Matthias Wielscher

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Employments

since 09/2022	**Senior Research Associate** Medical University of Vienna, Dept. of Laboratory Medicine
since 01/2020	**Senior Research Associate** Medical University of Vienna, Dept. of Dermatology
06/2018 -12/2019	**Bioinformatician / Senior Analyst** Genomics England (100k Genomes Project)
since 06/2018	**Consultant for Genetic Epidemiology** Imperial College, Dept. of Epidemiology and Biostatistics
07/2015- 05/2018	**Research Associate** Imperial College, Dept. of Epidemiology and Biostatistics
01/2011-04/2015	**PhD Student** AIT, Department of Health and Environment
2009-2011	**Research Assistant** AIT, Department of Health and Environment
2008-2009	**MSc Student** AIT, Department of Health and Environment

Education

2011-2015	**PhD thesis** University of Natural Resources and Life Science, *Title: Differential DNA methylation signatures in interstitial lung disease and lung cancer reveal novel biomarkers*
2012-2013	Training in Statistics at Technical University of Vienna
2008-2009	**MSc Thesis** University of Vienna, *Title: DNA methylation analysis for minimal-invasive Breast cancer diagnostic testing*
2002-2007	**Degree program Genetics/Microbiology** University of Vienna, Austria

Leaves

02/2014 – 10/2014	Paternal Leave 1
10/2009 – 06/2010	Paternal Leave 2

Main research area

My research is focused on genetics and diagnosis. At the Department of Dermatology, I set up a whole genome analysis and annotation pipeline for rare skin diseases, but I am also investigating the genomes of skin resident bacteria and phages. For this we combined several large scale publicly available data sets and novel data sets.

At the Department of Laboratory Medicine, I am managing the NGS pipelines for clinical Diagnosis, while testing new reporting standards and variant calling and annotation approaches to improve patient care.

I have also a strong interest in genome wide associations studies and Mendelian Randomization approaches, which I do in collaboration with Imperial College and the Department of Epidemiology of the MedUni Vienna, mainly using UK-Biobank data.

Major scientific contributions/additional research achievements

Research Data generation: While working in Genomics England Research environment I aggregated and quality controlled 640 million variants across 60.000 patients from individual whole genome datasets to one joint dataset. The dataset occupied 120 TB of storage and to our best knowledge was the largest call set outside of the US at the time.

Software: Most of my applications and code is publicly available on GitHub. (https://github.com/Mwielscher). This includes: scripts and instructions for GWAS, Mendelian Randomization, Triangulation analysis, GATK based germline variant calling, GATK somatic variant calling, setting up a secure cloud environment, RNA sequencing, single cell RNA sequencing.

Teaching: For my students I set up a JupyterHub hosted on a VM of the Medical University of Vienna. This allows enrolled students to start coding R from any Internet Browser window and use analysis-code and test datasets I provide for them. I teach R-Basics, and an advanced class for RNA sequencing analysis. Recently, I was invited to teach a class for “Spezielle Gebiete der angewandten AI” at Technikum Vienna.

Project Coordination: I have coordinated a large-scale epigenome wide association study with contributors from more than 100 different Institutions worldwide. I am also member of several genetic consortia such as GIANT, GLGC and Spirometer.

Research Grant preparation: I have been involved in successful Grant applications: PREcisE - A precision nutri-epigenetic approach to tackle the mother-to-child transmission of impaired glucose and GENEROSITY - A sustainable and multi-actor approach to prevent another generation of childhood obesity in Europe. More recently, I have submitted proposals to WWTF and Marie Curie program without funding success.

Scholarly activities: I am a topic Editor for Frontiers in Genetics and I am reviewing regularly for International Journal of Epidemiology and American Journal of Epidemiology as well as occasionally several other journals.

Patent: I hold a patent for serum DNA methylation markers. ***Diagnosis of Lung Cancer***. European Patent Application EP14194732.5, filed 25-11-2014 and U.S. application PCT /EP2015/077478

Publications

total number of publications: **52**
total number of citations: **2633**
h index: **26**
https://scholar.google.com/citations?user=tWrztwwAAAAJ&hl=de

Wielscher M, Pfisterer K, Samardzic D, Balsini P, Bangert C, Jaeger K, Buchberger M, Selitsch B, Pjevac P, Willinger B, Weninger W, **The Phageome in Normal and Inflamed Human Skin.** Available at SSRN: http://dx.doi.org/10.2139/ssrn.4294793 accepted at Science Advances|AAAS

Sunder-Plassmann R, Geusau A, Endler G, Weninger W and **Wielscher M**. **Identification of Genetic Risk Factors for Keratinocyte Cancer in Immunosuppressed Solid Organ Transplant Recipients: A Case-Control Study.**
Cancers (Basel). 2023 Jun 26;15(13):3354. doi: 10.3390/cancers15133354.

Wielscher M, Mandaviya P, Kuehnel B, Joehanes R, Mustafa R [..], Jarvelin MR. ***DNA methylation signature of chronic low-grade inflammation and its role in cardio-respiratory diseases.*** Nat Commun. 2022 May 3;13(1):2408. doi: 10.1038/s41467-022-29792-6.

Wielscher M, Amaral AFS, van der Plaat D, Sebert S, Wain L, Sebert S, Mosen-Ansorena D, Auvinen J, Herzig A, Dehghan A, Jarvis D, Jarvelin MR. ***Genetic correlation and causal relationships between cardio-metabolic traits and Lung function Impairment.*** Genome Med. 2021 Jun 21;13(1):104. doi: 10.1186/s13073-021-00914-x.

Wielscher M, Vierlinger K, Kegler U, Ziesche R, Gsur A, Weinhausel A. ***Diagnostic Performance of Plasma DNA Methylation Profiles in Lung Cancer, Pulmonary Fibrosis and COPD.*** EBioMedicine 2015, 2(8):929-936.

Wielscher M, Liou W, Pulverer W, Singer CF, Rappaport-Fuerhauser C, Kandioler D, Egger G, Weinhausel A. ***Cytosine 5-Hydroxymethylation of the LZTS1 Gene Is Reduced in Breast Cancer.*** Translational oncology 2013, 6(6):715-721.

Wielscher M, Pulverer W, Peham J, Hofner M, Rappaport CF, Singer C, Jungbauer C, Nohammer C, Weinhausel A. ***Methyl-binding domain protein-based DNA isolation from human blood serum combines DNA analyses and serum-autoantibody testing***. BMC clinical pathology 2011, 11:11.

100,000 Genomes Project Pilot Investigators, amongst authors **Wielscher M,** Caulfield M. ***100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care - Preliminary Report.*** N Engl J Med. 2021 Nov 11;385(20):1868-1880. doi: 10.1056/NEJMoa2035790.

Hawe JS, Wilson R, Schmid KT, Zhou L, Lakshmanan LN, Lehne BC, Kühnel B, Scott WR, **Wielscher M,** et al. ***Genetic variation influencing DNA methylation provides insights into molecular mechanisms regulating genomic function.*** Nat Genet. 2022 Jan;54(1):18-29. doi: 10.1038/s41588-021-00969-x. Epub 2022 Jan 3.

Imboden M*, **Wielscher M***, Rezwan FI*, Amaral AFS*, Schaffner E, Jeong A, et al. ***Epigenome-wide association study of lung function level and its change.*** Eur Respir J. 2019 Jul 4;54(1):1900457. doi: 10.1183/13993003.00457-2019. Print 2019 Jul. * contributed equally