# Oliver Hofmann

## Personalia

date of birth 12. August 1972

place of birth Cologne nationality: German

#### Education

2000–2004 **Doctor of Science**, Department of Biochemistry, University of Cologne, Cologne, Germany.

Determination of dependencies between enzymatic activities and disease related concepts by automatic

evaluation of the scientific literature.

1993–1999 **Diploma Biology**, *University of Cologne*, Cologne, Germany.

Major subject: genetics, minor subjects: developmental biology, biochemistry

# Experience

#### Academic

2022-Present **Professor**, University of Melbourne, Melbourne, Australia.

2016–2022 Associate Professor, University of Melbourne, Melbourne, Australia.

2016–Present **Head of Bioinformatics**, Collaborative Centre for Genomic Cancer Medicine, University of Melburne Center for Cancer Research (formerly Centre for Cancer Research), Melbourne, Australia.

- o Building a Genomics Platform Group to support clinically accredited sequencing with a focus on rapid whole genome/transcriptiome sequencing of cancer samples
- o Develop national genomics infrastructure
- o Co-leading Australian Genomics Data Working Group.
- o Co-lead for the National Approach to Genomic Information Management implementation phase.
- o Co-leading the Large Scale Genomnics working group for the Global Alliance for Genomics and Health.
- Co-lead for the Illumina-University of Melbourne Genomics Hub and the Advanced Genomics Collaboration, a public/private partnership to develop new genomic technologies, develop Victoria's workforce and drive clinical translation.
- o Appointed to the Expert Advisory Committee for Genomics Australia by the Federal Department of Health and Ages Care

2015–2016 Visiting Associate Professor, Department of Biostatistics, Harvard T.H. Chan School of Public Health, Boston, MA, US.

- o Provide strategic support to the Harvard T.H. Chan Bioinformatics Core on research development.
- Designed and taught a "Massive Open Online Course" on re-sequencing study design and analysis through the edX platform.
- 2015–2016 Reader, Institute of Cancer Sciences, University of Glasgow, Glasgow, UK.

2015–2016 Head of Bioinformatics, Wolfson-Wohl Cancer Research Center, Glasgow, UK.

- Planned and deployed the computational infrastructure and software required to support a local Illumina X10 system maintained by Dr. Sean Grimmond.
- Provided guidance and bioinformatics support for new initiatives such as the Scottish Genome Partnership (in collaboration with University of Edinburgh and Genomics England) and the Cancer Research UK Big Data Initiative.
- o Supported the NHS Scotland with the development of a clinical gene panel for patient diagnostics.
- Developed methods for improved interrogation of WGS data (structural variation, minority variant detection, prioritisation).

- 2014–2015 Director, Bioinformatics Core, Harvard T.H. Chan School of Public Health, Boston, MA, US.
  - o Raised funding from Harvard Medical School and the Harvard NeuroDiscovery Center, expanding our bioinformatics support to these communities.
  - Engaged industry through funded collaborations. Research topics included shorter projects such
    as optimising NGS workflows on AWS (Intel), benchmarking research computing infrastructure for
    large scale WGS analysis (Dell), validating gene panels (Color Genomics) and porting our NGS
    workflow to the Azure Cloud Computing environment (Microsoft) as well as an ongoing, multi-year
    collaboration with AstraZeneca to improve methods for processing cancer genomics data (structural
    variation, subclonal structure).
  - o Initiated a bioinformatics training program, hiring two full time trainers to develop and teach one day and ten day workshops on best practices in data management, scientific computing, programming and NGS data analysis; the program had 200 students in the first year and has been renewed.
- 2012–2015 **Senior Research Scientist**, Department of Biostatistics, Harvard T.H. Chan School of Public Health, Boston, MA, US.
- 2011-Present Affiliated Faculty, Harvard Stem Cell Institute, Cambridge, MA, US.
  - Expanded the HSPH Bioinformatics Core to serve as the bioinformatics support unit for stem cell researchers in the Harvard Community.
  - Helped to raise funding for the Harvard Stem Cell Commons, a data repository to support reproducible research and data dissemination.
  - 2010–2012 **Research Scientist**, Department of Biostatistics, Harvard T.H. Chan School of Public Health, Boston, MA, US.
  - 2008–2014 Associate Director, Bioinformatics Core, Harvard T.H. Chan School of Public Health, Boston, MA, US.
    - o Re-organized the Bioinformatics Core to focus on next-generation sequencing technology support.
    - o Increased the Core staff to a total of ten analysts, developers and research computing specialists.
    - o Achieved full self-sustainability for the Core through a recharge center model.
    - Expanded bioinformatics support to the HIV community (funds raised from the Harvard Center for AIDS Research) and translational medicine (as the only bioinformatics group to receive funding from Harvard Catalyst).
    - Supported >300 projects over six years, ranging from ChIP-Seq to large scale exome-sequencing, single-cell RNA-Seq and more customized technologie. Projects included large scale collaborative efforts to study predisposition to breast cancer (6000 patient exome-seq cohort) and early onset of Alzheimer's Disease (1500 WGS).
  - 2008–2009 Research Associate, Department of Biostatistics, Harvard T.H. Chan School of Public Health, Boston, MA, US.
    - o Started the T.H. Chan Bioinformatics Core, supporting the Director Dr. Winston Hide.
    - Studied the expression patterns of Cancer Testis Antigens in public expression data in collaboration with the Ludwig Institute for Cancer Research.
  - 2007–2008 **Research Fellow**, Department of Biostatistics, Harvard T.H. Chan School of Public Health, Boston, MA, US.
  - 2005–2007 **Post-doctoral researcher**, South African National Bioinformatics Institute (SANBI), University of the Western Cape, Cape Town, South Africa.
    - o Developed and maintained the eVOC controlled vocabulary system.
    - Analysed public EST data for splicing patterns and tissue specificity.
    - o Participated in FANTOM4 and FANTOM5, implementing new methods for CAGE sequence analysis.
  - 2000–2004 PhD student, Department of Biochemistry, University of Cologne, Cologne, Germany.
    - $\circ\,$  Adapted Natural Language Parsing (NLP) methods for data extraction from PubMed records.
    - Participated in the design and taught courses for the Cologone Post-Graduate Bioinformatics Program.

#### Industry

2001–2002 Research fellow, deCODE genetics, Reykajvik, Iceland.

Expanded existing tools for pattern matching and gene clustering for use with internal human genome builds. Advised by Dr. Daniel Oskarson, Head of Bioinformatics.

# Teaching

- 2015 **Introduction to Bioinformatics**, *Harvard Medical School*, Boston, MA, US. Co-organized 80h, in-depth bioinformatics and NGS course
- 2015 edX PH525: Case study: Variant Discovery and Genotyping, edX MOOC, Cambridge, MA, US.
  - NGS using the command line in a MOOC environment
- 2012–2015 NGS Workshops, Harvard T.H. Chan School of Public Health, Boston, MA, US. NGS in Galaxy (Exome-Seq, RNA-Seq, ChIP-Seq)
- 2011–2015 **Postgrad lectures**, Harvard T.H. Chan School of Public Health, Boston, MA, US. Bio 517, Introduction to next-generation sequencing
  - 2011 **Postgrad lectures**, Cold Spring Harbor Laboratories, Laurel Hollow, NY, US. Programming for biology
  - 2010 Tutorial, ISMB Conference, Boston, MA, US.
    Large scale biological data mining for functional genomics and metagenomics
- 2005–2007 Undergrad lectures, University of Cape Town and University of the Western Cape, Cape Town/Bellville, South Africa.

  Computational biology, information extration and data integration
  - 2005 **Student advisor**, South African National Bioinformatics Institute, University of the Western Cape, Bellville, South Africa.

    Supervisor of Honours' students Feziwe Mpondo and Thurayah Davids
- 2003–2004 **Post-grad lectures**, *CUBIC*, Cologne, Germany.

  Pattern recognition, hidden markov models, suffix trees and natural language processing
  - 2002 **Post-grad lectures**, *CUBIC*, Cologne, Germany. Genome analysis (assembly, comparison, annotation) and text mining
- 2002–2004 Curriculum development, CUBIC (Bioinformatics Competence Center Cologne), Cologne, Germany.

  Course planning for a one-year post-graduate school in computational biology
- 2001–2002 **Student advisor**, deCODE genetics, Reykajvik, Iceland. Co-supervision of Master's students Fionna Cunningham and Chrysi Kirtay
- 2000–2004 **Teaching assistant**, Department of Biochemistry, University of Cologne, Cologne, Germany. Graduate courses in protein homology modelling

# Professional Activities

#### Advisory Role

- $2010-2012 \quad \textbf{Technical Advisory Committee}, \textit{Harvard Institute for Quantitative Social Science}.$
- 2012–2015 Tools and Technology Committee, Harvard Medical School.
  - 2016 Informatics Guidance Group, Scottish Genome Partnership.
  - 2016 Big Data in Cancer Strategy Committee, Cancer Research UK.
- 2016–2022 Common Workflow Language Steering Group, Global Alliance for Genomics and Health.
- 2016–2022 Bioinformatics Commons Pathfinder Project Steering Committee, Bioplatforms Australia.
  - 2018 Rare Disease Data Advisory Group, Genome Canada.
- 2023–2025 Expert Advisory Group on Genomics Australia, Australian Government Department of Health and Aged Care.
- 2024-Present Strategic Advisory Board, Global Alliance for Genomics and Health.
- 2024-Present Data Science Steering Committee, Garvan Institute of Medical Research.
- 2024-Present Infrastructure Advisory Board, European Genomics Data Infrastructure.

#### Consortium Participation

2006-2012 **FANTOM 4-6**.

Functional Annotation of the Mammalian Genome.

2012–2016 Genome in a Bottle Consortium, National Institute of Standards and Technology.

Developing genomic reference standards for sequence analysis.

2013-2015 The Human Heredity and Health in Africa (H3Africa) Initiative.

Development of bioinformatics expertise in Africa.

2015-Present Global Alliance for Genomics and Health (GA4GH).

Interoperable approaches to genomic data analysis and sharing.

Consortium Leadership

2016–2025 Australian Genomic Health Alliance.

Bioinformatics Program Co-lead, Building re-usable genomics infrastructure for Australia.

2017-Present Global Alliance for Genomics and Health (GA4GH).

Workstream Co-lead, Large Scale Genomics; Member Steering Committee.

2019-Present GA4GH National Initiatives Forum (NIF).

Steering Committee.

# Funding

#### **HSPH** Bioinformatics Core.

Raised funding to move the Harvard T.H. Chan Bioinformatics Core from subsidies to a fully self-sustained recharge center. I was a named investigagtor (key personnel) on **more than 30 NIH grants** and secured funding to support bioinformatics infrastructure development from industry, including **Biogen**, **Color Genomics**, **Microsoft**, **Dell** and **Intel**. Notable other funding sources included:

- o since 2008: Bioinformatics Core for the **Harvard NIEHS Center for Environmental Health**, \$40,000/year
- o since 2009: Bioinformatics Consulting for Harvard Catalyst, \$50,000/year
- o since 2011: Center for Stem Cell Bioinformatics, Harvard Stem Cell Institute, \$150,000/year
- 2012: Archon Genomics X-Prize through EdgeBio to build a variant calling validation framework, \$90,000
- o since 2013: Bioinformatics Core for the Harvard Center for AIDS Research, \$40,000/year
- o 2013: Cure Alzheimer's Fund through Rudy Tanzi, Massachusetts General Hospital, \$500,000 to process and analyze 1500 whole genome samples from families with a history of Alzheimer's Disease
- o since 2014: Bioinformatics Support for the Harvard NeuroDiscovery Center, \$200,000/year
- o since 2014: Bioinformatics Support for Harvard Medical School, \$400,000/year
- o since 2014: Funding from **AstraZeneca** to develop novel methods for cancer genome analysis and RNA-Seq, \$200,000/year.

#### Wolfson Wohl Cancer Research Center.

Co-Investigator and co-lead bioinformatics (with Colin Semple, IGMM, Edinburgh) for the **Scottish Genome Partnership**, total budget of 6,000,000 GBP.

#### University of Melbourne.

Co-investigator for **The Australian Pancreatic Cancer Organoid Biobank** (Avner Foundation, \$916,000 AUD), **The Australian Genomics Health Alliance** (NHMRC, \$300,000 AUD/year), Dissecting mutational landscapes and cellular ecosystems of pancreatic cancer (NHMRC, \$884,000 AUD) and an ARC-LIEF Grant for Merit Allocation to the National Computational Infrastructure (\$4,000,000 AUD). Co-lead of the **Illumina - University of Melbourne Genomics Hub** (\$60,000,000 AUD) and **GUARDIANS** (NCRIS, National Genomics Infrastructure, \$25,000,000 AUD. CI on the Breast Cancer Atlas project (DOD, 5,800,000 USD).)

# **Publications**

- E. DeBortoli, E. McGahan, T. Yanes, J. Berkman, L.G. Aoude, A.K. Smit, A. Gokoolparsadh, A. Hermes, L. Newett, M. Bourke, S. Hanson, H. Hughes, <u>O. Hofmann</u>, I. Goranitis, R. McWhirter, V. Milch, J. Steinberg, A. McInerney-Leo, *Utility of Germline, Somatic and ctDNA Testing in Adults With Cancer*, Cancer Medicine, (2025)
- R.J. Rebello, A. Posner, R. Dong, O.W.J. Prall, T. Sivakumaran, C. B. Mitchell, A. Flynn, A. Caneborg, C. Mitchell, S. Kanwal, C. Fedele, S. Webb, K. Fisher, H-L. Wong, S. Balachander, W. Zhu, S. Nicolson, V. Dimitriadis, N. Wilcken, A. DeFazio, B. Gao, M. Singh, I.M. Collins, C. Steer, M. Warren, N. Karanth, H. Xu, A. Fellowes, R.J. Hicks, K.P. Stewart, C. Shale, P. Priestley, S-J. Dawson, J.H.A. Vissers, S.B. Fox, P. Schofield, D. Bowtell, O. Hofmann, S.M. Grimmond, L. Mileshkin, R.W. Tothill, Whole genome sequencing improves tissue-of-origin diagnosis and treatment options for cancer of unknown primary, Nature Comms, (2025)
- Z. Stark, D. Glazer, O. Hofmann, A. Rendon, C.R. Marshall, G.S. Ginsburg, C. Lunt, N. Allen, M. Effingham, J. Hastings Ward, S.L. Hill, R. Ali, P. Goodhand, A. Page, H.L. Rehm, K.N. North, R.H. Scott, A call to action to scale up research and clinical genomic data sharing, Nature Review Genetics, (2024).
- M. Vu, K. Degeling, G.L. Ryland, <u>O. Hofmann</u>, A.P. Ng, D. Westerman, M.J. IJzerman, *Economic Impact of Whole Genome Sequencing and Whole Transcriptome Sequencing Versus Routine Diagnostic Molecular Testing to Stratify Patients with B-Cell Acute Lymphoblastic Leukemia*, Journal of Molecular Diagnostics, (2024).
- A. Wickramarachchi, B. Hosking, Y. Jain, J. Grimes, M.J. O'Brien, T. Wright, M. Burgess, V.S.K. Lin, F. Reisinger, O. Hofmann, M. Lawley, L.O.W. Wilson, N.A. Twine, D.C. Bauer, Scalable genomic data exchange and analytics with sBeacon, Nature Biotechnology, (2024).
- A Matched Molecular and Clinical Analysis of the Epithelioid Haemangioendothelioma Cohort in the Stafford Fox Rare Cancer Program and Contextual Literature Review, A. Abdelmogod, L. Papadopoulos, S. Riordan, M. Wong, M. Weltman, R. Lim, C. McEvoy, A. Fellowes, S. Fox, J. Bedo, J. Penington, K. Pham, O. Hofmann, J.H.A. Vissers, S. Grimmond, G. Ratnayake, M. Christie, C. Mitchell, W. Murray, K. McClymont, P. Luk, A.T. Papenfuss, D. Kee, C.L. Scott, D. Goldstein, H.E. Barker, Cancers, (2023).
- Targeting homologous recombination deficiency in uterine leiomyosarcoma, I.M. Collins, G. Dall, C.J. Vandenberg, K. Nesic, G. Ratnayake, W. Zhu, J.H.A. Vissers, J. Bedo, J. Penington, M.J. Wakefield, D. Kee, A. Carmagnac, R. Lim, K. Shield-Artin, B. Milesi, A. Lobley, E.L. Kyran, E. O'Grady, J. Tram, W. Zhou, D. Nugawela, K.P. Stewart, R. Caldwell, L. Papadopoulos, A.P. Ng, H.E. Barker, A. Dobrovic, S.B. Fox, O. McNally, J. Power, T. Meniawy, T.H. Tan, O. Klein, S. Barnett, I. Olesen, A. Hamilton, O. Hofmann, S. Grimmond, A.T. Papenfuss, C.L. Scott, J Exp Clin Cancer Res, (2023).
- Z. Stark, T. Boughtwood, M. Haas, J. Braithwaite, C.L. Gaff, I. Goranitis, A.B. Spurdle, D.P. Hansen, O. Hofmann, N. Laing, S. Metcalfe, A.J. Newson, H.S. Scott, N. Thorne, R.L. Ward, M.E. Dinger, S. Best, J.C. Long, S.M. Grimmond, J. Pearson, N. Waddell, C.P. Barnett, M. Cook, M. Field, D. Fielding, S.B. Fox, J. Gecz, A. Jaffe, R.J. Leventer, P.J. Lockhart, S. Lunke, A.J. Mallett, J. McGaughran, L. Mileshkin, K. Nones, T. Roscioli, I.E. Scheffer, C. Semsarian, C. Simons, D.M. Thomas, D.R. Thorburn, R. Tothill, D. White, S. Dunwoodie, P.T. Simpson, P. Phillips, MJ Brion, K. Finlay, M.C.J. Quinn, T. Mattiske, E. Tudini, K. Boggs, S. Murray, K. Wells, J. Cannings, A.H. Sinclair, J. Christodoulou, K.N. North, Australian Genomics: Outcomes of a 5-year national program to accelerate the integration of genomics in healthcare, The American Journal of Human Genetics, (2023).
- ICGC/TCGA Pan-Cancer Analysis of Whole Genomes Consortium, Genomic and molecular analyses identify molecular subtypes of pancreatic cancer recurrence, Gastroenterology, (2022).
- A.D. Yates, J. Adams, S. Chaturvedi, R.M. Davies, M. Laird, R. Leinonen, R. Nag, N.C. Sheffield, O. Hofmann, T.M. Keane, *Refget: standardized access to reference sequences*, Bioinformatics, (2022).

- G.L. Ryland, A. Bajel, M. Dickinson, P.G. Ekert, <u>O. Hofmann</u>, M. IJzerman, A.P. Ng, A. Oshlack, B. Schmidt, I.S. Tiong, M. Vu, D.A. Westerman, S. Grimmond, P. Blombery, *Diagnostic Utility of Multimodal Genomic Profiling for Molecular Classification and MRD Assessment in Adult B-Cell Acute Lymphoblastic Leukemia*, Blood, (2021).
- The Global Alliance for Genomics and Health Consortium, GA4GH: International policies and standards for data sharing across genomic research and healthcare, Cell Genomics, (2021).
- Z. Stark, R.E. Foulger, E. Williams, B.A. Thompson, C. Patel, S. Lunke, C. Snow, I. Leong, A. Puzriakova, L.C. Daugherty, S. Leigh, C. Boustred, O. Niblock, A. Rueda-Martin, O. Gerasimenko, K. Savage, W. Bellamy, V. San Kho Lin, R. Valls, L. Gordon, H.K. Brittain, E.R.A. Thomas, A.L.T. Tavares, M. McEntagart, S.M. White, T.Y. Tan, A. Yeung, L. Downie, I. Macciocca, E. Savva, C. Lee, A. Roesley, P. De Fazio, J. Deller, Z.C. Deans, S.L. Hill, M.J. Caulfield, K.N. North, R.H. Scott, A. Rendon, O. Hofmann, E.M. McDonagh, Scaling national and international improvement in virtual gene panel curation via a collaborative approach to discordance resolution, The American Journal of Human Genetics, (2021).
- A. Senf, R. Davies, F. Haziza, J. Marshall, J. Troncoso-Pastoriza, <u>O. Hofmann</u>, T.M. Keane, *Crypt4GH: a file format standard enabling native access to encrypted data*, Bioinformatics, (2021).
- Z. Stark, R.E. Foulger, E. Williams, B. Thompson, C. Patel, I.U.S. Leong, L.C. Daugherty, S. Leigh, C. Snow, C. Boustred, O. Niblock, A. Rueda-Martin, O. Gerasimenko, E. Ivanov, K. Savage, W. Bellamy, V. San Kho Lin, R. Valls, L. Gordon, H. Brittain, R.H. Scott, E.R.A. Thomas, A. de Burca, A.L.T. Tavares, Z. Hyder, M. McEntagart, C. Turnbull, S.M. White, T.Y. Tan, A. Yeung, L. Downie, S. Lunke, J. Deller, Z.C. Deans, S.L. Hill, M.J. Caulfield, K.N. North, A. Rendon, O. Hofmann, E.M. McDonagh, PanelApp: accelerating international consensus on virtual gene panels through collaboration on a federated open platform, European Journal of Human Genetics, (2020).
- D. Prokopenko, S.L. Morgan, K. Mullin, <u>O. Hofmann</u>, B. Chapman, R. Kirchner, S. Amberkar, I. Wohlers, C. Lange, W. Hide, L. Bertram, R.E. Tanzi, *Whole-genome sequencing reveals new Alzheimer's disease-associated rare variants in loci related to synaptic function and neuronal development*, Alzheimer's and Dementia, (2020).
- S. Nakken, V. Saveliev, <u>O. Hofmann</u>, P. Moller, O. Myklebost, E. Hovig, *Cancer Predisposition Sequencing Reporter (CPSR): a flexible variant report engine for germline screening in cancer*, International Journal of Cancer, (2020).
- D. Prokopenko, J. Hecker, R. Kirchner, B.A. Chapman, <u>O. Hofmann</u>, K. Mullin, W. Hide, L. Bertram, N. Laird, D.L. DeMeo, C. Lange, R.E. Tanzi, *Identification of Novel Alzheimer's Disease Loci Using Sex-Specific Family-Based Association Analysis of Whole-Genome Sequence Data*, Sci Rep. (2020).
- L.K. Cuddy, D. Prokopenko, E.P. Cunningham, R. Brimberry, P. Song, R.RKirchner, B.A. Chapman, O. Hofmann, W. Hide, D. Procissi, T. Hanania, S.C. Leiser, R.E. Tanzi, R. Vassar, Ab-accelerated neurodegeneration caused by Alzheimer's-associated ACE variant R1279Q is rescued by angiotensin system inhibition in mice, Sci Transl Med, (2020).
- G.L. Ryland, A. Barraclough, C.Y. Fong, S. Fleming, A. Bajel, <u>O. Hofmann</u>, D. Westerman, S. Grimmond, P. Blombery, *Inotuzumab ozogamicin resistance associated with a novel CD22 truncating mutation in a case of B-acute lymphoblastic leukaemia*, Br J Haematol, (2020).
- O.W.J. Prall, V. Nastevski, H. Xu, C.R.E. McEvoy, J.H.A. Vissers, D.J. Byrne, E. Takano, S. Yerneni, S. Ellis, T. Green, C.A. Mitchell, W.K. Murray, C.L. Scott, S.M. Grimmond, O. Hofmann, A. Papenfuss, D. Kee, A. Fellowes, I.S. Brown, G. Miller, M.P. Kumarasinghe, A. Perren, C.B. Nahm, A. Mittal, J. Samra, M. Ahadi, S.B. Fox, A. Chou, A.J. Gill, *RAF1* rearrangements are common in pancreatic acinar cell carcinomas, Mod Pathol, (2020).
- ICGC/TCGA Pan-Cancer Analysis of Whole Genomes Consortium, *Pan-cancer analysis of whole genome*, Nature, (2020).

- J. Dahlberg, J. Hermansson, S. Sturlaugsson, M. Lysenkova, P. Smeds, C. Ladenvall, R.V. Guimera, F. Reisinger, <u>O. Hofmann</u>, P. Larsson, *Arteria: An automation system for a sequencing core facility*, GigaScience, (2019).
- D.P. Hansen, M.E. Dinger, O. Hofmann, N. Thorne, T.F. Boughtwood, *Preparing Australia for genomic medicine: data, computing and digital health*, Med J Aust (2019).
- G. Macintyre, T.E. Goranova, D. De Silva, D. Ennis, A.M. Piskorz, M. Eldridge, D. Sie, L.A. Lewsley, A. Hanif, C. Wilson, S. Dowson, R.M. Glasspool, M. Lockley, E. Brockbank, A. Montes, A. Walther, S. Sundar, R. Edmondson, G.D. Hall, A. Clamp, C. Gourley, M. Hall, C. Fotopoulou, H. Gabra, J. Paul, A. Supernat, D. Millan, A. Hoyle, G. Bryson, C. Nourse, L. Mincarelli, L.N. Sanchez, B. Ylstra, M. Jimenez-Linan, L. Moore, O. Hofmann, F. Markowetz, I. McNeish, J.D. Brenton, Copy number signatures and mutational processes in ovarian carcinoma, Nat Genet, (2018).
- A. Li, B. Hooli, K. Mullin, R.E. Tate, A. Bubnys, R. Kirchner, B. Chapman, <u>O. Hofmann</u>, W. Hide, R.E. Tanzi, *Silencing Of The Drosophila Ortholog Of Sox5 Leads To Abnormal Neuronal Development And Behavioral Impairment*, Hum Mol Genet, (2017).
- J.L. Humphris, A.M. Patch, K. Nones, P.J. Bailey, A.L. Johns, S. McKay, D.K. Chang, D.K. Miller, M. Pajic, K.S. Kassahn, M.C. Quinn, T.J. Bruxner, A.N. Christ, I. Harliwong, S. Idrisoglu, S. Manning, C. Nourse, E. Nourbakhsh, A. Stone, P.J. Wilson, M. Anderson, J.L. Fink, O. Holmes, S. Kazakoff, C. Leonard, F. Newell, N. Waddell, S. Wood, R.S. Mead, Q. Xu, J. Wu, M. Pinese, M.J. Cowley, M.D. Jones, A.M. Nagrial, V.T. Chin, L.A. Chantrill, A. Mawson, A. Chou, C.J. Scarlett, A.V. Pinho, I. Rooman, M. Giry-Laterriere, J.S. Samra, J.G. Kench, N.D. Merrett, C.W. Toon, K. Epari, N.Q. Nguyen, A. Barbour, N. Zeps, N.B. Jamieson, C.J. McKay, C.R. Carter, E.J. Dickson, J.S. Graham, F. Duthie, K. Oien, J. Hair, J.P. Morton, O.J. Sansom, R. Gretzmann, R.H. Hruban, A. Maitra, C.A. Iacobuzio-Donahue, R.D. Schulick, C.L. Wolfgang, R.A. Morgan, R.T. Lawlor, B. Rusev, V. Corbo, R. Salvia, I. Cataldo, G. Tortora, M.A. Tempero; Australian Pancreatic Cancer Genome Initiative, O. Hofmann, J.R. Eshleman, C. Pilarsky, A. Scarpa, E.A. Musgrove, A.J. Gill, J.V. Pearson, S.M. Grimmond, N. Waddell, A.V. Biankin, Hypermutation In Pancreatic Cancer, Gastroenterology, (2017).
- M.J. Ahdesmaki, B. Chapman, P.E. Cingolani, <u>O. Hofmann</u>, A. Sidoruk, Z. Lai, G. Zakharov, M. Rodichenko, M. Alperovich, D. Jenkins, T.H. Carr, D. Stetson, B. Dougherty, J.C. Barrett, J. Johnson, *Prioritisation of Structural Variant Calls in Cancer Genomes*, PeerJ, (2017).
- S. Lindstrom, A. Ablorh, B. Chapman, A. Gusev, G. Chen, C. Turman, A.H. Eliassen, A.L. Price, B.E. Henderson, L. Le Marchand, O. Hofmann, C.A. Haiman, P. Kraft, *Deep targeted sequencing of 12 breast cancer susceptibility regions in 4611 women across four different ethnicities*, Breast Cancer Res, (2016).
- C. Grasso, M. Anaka, <u>O. Hofmann</u>, R. Sompallae, K. Broadley, W. Hide, M.V. Berridge, J. Cebon, A. Behren, M.J. McConnell, *Iterative sorting reveals CD133+ and CD133- melanoma cells as phenotypically distinct populations*, BMC Cancer, (2016).
- L.D. Wang, S.B. Ficarro, J.N. Hutchinson, R. Csepanyi-Komi, P.T. Nguyen, E. Wisniewski, J. Sullivan, <u>O. Hofmann</u>, E. Ligeti, J.A. Marto, A.J. Wagers, *Phosphoproteomic profiling of mouse primary HSPCs reveals new regulators of HSPC mobilization*, Blood, (2016)
- Z. Lai, A. Markovets, M. Ahdesmaki, B. Chapman, <u>O. Hofmann</u>, R. McEwen, J. Johnson, B. Dougherty, J.C. Barrett, J.R. Dry, *VarDict: a novel and versatile variant caller for next-generation sequencing in cancer research*, Nucleic Acids Res, (2016).
- S. Sunshine, R. Kirchner, S.S. Amr, L. Mansur, R. Shakhbatyan, M. Kim, A. Bosque, R.F. Siliciano, V. Planelles, <u>O. Hofmann</u>, S. Ho Sui, J.Z. Li, *HIV Integration Site Analysis of Cellular Models of HIV Latency with a Probe-Enriched Next-Generation Sequencing Assay*, J Virol, (2016).

- P. Bailey, D.K. Chang, K. Nones, A.L. Johns, A. Patch, M. Gingras, D.K. Miller, A.N. Christ, T.J.C. Bruxner, M.C. Quinn, C. Nourse, L.C. Murtaugh, I. Harliwong, S. Idrisoglu, S. Manning, E. Nourbakhsh, S. Wani, L. Finkl, O. Holmes, V. Chin, M.J. Anderson, S. Kazakoff, C. Leonard, F. Newell, N. Waddell, S. Wood, Q. Xu, P.J. Wilson, N. Cloonan, K. S. Kassahn, D. Taylor, K. Quek, A. Robertson, L. Pantano, L. Mincarelli, L.N. Sanchez, L. Evers, J. Wu, M. Pinese, M.J. Cowley, M.D. Jones, E.K. Colvin2, A.M. Nagrial, E.S. Humphrey, L.A. Chantrill, A. Mawson, J. Humphris, A. Chou, M. Pajic, C.J. Scarlett, A.V. Pinho, M. Giry-Laterriere, I. Rooman, J.S. Samra, J.G. Kench, J.A. Lovell, N.D. Merrett, C.W. Toon, K. Epari, N.Q. Nguyen, A. Barbour, N. Zeps15, K. Moran-Jones, N.B. Jamieson, J.S. Graham, F. Duthie, K. Oien, J. Hair, R. Gratzmann, A. Maitra, C.A. Iacobuzio-Donahue, C.L. Wolfgang, R.A. Morgan, R.T. Lawlor, V. Corbo, C. Bassi, B. Rusev, P. Capelli, R.Salvia, G. Tortora, D. Mukhopadhyay, G.M. Petersen, Australian Pancreatic Cancer Genome Initiative, D.M.Munzy, W.E. Fisher, S.A. Karim, J.R. Eshleman, R.H. Hruban, C. Pilarsky, J.P. Morton, O.J. Sansom, A. Scarpa, E.A. Musgrove, U.H. Bailey, O. Hofmann, D.A. Wheeler 31, 32, A.J. Gill, R.A. Gibbs, J.V. Pearson, N. Waddell, A.V. Biankin and S.M. Grimmond, Integrative genomic analysis of pancreatic cancer identifies  $subtypes\ with\ distinct\ histopathological\ characteristics,\ Nature,\ (2016).$
- F.L. Craciun, V. Bijol, A.K. Ajay, P. Rao, R.K. Kumar, J. Hutchinson, <u>O. Hofmann</u>, N. Joshi, J.P. Luyendyk, U. Kusebauch, C.L. Moss, A. Srivastava, J. Himmelfarb, S.S. Waikar, R.L. Moritz, V.S. Vaidya, *RNA Sequencing Identifies Novel Translational Biomarkers of Kidney Fibrosis*, J Am Soc Nephrol, (2015).
- E. O'Day, M.T. Le, S. Imai, S.M. Tan, R. Kirchner, H. Arthanari, <u>O. Hofmann</u>, G. Wagner, J. Lieberman, An RNA-binding Protein, Lin28, Recognizes and Remodels G-quartets in the MicroRNAs (miRNAs) and mRNAs It Regulates, J Biol Chem, (2015).
- C. Gedye, T. Cardwell, N. Dimopoulos, B.S. Tan, H. Jackson, S. SvobodovÃ-¿Â½, M. Anaka, A. Behren, C. Maher, O. Hofmann, W. Hide, O. Caballero, I.D. Davis, J. Cebon *Mycoplasma Infection Alters Cancer Stem Cell Properties in Vitro*, Stem Cell Rev, (2015).
- J. Sun, A. Ramos, B. Chapman, J.B. Johnnidis, L. Le, Y.J. Ho, A. Klein, <u>O. Hofmann</u>, F.D. Camargo, *Clonal dynamics of native haematopoiesis*, Nature, (2014).
- S.M. Tan, R. Kirchner, J. Jin, O. Hofmann, L. MCReynolds, W. Hide, J. Lieberman, Sequencing of captive target transcripts identifies the network of regulated genes and functions of primate-specific miR-522, Cell Reports, (2014).
- The Fantom5 Consortium, A promoter level mammalian expression atlas, Nature, (2014).
- E. Dimont, <u>O. Hofmann</u>, A.R.R. Forrest, H. Kawaji, The Fantom Consortium, W. Hide, *CAGExploreR*: an R package for the analysis and visualization of promoter dynamics across multiple experiments, Bioinformatics, (2014).
- J.M. Zook, B. Chapman, J. Wang, D. Mittelman, <u>O. Hofmann</u>, W. Hide, M. Salit, *Integrating human sequence data sets provides a resource of benchmark SNP and indel genotype calls*, Nature Biotechnology, (2014).
- J.Z. Li, B. Chapman, P. Charlebois, <u>O. Hofmann</u>, B. Weiner, A. Porter, R. Samuel, S. Vardhanabhuti , L. Zheng, J. Eron, B. Taiwo, M.C. Zody, M.R. Henn, D.R. Kuritzkes, W. Hide, Comparison of illumina and 454 deep sequencing in participants failing raltegravir-based antiretroviral therapy, PLOS One, (2014).
- J. Lui, A.M. Krautzberger, S. Ho Sui, <u>O. Hofmann</u>, Y. Chen, M. Baetscher, I. Grgic, S. Kumar, B. Humphreys, W. Hide, A.P. McMahon, *Cell-specific translational profiling in acute kidney injury*, Journal of Clinical Investigation, (2014).
- J.J. Carmona, B. Izzi, A.C. Just, J Barupal, A.M. Binder, J. Hutchinson, <u>O. Hofmann</u>, J. Schwartz, A. Baccarelli, K.B. Michels, *Comparison of multiplexed reduced representation bisulfite sequencing (mRRBS) with the 450K Illumina Human BeadChip: from concordance to*

- practical applications for methylomic profiling in epigenetic epidemiologic studies, Epigenetics Chromatin, (2013).
- H.T. Huang, K.L. Kathrein, A. Barton, Z. Gitlin, Y.H. Huang, T.P. Ward, <u>O. Hofmann</u>, A. Dibiase, A. Song, S. Tyekucheva, W. Hide, Y. Zhou, L. Zon, *A network of epigenetic regulators guides developmental haematopoiesis in vivo*, Nature Cell Biology, (2013).
- O. Hofmann, R. Sompallae, C.A. Maher, C. Gedye, A. Behren, M. Vitezic, C.O. Daub, S. Devalle, O.L. Caballero, P. Carninci, Y. Hayashizaki, E.R. Lawlor, J. Cebon, and W. Hide, A comprehensive promoter landscape identifies a novel promoter for CD133 in restricted tissues, cancers and stem cells, Frontiers in Genetics, (2013).
- G.M. Altschuler, O. Hofmann, I. Kalatskaya, R. Payne, S. Ho Sui, U. Saxena, A. Krivtsov, S.A. Armstrong, T. Cai, L. Stein, and Hide W A, *Pathway Fingerprinting: An integrative approach to understand the functional basis of disease*, Genome Medicine, (2013).
- S.H. Sui, E. Merrill, N. Gehlenborg P. Haseley, I. Sytchev, R. Park, P. Rocca-Serra, S. Corlosquet, A. Gonzalez-Beltran, E. Maguire, <u>O. Hofmann</u>, P. Park, S. Das, S.A. Sansone, W. Hide W., *The Stem Cell Commons: an exemplar for data integration in the biomedical domain driven by the ISA framework*, AMIA Summits on Translational Science Proceedings, (2013).
- P.S. Lai, <u>O. Hofmann</u>, R.M. Baron, Q.R. Meng, H.S. Bresler, D.M. Brass, I.V. Yang, D.A. Schwartz, D.C. Christiani, and W. Hide, *Integrating Murine Gene Expression Studies To Understand Obstructive Lung Disease Due To Chronic Inhaled Endotoxin*, PLOS One, (2013).
- J.W. Froehlich, A.R. Vaezzadeh, M.K. Kirchner, A.C. Briscoe, <u>O. Hofmann</u>, W. Hide, H. Steen, and R.S. Lee, *An In-Depth Comparison Of The Male Pediatric And Adult Urinary Proteomes*, Biochimica Et Biophysica Acta, (2013).
- M. Schwede, D. Spentzos, S. Bentink, <u>O. Hofmann</u>, B. Haibe-Kains, D. Harrington, J. Quackenbush, and A.C. Culhane, *Stem Cell-Like Gene Expression In Ovarian Cancer Predicts Type II Subtype And Prognosis*, PLOS One, (2013).
- P.S. Lai, J.M. Fresco, M.A. Pinilla, A.A. Macias, R.D. Brown, J.A. Englert, <u>O. Hofmann</u>, J.A. Lederer, W. Hide, D.C. Christiani, M. Cernadas, and R.M. Baron, *Chronic endotoxin exposure produces airflow obstruction and lung dendritic cell expansion*, Am J Respir Cell Mol Biol, (2012).
- S-A. Sansone, P. Rocca-Serra, D. Field, E. Maguire, C. Taylor, <u>O. Hofmann</u>, H. Fang, S. Neumann, W. Tong, L. Amaral-Zettler, K. Begley, T. Booth, L. Bougueleret, G. Burns, B. Chapman, T. Clark, L-A. Coleman, J. Copeland, S. Das, A. de Daruvar, P. de Matos, I. Dix, S. Edmunds, C.T. Evelo, M.J. Forster, P. Gaudet, J. Gilbert, C. Goble, J.L. Griffin, D. Jacob, J. Kleinjans, L. Harland, K. Haug, H. Hermjakob, S.J. Ho Sui, A. Laederach, S. Liang, S. Marshall, A. McGrath, E. Merrill, D. Reilly, M. Roux, C.E. Shamu, C.A. Shang, C. Steinbeck, A. Trefethen, B. Williams-Jones, K. Wolstencroft, I. Xenarios, and W. Hide, *Toward interoperable bioscience data*, Nat Genet, (2012).
- P.R. Bushel, R. McGovern, L. Liu, <u>O. Hofmann</u>, A. Huda, J. Lu, W. Hide, and X. Lin, *Population differences in transcript-regulator expression quantitative trait Loci*, PLOS One, (2012).
- M. Mazumdar, W. Xia, <u>O. Hofmann</u>, M. Gregas, S. Ho Sui, W. Hide, T. Yang, H.L. Needleman, and D.C. Bellinger, *Prenatal Lead Levels, Plasma Amyloid Levels and Gene Expression in Young Adulthood*, Environ Health Perspect, (2012).
- S.J. Ho Sui, K. Begley, D. Reilly, B. Chapman, R. McGovern, P. Rocca-Sera, E. Maguire, G.M. Altschuler, T.A.A. Hansen, R. Sompallae, A. Krivtsov, R.A. Shivdasani, S.A. Armstrong, A.C. Culhane, M. Correll, S-A. Sansone, <u>O. Hofmann</u>, and W. Hide., *The Stem Cell Discovery Engine: an integrated repository and analysis system for cancer stem cell comparisons*, Nucleic Acids Res, (2011).

- A. Lal, M.P. Thomas, G. Altschuler, F. Navarro, E. O'Day, X.L. Li, C. Concepcion, Y-C. Han, J. Thiery, D.K. Rajani, A. Deutsch, <u>O. Hofmann</u>, A. Ventura, W. Hide, and J. Lieberman, Capture of MicroRNA-Bound mRNAs Identifies the Tumor Suppressor miR-34a as a Regulator of Growth Factor Signaling, PLOS Genet, (2011).
- H. Zhao, W. Lin, K. Kumthip, D. Cheng, D.N. Fusco, O. Hofmann, N. Jilg, A.W. Tai, K. Goto, L. Zhang, W. Hide, J.Y. Jang, L.F. Peng, and R.T. Chung, A functional genomic screen reveals novel host genes that mediate interferon-alpha's effects against hepatitis C virus, J Hepatol, (2011).
- S. Fu, L. Yang, P. Li, <u>O. Hofmann</u>, L. Dicker, W. Hide, X. Lin, S.M. Watkins, A.R. Ivanov, and G.S. Hotamisligil, *Aberrant Lipid Metabolism Disrupts Calcium Homeostasis Causing Liver Endoplasmic Reticulum Stress in Obesity*, Nature, (2011).
- C. Huttenhower and <u>O. Hofmann</u>, A quick guide to large-scale genomic data mining, PLOS Comput Biol, (2010).
- P. Rocca-Serra, M. Brandizi, E. Maguire, N. Sklyar, C. Taylor, K. Begley, D. Field, S. Harris, W. Hide, <u>O. Hofmann</u>, S. Neumann, P. Sterk, W. Tong, and S.A. Sansone, *ISA software suite: supporting standards-compliant experimental annotation and enabling curation at the community level*, Bioinformatics, (2010).
- <u>DIAGRAM consortium</u>, Twelve type 2 diabetes susceptibility loci identified through large-scale association analysis, Nat Genet, (2010).
- T. Ravasi, H. Suzuki, C.V. Cannistraci, S. Katayama, V.B. Bajic, K. Tan, A. Akalin, S. Schmeier, M. Kanamori-Katayama, N. Bertin, P. Carninci, C. Daub, A.R.R. Forrest, J. Gough, S. Grimmond, J-H. Han, T. Hashimoto, W. Hide, <u>O. Hofmann</u>, A. Kamburov, M. Kaur, H. Kawaji, A. Kubosaki, T. Lassmann, E. van Nimwegen, C.R. MacPherson, C. Ogawa, A. Radovanovic, A. Schwartz, R.D. Teasdale, J. Tegner, B. Lenhard, S.A. Teichmann, T. Arakawa, N. Ninomiya, K. Murakami, M. Tagami, S. Fukuda, K. Imamura, C. Kai, R. Ishihara, Y. Kitazume, J. Kawai, D.A. Hume, T. Ideker, and Y. Hayashizaki, *An atlas of combinatorial transcriptional regulation in mouse and man*, Cell, (2010).
- A. Lister, R.S. Datta, <u>O. Hofmann</u>, R. Krause, M. Kuhn, B. Roth, and R. Schneider, *Live coverage of scientific conferences using web technologies*, PLOS Comp Bio, (2010).
- A. Lal, F. Navarro, C.A. Maher, L.E. Maliszewski, N. Yan, E. O'Day, D. Chowdhury, D.M. Dykxhoorn, P. Tsai, <u>O. Hofmann</u>, K.G. Becker, M. Gorospe, W. Hide, and J. Lieberman, miR-24 Inhibits cell proliferation by targeting E2F2, MYC, and other cell-cycle genes via binding to "seedless" 3'UTR microRNA recognition elements, Mol Cell (2009).
- The Fantom4 consortium, A complex transcriptional network controls growth arrest and differentiation in a human myeloid leukemia cell line, Nat Genet, (2009).
- O. Hofmann, O. Caballero, B. Stevenson, Y. Chen, T. Cohen, R. Chua, C. Maher, S. Panji, U. Schaefer, U. Kruger, M. Lehvaslaiho, P. Carninci, Y. Hayashizaki, C. V. Jongeneel, A. J. G. Simpson, L. J. Old, and W. Hide, *Genome-wide analysis of cancer/testis gene-expression*, Proc Natl Acad Sci USA, (2008).
- G. Koscienlny, V. LeTexier, C. Gopalakrishnan, V. Kumanduri, J. Riethoven, F. Nardone, E. Whitfield, C. Fallsehr, <u>O. Hofmann</u>, M. Kull, H. E., S. Boue, E. Eyras, M. Plass, F. Lopez, W. Ritchie, V. Moucadel, T. Ara, H. Pospisil, A. Hermann, J. Reich, R. Guigo, P. Bork, M. von Knebel Doeberitz, J. Vilo, W. Hide, R. Apweiler, A. Thanaraj, and D. Gautheret, *ASTD: the alternative splicing and transcript diversity database*, Genomics, (2008).
- M. Kaur, S. Schmeier, C. R. MacPherson, <u>O. Hofmann</u>, W. A. Hide, S. Taylor, N. Willcox and V. B. Bajic, *Prioritizing genes of potential relevance to diseases affected by sex hormones: An example of Myasthenia Gravis*, BMC Genomics, (2008).

- HPRD Consortium, Human proteinpedia enables sharing of human protein data, Nat Biotechnol, (2008).
- Z. Lombard, N. Tiffin, <u>O. Hofmann</u>, V. B. Bajic, W. Hide, and M. Ramsay, *Computational selection and prioritization of candidate genes for fetal alcohol syndrome*, BMC Genomics, (2007).
- A. Kruger, O. Hofmann, P. Carninci, Y. Hayashizaki, and W. Hide, *Simplified ontologies allowing comparison of developmental mammalian gene expression*, Genome Biol, (2007).
- V. B. Bajic, S. L. Tan, A. Christoffels, C. Schönbach, L. Lipovich, L. Yang, <u>O. Hofmann</u>, A. Kruger, W. Hide, C. Kai, J. Kawai, D. A. Hume, P. Carninci, and Y. Hayashizaki, *Mice and men: their promoter properties*, PLOS Genet, (2006).
- A. Mehrle, H. Rosenfelder, I. Schupp, C. del Val, D. Arlt, F. Hahne, S. Bechtel, J. Simpson, O. Hofmann, W. Hide, K.-H. Glatting, W. Huber, R. Pepperkok, A. Poustka, and S. Wiemann, *The LIFEdb database in 2006*, Nucleic Acids Res, (2006).
- O. Hofmann and D. Schomburg, Concept-based annotation of enzyme classes, Bioinformatics, (2005).
- I. Schomburg, A. Chang, <u>O. Hofmann</u>, C. Ebeling, F. Ehrentreich, and D. Schomburg, *BRENDA:* a resource for enzyme data and metabolic information, Trends Biochem Sci, (2002).
- I. Schomburg, <u>O. Hofmann</u>, C. Baensch, A. Chang, and D. Schomburg, *Enzyme data and metabolic information: BRENDA*, a resource for research in biology, biochemistry and medicine, Gene Function & Disease, (2000).

# Book chapters

O. Hofmann, and W. Hide, Gene-expression ontologies and tag-based expression profiling, in: Cap-Analysis Gene Expression (CAGE): The Science of Decoding Genes Transcription (2009). Edited by Piero Carninci. Pan Stanford Publishing Pte Ltd.

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