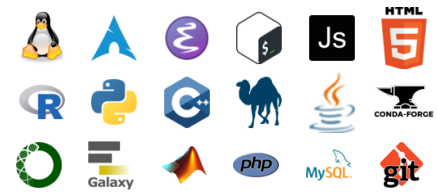


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Post-Doc Bioinformatician / Software Developer with experience in single-cell RNA cluster analysis, DNA variant and haplotype analysis, and open-source automation pipelines developed in rapport with scientists and clinicians alike

Education, Qualifications, and Work Experience

London, UK	Whitgift School	09.2000 - 10.2007	Private school, top 10 in UK	4 As
	University College London (UCL)	09.2007 - 08.2011	MSci Astrophysics	Grade - 2:1 Honours
		09.2011 - 08.2012	MSc Computer Science	Distinction
	Royal Free Hospital (UCL)	09.2012 - 10.2016	PhD Biomedical Science	(No corrections)
		10.2016 - 09.2017	Post-Doc Biomedical Scientist	DNA Variant Analysis Linkage and Haplotypes Teaching Assistant
Freiburg, DE	Galaxy Team (Backofen Group)	09.2017 - (current)	Post-Doc Bioinformatician	Galaxy Tool Development Galaxy Training Workshops scRNA Analysis Teaching / Mentoring
	Max-Planck Institute (Grün Lab)	10.2018 - (current)	Guest Scientist	scRNA Analysis

Notable Works

- High-Throughput Linkage Analysis Pipeline** Performs comprehensive quality linkage analysis through a series of curated collection of quality checking utilities modified accordingly to handle large-data. (Github)
- High-Throughput Sequence Analysis Pipeline** VCF variant annotation and filtering utility that makes additional use of family data and penetrance models, and multiple annotation sources to produce an interactive web-report. (Bitbucket)
- Haplotype Visualization and Pedigree Tool** Web-based pedigree creation application, with haplotype visualization and partial reconstruction for genotype analysis and comparison. (Github)
- Single Cell Training Materials on Galaxy** Collection of trainings and wrappers for scRNA-seq analysis, incorporated into the Galaxy Training Network for both 10x and custom protocols, from pre-processing to downstream analysis. (GTN)

Publications

2014	- S. Drury, C. Boustred, M. Tekman, H. Stanescu, R. Kleta, N. Lench, L. S. Chitty, and R. H. Scott. A novel homozygous ERCC5 truncating mutation in a family with prenatal arthrogryposis—further evidence of genotype-phenotype correlation. <i>Am. J. Med. Genet. A</i> , 164A(7):1777–1783, Jul 2014
2015	- N. E. Mencacci, I. Rubio-Agusti, A. Zdebik, F. Asmus, M. H. Ludtmann, M. Ryten, V. Plagnol, A. K. Hauser, S. Bandres-Ciga, C. Bettencourt, P. Forabosco, D. Hughes, M. M. Soutar, K. Peall, H. R. Morris, D. Trabzuni, M. Tekman, H. C. Stanescu, R. Kleta, M. Carecchio, G. Zorzi, N. Nardocci, B. Garavaglia, E. Lohmann, A. Weissbach, C. Klein, J. Hardy, A. M. Pittman, T. Foltynie, A. Y. Abramov, T. Gasser, K. P. Bhatia, and N. W. Wood. A missense mutation in KCTD17 causes autosomal dominant myoclonus-dystonia. <i>Am. J. Hum. Genet.</i> , 96(6):938–947, Jun 2015
	- P. Le Quesne Stabej, H. J. Williams, C. James, M. Tekman, H. C. Stanescu, R. Kleta, L. Ocaka, F. Lescai, H. L. Storr, M. Bitner-Glindzicz, C. Bacchelli, G. S. Conway, G. E. Moore, B. G. Gaspar, M. Hubank, R. H. Scott, E. Chanudet, and E. Stupka. STAG3 truncating variant as the cause of primary ovarian insufficiency. <i>Eur. J. Hum. Genet.</i> , 24(1):135–138, Jun 2015
2016	- D. M. Rowczenio, D. S. Iancu, H. Trojer, J. A. Gilbertson, J. D. Gillmore, A. D. Wechalekar, M. Tekman, H. C. Stanescu, R. Kleta, T. Lane, P. N. Hawkins, and H. J. Lachmann. Autosomal dominant familial Mediterranean fever in Northern European Caucasians associated with deletion of p.M694 residue—a case series and genetic exploration. <i>Rheumatology (Oxford)</i> , May 2016
	- O. Abdelhadi, D. Iancu, M. Tekman, H. Stanescu, D. Bockenhauer, and R. Kleta. Founder mutation in KCNJ10 in Pakistani patients with EAST syndrome. <i>Mol Genet Genomic Med</i> , 4(5):521–526, Jun 2016
2017	- P. Stabej, C. James, L. Ocaka, M. Tekman, S. Grunewald, E. Clement, H. Stanescu, R. Kleta, D. Morrogh, A. Calder, et al. An example of the utility of genomic analysis for fast and accurate clinical diagnosis of complex rare phenotypes. <i>Orphanet Journal of Rare Diseases</i> , 12(1):24, 2017
	- O. Cabezas, S. Flanagan, H. Stanescu, E. Garcia-Martinez, R. Caswell, H. Lango-Allen, M. Anton-Gamero, J. Argente, A. Bussell, A. Brandli, et al. Polycystic kidney disease with hyperinsulinemic hypoglycemia caused by a promoter mutation in pmm2. <i>Journal of the American Society of Nephrology</i> , 2017
	- M. Tekman, A. Medlar, M. Mozere, R. Kleta, and H. Stanescu. Haploforge: a comprehensive pedigree drawing and haplotype visualization web application. <i>Bioinformatics</i> , 33(24):3871–3877, 2017
2018	- M. Tekman, M. Mozere, J. Kari, D. Bockenhauer, R. Kleta, and H. Stanescu. Ovas: an open-source variant analysis suite with inheritance modelling. <i>BMC bioinformatics</i> , 19(1):46, 2018
	- M. Reichold, E. Klootwijk, J. Reinders, E. Otto, M. Milani, C. Broeker, C. Laing, J. Wiesner, S. Devi, W. Zhou, et al. Glycine amidinotransferase (gatl), renal fanconi syndrome, and kidney failure. <i>Journal of the American Society of Nephrology</i> , 29(7):1849–1858, 2018
2019	- Danit Oz-Levi, Tsviya Olender, Ifat Bar-Joseph, Yiwen Zhu, Dina Marek-Yagel, Iros Barozzi, Marco Osterwalder, Anna Alkelai, Elizabeth K Ruzzo, Yujun Han, et al. Noncoding deletions reveal a gene that is critical for intestinal function. <i>Nature</i> , 571(7763):107–111, 2019