2 levels predicts progression-free survival advantage in bevacizumab-treated patients with ovarian cancer. Clinical cancer research : an official journal of the American Association for Cancer Research 2014, PMID: 24947924PMC: PMC4154862.
- Association between variants of PRDM1 and NDP52 and Crohn's disease, based on exome sequencing and functional studies. Gastroenterology 2013, PMID: 23624108, DOI: 10.1053/j.gastro.2013.04.040
- Maternal breastfeeding history and Alzheimer's disease risk. Journal of Alzheimer's disease : JAD 2013, PMID: 23948914
- Deep determinism and the assessment of mechanistic interaction. Biostatistics (Oxford, England) 2012, PMID: 23255363PMC: PMC3677734, DOI: 10.1093/biostatistics/kxs049
- Direct genetic effects and their estimation from matched case-control data. Genetic epidemiology 2012, PMID: 22829130, DOI: 10.1002/gepi.21660
- Analysis of Interaction for Identifying Causal Mechanisms in the above ref book Causality , 2012-06 DOI: 10.1002/9781119945710.ch14
- ibidem: Assessing Dynamic Treatment Strategies DOI:10.1002/9781119945710.ch8
- ibidem: Ion Channels as a Possible Mechanism of Neurodegeneration in Multiple Sclerosis 2012-06, DOI: 10.1002/9781119945710.ch15

- Influence of 9p21.3 genetic variants on clinical and angiographic outcomes in early-onset myocardial infarction. *Journal of the American College of Cardiology*, 2011, PMID: 21757122DOI: 10.1016/j.jacc.2010.11.075
- Genetic association between NLRP3 variants and Crohn's disease does not replicate in a large UK panel. *Inflammatory bowel diseases* 2010, PMID: 21560198. DOI: 10.1002/ibd.21499
- Association between protective and deleterious HLA alleles with multiple sclerosis in Central East Sardinia. *PloS one* 2009, PMID: 19654877PMC: PMC2716537DOI: 10.1371/journal.pone.0006526 Source: Europe PubMed Central
- Genome-wide association of early-onset myocardial infarction with single nucleotide polymorphisms and copy number variants. *Nature genetics* 2009, PMID: 19198609PMC: PMC2681011DOI: 10.1038/ng.327
- Contribution of TNFSF15 gene variants to Crohn's disease susceptibility confirmed in UK population. *Inflammatory bowel diseases* 2008m PMID: 18338776DOI: 10.1002/ibd.20399
- Genetic determinants of ulcerative colitis include the ECM1 locus and five loci implicated in Crohn's disease. *Nature genetics* 2008, PMID: 18438406PMC: PMC2719289DOI: 10.1038/ng.145
- Mapping the platelet profile for functional genomic studies and demonstration of the effect size of the GP6 locus. *Journal of thrombosis and haemostasis* : JTH 2007m PMID: 17663743.
- Association between the ACCN1 gene and multiple sclerosis in Central East Sardinia. *PloS one* 2007, PMID: 17534430PMC
- IL23R variation determines susceptibility but not disease phenotype in inflammatory bowel disease. *Gastroenterology* 2007, PMID: 17484863PMC: PMC2696256.
- A novel prognostic index to determine the impact of cardiac conditions and co-morbidities on one-year outcome in patients with heart failure. *The American journal of cardiology* 2006, PMID: 17027575.
- Predicting the strongest domain-domain contact in interacting protein pairs. *Statistical applications in genetics and molecular biology* 2006, PMID: 16646869. DOI: 10.2202/1544-6115.1195
- Effectiveness of potent antiretroviral therapy on progression of human immunodeficiency virus: Bayesian modelling and model checking via counterfactual replicates *Journal of the Royal Statistical Society: Series C (Applied Statistics)* 2004. DOI: 10.1111/j.1467-9876.2004.04985.x

- Statistical analysis of domains in interacting protein pairs. *Bioinformatics* 2004, PMID: 15509600
- Supra-domains: evolutionary units larger than single protein domains. *Journal of molecular biology* 2004, PMID: 15095989.
- Bayesian trio models for association in the presence of genotyping errors. *Genetic epidemiology* 2004m PMID: 14691958DOI: 10.1002/gepi.10291
- RESAMPLE-MOVE Filtering with Cross-Model Jumps Sequential Monte Carlo Methods in Practice 2001, DOI: 10.1007/978-1-4757-3437-9 6Part of ISBN: 9781441928870
- Value of right ventricular ejection fraction in predicting short-term prognosis of patients with severe chronic heart failure. *The Journal of heart and lung transplantation : the official publication of the International Society for Heart Transplantation* 1997, PMID: 9257260
- Dynamic Conditional Independence Models and Markov Chain Monte Carlo Methods *Journal of the American Statistical Association* 1997 DOI: 10.1080/01621459.1997.10473661Part of ISSN: 0162-1459
- Dynamic Conditional Independence Models and Markov Chain Monte Carlo Methods *Journal of the American Statistical Association* 1997, DOI: 10.2307/2965410Part of ISSN: 0162-1459
- Interaction between exercise training and ejection fraction in predicting prognosis after a first myocardial infarction. *Circulation* 1996, PMID: 8790035DOI: 10.1161/01.cir.94.5.978
- Bayesian analysis of survival on multiple time scales. *Statistics in medicine* 1994. PMID: 8047738DOI: 10.1002/sim.4780130804
- Predictors of prognosis in patients awaiting heart transplantation. *The Journal of heart and lung transplantation : the official publication of the International Society for Heart Transplantation* 1993, PMID: 8241212
- Bayesian networks for patient monitoring *Artificial Intelligence in Medicine* 1992, DOI: 10.1016/0933-3657(92)90030