Professor Tom Gaunt
B.Sc., Ph.D.(Soton.)

Professor of Health and Biomedical Informatics

Area of research

Molecular genetics and bioinformatics

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Summary

Biography

Following an undergraduate degree in Biology, I worked as a researcher in the Human Genetics Division at the University of Southampton, during which time I completed my PhD in Human Genetics. In 2005 I was awarded a British Heart Foundation Intermediate Fellowship to pursue my interests in cardiovascular genetic epidemiology, and in 2006 I secured a lectureship at the University of Bristol. I have subsequently built an interdisciplinary research team in the MRC Integrative Epidemiology Unit working on the application of bioinformatics and data mining approaches in epidemiology.

Research interests

My research interests lie in the development and application of informatics approaches to molecular and non-molecular data in population health sciences. I am involved in a wide range of different projects and am always interested in hearing from potential PhD students or postdoctoral researchers. A selection of my research interests:

Causal inference

The MR-Base platform aims to systematise causal inference using Mendelian randomization [Gib Hemani, Philip Haycock and Jie (Chris) Zheng]. MR-Base integrates an extensive database of genome-wide association study data with Mendelian randomization methods in both a user-friendly web application and a comprehensive R package. We are extending this work to include systematic causal analysis of a wide array of risk factors and diseases, and integration with other types of data to improve causal inference.

Literature mining

The MELODI platform aims to mine mechanistic pathways from the biomedical literature [Ben Elsworth]. The software searches for overlapping terms between two literature sets that represent two different entities (eg a risk factor and a disease). Enriched overlapping terms may represent candidate mechanisms for further investigation. MELODI is paralleled by the TeMMPo platform (developed in collaboration with WCRF), which assesses the literature for number of publications underpinning hypothesised mechanistic pathways.

Epigenetics

As co-I of the BBSRC-funded ARIES project I led the bioinformatics workpackage in generating, QC'ing and normalizing the data, and have subsequently been involved in over 20 papers utilizing these data (including a major methylation QTL analysis published in Genome Biology in 2016). The methylation QTL derived from the ARIES data are presented in our online mQTLdb, and ongoing work with the GoDMC consortium will substantially extend the scale of this analysis.

Machine learning

I have interests in the application of machine learning approaches to molecular data, and (with Colin Campbell) have published tools that predict the functional effects of genetic variants (the widely-used FATHMM suite of tools), haploinsufficiency (HIPred) and breast cancer survival (FS-MKL).

Other software

Other software tools I have overseen include: FATHMM (Shihab), mQTLdb (Shihab), TeMMPo and GTB (Shihab) (see MRC-IEU software page).

See my Scopus and Google Scholar pages for publications.

Research group and funding

My group currently comprises 5 postdoctoral research associates and 3 PhD students. I co-lead the Bioinformatics and Data Mining theme in the MRC Integrative Epidemiology Unit, and have PI funding for collaborative projects with GlaxoSmithKline, Biogen and the CHDI foundation. As co-investigator on the CRUK Integrative Cancer Epidemiology programme I lead a bioinformatics cross-cutting strand, and as a co-investigator on the Bristol NIHR Biomedical Research Centre I co-lead a work-strand within the Translational Population Sciences theme. I am a co-investigator and Executive Board member on the Wellcome Trust strategic award for the ALSPAC cohort.

Teaching

MB ChB programme

- 1st year *Clinical Epidemiology* (Human Basis of Medicine)
- 4th year *Community Orientated Medical Practice I*: Genetic Epidemiology and Critical Appraisal

Short courses

- Basic Genetics and Genomics
- Genetic Association Studies
- Advanced Genetics and Genomics

Expertise

Molecular Genetics and Genetic Epidemiology of cardiovascular disease and obesity: I am funded by the British Heart Foundation and Medical Research Council to investigate the role of genes in heart disease and related traits in the British Women's Heart and Health Study. Bioinformatics and computational biology, including: integration of genome-wide association study data, identification of Splicing and Translational Efficiency Polymorphisms (STEPs), pathway informatics and LD/haplotype analysis.

- molecular genetics
- cardiovascular disease
- obesity
- genes
- bioinformatics
- genome-wide association

Memberships

Organisations

Bristol Medical School (PHS)

Other sites

- Bhi
- Populationhealth

Centres, collaborations and units

- MRC Centre for Causal Analyses in Translational Epidemiology
- MRC Integrative Epidemiology Unit

Links

- Group website

Recent publications

- Kazmi, N, Gaunt, T, Koda, Y, Ndiaye, N, Visvikis-Siest, S, Morton, M & Galea, I, 2019, 'Genetic determinants of circulating haptoglobin concentration'. *Clinica Chimica Acta*.
- Hatcher, C, Relton, C, Gaunt, T & Richardson, T, 2019, 'Leveraging brain cortex-derived molecular data to elucidate epigenetic and transcriptomic drivers of complex traits and disease'. *Translational Psychiatry*, vol 9.
- Taylor, K, Smith, GD, Relton, C, Gaunt, T & Richardson, T, 2019, 'Prioritizing putative influential genes in cardiovascular disease susceptibility by applying tissue-specific Mendelian randomization'. *Genome Medicine*, vol 11.
- Guyatt, AL, Brennan, RR, Burrows, K, Guthrie, PAI, Ascione, R, Ring, SM, Gaunt, TR, Pyle, A, Cordell, HJ, Lawlor, DA, Chinnery, PF, Hudson, G & Rodriguez, S, 2019, 'A genome-wide association study of mitochondrial DNA copy number in two population-based cohorts'. *Human Genomics*, vol 13.
- Millard, L, Davies, N, Tilling, K, Gaunt, T & Smith, GD, 2019, 'Searching for the causal effects of body mass index in over 300 000 participants in UK Biobank, using Mendelian randomization'. *PLoS Genetics*, vol 15.
- , Tachmazidou, I, Hatzikotoulas, K, Southam, L, Esparza-Gordillo, J, Haberland, V, Zheng, J, Johnson, T, Koprulu, M, Zengini, E, Steinberg, J, Wilkinson, JM, Bhatnagar, S, Hoffman, JD, Buchan, N, Süveges, D, Yerges-Armstrong, L, Smith, GD, Gaunt, TR, Scott, RA, McCarthy, LC & Zeggini, E, 2019, 'Identification of new therapeutic targets for osteoarthritis through genome-wide analyses of UK Biobank data'. *Nature Genetics*, vol 51., pp. 230-236
- Haworth, S, Shapland, CY, Hayward, C, Prins, BP, Felix, JF, Medina-Gomez, C, Rivadeneira, F, Wang, C, Ahluwalia, TS, Vrijheid, M, Guxens, M, Sunyer, J, Tachmazidou, I, Walter, K, Iotchkova, V, Jackson, A, Cleal, L, Huffmann, J, Min, JL, Sass, L, Timmers, PR, , Al Turki, S, Anderson, CA, Boustred, C, Curran, S, Day, IN, Evans, DM, Gaunt, TR, Kemp, JP, Lawson, D, Northstone, K, Paternoster, L, Ring, S, Shihab, HA, Shin, SY, Soranzo, N, Tian, J, Tobin, MD, Wain, LV & others 2019, 'Low-frequency variation in TP53 has large effects on head circumference and intracranial volume'. *Nature Communications*, vol 10.
- Richardson, TG, Haycock, PC, Zheng, J, Timpson, NJ, Gaunt, TR, Smith, GD, Relton, CL & Hemani, G, 2018, 'Systematic Mendelian randomization framework elucidates hundreds of CpG sites which may mediate the influence of genetic variants on disease'. *Human Molecular Genetics*, vol 27., pp. 3293-3304
- Rogers, M, Shihab, HA, Mort, M, Cooper, DN, Gaunt, T & Campbell, C, 2018, 'FATHMM-XF: accurate prediction of pathogenic point mutations via extended features'. *Bioinformatics*, vol 34., pp. 511-513
- Hemani, G, Zheng, J, Elsworth, B, Wade, K, Haberland, V, Baird, D, Laurin, C, Burgess, S, Bowden, J, Langdon, R, Tan, V, Yarmolinsky, J, Shihab, H, Timpson, N, Evans, D, Relton, C, Martin, R, Smith, GD, Gaunt, T & Haycock, P, 2018, 'The MR-Base platform supports systematic causal inference across the human phenome'. *eLife*, vol 7.

View complete publications list in the University of Bristol publications system

Projects

- Accessible Resource for Integrated Epigenomics Studies (ARIES)
- Avon Longitudinal Study of Parents and Children (ALSPAC)
- Epigenetics and Social Science Network
- Epigenetics: Environment, Embodiment and Equality (E4 project)
- Interpreting epigenetic signatures in studies of early life adversity (Interstela project)